Publication No.	Study title	Pubmed ID	Year of publication	Institution	IRD-affected Subjects numbers	Age at reporting (years)	Gender	Consanguinity	Clinical diagnosis	Panel/Platform	Report gene	Mutation Type	Reported Novelty
Non-Syndromic IRDs													
RP													
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	1	5 Years	F	Yes	Retinitis pigmentosa	Clinical Exome	RPE65	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2	28 Years	M	No	Retinitis pigmentosa	Clinical Exome	RPGR	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	3	22 Years	F	Yes	Retinitis pigmentosa	Clinical Exome	CNGB1	Duplication FS	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	4	44 Years	F	No	Retinitis pigmentosa	Clinical Exome	PRPH2	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	5	19 Years	M	No	Retinitis pigmentosa	Clinical Exome	CERKL	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	6	24 Years	F	Yes	Rod cone dystrophy	Clinical Exome	IMPG2	5' splice site	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	7	31 years	M	Yes	Retinitis pigmentosa	Clinical Exome	PDE6A	Insertion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	8	12 Years	F	Yes	Retinitis pigmentosa .	Clinical Exome	NR2E3	Indel	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	9	27 Years	M	No	Retinitis pigmentosa	Clinical Exome	CERKL	Deletion FS	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	10	29 Years	M	Yes	Retinitis pigmentosa	Clinical Exome	TULP1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	11	19 Years	F	No	Retinitis pigmentosa	Clinical Exome	CERKL	Deletion FS	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	12	43 Years	M	No	Retinitis pigmentosa	Clinical Exome	PROM1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	13	22 Years	F	Yes	Retinitis pigmentosa	Clinical Exome	RP1	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	14	33 Years	M	No	Retinitis pigmentosa	Clinical Exome	AHI1	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	14	33 Years	M	No	Retinitis pigmentosa	Clinical Exome	AHI1	Duplication	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	15	23 Years	M	No	Retinitis pigmentosa	Clinical Exome	EYS	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	16	9 Years	F	Yes	Rod cone dystrophy	Clinical Exome	RPE65	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	17	31 Years	M	Yes	Rod cone dystrophy	Clinical Exome	PRPF31	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	18	6 Years	M	No	Retinitis Pigmentosa	Clinical Exome	SPATA7	Insertion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	19	76 Years	M	No	Retinitis pigmentosa	Clinical Exome	USH2A	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	19	76 Years	M	No	Retinitis pigmentosa	Clinical Exome	USH2A	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	20	45 Years	M	No	Retinitis pigmentosa	Clinical Exome	USH2A	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	20	45 Years	M	No	Retinitis pigmentosa	Clinical Exome	USH2A	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	21	3 Years	M	No	Rod dystrophy or Retinitis pigmentosa	Clinical Exome	PDE6A	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	22	20 Years	M	No	Retinitis pigmentosa	Clinical Exome	FSCN2	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	23	27 Years	M	Yes	Retinitis pigmentosa	Clinical Exome	RIMS1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	23	27 Years	M	Yes	Retinitis pigmentosa	Clinical Exome	RBP3	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	23	27 Years	M	Yes	Retinitis pigmentosa	Clinical Exome	CEP290	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	24	7 Years	M	No	Rod cone dystrophy	Clinical Exome	RPE65	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	24	7 Years	M	No	Rod cone dystrophy	Clinical Exome	RPE65	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	24	7 Years	M	No	Rod cone dystrophy	Clinical Exome	RP9	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	25	22 Years	M	No	Rod cone dystrophy	Clinical Exome	CRB1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	25	22 Years	M	No	Rod cone dystrophy	Clinical Exome	CRB1	3' splice site	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	26	4 Years	F	No	Rod cone dystrophy	Clinical Exome	CRX	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	26	4 Years	F	No	Rod cone dystrophy	Clinical Exome	TRPM1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	27	33 Years	F	No	Retinitis pigmentosa	Clinical Exome	USH2A	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	27	33 Years	F	No	Retinitis pigmentosa	Clinical Exome	USH2A	Missense	NA

IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	27	33 Years	F	No	Retinitis pigmentosa	Clinical Exome	PROM1	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	27	33 Years	F	No	Retinitis pigmentosa	Clinical Exome	PROM1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	27	33 Years	F	No	Retinitis pigmentosa	Clinical Exome	RPE65	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	28	8 Years	M	Yes	Rod cone dystrophy	Clinical Exome	TULP1	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	28	8 Years	M	Yes	Rod cone dystrophy	Clinical Exome	SPATA7	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	29	58 years	M	No	Retinitis pigmentosa	Clinical Exome	KCNJ13	missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	30	15 Years	F	Yes	Retinitis pigmentosa	Clinical Exome	RDH12	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	31	33 years	M	No	Retinitis pigmentosa	Clinical Exome	PRPF31	Deletion	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	32	19 Years	M	No	Retinitis Pigmentosa	Clinical Exome	CNGB1	Deletion	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	33	13 Years	M	No	Retinitis pigmentosa	Clinical Exome	EYS	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	33	13 Years	M	No	Retinitis pigmentosa	Clinical Exome	ZNF408	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	34	6 Years	F	No	Rod retinal dystrophy	Clinical Exome	PRPF8	3' splice site	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	34	6 Years	F	No	Rod retinal dystrophy	Clinical Exome	RP1L1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	34	6 Years	F	No	Rod retinal dystrophy	Clinical Exome	GRM6	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	35	36 Years	F	No	Retinitis pigmentosa	Clinical Exome	EYS	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	35	36 Years	F	No	Retinitis pigmentosa	Clinical Exome	EYS	Deletion FS	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	35	36 Years	F	No	Retinitis pigmentosa	Clinical Exome	USH2A	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	35	36 Years	F	No	Retinitis pigmentosa	Clinical Exome	USH2A	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	36	33 Years	M	Yes	Retinitis pigmentosa	Clinical Exome	CDHR1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	36	4 Years	F	Yes	Rod cone dysfunction? or Juvenile Retinitis pigmentosa	Clinical Exome	RAX2	Duplication	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	36	4 Years	F	Yes	Rod cone dysfunction? or Juvenile Retinitis pigmentosa	Clinical Exome	CEP290	3' splice site	Novel
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	37	5	F	NA	RP	TP	*RPE65*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	38	4	F	NA	Rod cone dysfunction ?RP	TP	*RAX2*	Duplication	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	39	38	M	NA	RP	TP	*USH2A*	splice site	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	39	38	M	NA	RP	TP	*USH2A*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	40	22	M	NA	RP/Ciliopathy disorder	TP	*EYS*	deletion FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	41	22	F	NA	RP	TP	*CNGB1*	duplication FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	42	28	M	NA	RP	TP	*RPGR*	deletion FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	43	44	F	NA	RP	TP	*PRPH2*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	44	4	M	NA	Rod cond dystrophy	TP	*TRPM1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	45	33	M	NA	RP	TP	*CDHR1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	46	3	F	NA	Rod cone dysfunction	TP	*TULP1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	47	32	M	NA	retinitis pigmentosa	TP	*PDE6A*	insertion FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	48	25	F	NA	rod cone dystrophy	TP	*IMPG2*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	49	19	M	NA	retinitis pigmentosa	TP	*CERKL*	deletion FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	50	7	M	NA	rod cone dystrophy	TP	*RPE65*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	50	7	M	NA	rod cone dystrophy	TP	*RPE65*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	50	7	M	NA	rod cone dystrophy	TP	*RP9*	deletion FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	51	33	F	NA	Retinitis pigmentosa	TP	*RPE65*	missense	NA

IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	51	33	F	NA	Retinitis pigmentosa	TP	*PROM1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	51	33	F	NA	Retinitis pigmentosa	TP	*PROM1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	51	33	F	NA	Retinitis pigmentosa	TP	*USH2A*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	51	33	F	NA	Retinitis pigmentosa	TP	*USH2A*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	52	5	M	NA	Rod cone dystrophy	WES	*RDH12*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	53	28	M	NA	RP	WES	*SNRNP200*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	54	11	M	NA	RP	WES	*CEP290*	splice site	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	55	2	M	NA	RP	WES	*CEP290*	splice site	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	56	0.9	F	NA	atypical RP	WES	*RP1*	missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	57	14 years	M	NA	B/L retinitis pigmentosa	exome sequencing/panel-based sequencing/chromosomal microarray	CERKL	NA	NA
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	58	27	F	C	AR-RP	Homozygosity mapping	CDHR1	deletion FS	Novel
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	58	27	F	C	AR-RP	Homozygosity mapping	MERTK	missense	NA
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	58	27	F	C	AR-RP	Homozygosity mapping	ARL6	missense	NA
IRD 8	Choroidal structural analysis and vascularity index in retinal dystrophies	30178525	2019	Aravind Eye Hospital, Madurai, India.	59-75	NA	NA	NA	RP	NA	NA	NA	NA
IRD 9	Focal Choroidal Excavation in Retinal Dystrophies	27533784	2018	L. V. Prasad Eye Institute	76-168	NA	NA	NA	RP	NA	NA	NA	NA
IRD10	Choroidal hyper-reflective foci and vascularity in retinal dystrophy	31856490	2020	Moorfields Eye Hospital NHS Foundation Trust, London, United Kingdom, Srimati Kanuri Santhamma Centre for Vitreo-Retinal Diseases, Hyderabad Eye Research LVPEI	169	NA	FEMALE	NA	RP	NA	NA	NA	NA
IRD10	Choroidal hyper-reflective foci and vascularity in retinal dystrophy	31856490	2020	Moorfields Eye Hospital NHS Foundation Trust, London, United Kingdom, Srimati Kanuri Santhamma Centre for Vitreo-Retinal Diseases, Hyderabad Eye Research LVPEI	170	NA	MALE	NA	RP	NA	NA	NA	NA
IRD11	Evaluation of photoreceptor function in inherited retinal diseases using rod- and cone-enhanced flicker stimuli	33834501	2021	L V Prasad Eye Institute, Hyderabad, India.	171-177	32.4 - 13.5 years	5M, 2F	NA	RP	NA	NA	NA	NA
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	178	25	F	C	arRP	Homozygosity mapping	MERTK	nonsense	Novel
LCA 18	RPE65 gene: multiplex PCR and mutation screening in patients from India with retinal degenerative diseases.	12357075	2002	Department of Genetics and Molecular Biology, Medical and Vision Research Foundations, Sankara Nethralaya, Chennai 600 006, India.	179	NA	NA	C	arRP	multiplex PCR follwed by sequencing for RPE65	RPE65	missense	Reported
LCA 18	RPE65 gene: multiplex PCR and mutation screening in patients from India with retinal degenerative diseases.	12357075	2002	Department of Genetics and Molecular Biology, Medical and Vision Research Foundations, Sankara Nethralaya, Chennai 600 006, India.	180	NA	NA	NA	arRP	multiplex PCR follwed by sequencing for RPE65	RPE65	isocoding	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	181	NA	NA	C	RP	Targeted Exome Sequencing	ABCA4	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	182	NA	NA	NC	RP	Targeted Exome Sequencing	ABCA4	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	183	NA	NA	NC	RP	Targeted Exome Sequencing	C2orf71	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	184	NA	NA	C	RP	Targeted Exome Sequencing	C8orf37	DELETION	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	185	NA	NA	C	RP	Targeted Exome Sequencing	CDHR1	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	186	NA	NA	NC	RP	Targeted Exome Sequencing	CEP290	STOP	Reported

RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	187	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	188	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	189	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	190	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	191	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	192	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	193	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	194	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	195	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	196	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	197	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	198	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	199	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	200	NA	NA	NC	RP	Targeted Exome Sequencing	CERKL	SPLICING VARIANT	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	201	NA	NA	C	RP	Targeted Exome Sequencing	CNGB1	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	202	NA	NA	C	RP	Targeted Exome Sequencing	CNGB1	STOP	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	203	NA	NA	C	RP	Targeted Exome Sequencing	CRB1	SPLICING VARIANT	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	204	NA	NA	NC	RP	Targeted Exome Sequencing	CRB1	DELETION	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	205	NA	NA	NC	RP	Targeted Exome Sequencing	CRB1	DELETION	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	206	NA	NA	NC	RP	Targeted Exome Sequencing	CRB1	STOP	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	207	NA	NA	NC	RP	Targeted Exome Sequencing	CRX	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	208	NA	NA	NC	RP	Targeted Exome Sequencing	EYS	SPLICING VARIANT	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	209	NA	NA	NC	RP	Targeted Exome Sequencing	EYS	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	210	NA	NA	NC	RP	Targeted Exome Sequencing	EYS	SPLICING VARIANT	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	211	NA	NA	C	RP	Targeted Exome Sequencing	EYS	DELETION	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	212	NA	NA	NC	RP	Targeted Exome Sequencing	EYS	DELETION	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	213	NA	NA	C	RP	Targeted Exome Sequencing	FAM161A	frameshift_del	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	214	NA	NA	NC	RP	Targeted Exome Sequencing	FAM161A	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	215	NA	NA	NA	RP	Targeted Exome Sequencing	FAM161A	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	216	NA	NA	C	RP	Targeted Exome Sequencing	IMPG2	frameshift_del	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	217	NA	NA	C	RP	Targeted Exome Sequencing	MERTK	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	218	NA	NA	C	RP	Targeted Exome Sequencing	MERTK	SPLICING VARIANT	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	219	NA	NA	C	RP	Targeted Exome Sequencing	PDE6A	SPLICING VARIANT	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	220	NA	NA	NC	RP	Targeted Exome Sequencing	PDE6A	frameshift_del	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	221	NA	NA	NA	RP	Targeted Exome Sequencing	PDE6B	SPLICING VARIANT	Reported

RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	222	NA	NA	C	RP	Targeted Exome Sequencing	PDE6B	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	223	NA	NA	NC	RP	Targeted Exome Sequencing	PDE6B	SPLICING VARIANT	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	224	NA	NA	C	RP	Targeted Exome Sequencing	PDE6B	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	225	NA	NA	NC	RP	Targeted Exome Sequencing	PROM1	STOP	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	226	NA	NA	C	RP	Targeted Exome Sequencing	PROM1	DELETION	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	227	NA	NA	NC	RP	Targeted Exome Sequencing	PROM1	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	228	NA	NA	NC	RP	Targeted Exome Sequencing	PROM1	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	229	NA	NA	NC	RP	Targeted Exome Sequencing	PRPF31	SPLICING VARIANT	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	230	NA	NA	NC	RP	Targeted Exome Sequencing	PRPF31	SPLICING VARIANT	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	231	NA	NA	NC	RP	Targeted Exome Sequencing	PRPF31	DELETION	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	232	NA	NA	NC	RP	Targeted Exome Sequencing	RDH12	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	233	NA	NA	NA	RP	Targeted Exome Sequencing	RDH12	MISSENSE	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	234	NA	NA	C	RP	Targeted Exome Sequencing	RDH12	MISSENSE	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	235	NA	NA	NC	RP	Targeted Exome Sequencing	RDH12	MISSENSE	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	236	NA	NA	C	RP	Targeted Exome Sequencing	RDH12	STOP	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	237	NA	NA	C	RP	Targeted Exome Sequencing	RP1	frameshift_del	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	238	NA	NA	C	RP	Targeted Exome Sequencing	RP1	frameshift_del	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	239	NA	NA	NA	RP	Targeted Exome Sequencing	RPE65	MISSENSE	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	240	NA	NA	NA	RP	Targeted Exome Sequencing	RPE65	STOP	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	241	NA	NA	NC	RP	Targeted Exome Sequencing	RPE65	MISSENSE	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	242	NA	NA	NC	RP	Targeted Exome Sequencing	RPGR1P1	DUPLICATION	Novel
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	243	NA	NA	C	RP	Targeted Exome Sequencing	TTC8	SPLICING VARIANT	Reported
RP 1	Next-generation sequencing-based genetic testing and phenotype correlation in retinitis pigmentosa patients from India	37322672	2023	Vision Research Foundation	244	NA	NA	C	RP	Targeted Exome Sequencing	TULP1	MISSENSE	Reported
RP 2	Genetic analysis of Indian families with autosomal recessive retinitis pigmentosa by homozygosity screening	19339744	2009	L. V. Prasad Eye Institute,	245	21	F	C	RP	Homozygosity mapping, Sanger sequencing	ABCA4	Nonsense	Reported
RP 2	Genetic analysis of Indian families with autosomal recessive retinitis pigmentosa by homozygosity screening	19339744	2009	L. V. Prasad Eye Institute,	246	16	F	C	RP	Homozygosity mapping, Sanger sequencing	RLBP1	MISSENSE	Novel
RP 2	Genetic analysis of Indian families with autosomal recessive retinitis pigmentosa by homozygosity screening	19339744	2009	L. V. Prasad Eye Institute,	247	19	M	NC	RP	Homozygosity mapping, Sanger sequencing	RP1	frameshift_del	Novel
RP 2	Genetic analysis of Indian families with autosomal recessive retinitis pigmentosa by homozygosity screening	19339744	2009	L. V. Prasad Eye Institute,	248	7	M	C	RP	Homozygosity mapping, Sanger sequencing	RPE65	frameshift_del	Reported
RP 2	Genetic analysis of Indian families with autosomal recessive retinitis pigmentosa by homozygosity screening	19339744	2009	L. V. Prasad Eye Institute,	249	22	M	C	RP	Homozygosity mapping, Sanger sequencing	TULP1	MISSENSE	Novel
RP 3	Homozygous null mutations in the ABCA4 gene in two families with autosomal recessive retinal dystrophy	16546111	2006	L. V. Prasad Eye Institute,	250	28	M	C	RP	Homozygosity mapping, Sanger sequencing	ABCA4	Nonsense	Reported
RP 3	Homozygous null mutations in the ABCA4 gene in two families with autosomal recessive retinal dystrophy." American journal of ophthalmology 141.5 (2006): 906-913	16546111	2006	L. V. Prasad Eye Institute,	251	32	M	C	RP	Homozygosity mapping, Sanger sequencing	ABCA4	Deletion	Novel
RP 4	A splicing mutation in aryl hydrocarbon receptor associated with retinitis pigmentosa	29726989	2018	Aravind Medical Research Foundation,	252	10	M	NC	RP	WES and Sanger	AHR	SPLICING VARIANT	Novel
RP 5	A nonsense mutation in C8orf37 linked with retinitis pigmentosa, early macular degeneration, cataract, and myopia in an arRP family from North India	37170250	2023	Department of Human Genetics, Guru Nanak Dev University	253	20	M	C	RP	WES and Sanger	C8orf37	STOP	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	254	59	F	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	255	27	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	256	32	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported

RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	257	20	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	258	26	M	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	259	55	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	260	63	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	261	33	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	262	33	M	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	263	30	M	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	264	51	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	265	35	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	266	23	NA	NC	RP	Targeted Exome Sequencing	CERKL	DELETION	Reported
RP 6	CERKL mutation causing retinitis pigmentosa(RP) in Indian population - a genotype and phenotype correlation study	32865075	2020	Vision Research Foundation	267	28	NA	NC	RP	Targeted Exome Sequencing	CERKL	SPLICING VARIANT	Reported
RP 7	Whole exome sequencing identified novel CRB1 mutations in Chinese and Indian populations with autosomal recessive retinitis pigmentosa.	27670293	2016	Aravind Medical Research Foundation,	268	NA	M	NC	RP	WES and Sanger	CRB1	MISSENSE	Reported
RP 7	Whole exome sequencing identified novel CRB1 mutations in Chinese and Indian populations with autosomal recessive retinitis pigmentosa.	27670293	2016	Aravind Medical Research Foundation,	269	NA	NA	NA	RP	WES and Sanger	CRB1	MISSENSE	Novel
RP 7	Whole exome sequencing identified novel CRB1 mutations in Chinese and Indian populations with autosomal recessive retinitis pigmentosa.	27670293	2016	Aravind Medical Research Foundation,	270	NA	NA	NA	RP	WES and Sanger	CRB1	MISSENSE	Novel
RP 7	Whole exome sequencing identified novel CRB1 mutations in Chinese and Indian populations with autosomal recessive retinitis pigmentosa.	27670293	2016	Aravind Medical Research Foundation,	271	NA	NA	NA	RP	WES and Sanger	CRB1	MISSENSE	Novel
RP 8	Whole-exome Sequencing Analysis Identifies Mutations in the EYS Gene in Retinitis Pigmentosa in the Indian Population.	26787102	2016	Aravind Medical Research Foundation,	272	8	M	NA	RP	WES and Sanger	EYS	frameshift_del	Novel
RP 8	Whole-exome Sequencing Analysis Identifies Mutations in the EYS Gene in Retinitis Pigmentosa in the Indian Population.	26787102	2016	Aravind Medical Research Foundation,	273	38	M	NA	RP	WES and Sanger	EYS	STOP	Novel
RP 8	Whole-exome Sequencing Analysis Identifies Mutations in the EYS Gene in Retinitis Pigmentosa in the Indian Population.	26787102	2016	Aravind Medical Research Foundation,	274	44	F	NA	RP	WES and Sanger	EYS	missense	Novel
RP 8	Whole-exome Sequencing Analysis Identifies Mutations in the EYS Gene in Retinitis Pigmentosa in the Indian Population.	26787102	2016	Aravind Medical Research Foundation,	275	25	M	NA	RP	WES and Sanger	EYS	SPLICING VARIANT	Novel
RP 8	Whole-exome Sequencing Analysis Identifies Mutations in the EYS Gene in Retinitis Pigmentosa in the Indian Population.	26787102	2016	Aravind Medical Research Foundation,	276	20	F	NA	RP	WES and Sanger	EYS	STOP	Novel
RP 9	Whole-exome sequencing reveals a novel frameshift mutation in the FAM161A gene causing autosomal recessive retinitis pigmentosa in the Indian population	26246154	2015	Aravind Medical Research Foundation,	277	NA	F	C	RP	WES and Sanger	FAM161A	frameshift_del	Reported
RP 9	Whole-exome sequencing reveals a novel frameshift mutation in the FAM161A gene causing autosomal recessive retinitis pigmentosa in the Indian population	26246154	2015	Aravind Medical Research Foundation,	278	NA	M	C	RP	WES and Sanger	FAM161A	frameshift_del	Reported
RP 9	Whole-exome sequencing reveals a novel frameshift mutation in the FAM161A gene causing autosomal recessive retinitis pigmentosa in the Indian population	26246154	2015	Aravind Medical Research Foundation,	279	NA	NA	NC	RP	WES and Sanger	FAM161A	frameshift_del	Reported
RP 10	A novel mutation in MERTK for rod-cone dystrophy in a North Indian family	30851773	2019	Department of Human Genetics, Guru Nanak Dev University	280	18	F	C	RP	WES and Sanger	METRK	STOP	Novel
RP 11	Mutations in TULP1, NR2E3, and MFRP genes in Indian families with autosomal recessive retinitis pigmentosa	22605927	2012	Kallam Anji Reddy Molecular Genetics Laboratory,	281	29	M	NC	RP	Homozygosity mapping, Sanger sequencing	MFRP	STOP	Reported
RP 11	Mutations in TULP1, NR2E3, and MFRP genes in Indian families with autosomal recessive retinitis pigmentosa	22605927	2012	Kallam Anji Reddy Molecular Genetics Laboratory,	282	10	M	C	RP	Homozygosity mapping, Sanger sequencing	NR2E3	frameshift_del	Reported
RP 11	Mutations in TULP1, NR2E3, and MFRP genes in Indian families with autosomal recessive retinitis pigmentosa	22605927	2012	Kallam Anji Reddy Molecular Genetics Laboratory,	283	14	M	C	RP	Homozygosity mapping, Sanger sequencing	TULP1	MISSENSE	Reported
RP 11	Mutations in TULP1, NR2E3, and MFRP genes in Indian families with autosomal recessive retinitis pigmentosa	22605927	2012	Kallam Anji Reddy Molecular Genetics Laboratory,	284	14	F	C	RP	Homozygosity mapping, Sanger sequencing	TULP1	MISSENSE	Novel
RP 12	A novel mutation in the PRPF31 in a North Indian adRP family with incomplete penetrance	30099644	2108	Department of Human Genetics, Guru Nanak Dev University (GNDU),	285	35	F	NC	RP	Microsatellite-marker analysis AND SANGER	PRPF31	MISSENSE	Novel
RP 13	A novel 7 bp deletion in PRPF31 associated with autosomal dominant retinitis pigmentosa with incomplete penetrance in an Indian family	23041261	2012	Department of Human Genetics, Guru Nanak Dev University	286	17	M	NC	RP	Microsatellite-marker analysis AND SANGER	PRPF31	DELETION	Novel
RP 14	Retinitis pigmentosa: mutation analysis of RHO, PRPF31, RP1, and IMPDH1 genes in patients from India	18552984	2008	Vision Research Foundation	287	NA	F	NC	RP	SANGER	PRPF31	DELETION	Novel
RP 14	Retinitis pigmentosa: mutation analysis of RHO, PRPF31, RP1, and IMPDH1 genes in patients from India	18552984	2008	Vision Research Foundation	288	NA	F	NC	RP	SANGER	PRPF31	DELETION	Novel
RP 14	Retinitis pigmentosa: mutation analysis of RHO, PRPF31, RP1, and IMPDH1 genes in patients from India	18552984	2008	Vision Research Foundation	289	44	F	C	RP	SANGER	RHO	MISSENSE	Reported

RP 15	Mutation analysis of codons 345 and 347 of rhodopsin gene in Indian retinitis pigmentosa patients	11910130	2001	Division of Biochemistry, Department of Chemistry, University of Pune	290	NA	NA	NC	RP	Allele-specific assays for codons 345, 347 in RHO gene	RHO	MISSENSE	Reported
RP 15	Mutation analysis of codons 345 and 347 of rhodopsin gene in Indian retinitis pigmentosa patients	11910130	2001	Division of Biochemistry, Department of Chemistry, University of Pune	291	NA	NA	NC	RP	Allele-specific assays for codons 345, 347 in RHO gene	RHO	MISSENSE	Reported
RP 16	Targeted next-generation sequencing reveals novel RP1 mutations in autosomal recessive retinitis pigmentosa	29425069	2018	Aravind Medical Research Foundation	292	38	M	C	RP	Targeted NGS and Sanger	RP1	frameshift_del	Novel
RP 17	Mutations within the cGMP-binding domain of CNGA1 causing autosomal recessive retinitis pigmentosa in human and animal model	36115851	2022	Dr. ALM PG IBMS, University of Madras, Taramani Campus,	293	22	M	NC	RP	TES	CNGA1	MISSENSE	Reported
RP 18	A novel homozygous missense mutation p.P388S in TULP1 causes protein instability and retinitis pigmentosa	33907372	2021	All India Institute Medical Sciences, Mangalagiri, Andhra Pradesh, India.	294	25	M	C	RP	Exome sequencing and sanger	TULP1	MISSENSE	Reported
RP 19	Mutation of SPATA7 in a family with autosomal recessive early-onset retinitis pigmentosa	23300508	2012	Kallam Anji Reddy Molecular Genetics Laboratory, Prof Brien Holden Eye Research Centre	295	37	M	C	RP	homozygosity mapping and sanger	SPATA7	DELETION	Novel
RP 20	Retinitis pigmentosa and congenital toxoplasmosis: a rare coexistence	17595483	2007	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, Ansari Nagar, India	296	8	M	NC	RP withcongenital toxoplasmosis	NA	NA	NA	NA
RP 21	Weill-Marchesani syndrome associated with retinitis pigmentosa	17322607	2007	Aravind Eye Hospitals	297	14	F	C	RP with Weill-Marchesani syndrome	NA	NA	NA	NA
RP 22	Prevalence of retinitis pigmentosa in South Indian population aged above 40 years	18780262	2008	Bhagwan Mahavir Vitreo-Retinal Services, Medical Research Foundation, Tamil Nadu, India.	298-310	NA	4M/9F	NA	RP	NA	NA	NA	NA
RP 23	Principal components' analysis of multifocal electroretinogram in retinitis pigmentosa	21836339	2011	Department of Vitreo-Retinal Services, Aditya Jyot Eye Hospital	311-334	NA	M (n=22)/F	NA	RP	NA	NA	NA	NA
RP 24	Effect of short-term oral valproic Acid on vision and visual field in retinitis pigmentosa	25135586	2012	Sankara Eye Hospital Varthur, Whitefield Road, Kundalahalli, Bangalore, 560037 India	335-344	NA	M (n=6)/F	NA	RP	NA	NA	NA	NA
RP 25	Efficacy of oral valproic acid in patients with retinitis pigmentosa	24955739	2014	Sankara Eye Hospital Varthur, Whitefield Road, Kundalahalli, Bangalore, 560037 Karnataka India	345-374	NA	M (n=22)/F	NA	RP	NA	NA	NA	NA
RP 26	Correlation of structure and function of the macula in patients with retinitis pigmentosa	25952950	2015	Department of Vitreoretina, Narayana Nethralaya, Bangalore, India.	375-404	NA	M (n=23)/F	NA	RP	NA	NA	NA	NA
RP 27	Acute unilateral vision loss with optic disc oedema in retinitis pigmentosa	26240107	2015	Department of Neuro-ophthalmology, L V Prasad Eye Institute, Hyderabad, , India	405	36	F	NA	optic disc oedema	NA	NA	NA	NA
RP 28	Bilateral retinitis pigmentosa with unilateral choroidal nevus: A hitherto unreported association	28210186	2016	Aditya Birla Sankara Nethralaya, 147, Mukundapur, E.M.Bypass, Kolkata 700 099, , India	406	50	M	NA	unilateral choroidal nevus	NA	NA	NA	NA
RP 29	Ultra-wide Field Fluorescein Angiography in Retinitis Pigmentosa with Intermediate Uveitis	27413510	2016	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, Ansari Nagar, India	407	18	M	NA	Intermediate Uveitis	NA	NA	NA	NA
RP 30	Choroidal thickness profile in Retinitis Pigmentosa - Correlation with outer retinal structures	26949351	2016	Smt. Kanuri Santhamma Retina Vitreous Centre, L.V. Prasad Eye Institute, Kallam Anji Reddy Campus, L.V. Prasad Marg	408-476	NA	M (n=47)/F	NA	RP	NA	NA	NA	NA
RP 31	Phenotypic heterogeneity in family members of patients with retinitis pigmentosa	37322671	2023	Shri Bhagwan Mahavir Vitreoretinal Services, Sankara Nethralaya, Tamil Nadu, India.	477	NA	NA	NA	RP	NA	NA	NA	NA
RP32	Prevalence of retinitis pigmentosa in India: the Central India Eye and Medical Study	22594809	2012	Suraj Eye Institute, Nagpur,	478	NA	NA	NA	RP	NA	NA	NA	NA
RP 33	Visual impairment and blindness due to retinitis pigmentosa in India: 15-year follow-up of the Andhra Pradesh Eye Disease Study cohort	36872706	2023	Allen Foster Community Eye Health Research Centre, Gullapalli Pratibha Rao International Centre for Advancement of Rural Eye care, L V Prasad Eye Institute	479	NA	NA	NA	RP	NA	NA	NA	NA
RP 34	DNA polymorphisms, identified by an X-chromosome short-arm probe L 1.28 (DXS7), in different racial groups	2897946	1988	Department of Human Genetics, University of Newcastle upon Tyne, UK.	480-537	NA	M (n=38)/F	NA	RP	NA	NA	NA	NA
RP 35	A genetic analysis of retinitis pigmentosa	8225518	1993	Department of Genetics, Post Graduate Institute of Basic Medical Sciences, Taramani, India	538	NA	NA	NA	RP	NA	NA	NA	NA
RP 36	Retinitis pigmentosa in India: a genetic and segregation analysis	7606847	1995	Department of Genetics & Molecular Biology, Vision Research Foundation, India.	539	NA	NA	NA	RP	NA	NA	NA	NA
RP 37	Retinitis pigmentosa genetics: a study in Indian population	8916593	1996	Division of Biochemistry, University of Poona,	540	NA	NA	NA	RP	NA	NA	NA	NA
RP 38	Prothrombin time in retinitis pigmentosa	9847484	1998	Division of Biochemistry, University of Poona,	541	NA	NA	NA	RP	NA	NA	NA	NA
RP 39	Capsulorhexis phimosis in retinitis pigmentosa despite capsular tension ring implantation	11687372	2001	Sankara Nethralaya, Chennai, Tamil Nadu, India.	542	56	M	NA	retinitis pigmentosa presented with dense nuclear sclerosis and	NA	NA	NA	NA
RP 40	Screening for homozygosity by descent in families with autosomal recessive retinitis pigmentosa	12532037	2002	L. V. Prasad Eye Institute, L. V. Prasad Marg, Banjara Hills,	543	NA	NA	NA	RP	NA	NA	NA	NA
RP 41	Mapping of locus for autosomal dominant retinitis pigmentosa on chromosome 6q23	22083234	2012	Molecular Genetics Laboratory, Hyderabad Eye Research Foundation, LV Prasad Eye Institute, LV Prasad Marg, Banjara Hills,	544	NA	NA	NA	RP	NA	NA	NA	NA
RP 42	Severity of familial isolated retinitis pigmentosa across different inheritance patterns among an Asian Indian cohort	23463886	2012	epartments of Genetics and Molecular Biology (PB, KN, JM) and Biostatistics (TM), Vision Research Foundation,	545	NA	NA	NA	RP	NA	NA	NA	NA
RP 43	A Family of Congenital Hepatic Fibrosis and Atypical Retinitis Pigmentosa	26918098	2015	Department of Pathology, Topiwala National Medical College and Bai Yamunabai Laxman Nair Hospital,	546	NA	NA	NA	Retinitis Pigmentosa with Congenital Hepatic Fibrosis	NA	NA	NA	NA
RP 44	CLINICAL PRESENTATIONS AND OUTCOMES OF RHEGMATOGENOUS RETINAL DETACHMENT IN RETINITIS PIGMENTOSA	26655616	2016	Department of Retina and Vitreous, GMR Varalakshmi Campus, LV Prasad Eye Institute, Visakhapatnam, India.	547	NA	NA	NA	RETINAL DETACHMENT IN RETINITIS PIGMENTOSA	NA	NA	NA	NA
RP 45	Concurrent retinitis pigmentosa and pigmented paravenous retinochoroidal atrophy phenotypes in the same patient	27905344	2016	Medical Research Foundation, Sankara Nethralaya, Tamil Nadu, India	548	32	F	NA	rp with pigmented paravenous retinochoroidal atrophy	NA	NA	NA	NA
RP 46	Validity and cost-effectiveness of cone adaptation test as a screening tool to detect retinitis pigmentosa	27843226	2016	Department of Retina, H. V. Desai Eye Hospital, Pune, India.	549-576	NA	NA (n=28)	NA	RP	NA	NA	NA	NA
RP 47	Ultra-wide field imaging of retinal haemangioma in retinitis pigmentosa	27553894	2017	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences,	577	55	NA	NA	retinal haemangioma in retinitis pigmentosa	NA	NA	NA	NA
RP 48	Unilateral retinitis pigmentosa: clinical and electrophysiological diagnosis	29784186	2018	Sri Sankaradeva Nethralaya,	578-580	43	M	NA	Unilateral retinitis pigmentosa	NA	NA	NA	NA
RP 48	Unilateral retinitis pigmentosa: clinical and electrophysiological diagnosis	29784186	2018	Sri Sankaradeva Nethralaya,	581-583	44	F	NA	Unilateral retinitis pigmentosa	NA	NA	NA	NA
RP 48	Unilateral retinitis pigmentosa: clinical and electrophysiological diagnosis	29784186	2018	Sri Sankaradeva Nethralaya,	584-586	23	M	NA	Unilateral retinitis pigmentosa	NA	NA	NA	NA
RP 49	Ultra-widefield imaging in Coats'-type retinitis pigmentosa	29941749	2018	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, , India	587	35	F	NA	Coats'-type retinitis pigmentosa	NA	NA	NA	NA
RP 50	Retinal Detachment in 31 Eyes with Retinitis Pigmentosa	31047295	2018	Shri BhagwanMahavir Vitreoretinal Services, Sankara Nethralaya, India	588-618	NA	M (n=29)/F (n=2)	NA	Retinal detachment (RD with RP	NA	NA	NA	NA

RP 51	Subluxated Soemmering ring in retinitis pigmentosa	31238452	2019	Cornea and Anterior Services, MGM Eye Institute, Raipur, , India	619	54	M	NA	Subluxated Soemmering ring in retinitis pigmentosa	NA	NA	NA	NA
RP 52	Quantifying microstructural changes in retinitis pigmentosa using spectral domain - optical coherence tomography	31123686	2019	Department of Vitreo Retina, Narayana Nethralaya Eye Institute,	620-732	NA	M (n=71)/F (n=42)	NA	RP	NA	NA	NA	NA
RP 53	The safety of modified bilateral electroconvulsive therapy in retinitis pigmentosa	31302437	2019	Advanced Eye Center, Postgraduate Institute of Medical Education and Research,	733	46	M	NA	RP	NA	NA	NA	NA
RP 54	Focal choroidal excavation with macular hole in a case of advanced retinitis pigmentosa	32971680	2020	Department of Vitreo-Retina, B. W. Lions Superspeciality Eye Hospital, Bengaluru, Karnataka, India	734	32	F	NA	Focal choroidal excavation with macular hole with RP	NA	NA	NA	NA
RP 55	Clinical Presentation and Demographic Distribution of Retinitis Pigmentosa in India and Implications for Potential Treatments: Electronic Medical Records Driven Big Data Analytics: Report I	34404308	2022	Center for Vitreo - Retinal Diseases, L V Prasad Eye Institute, Telangana, India.	735-15796	NA	M (n=9290) /F (n=5772)	NA	RP	NA	NA	NA	NA
RP 56	UNILATERAL RETINITIS PIGMENTOSA	18170782	1962	Gandhi Eye Hospital, India	15797	42	M	NA	UNILATERAL RETINITIS PIGMENTOSA	NA	NA	NA	NA
RP 57	Multimodal imaging in a case of bilateral astrocytic hamartoma with retinitis pigmentosa	36875628	2023	Department of Vitreo Retina, Aditya Birla Sankara Nethralaya, India	15798	15	M	NA	bilateral astrocytic hamartoma with RP	NA	NA	NA	NA
RP 58	Bilateral astrocytic hamartoma with vasoproliferative tumour in retinitis pigmentosa	36814168	2023	Retina and Vitreous Services, LV Prasad Eye Institute, Mithu Tulsi Chanri Campus,	15799	32	M	NA	Bilateral astrocytic hamartoma with vasoproliferative tumour	NA	NA	NA	NA
RP 59	Bilateral Occlusive Vasculitis Associated with Retinitis Pigmentosa - A Case Report	34797751	2023	Uvea Services, Aravind Eye Hospital, Chennai, India.	15800	34	M	NA	bilateral RP with OD inflammatory central retinal vein occlusion (CRVO) and OS occlusive vasculitis with bilateral macular edema	NA	NA	NA	NA
RP 60	POLYPOIDAL CHOROIDAL VASCULOPATHY ASSOCIATED WITH SECTOR RETINITIS PIGMENTOSA	37643040	2023	Department of Retina and Vitreous Services, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Coimbatore, India.	15801	63	F	NA	Polypoidal choroidal vasculopathy WITH RP	NA	NA	NA	NA
RP 61	Unilateral retinitis pigmentosa and macular coloboma with fellow eye pigmented paravenous chorioretinal atrophy	36598140	2023	Department of Ophthalmology, Institute of Medical Sciences & SUM Hospital, Siksha O Anusandhan University, India	15802	NA	NA	NA	RP	NA	NA	NA	NA
RP 62	Prevalence of primary angle-closure disease in retinitis pigmentosa	35791130	2022	Narayana Nethralaya, Bengaluru, Karnataka, India.	15803-16183	NA	M (n=381)	NA	RP	NA	NA	NA	NA
RP 63	Enhanced tapetal-like reflex in sector retinitis pigmentosa	34353842	2021	Retina & Vitreous, Aravind Eye Hospital Coimbatore, Coimbatore, India	16184	55	F	NA	Enhanced tapetal-like reflex with RP	NA	NA	NA	NA
RP 64	Diagnostic and Therapeutic Challenges due to Psychosis and Catatonia in Non-syndromic Retinitis Pigmentosa: A Case Report	34584317	2021	Dept of Psychiatry, Jawaharlal Institute of Postgraduate Medical Education and Research (JIPMER), India	16185	20	F	NA	Psychosis and Catatonia in RP	NA	NA	NA	NA
RP 65	Cataract surgery in retinitis pigmentosa	34146021	2021	Pediatric Ophthalmology and Strabismus Services, MGM Eye Institute, Raipur, India	16186	NA	NA	NA	RP	NA	NA	NA	NA
RP 66	Hashimoto's encephalopathy in association with retinitis pigmentosa - First reported case	32509688	2020	Department of General Medicine, Rajendra Institute of Medical Sciences, Ranchi, India	16187	NA	NA	NA	Hashimoto's Encephalopathy with RP	NA	NA	NA	NA
RP 67	Retinitis pigmentosa with bilateral irido-fundal coloboma	32971656	2020	Department of Retina, Sri Kiran Institute of Ophthalmology, Kakinada, India	16188	40	male	yes	bilateral irido-fundal coloboma with RP	NA	NA	NA	NA
RP 68	Rare case of simultaneous manifestation of pigmented paravenous retinochoroidal atrophy and retinitis pigmentosa in contralateral eye	31638054	2019	Department of Retina and Vitreous, Retina Foundation, Ahmedabad, India	16189	22	F	NA	pigmented paravenous retinochoroidal atrophy with RP	NA	NA	NA	NA
RP 69	Isolated microspherophakia with retinitis pigmentosa	31238436	2019	Advanced Eye Centre, Postgraduate Institute of Medical Education and Research, India	16190	42	F	NA	microspherophakia with retinitis pigmentosa	NA	NA	NA	NA
RP 70	Double trouble: exudative hypertensive retinopathy in a patient with retinitis pigmentosa	30413460	2018	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences,	16191	24	F	NA	exudative hypertensive retinopathy with RP	NA	NA	NA	NA
RP 71	Uveitis in Patients with Retinitis Pigmentosa: 30 Years' Consecutive Data	28960116	2018	Shri Bhagwan Mahavir Vitreoretinal Services , Sankara Nethralaya, India.	16192	NA	NA	NA	Uveitis with RP	NA	NA	NA	NA
RP 72	Unilateral Macular Star in a Case of Hypertension and Retinitis Pigmentosa	27128810	2017	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences , India	16193	17	F	NA	Unilateral Macular Star HT with RP	NA	NA	NA	NA
RP 73	Intravitreal dexamethasone implant for recalcitrant cystoid macular edema secondary to retinitis pigmentosa: a pilot study	28378252	2015	Eye Hospital and Retina Centre, Mahajan Lane, Raopura,	16194	NA	NA	NA	Cystoid macular edema with RP	NA	NA	NA	NA
RP 74	Proliferative diabetic retinopathy in typical retinitis pigmentosa	26021380	2015	Cataract & Medical Retina Services, Vasan Eye Care, Tamil Nadu, India.	16195	39	F	NA	PDR with RP	NA	NA	NA	NA
RP 75	Retinitis pigmentosa patients with sickle cell disease and dextrocardia and situs inversus syndrome	15887731	2001	Department of Genetics and Molecular Biology, Vision Research Foundation, Sankara Nethralaya,	16196	20	M	NA	RP with SCD	NA	NA	NA	NA
RP 75	Retinitis pigmentosa patients with sickle cell disease and dextrocardia and situs inversus syndrome	15887731	2001	Department of Genetics and Molecular Biology, Vision Research Foundation, Sankara Nethralaya,	16197	13	M	NA	RP with SCD	NA	NA	NA	NA

BBS 6	Goyal S, Jäger M, Robinson PN, Vanita V. Confirmation of TTC8 as a disease gene for nonsyndromic autosomal recessive retinitis pigmentosa (RP51). Clin Genet. 2016 Apr;89(4):454-460. doi: 10.1111/cge.12644. Epub 2015 Aug 25. PMID: 26195043.	26195043	2016	1 Department of Human Genetics, Guru Nanak Dev University, Amritsar, India. 2 Institute for Medical and Human Genetics, Berlin, Germany. 3 Berlin Brandenburg Center for Regenerative Therapies (BCRT), Charité-Universitätsmedizin Berlin, Berlin, Germany. 4 Max Planck Institute for Molecular Genetics, Berlin, Germany. 5 Department of Mathematics and Computer Science, Institute for Bioinformatics, Freie Universität Berlin, Berlin, Germany.	16198	22	Male	C	AR-RP	WES	TTC8 (BBS8)	Missense	Novel
LCA													
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	1	15 Years	M	No	LCA	Clinical Exome	SPATA7	3' splice site	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	2	10 Years	M	No	LCA	Clinical Exome	AIPL1	Frame shift	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	3	2 Years	M	Yes	LCA	Clinical Exome	CRB1	Non-sense	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	4	9 Years	F	No	LCA	Clinical Exome	GUCY2D	Substitution	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	5	1 Year	M	Yes	LCA	Clinical Exome	GUCY2D	Non-sense	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	6	1 Year	M	No	LCA	Clinical Exome	AIPL1	5' splice site	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	7	6 Years	F	Yes	LCA	Clinical Exome	RPE65	Missense	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	8	8 Months	F	No	LCA	Clinical Exome	CRX	Deletion FS	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	9	12 Years	F	No	LCA	Clinical Exome	AIPL1	Substituion	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	10	3 Years	M	Yes	LCA	Clinical Exome	GUCY2D	5' splice site	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	11	4 Months	F	No	LCA	Clinical Exome	USH2A	Missense	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	11	4 Months	F	No	LCA	Clinical Exome	CEP290	Non-sense	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	12	2 Years	M	Yes	LCA	Clinical Exome	MERTK	Missense	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	13	3 Years	M	Yes	LCA	Clinical Exome	CRB1	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	13	3 Years	M	Yes	LCA	Clinical Exome	RIMS1	Missense	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	14	5 Years	F	No	LCA	Clinical Exome	ALMS1	Deletion	Reported
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2023	Narayana Netralaya Foundataion	14	5 Years	F	No	LCA	Clinical Exome	ALMS1	Deletion	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	15	15	M	NA	LCA	TP	*SPATA7*	missense	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	16	0.5	M	NA	LCA	TP	*EYS*	missense	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	17	10	M	NA	LCA	TP	*AIPL1*	deletion FS	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	18	7	F	NA	LCA	TP	*CRX*	missense	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	19	2	M	NA	LCA	TP	*CRB1*	missense	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	20	9	F	NA	LCA	TP	*GUCY2D*	missense	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	21	1	M	NA	LCA	TP	*AIPL1*	splice site	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	22	1	M	NA	LCA	TP	*GUCY2D*	missense	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	23	7	F	NA	LCA	WES	*CEP290*	missense	Reported
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	24	5.5	F	NA	LCA	WES	*RPGRIP1*	inertion FS	Reported
LCA 1	A novel c.1937T>C (p.Leu646Pro) missense mutation in a patient with Leber congenital amaurosis.	35101627	2022	Department of Ophthalmology, Post Graduate Institute of Medical Education and Research, Chandigarh, India. Department of Paediatrics, Post Graduate Institute of Medical Education and Research, Chandigarh, India.	25	10 M	F	NA	LCA	Focussed exome sequencing	GUCY2D	Missense	Reported
LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	26	1	F	C	LCA	Clinical Exome	RPE65	duplication FS	Reported

LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	27	1	M	C	LCA	Clinical Exome	LCA5	deletion FS	Reported
LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	28	1	F	C	LCA	Clinical Exome	LCA5	deletion FS	Novel
LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	29	0.75	M	NC	LCA	Clinical Exome	CRX	deletion FS	Novel
LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	30	2	F	C	LCA	Clinical Exome	PRPH2	Missense	Reported
LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	31	0.5	F	C	LCA	Clinical Exome	CEP290	Missense	Novel
LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	32	6	M	C	LCA	Clinical Exome	ALMS1	deletion FS	Novel gene for the phenotype
LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	33	2	F	C	LCA	Clinical Exome	IFT80	Missense	Novel gene
LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	34	2	F	C	LCA	Clinical Exome	RP1	deletion FS	Novel gene for the phenotype

LCA 2	Clinical exome sequencing facilitates the understanding of genetic heterogeneity in Leber congenital amaurosis patients with variable phenotype in southern India	33957996	2021	Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. Department of Molecular Biology, Aravind Medical Research Foundation - Affiliated to Alagappa University, Karaikudi, Tamil Nadu, India. Department of Paediatric and Adult strabismus, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Bioinformatics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India. Department of Molecular Genetics, Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, 625020, India. sundar@aravind.org.	35	2	M	C	LCA	Clinical Exome	No variant of interest	NA	NA
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	36	15	M	C	LCA	GeneChip Human Mapping 250K Nspl Array/SNP Genotyping/homozygosity mapping	RPE65	splice site	Reported
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	37	19	M	C	LCA	GeneChip Human Mapping 250K Nspl Array/SNP Genotyping/homozygosity mapping	CRB1	Missense	Reported
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	38	19	M	C	LCA	GeneChip Human Mapping 250K Nspl Array/SNP Genotyping/homozygosity mapping	GUCY2D	deletion FS	Novel
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	39	34	F	C	LCA	GeneChip Human Mapping 250K Nspl Array/SNP Genotyping/homozygosity mapping	IQCB1	splice site	Novel
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	40	14	M	C	LCA	GeneChip Human Mapping 250K Nspl Array/SNP Genotyping/homozygosity mapping	AIPL1	Missense	Reported
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	41	14	F	C	LCA	GeneChip Human Mapping 250K Nspl Array/SNP Genotyping/homozygosity mapping	Unidentified	NA	Reported
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	42	5	F	C	LCA	GeneChip Human Mapping 250K Nspl Array/SNP Genotyping/homozygosity mapping	AIPL1	Missense	Novel

LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	43	26	M	C	LCA	GeneChip Human Mapping 250K NspI Array/SNP Genotyping/homozygosity mapping	RPE65	Missense	Reported
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	44	29	F	C	LCA	GeneChip Human Mapping 250K NspI Array/SNP Genotyping/homozygosity mapping	AIPL1	nonsense	Novel
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	45	8	M	C	LCA	GeneChip Human Mapping 250K NspI Array/SNP Genotyping/homozygosity mapping	SPATA7	Missense	Novel
LCA 3	Homozygosity Mapping in Leber Congenital Amaurosis and Autosomal Recessive Retinitis Pigmentosa in South Indian Families.	26147992	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India; Ph.D Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. Head, Centre for Bioinformatics, Vision Research Foundation, Chennai, India. Department of Peadiatric ophthalmology and starbismus, Medical Research Foundation, Chennai, India. Department of Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Head, Department of Biological Science, Birla Institute of Technology & Science (BITS), Hyderabad, India.	46	26	F	C	LCA	GeneChip Human Mapping 250K NspI Array/SNP Genotyping/homozygosity mapping	RPGRIP1RDH12	splice site	Novel
LCA 5	Repetitive Eye Poking in an Infant - A Diagnostic Conundrum	33452791	2021	All India Institute of Medical Sciences (AIIMS), Mangalagiri, AP, India	47	1Y	F	C	LCA	Clinical Exome	GUCY2D	Missense	Reported
LCA 6	Multiple retinal astrocytic hamartomas in siblings with lebers congenital amaurosis: a case series and review of literature.	32967644	2020	Dr. Shroff Charity Eye Hospital, 5027, Kedarnath Lane, Daryaganj, Delhi, New Delhi, 110002, India. laganpaul@gmail.com.	48	8	M	NA	LCA+RAH	NA	NA	NA	NA
LCA 7	Co-Occurrence of Leber Congenital Amaurosis and Meckel Syndrome Type 1 in a Fetus: Is There a Lesson to Be Learned?	31191208	2019	Department of Medical Genetics, Nizam's Institute of Medical Sciences, Hyderabad, India. Diagnostics Division, Centre for DNA Fingerprinting and Diagnostics (CDFD), Hyderabad, India.	49	Fetus	F	C	MKS+LCA	WES	MKS1	Missense	Reported
LCA 7	Co-Occurrence of Leber Congenital Amaurosis and Meckel Syndrome Type 1 in a Fetus: Is There a Lesson to Be Learned?	31191208	2019	Department of Medical Genetics, Nizam's Institute of Medical Sciences, Hyderabad, India. Diagnostics Division, Centre for DNA Fingerprinting and Diagnostics (CDFD), Hyderabad, India.	50	Fetus	F	C	MKS+LCA	WES	RPGRIP1	deletion	Novel
LCA 8	Retinal detachment in a child with severe early childhood onset retinal dystrophy.	30150358	2018	Dr Rajendra Prasad center for ophthalmic sciences, New Delhi, All India Institute of Medical Science, Bhopal, Madhya Pradesh,	51	10Y	M	NA	severe early childhood onset retinal dystrophy (SECORD) with macular coloboma in BE, with a rhegmatogenous retinal detachment (RRD) in LE (in lieu of subretinal proliferative vitreoretinopathy (PVR) and pigments behind the lens}	NA	NA	deletion	Reported
LCA 9	Retinal astrocytic hamartoma in a patient with Leber's congenital amaurosis.	25737223	2015	LV Prasad Eye Institute, Hyderabad, Telangana, India. UC Irvine, Irvine, California, USA.	52	NA	F	C	RAH+LCA	NA	NA	NA	NA
LCA 10	Leber's congenital amaurosis as the retinal degenerative phenotype in thiamine responsive megaloblastic anemia: a case report.	23638917	2014	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation , Chennai, Tamil Nadu , India DEPT. of pediatric ophthalmology, medical research foundation chennai	53	10M	F	C	TRMA+A female child presented with Leber's congenital amaurosis at 10 months of age, later diagnosed with hearing	NR	SLC19A2	missense	NOVEL
LCA 11	Ultra wide field imaging of coats like response in Leber's congenital amaurosis.	28559726	2017	Dr. RP Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India	54	12	M	NA	LCA	NA	NA	NA	NA
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	55	3	M	N	LCA	Direct RPE65 seq	RPE65	insertion	novel
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	56	4	F	Y	LCA	Direct RPE65 seq	RPE65	missense	novel
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	57	7	M	Y	LCA	Direct RPE65 seq	RPE65	deletion FS	reported
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	58	6	F	Y	LCA	Direct RPE65 seq	RPE65	deletion FS	reported
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	59	4	M	N	LCA	Direct RPE65 seq	RPE65	missense	reported

LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	60	4	M	Y	LCA	Asper chip analysis confirmed by directional sequencing	GUCY2D	missense	reported
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	61	11	F	Y	LCA	Asper chip analysis confirmed by directional sequencing	GUCY2D	missense	reported
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	62	2	M	N	LCA	Asper chip analysis confirmed by directional sequencing	GUCY2D	duplication	novel
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	63	5	M	Y	LCA	Asper chip analysis confirmed by directional sequencing	AIPL1	nonsense	reported
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	64	7	M	Y	LCA	Asper chip analysis confirmed by directional sequencing	CRX	missense	reported
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	64	7	M	Y	LCA	Asper chip analysis confirmed by directional sequencing	IQCB1	nonsense	reported
LCA 12	Mutational screening of LCA genes emphasizing RPE65 in South Indian cohort of patients.	24066033	2013	Aravind Medical Research Foundation, Aravind Eye Hospital, Madurai, Tamil Nadu, India.	65	3	F	Y	LCA	Asper chip analysis confirmed by directional sequencing	RPGRIP1	nonsense	reported
LCA 13	Mutations that are a common cause of Leber congenital amaurosis in northern America are rare in southern India.	19753312	2009	Department of Genetics, Dr. G. Venkataswamy Eye Research Institute, Aravind Medical Research Foundation, Madurai, Tamilnadu, India.	66	3	F	Y	LCA	allele-specific assay	RPE65	missense	reported
LCA 13	Mutations that are a common cause of Leber congenital amaurosis in northern America are rare in southern India.	19753312	2009	Department of Genetics, Dr. G. Venkataswamy Eye Research Institute, Aravind Medical Research Foundation, Madurai, Tamilnadu, India.	67	4 months	F	N	LCA	Bidirectional sequencing	RPE65	missense	reported
LCA 14	Screening of the RPE65 gene in the Asian Indian patients with leber congenital amaurosis.	18484312	2008	Vision Research Foundation, Sankara Nethralaya, Chennai, India	68	3	F	Y	LCA	direct sequencing of PCR	RPE65	Missense	novel
LCA 14	Screening of the RPE65 gene in the Asian Indian patients with leber congenital amaurosis.	18484312	2008	Vision Research Foundation, Sankara Nethralaya, Chennai, India	69	NA	NA	NA	LCA	direct sequencing of PCR	RPE65	Missense	reported
LCA 14	Screening of the RPE65 gene in the Asian Indian patients with leber congenital amaurosis.	18484312	2008	Vision Research Foundation, Sankara Nethralaya, Chennai, India	70	NA	NA	NA	LCA	direct sequencing of PCR	RPE65	isocoding	reported
LCA 15	Identification of a novel splice-site mutation in the Lebercilin (LCA5) gene causing Leber congenital amaurosis.	18334959	2008	Department of Genetics and Molecular Biology, Vision Research Foundation, Sankara Nethralaya Chennai, India.	71	7m	F	Y	LCA	Homozygosity mapping	LCA5	splice site	Novel
LCA 16	Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration.	17186464	2006	From the Departments of Ophthalmology and Visual Sciences (J.S.F.; K.B.; M.O.; E.F.; D.A.T.; J.R.H. A.S.) and Human Genetics (A.S.), W. K. Kellogg Eye Center, University of Michigan, Ann Arbor; the Jackson Laboratory, Bar Harbor, Maine (B.C.; N.L.H.); Kallam Anji Reddy Molecular Genetics Laboratory (C.K.; H.P.S.) and Kannuri Santhamma Retina-Vitreous Services (S.J.), L. V. Prasad Eye Institute, Banjara Hills, Hyderaba, India; Department of Molecular Genetics, Institute of Ophthalmology, University College London (C.C.; A.R.W.; S.S.B.), and Moorfield Eye Hospital (A.R.W.), London; Department of Ophthalmology, University Hospital of Lund, Lund, Sweden (S.A.); and Department of Ophthalmology, Scheie Eye Institute, University of Pennsylvania, Philadelphia (S.G.J.)	72	NA	NA	NA	LCA	PCR	RD3	splice site	Novel gene
LCA 17	Electroretinographic assessment and diagnostic reappraisal of children with visual dysfunction: a prospective study.	17322600	2007	Department of Retina-Vitreous Services, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamil Nadu, India. drvasumathy@yahoo.com	73	4.4+/- 3.2	20 M/27 F	NA	LCA	NA	NA	NA	NA
LCA 18	RPE65 gene: multiplex PCR and mutation screening in patients from India with retinal degenerative diseases.	12357075	2002	Department of Genetics and Molecular Biology, Medical and Vision Research Foundations, Sankara Nethralaya, Chennai 600 006, India.	74	NA	NA	NA	LCA	multiplex PCR follwed by sequencing for RPE65	RPE65	missense	Reported
LCA 18	RPE65 gene: multiplex PCR and mutation screening in patients from India with retinal degenerative diseases.	12357075	2002	Department of Genetics and Molecular Biology, Medical and Vision Research Foundations, Sankara Nethralaya, Chennai 600 006, India.	75	NA	NA	NA	LCA	multiplex PCR follwed by sequencing for RPE65	RPE65	isocoding	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	76	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	missense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	77	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	missense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	78	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	missense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	79	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	80	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	nonsense	Reported

LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	81	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	81	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	82	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	83	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	deletion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	84	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	deletion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	85	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	86	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	nonsense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	87	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	87	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	GUCY2D	deletion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	88	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	AIPL1	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	89	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	AIPL1	missense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	90	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	AIPL1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	91	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	AIPL1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	92	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	AIPL1	nonsense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	93	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	AIPL1	nonsense	Novel

LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	94	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	AIPL1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	95	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPE65	deletion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	96	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPE65	splice site	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	97	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPE65	duplication FS	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	98	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPE65	duplication FS	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	99	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPE65	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	100	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPE65	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	101	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPE65	missense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	101	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPE65	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	102	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPGRIP1	missense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	103	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPGRIP1	nonsense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	104	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPGRIP1	nonsense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	105	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPGRIP1	deletion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	106	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RPGRIP1	missense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	107	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	LCA5	nonsense	Reported

LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	108	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	LCA5	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	109	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	LCA5	deletion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	110	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	LCA5	deletion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	111	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	LCA5	deletion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	112	between 4 and 11	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	IQCB1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	113	7months	M	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	IQCB1	nonsense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	114	between 4 and 11	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	IQCB1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	115	between 4 and 11	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	IQCB1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	116	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	CRB1	insertion FS	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	117	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	CRB1	splice site	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	118	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	CRB1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	119	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	CRB1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	120	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RDH12	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	121	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RDH12	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	122	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RDH12	missense	Novel

LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	123	NA	NA	C	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	SPATA7	splice site/silent	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	124	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	SPATA7	splice site	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	125	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	SPATA7	splice site	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	126	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	NMNAT1	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	127	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	NMNAT1	missense	Novel
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	128	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	CRX	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	129	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	RD3	splice site	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	130	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	TULP1	missense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	131	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	AIPL1	nonsense	Reported
LCA 19	Genetic profile and mutation spectrum of Leber congenital amaurosis in a larger Indian cohort using high throughput targeted re-sequencing.	29068479	2018	Vision Research Foundation, Chennai, India. Birla Institute of Technology & Science (BITS), Hyderabad, India Medical Research Foundation, Chennai, India.	132	NA	NA	NA	LCA	Targeted re-sequencing of 20 candidate genes was performed using Agilent HaloPlex target enrichment assay and sequenced on Ilumina MiSeq platform	KCNJ13	missense	Reported
AS 1	Srikrupa NN, Sripriya S, Pavithra S, Sen P, Gupta R, Mathavan S. Whole-exome sequencing identifies two novel ALMS1 mutations in Indian patients with Leber congenital amaurosis. Hum Genome Var. 2021 Mar 29;8(1):12. doi: 10.1038/s41439-021-00143-z. PMID: 33782391; PMCID: PMC8007799.	33782391	2021	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Sankara Nethralaya, Chennai, India	133	18 months	Female	NC	LCA, ALMS1	Whole Exome Sequencing	ALMS1	Nonsense	Yes
AS 1	Srikrupa NN, Sripriya S, Pavithra S, Sen P, Gupta R, Mathavan S. Whole-exome sequencing identifies two novel ALMS1 mutations in Indian patients with Leber congenital amaurosis. Hum Genome Var. 2021 Mar 29;8(1):12. doi: 10.1038/s41439-021-00143-z. PMID: 33782391; PMCID: PMC8007799.	33782391	2021	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Sankara Nethralaya, Chennai, India	134	2.5 years	Male	NC	LCA, ALMS1	Whole Exome Sequencing	ALMS1	Frameshift	Yes
STGD													
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	1	NA	NA	NA	STGD	Sanger	ABCA4	Stop Codon	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	2	NA	NA	NA	STGD	Sanger	ABCA4	Stop Codon	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	3	NA	NA	NA	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	4	NA	NA	NA	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	5	NA	NA	NA	STGD	Sanger	ABCA4	Missense	Novel
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	6	NA	NA	NA	STGD	Sanger	ABCA4	Stop Codon	Reported

STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	7	NA	NA	NA	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	8	16	M	C	STGD	Sanger	ABCA4	Stop Codon	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	9	8	M	C	STGD	Sanger	ABCA4	Stop Codon	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	10	7	M	C	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	11	10	F	NC	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	12	30	F	C	STGD	NGS	ABCA4	Missense	Novel
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	12	30	F	C	STGD	NGS	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	13	10	M	C	STGD	NGS	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	14	16	F	C	STGD	NGS	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	15	16	M	C	STGD	NGS	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	16	17	F	NC	STGD	Sanger	ABCA4	Stop Codon	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	16	17	F	NC	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	16	17	F	NC	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	17	12	M	NC	STGD	Sanger	ABCA4	DELETION	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	17	12	M	NC	STGD	Sanger	ABCA4	Stop Codon	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	18	14	M	NC	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	18	14	M	NC	STGD	Sanger	ABCA4	SPLICING VARAINT	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	19	NA	NA	NA	STGD	Sanger	ABCA4	Stop Codon	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	20	NA	NA	NA	STGD	Sanger	ABCA4	Missense	Reported
STGD 1	Genetic characterization of Stargardt clinical phenotype in South Indian patients using sanger and targeted sequencing	31934596	2020	Department of Genetics, Aravind Medical Research Foundation	21	NA	NA	NA	STGD	Sanger	ABCA4	Ins/Del	Nocel
STGD 2	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease	25922843	2015	GROW Research Laboratory, Narayana Nethralaya, Narayana Health City,	22	27	M	NC	group 1 STGD.	NGS and Sanger	ABCA4	Missense	Reported
STGD 2	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease	25922843	2015	GROW Research Laboratory, Narayana Nethralaya, Narayana Health City,	22	27	M	NC	group 1 STGD.	NGS and Sanger	ABCA4	Missense	Reported
STGD 2	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease	25922843	2015	GROW Research Laboratory, Narayana Nethralaya, Narayana Health City,	23	31	M	NC	group 1 STGD.	NGS and Sanger	ABCA4	SPLICING VARAINT	Novel
STGD 2	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease	25922843	2015	GROW Research Laboratory, Narayana Nethralaya, Narayana Health City,	23	31	M	NC	group 1 STGD.	NGS and Sanger	ABCA4	Missense	Novel
STGD 2	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease	25922843	2015	GROW Research Laboratory, Narayana Nethralaya, Narayana Health City,	24	22	M	NC	group 3 STGD	NGS and Sanger	ABCA4	Missense	Reported

STGD 2	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease	25922843	2015	GROW Research Laboratory, Narayana Nethralaya, Narayana Health City,	25	26	M	NC	group 1 STGD.	NGS and Sanger	ABCA4	STOP MUTATION	Novel
STGD 2	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease	25922843	2015	GROW Research Laboratory, Narayana Nethralaya, Narayana Health City,	25	26	M	NC	group 1 STGD.	NGS and Sanger	ABCA4	Missense	Reported
STGD 2	Identification of Novel Mutations in ABCA4 Gene: Clinical and Genetic Analysis of Indian Patients with Stargardt Disease	25922843	2015	GROW Research Laboratory, Narayana Nethralaya, Narayana Health City,	26	16	F	C	group 2 STGD	NGS and Sanger	ABCA4	STOP MUTATION	Reported
STGD 3	Whole exome sequencing identifies a novel splice-site mutation in IMPG2 gene causing Stargardt-like juvenile macular dystrophy in a north Indian family	34990796	2022	Department of Molecular and Human Genetics, Institute of Science, Banaras Hindu University, Varanasi	27	NA	NA	NC	Stargardt-like juvenile macular dystrophy.	-	IMPG2	SPLICING VARAINT	Novel
STGD 4	Focal choroidal excavation in Stargardt's dystrophy	32843395	2020	Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India	28	35	F	NA	Focal choroidal excavation in Stargardt's	NA	NA	NA	NA
STGD 5	Visual rehabilitation using video game stimulation for Stargardt disease	30886944	2019	Department of Vitreoretinal diseases, Medical Research Foundation, Sankara Nethralaya, Chennai, India	29-36	NA	F (n=6)/M (n=2)	NA	Stargardt	NA	NA	NA	NA
STGD 6	Swept-source optical coherence tomography study of choroidal morphology in Stargardt disease	29930450	2018	Department of General Ophthalmology, Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	37-62	NA	NA	NA	Stargardt	NA	NA	NA	NA
STGD 7	Clinical profile and demographic distribution of Stargardt disease phenotypes: An Electronic medical record-driven big data analytics from a multitier eye care network	37787244	2023	Department of Ophthalmology, Anant Bajaj Retina Institute, L V Prasad Eye Institute, Bhubaneswar, Odisha, India	62-1996	NA	NA	NA	Stargardt	NA	NA	NA	NA
STGD 8	Insights into autofluorescence patterns in Stargardt macular dystrophy using ultra-wide-field imaging	28689222	2017	Dr. R.P. Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, 57, Sadar Apartments, Mayur Vihar Phase 1 extension, New Delhi	1997-2007	NA	NA	NA	Stargardt	NA	NA	NA	NA
STGD 9	CHOROIDAL STRUCTURAL CHANGES AND VASCULARITY INDEX IN STARGARDT DISEASE ON SWEPT SOURCE OPTICAL COHERENCE TOMOGRAPHY	29016459	2018	Department of Ophthalmology, Medical Research Foundation, Sankara Netralya, Chennai, India.	2008-2054	NA	NA	NA	Stargardt	NA	NA	NA	NA
STGD 10	Comparative analysis of autofluorescence and OCT angiography in Stargardt disease	29074493	2018	Department of Smt Kanuri Santhamma Centre for Vitreo Retinal Diseases, LV Prasad Eye Institute, Hyderabad, India.	2055-2080	NA	NA	NA	Stargardt	NA	NA	NA	NA
STGD 11	Epiretinal membrane removal in patients with Stargardt disease	25686068	2015	Department of Vitreoretina, Shri Bhagwan Mahavir Vitreoretinal Services, Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	2081-2082	31	M	NA	Stargardt	NA	NA	NA	NA
STGD 11	Epiretinal membrane removal in patients with Stargardt disease	25686068	2015	Department of Vitreoretina, Shri Bhagwan Mahavir Vitreoretinal Services, Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	2083-2084	24	M	NA	Stargardt	NA	NA	NA	NA
STGD 12	Three-dimensional spectral domain optical coherence tomography in Stargardt disease and fundus flavimaculatus	24319520	2012	Department of Ophthalmology, King George's Medical University, Lucknow, India	2085-2086	16	M	NA	Stargardt	NA	NA	NA	NA
STGD 12	Three-dimensional spectral domain optical coherence tomography in Stargardt disease and fundus flavimaculatus	24319520	2012	Department of Ophthalmology, King George's Medical University, Lucknow, India	2087-2088	36	M	NA	Stargardt	NA	NA	NA	NA
STGD 13	Choroidal osteoma and pattern dystrophy of retinal pigment epithelium	29260499	2019	Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi,	2089	30	F	NA	Stargardt	NA	NA	NA	NA
STGD 14	Multimodal imaging in multifocal pattern dystrophy simulating fundus flavimaculatus	27380982	2016	Shri Bhagwan Mahavir Vitreoretinal Services, Sankara Nethralaya, Chennai, Tamil Nadu, India	2090	55	M	NA	dystrophy simulating fundus flavimaculatus	NA	NA	NA	NA
STGD 15	Visual rehabilitation using microperimetric acoustic biofeedback training in individuals with central scotoma	30253443	2019	Department of Comprehensive Ophthalmology, Medical Research Foundation, Chennai, India.	2091	NA	NA	NA	Stargardt	NA	NA	NA	NA
STGD 16	Choroidal thickness profile in inherited retinal diseases in Indian subjects	26139798	2015	Smt. Kanuri Santhamma Retina Vitreous Centre, L. V. Prasad Eye Institute, Hyderabad, Telangana, India	2092	NA	NA	NA	Stargardt	NA	NA	NA	NA
STGD 17	Morphological features of focal choroidal excavation and its association with macular pathology in Asian Indian eyes	33727453	2021	Department of Vitreo-Retina, Narayana Nethralaya Eye Institute, Bangalore, Karnataka, India	2093	NA	NA	NA	Stargardt	NA	NA	NA	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2094	30 Years	F	C	Stargardt disease	Clinical Exome	ABCA4	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2095	6 Years	M	C	Stargardt disease	Clinical Exome	ABCA4	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2096	12 Years	M	NC	Stargardt disease	Clinical Exome	ABCA4	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2096	12 Years	M	NC	Stargardt disease	Clinical Exome		Non-sense	NA

IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2097	16 Years	M	C	Stargardt disease	Clinical Exome	ABCA4	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2098	11 Years	F	C	Stargardt disease	Clinical Exome	ABCA4	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2098	11 Years	F	C	Stargardt disease	Clinical Exome	ABCA4	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2099	30 Years	M	C	Stargardt disease	Clinical Exome	ABCA4	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2100	20 Years	F	C	Stargardt disease	Clinical Exome	ABCA4	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2101	28 Years	M	C	Stargardt disease	Clinical Exome	C2orf71	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2102	28 Years	F	NC	Stargardt disease	Clinical Exome	CERKL	Deletion FS	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2103	8 Years	M	NC	Stargardt disease	Clinical Exome	ABCA4	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2103	8 Years	M	NC	Stargardt disease	Clinical Exome	ABCA4	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2103	8 Years	M	NC	Stargardt disease	Clinical Exome	ABCA4	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2103	8 Years	M	NC	Stargardt disease	Clinical Exome	ABCA4	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2104	24 Years	F	NC	Stargardt disease	Clinical Exome	ABCA4	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2104	24 Years	F	NC	Stargardt disease	Clinical Exome	ABCA4	3' splice site	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	2105	30	F	NA	Stargardt disease	TP	*ABCA4*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	2106	5	M	NA	Stargardt disease	TP	*PROM1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	2107	6	M	NA	Stargardt disease	TP	*ABCA4*	splice site	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	2108	24 years	F	NA	Stargardt disease	exome sequencing/panel-based sequencing/chromosomal microarray	ABCA4	missense	NA
IRD 5	Derivation of three induced pluripotent stem cell lines under feeder-free culture conditions from peripheral blood mononuclear cells (PBMC) of Indian patients suffering from inherited retinal diseases carrying different mutations.	32278301	2020	Eyestem Research Private Limited, C-CAMP, GKVK Post, Bangalore 560065, Karnataka, India. Centre for Eye Genetics and Research, Cytecare Hospital, Bangalore 560064, Karnataka, India. Eyestem Research Private Limited, C-CAMP, GKVK Post, Bangalore 560065, Karnataka, India.	2109	19	F	NA	STGD	NA	ABCA4	missense	NA
IRD 5	Derivation of three induced pluripotent stem cell lines under feeder-free culture conditions from peripheral blood mononuclear cells (PBMC) of Indian patients suffering from inherited retinal diseases carrying different mutations.	32278301	2020	Eyestem Research Private Limited, C-CAMP, GKVK Post, Bangalore 560065, Karnataka, India. Centre for Eye Genetics and Research, Cytecare Hospital, Bangalore 560064, Karnataka, India. Eyestem Research Private Limited, C-CAMP, GKVK Post, Bangalore 560065, Karnataka, India.	2109	19	F	NA	STGD	NA	ABCA4	missense	NA
IRD 8	Choroidal structural analysis and vascularity index in retinal dystrophies	30178525	2019	Aravind Eye Hospital, Madurai, India.	2110-2113	NA	NA	NA	Stargardt	NA	NA	NA	NA
IRD 9	Focal Choroidal Excavation in Retinal Dystrophies	27533784	2018	L. V. Prasad Eye Institute	2114-2160	NA	NA	NA	Stargardt	NA	NA	NA	NA
IRD 10	Choroidal hyper-reflective foci and vascularity in retinal dystrophy	31856490	2020	Moorfields Eye Hospital NHS Foundation Trust, London, United Kingdom, Srimati Kanuri Santhamma Centre for Vitreo-Retinal Diseases, Hyderabad Eye Research LVPEI	2161	10	FEMALE	NA	Stargardt	NA	NA	NA	NA

IRD 10	Choroidal hyper-reflective foci and vascularity in retinal dystrophy	31856490	2020	Moorfields Eye Hospital NHS Foundation Trust, London, United Kingdom, Srimati Kanuri Santhamma Centre for Vitreo-Retinal Diseases, Hyderabad Eye Research LVPEI	2162	18	MALE	NA	Stargardt	NA	NA	NA	NA
IRD 11	Evaluation of photoreceptor function in inherited retinal diseases using rod- and cone-enhanced flicker stimuli	33834501	2021	L. V. Prasad Eye Institute	2162-2175	23.3 - 12.2 years	8 M, 5 F	NA	Stargardt	NA	NA	NA	NA
CDD													
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	1	23 Years	F	Yes	Cone rod dystrophy	Clinical Exome	ABCA4	Non-sense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2	3 Years	M	Yes	Cone-Rod Dystrophy	Clinical Exome	RPGR	Deletion	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	3	9 Years	M	No	Cone rod dystrophy	Clinical Exome	CRX	Insertion	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	4	25 Years	F	No	Cone rod dystrophy	Clinical Exome	PROM1	Missense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	5	9 Years	F	Yes	Cone rod dystrophy	Clinical Exome	CNGA3	Missense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	6	26 Years	M	No	Cone dystrophy	Clinical Exome	ABCA4	Duplication	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	6	26 Years	M	No	Cone dystrophy	Clinical Exome	ABCA4	Missense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	7	4 Years	F	Yes	Cone dystrophy	Clinical Exome	ATF6	Non-sense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	8	14 Years	F	No	Cone dystrophy	Clinical Exome	ABCA4	Missense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	8	14 Years	F	No	Cone dystrophy	Clinical Exome	ABCA4	Non-sense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	9	8 Years	M	No	Cone or cone-rod dystrophy.	Clinical Exome	CHM	5' splice site	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	10	5 Years	F	Yes	Cone rod dystrophy	Clinical Exome	CNGA3	Deletion	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	10	5 Years	F	Yes	Cone rod dystrophy	Clinical Exome	CNGA3	Missense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	10	5 Years	F	Yes	Cone rod dystrophy	Clinical Exome	CACNA2D4	Missense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	10	5 Years	F	Yes	Cone rod dystrophy	Clinical Exome	CACNA2D4	5' splice site	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	11	9 Years	F	No	Cone dystrophy	Clinical Exome	CRX	Missense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	12	1 Year	F	No	Cone Dystrophy	Clinical Exome	PDE6B	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	12	1 Year	F	No	Cone Dystrophy	Clinical Exome	IFT172	Missense	R
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	12	1 Year	F	No	Cone Dystrophy	Clinical Exome	IFT172	Missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	13	22	M	NA	Cone dystrophy	TP	*GUCY2D*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	14	27	M	NA	Photophobia, cone rod dystrophy	TP	*RIMS1*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	15	23	F	NA	Cone rod dystrophy	TP	*ABCA4*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	16	20	M	NA	Cone rod dystrophy	TP	*RPGRIP1*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	17	2.75	F	NA	Cone rod dystrophy	TP	None	NA	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	18	6	M	NA	Cone dystrophy	TP	*CNGA3*	indel FS	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	19	26	M	NA	Cone dystrophy	TP	*ABCA4*	duplication	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	19	26	M	NA	Cone dystrophy	TP	*ABCA4*	missense	R

IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	20	5	F	NA	Cone rod dystrophy	TP	*CNGA3*	deletion FS	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	20	5	F	NA	Cone rod dystrophy	TP	*CNGA3*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	20	5	F	NA	Cone rod dystrophy	TP	*CACNA2D4*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	20	5	F	NA	Cone rod dystrophy	TP	*CACNA2D4*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	21	9	F	NA	Cone rod dystrophy	TP	*CNGA3*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	22	24	M	NA	cone rod degeneration	TP	*PROM1*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	22	24	M	NA	cone rod degeneration	TP	*SNRNP200*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	23	11	M	NA	Cone rod dystrophy	WES	*ABCA4*	missense	R
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	24	32	M	NA	Cone dystrophy	WES	None	NA	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	25	7	M	NA	Incomplete achromatopsia	WES	None	NA	NA
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	26	15	F	C	CRD	Homozygosity mapping	ABCA4	nonsense	Novel
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	26	15	F	C	CRD	Homozygosity mapping	ZNF513	NA	NA
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	26	15	F	C	CRD	Homozygosity mapping	ROM1	NA	NA
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	26	15	F	C	CRD	Homozygosity mapping	OPA1	NA	NA
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	26	15	F	C	CRD	Homozygosity mapping	HMCN1	NA	NA
IRD 8	Choroidal structural analysis and vascularity index in retinal dystrophies	30178525	2019	Aravind Eye Hospital, Madurai, India.	27-29	NA	NA	NA	CRD	NA	NA	NA	NA

LCA 17	Electroretinographic assessment and diagnostic reappraisal of children with visual dysfunction: a prospective study.	17322600	2007	Department of Retina-Vitreous Services, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamil Nadu, India. drvasumathy@yahoo.com	30-76	NA	9 M/16 F ACHM only		ACHM	NA	NA	NA	NA
CDD 1	Whole exome sequencing unveils a frameshift mutation in CNGB3 for cone dystrophy: A case report of an Indian family	28746191	2017	Department of Bioinformatics and Department of Molecular and Human Genetics, Institute of Science,Banaras Hindu University, Varanasi, India Premas Life Sciences Private Limited, Delhi R. K. Netralaya Eye Hospital and Research Centre, Varanasi, Uttar Pradesh, India.	77	9	F	Y	cone dystrophy	WES for siblings Sanger for seggregation	CNGB3	deletion FS	R
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	78	23	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	79	30	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	80	20	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	81	14	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	82	38	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	83	36	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	84	25	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	85	32	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	86	20	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	87	31	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	88	30	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	89	40	M	NA	colour vision defeciency	NA	NA	NA	NA
CDD 2	The impact of display saturation on visual search performance in congenital colour vision deficiency.	37682873	2023	Brien Holden Institute of Optometry and Vision Sciences, L V Prasad Eye Institute, Hyderabad, India.	90	15	F	NA	colour vision defeciency	NA	NA	NA	NA
CDD 3	Unilateral cone dysfunction with asymmetric maculopathy - Clinical features, multimodal imaging and genetic analysis of a novel phenotype.	33120657	2020	Narayana Nethralaya Eye Institute. Bangalore, Karnataka, India	91	14	M	No	Unilateral cone dysfunction with asymmetric maculopathy	WES	USH2A	missense	Reported
CDD 3	Unilateral cone dysfunction with asymmetric maculopathy - Clinical features, multimodal imaging and genetic analysis of a novel phenotype.	33120657	2020	Narayana Nethralaya Eye Institute. Bangalore, Karnataka, India	91	14	M	No	Unilateral cone dysfunction with asymmetric maculopathy	WES	PDE6B	missense	Reported
CDD 3	Unilateral cone dysfunction with asymmetric maculopathy - Clinical features, multimodal imaging and genetic analysis of a novel phenotype.	33120657	2020	Narayana Nethralaya Eye Institute. Bangalore, Karnataka, India	91	14	M	No	Unilateral cone dysfunction with asymmetric maculopathy	WES	PRPF4	missense	Reported
CDD 3	Unilateral cone dysfunction with asymmetric maculopathy - Clinical features, multimodal imaging and genetic analysis of a novel phenotype.	33120657	2020	Narayana Nethralaya Eye Institute. Bangalore, Karnataka, India	91	14	M	No	Unilateral cone dysfunction with asymmetric maculopathy	WES	PDE6G	missense	Reported
CDD 3	Unilateral cone dysfunction with asymmetric maculopathy - Clinical features, multimodal imaging and genetic analysis of a novel phenotype.	33120657	2020	Narayana Nethralaya Eye Institute. Bangalore, Karnataka, India	91	14	M	No	Unilateral cone dysfunction with asymmetric maculopathy	WES	CRX	deletion	Reported
CDD 4	Prevalence and gene frequency of color vision impairments among children of six populations from North Indian region.	30258865	2015	Human Genetics and Toxicology Laboratory, Section of Genetics, Department of Zoology, Faculty of Life Sciences, Aligarh Muslim University, Aligarh, 202002, Uttar Pradesh, India.	92-171	NA	72 M; 8 F	Not to consider for analysis: 815 NC	CVD	Hardy Weinberg equilibrium test	NA	NA	NA
CDD 5	Prevalence of Red-Green Color Vision Defects among Muslim Males and Females of Manipur, India.	23515069	2013	Human Genetics and Toxicology Laboratory, Section of Genetics, Department of Zoology, Aligarh Muslim University, Aligarh, Uttar Pradesh, India.	169-308	NA	118/1352 MALES 22/1280 FEMALES	NA	NA	NA	NA	NA	NA
CDD 6	Study of colour blindness in Jat Sikhs	7649599	1995	Department of Physiology, Govt. Medical College, Patiala.	309-359	10-60 y	50 M/ 1 F CB	NA	NA	NA	NA	NA	NA
CDD 7	Colour blindness--a rural prevalence survey. Indian J Ophthalmol.	3502467	1987	Laxmi Smruti, 773/1-A, Erandawana, Near Kamla Nehru Park, Pune-41 004, India	360-955	NA	596 males / 0	NA	NA	NA	NA	NA	NA
CDD 8	Bilateral focal choroidal excavation in cone dystrophy	27012694	2016	Sankara Nethralaya, Kolkata, West Bengal, India.	956	28	M	NA	bilateral cone dystrophy with focal choroidal excavation		NA	NA	NA
CDD 9	Keratoconus associated with cone-rod dystrophy	11917188	2002	Sankara Nethralaya Medical and Vision Research Foundations, Chennai, Tamil Nadu, India. mrf@sankaranethralaya.org	957	31	M	NA	bilateral keratoconus associated with apical corneal scarring and Cone-rod dystrophy		NA	NA	NA

XLRS 1	Identification and molecular characterization of two recurrent missense mutations in the RS1 gene in two families with X-linked retinoschisis from North India	37317958	2023	Department of Molecular and Human Genetics, Institute of Science, Banaras Hindu University, Varanasi,	1	NA	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 1	Identification and molecular characterization of two recurrent missense mutations in the RS1 gene in two families with X-linked retinoschisis from North India	37317958	2023	Department of Molecular and Human Genetics, Institute of Science, Banaras Hindu University, Varanasi,	2	NA	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	3	4	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Novel
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	4	6	MALE	NA	X-linked retinoschisis	SANGER	RS1	DUPLICATION	Novel
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	5	9	MALE	NA	X-linked retinoschisis	SANGER	RS1	DUPLICATION	Novel
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	6	7	MALE	NA	X-linked retinoschisis	SANGER	RS1	DUPLICATION	Novel
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	7	9	MALE	NA	X-linked retinoschisis	SANGER	RS1	FRMAESHIFT DELETION	Novel
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	8	21	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Novel
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	9	40	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	10	25	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	11	45	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	12	41	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	13	14	MALE	NA	X-linked retinoschisis	SANGER	RS1	DUPLICATION	Novel
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	14	9	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	15	7	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	16	27	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	17	25	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	18	11	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	19	12	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	20	19	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	21	26	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	22	8	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Novel
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	23	12	MALE	NA	X-linked retinoschisis	SANGER	RS1	STOP	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	24	2	MALE	NA	X-linked retinoschisis	SANGER	RS1	SPLICING	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	25	11	MALE	NA	X-linked retinoschisis	SANGER	RS1	SPLICING	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	26	39	MALE	NA	X-linked retinoschisis	SANGER	RS1	SPLICING	Reported
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	27	20	MALE	NA	X-linked retinoschisis	SANGER	RS1	ND	NA
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	28	37	MALE	NA	X-linked retinoschisis	SANGER	RS1	ND	NA
XLRS 2	Understanding variable disease severity in X-linked retinoschisis: Does RS1 secretory mechanism determine disease severity?	29851975	2018	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation,	29	18	MALE	NA	X-linked retinoschisis	SANGER	RS1	ND	NA
XLRS 3	Genetic variations in the hotspot region of RS1 gene in Indian patients with juvenile X-linked retinoschisis	17515881	2007	Department of Genetics, Aravind Medical Research Foundation, Madurai, India.	30	10	MALE	NA	X-linked retinoschisis	PCR-SSCP and sanger	RS1	MISSENSE	Reported

XLRS 3	Genetic variations in the hotspot region of RS1 gene in Indian patients with juvenile X-linked retinoschisis	17515881	2007	Department of Genetics, Aravind Medical Research Foundation, Madurai, India.	31	11	MALE	NA	X-linked retinoschisis	PCR-SSCP and sanger	RS1	MISSENSE	Novel
XLRS 3	Genetic variations in the hotspot region of RS1 gene in Indian patients with juvenile X-linked retinoschisis	17515881	2007	Department of Genetics, Aravind Medical Research Foundation, Madurai, India.	32	20	MALE	NA	X-linked retinoschisis	PCR-SSCP and sanger	RS1	MISSENSE	Reported
XLRS 3	Genetic variations in the hotspot region of RS1 gene in Indian patients with juvenile X-linked retinoschisis	17515881	2007	Department of Genetics, Aravind Medical Research Foundation, Madurai, India.	33	10	MALE	NA	X-linked retinoschisis	PCR-SSCP and sanger	RS1	STOP	Novel
XLRS 3	Genetic variations in the hotspot region of RS1 gene in Indian patients with juvenile X-linked retinoschisis	17515881	2007	Department of Genetics, Aravind Medical Research Foundation, Madurai, India.	34	17	MALE	NA	X-linked retinoschisis	PCR-SSCP and sanger	RS1	MISSENSE	Reported
XLRS 3	Genetic variations in the hotspot region of RS1 gene in Indian patients with juvenile X-linked retinoschisis	17515881	2007	Department of Genetics, Aravind Medical Research Foundation, Madurai, India.	35	30	MALE	NA	X-linked retinoschisis	PCR-SSCP and sanger	RS1	MISSENSE	Reported
XLRS 4	Proteomic profiling of human intraschisis cavity fluid	28450823	2017	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India	36	4	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Novel
XLRS 4	Proteomic profiling of human intraschisis cavity fluid	28450823	2017	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India	37	40	MALE	NA	X-linked retinoschisis	SANGER	RS1	MISSENSE	Reported
XLRS 5	Vasoproliferative Tumor in X-Linked Retinoschisis	37204368	2023	Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Ophthalmic Sciences, New Delhi, India	38	26	MALE	NA	X-linked retinoschisis	NA	RS1	MISSENSE	Reported
XLRS 6	Genetic studies in a patient with X-linked retinoschisis coexisting with developmental delay and sensorineural hearing loss	28574807	2022	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation , Chennai	39	NA	MALE	NA	X-linked retinoschisis	Targeted gene sequencing,SNP 6.0 array-based whole genome	RS1	SPLICING	Reported
XLRS 7	Molecular characterization of a rare phenotype of X-linked retinoschisis with angle-closure glaucoma	31238476	2019	Dr Rajendra Prasad Centre for Ophthalmic Sciences, Laboratory of Cuto-Molecular Genetics, All India Institute of Medical Sciences, New Delhi, India	40	11	MALE	NA	XLRS) with angle-closure glaucoma	SANGER	RS1	DELETION	Novel
XLRS 8	X-linked retinoschisis	31856528	2020	Department of Vitreo-Retina, All India Institute of Medical Sciences, New Delhi, India	41	36	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 9	Bullous retinoschisis	31546543	2019	Advanced Eye Centre, Department of Ophthalmology, Post Graduate Institute of Medical Education and Research, Chandigarh, Punjab, India	42	59	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 10	Dramatic response to topical dorzolamide in X-linked retinoschisis	32587200	2020	Vitreo-Retina, Trauma and Uvea Services, Dr. Rajendra Prasad Center for Ophthalmic Sciences, All India Institute of Medical Sciences, Ansari Nagar, New Delhi, India	43	8	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	44	11	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	45	6	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	46	6	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	47	15	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	48	10	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	49	11	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	50	14	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	51	5	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	52	7	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	53	11	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	54	8	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	55	9	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	56	9	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	57	9	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	58	6	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	59	16	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	60	12	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA

XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	61	3	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	62	6	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	63	11	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	64	7	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	65	4	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	66	13	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	67	13	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	68	4	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	69	9	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	70	10	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	71	12	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	72	6	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	73	6	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	74	5	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	75	14	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	76	14	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 11	Outcome of vitreoretinal surgery for rhegmatogenous retinal detachment in X-linked juvenile retinoschisis	30451188	2018	Medical Research Foundation, Sankara Nethralaya, Chennai, Tamil Nadu, India	77	10	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 12	Traumatic Macular Retinoschisis	30090201	2018	ICARE Eye Hospital and Postgraduate Institute, Noida, Uttar Pradesh, India	78	30	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 13	Lamellar macular hole in X linked retinoschisis	27170611	2016	Dr Rajendra Prasad Centre for Ophthalmic Sciences, AIIMS, Delhi, India	79	10	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 13	Lamellar macular hole in X linked retinoschisis	27170611	2016	Dr Rajendra Prasad Centre for Ophthalmic Sciences, AIIMS, Delhi, India	80	13	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 14	A paradigm shift in the treatment of refractory angle closure glaucoma in a patient with X-linked juvenile retinoschisis	36927170	2023	VST Center for Glaucoma Care, L V Prasad Eye Institute, Hyderabad, India	81	NA	MALE	NA	XLRS with glaucoma.	NA	NA	NA	NA
XLRS 15	Multimodal imaging of bilateral macular hole in X-linked retinoschisis	32907874	2020	Smt. Kanuri Santhamma Centre for Vitreo-Retinal Diseases, L V Prasad Eye Institute, Hyderabad, Telengana, India.	82	25	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 16	Bilateral macular holes in X-linked retinoschisis: now the spectrum is wider	22011501	2011	Retina Institute of Karnataka, 122, 5th Main Road, Chamarajpet, Bangalore-560 018, India	83	25	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 17	Nasal involvement in X-linked retinoschisis	28820162	2017	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All Institute of Medical Sciences, New Delh	84	11	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 18	Congenital x-linked retinoschisis: a novel approach for management of a large schitic cavity overhanging the macula	25390856	2009	Shri Bhagwan Mahavir Vitreoretinal Services, Medical Research Foundation, Sankara Nethralaya, Chennai, India.	85	8	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 19	Comments on temporary resolution of foveal schisis following vitrectomy with silicon oil tamponade in X-linked retinoschisis with retinal detachment	27146945	2016	ICARE Eye Hospital and Post Graduate Institute, Noida, Uttar Pradesh, India	86	22	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 20	Evaluation of focal retinal function using multifocal electroretinography in patients with X-linked retinoschisis	20648073	2010	Shri Bhagwan Mahavir Vitreo-Retinal Services, Medical Research Foundation, Sankara Nethralaya, Chennai,	87	NA	NA	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 21	Surgical management of a large retinal cyst in X-linked retinoschisis with internal drainage: Report of an unusual case	32971698	2020	Shri Bhagwan Mahavir Vitreo-Retinal Services, Medical Research Foundation, Sankara Nethralaya, Chennai,	88	9	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 22	Multimodal imaging in retinoschisis	30787539	2019	Department of Vitreoretina, Narayana Nethralaya Eye Hospital, Bengaluru, Karnataka, India	89	22	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 23	Three-dimensional spectral domain optical coherence tomography in X linked foveal retinoschisis	23563673	2013	Department of Ophthalmology, KG's Medical University, Lucknow, Uttar Pradesh, India	90	24	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA

XLRS 23	Three-dimensional spectral domain optical coherence tomography in X linked foveal retinoschisis	23563673	2013	Department of Ophthalmology, KG's Medical University, Lucknow, Uttar Pradesh, India	91	14	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 24	Unilateral giant peripapillary drusen and retinal drusenoid deposits in a case of X-linked retinoschisis	26907824	2016	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India	92	25	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 25	Unusual manifestations of x-linked retinoschisis: clinical profile and diagnostic evaluation	17631851	2007	Aravind Eye Hospital & Postgraduate Institute of Ophthalmology, Madurai, Tamil Nadu,	93	NA	NA	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 26	Juxtapapillary retinochoroidal coloboma presenting with macular retinoschisis	35791246	2022	Department of Vitreo-Retina, B. W. Lions Eye Hospital, Bengaluru, Karnataka, India	94	50	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 27	Temporary resolution of foveal schisis following vitrectomy with silicon oil tamponade in X-linked retinoschisis with retinal detachment	26669343	2015	ICARE Eye Hospital and Post Graduate Institute, Noida, Uttar Pradesh, India	95	22	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 28	Foveal schisis as a cause of retinal detachment secondary to macular hole in juvenile X-linked retinoschisis	15805920	2005	Medical and Vision Research Foundation, Sankara Nethralaya, Tamil Nadu, India.	96	32	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 29	Peripapillary and macular retinoschisis - A vision-threatening sequelae of advanced glaucomatous cupping	34826994	2021	Department of Glaucoma, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamilnadu, India	97	29	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 29	Peripapillary and macular retinoschisis - A vision-threatening sequelae of advanced glaucomatous cupping	34826994	2021	Department of Glaucoma, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamilnadu, India	98	75	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 29	Peripapillary and macular retinoschisis - A vision-threatening sequelae of advanced glaucomatous cupping	34826994	2021	Department of Glaucoma, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamilnadu, India	99	32	FEMALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 29	Peripapillary and macular retinoschisis - A vision-threatening sequelae of advanced glaucomatous cupping	34826994	2021	Department of Glaucoma, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamilnadu, India	100	72	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 29	Peripapillary and macular retinoschisis - A vision-threatening sequelae of advanced glaucomatous cupping	34826994	2021	Department of Glaucoma, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamilnadu, India	101	34	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 30	Laser Barricade in Progressive Vision-Threatening Peripheral Retinoschisis	32273109	2020	Department of Vitreo-Retina Services, Sri Sankaradeva Nethralaya, Guwahati, Assam, India	102	3	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 31	Nasal retinoschisis in a case of high myopia	31238459	2019	Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India	103	32	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 32	Acquired tractional retinoschisis with giant outer - layer break underneath macula	32971663	2020	Srimati Kannuri Santhamma Center for Vitreo-retinal Diseases, LV Prasad Eye Institute, Hyderabad, Telangana, India	104	20	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 33	Myopic retinoschisis with intraretinal emulsified silicone oil appearing as a macular hyperoleon	32709804	2020	Shri Bhagwan Mahavir Department of Vitreo Retinal Services, Medical Research Foundation, Sankara Nethralaya, Chennai,	105	51	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 34	En-face optical coherence tomography of unilateral myopic retinoschisis	31436204	2019	Department of Ophthalmologist and Vitreoretina, Centre for Sight Eye Institute, New Delhi, India	106	56	FEMALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 35	Multimodal imaging in a case of stellate nonhereditary idiopathic foveomacular retinoschisis	35791213	2022	Vitreo Retina Services, Aravind Eye Hospital, Thavalakuppam, Pondicherry, India	107	64	FEMALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 36	Optic disc pit with multiquadrant peripapillary retinoschisis and choroidal coloboma	34266823	2021	Ophthalmology, All India Institute of Medical Sciences, New Delhi, India	108	18	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 37	Congenital retinoschisis: successful collapse with photocoagulation	12930120	2001	Vision and Medical Research Foundation, Chennai, India.	109	21	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
XLRS 38	Microscope-integrated optical coherence tomography-aided intraoperative diagnosis and management of peripheral tractional retinoschisis	30127155	2018	Department of Ophthalmology, Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India	110	48	MALE	NA	X-linked retinoschisis	NA	NA	NA	NA
Syndromic IRDs													
JS													
JS 1	Sharawat IK, Naresh Singh RK, Panda PK. Distinctive Imaging in a Toddler with Joubert's Syndrome. Ann Indian Acad Neurol. 2021 Mar-Apr;24(2):253-254	34220074	2020	Department of Pediatrics, Pediatric Neurology Division, All India Institute of Medical Sciences, Rishikesh, Uttarakhand, India	1	2 year	Male	NC	Joubert Syndrome	Whole-exome sequencing	AHI1	Missense	Reported
JS 2	Ahmetgjekaj I, Rahman M, Hyseni F, Guy A, Madani K, Saliaj K, Guy A, Vokshi V, Kola I, Musa J. A case report of Joubert syndrome with renal involvement and seizures in a neonate. Radiol Case Rep. 2021 Feb 24;16(5):1075-1079. doi: 10.1016/j.radcr.2021.02.031. Erratum in: Radiol Case Rep. 2022 Sep 29;17(12):4933. PMID: 33717386; PMCID: PMC7921194.	33717386	2022	Deccan College Of Medical Sciences (DCMS), Telangana, India	2	39 weeks	Female	NC	Joubert Syndrome	Genome Sequencing	CEP290	NA	NA
JS 3	Sivathanu D, Vetrichelvan D, Balakrishnan U, Manokaran RK. An Atypical Presentation of Joubert Syndrome Due to a Novel Mutation in ZNF423 Gene. J Pediatr Neurosci. 2020 Jul-Sep;15(3):294-296. doi: 10.4103/jpn.JPN_168_19. Epub 2020 Nov 6.	33531950	2020	1 Department of Paediatrics Sri Ramachandra Institute of Higher Education and Research, Chennai, Tamil Nadu, India. 2 Department of Neonatology Sri Ramachandra Institute of Higher Education and Research, Chennai, Tamil Nadu, India. 3 Division of Paediatric Neurology, Department of Neurology, Sri Ramachandra Institute of Higher Education and Research, Chennai, Tamil Nadu, India.	3	8 months	Male	NA	Joubert Syndrome, West Syndrome	Clinical Exome Sequencing	ZNF423	Missense	Novel
JS 4	Sreedevi N, Swapna N, Maruthy S, Jayakumar T, Sylvester C. Molecular Evaluation of Joubert Syndrome and Hearing Impairment in a Patient with Ataxic Cerebral Palsy. Glob Med Genet. 2023 Jul 17;10(3):190-193. doi: 10.1055/s-0043-1771184. PMID: 37501760; PMCID: PMC10370468.	37501760	2023	1 Department of Speech-Language Sciences, All India Institute of Speech and Hearing, Mysore, India. 2 Department of Speech-Language Pathology, All India Institute of Speech and Hearing, Mysore, India. 3Unit for Human Genetics, All India Institute of Speech and Hearing, Mysore, India.	4	3.5 Year	Male	C	Joubert Syndrome, Ataxic Cerebral Palsy (CP)	Whole-exome sequencing (WES)	GJB2	Missense	NA
JS 4	Sreedevi N, Swapna N, Maruthy S, Jayakumar T, Sylvester C. Molecular Evaluation of Joubert Syndrome and Hearing Impairment in a Patient with Ataxic Cerebral Palsy. Glob Med Genet. 2023 Jul 17;10(3):190-193. doi: 10.1055/s-0043-1771184. PMID: 37501760; PMCID: PMC10370468.	37501760	2023	1 Department of Speech-Language Sciences, All India Institute of Speech and Hearing, Mysore, India. 2 Department of Speech-Language Pathology, All India Institute of Speech and Hearing, Mysore, India. 3Unit for Human Genetics, All India Institute of Speech and Hearing, Mysore, India.	4	3.5 Year	Male	C	Joubert Syndrome, Ataxic Cerebral Palsy (CP)	Whole-exome sequencing (WES)	AHI1	Missense	NA
JS 5	Shetty M, Ramdas N, Sahni S, Mullapudi N, Hegde S. A Homozygous Missense Variant in INPP5E Associated with Joubert Syndrome and Related Disorders. Mol Syndromol. 2017 Nov;8(6):313-317. doi: 10.1159/000479673. Epub 2017 Sep 8. PMID: 29230161; PMCID: PMC5701266.	29230161	2017	1 Department of Medical Genetics, Manipal Hospital, Bangalore, India. 2 Dhiti Omics Technologies Pvt. Ltd., Bangalore, India.	5	6 Year	Female	C	JSRD	NGS-based clinical exome sequencing	INPP5E	Missense	Novel

JS 6	Patra S, Goyal G, Lone YA, Gupta G. Novel variant CPLANE 1: c.5051C>A (p.Ser1684Ter) in an Indian neonate with Joubert syndrome. BMJ Case Rep. 2023 May 31;16(5):e255561. doi: 10.1136/bcr-2023-255561. PMID: 37258045; PMCID: PMC10255207.	37258045	2023	1 Neonatology, Himalayan Institute of Medical Sciences, Dehradun, Uttarakhand, India. 2 Paediatric Surgery, Himalayan Institute of Medical Sciences, Dehradun, Uttarakhand, India. 3 Neonatology, Himalayan Institute of Medical Sciences, Dehradun, Uttarakhand, India	6	Neonate	NA	NA	JS	Clinical Exome Sequencing	CPLANE 1/C5ORF42	Missense	Novel
JS7	Hebbar M, Kanthi A, Shukla A, Bielas S, Girisha KM. A biallelic 36-bp insertion in PIBF1 is associated with Joubert syndrome. J Hum Genet. 2018 Jul;63(8):935-939. doi: 10.1038/s10038-018-0462-7. Epub 2018 Apr 25. PMID: 29695797; PMCID: PMC6060014.	29695797	2018	1 Department of Medical Genetics, Kasturba Medical College, Manipal, Manipal Academy of Higher Education, Manipal, India. 2 Department of Human Genetics, University of Michigan, Ann Arbor, MI, USA. 3 Department of Medical Genetics, Kasturba Medical College, Manipal, Manipal Academy of Higher Education, Manipal, India.	7	2 Year	Female	C	Joubert Syndrome	Exome Sequencing	PIBF1	Insertion	Novel
JS8	Kar A, Phadke SR, Das Bhowmik A, Dalal A. Whole exome sequencing reveals a mutation in ARMC9 as a cause of mental retardation, ptosis, and polydactyly. Am J Med Genet A. 2018 Jan;176(1):34-40. doi: 10.1002/ajmg.a.38537. Epub 2017 Nov 21. PMID: 29159890.	29159890	2018	1 Diagnostics Division, Centre for DNA Fingerprinting and Diagnostics, Hyderabad, India. 2 Graduate Studies, Manipal University, Manipal, Hyderabad, India. 3 Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, India.	8	NA	NA	NA	Joubert Syndrome	Homozygosity Mapping and Whole Exome Sequencing	ARMC9	splice-site	Novel
JS 9	Sanjeev RK, Kapoor S, Goyal M, Kapur R, Gleeson JG. Molar Tooth Sign with Deranged Liver Function Tests: An Indian Case with COACH Syndrome. Case Rep Pediatr. 2015;2015:385910. doi: 10.1155/2015/385910. Epub 2015 May 17. PMID: 26075130; PMCID: PMC4449927.	26075130	2015	1 Department of Pediatrics, ACMS, India. 2 Division of Genetics, Lok Nayak & Maulana Azad Medical College, New Delhi, India. 3 Department of Paediatrics, Maulana Azad Medical College, New Delhi, India. 4 Department of Radiology, ACMS, New Delhi, India. 5 Neurogenetics Laboratory, Department of Neurosciences and Paediatrics, USA ; Rady Children's Hospital, USA ; Howard Hughes Medical Institute, CA, USA.	9	6 Year	Female	NC	Joubert Syndrome and COACH syndrome	Genetic Analysis	TMEM67 (MKS3)	Missense	NA
JS 9	Srivastava S, Manisha R, Dwivedi A, Agarwal H, Saxena D, Agrawal V, Mandal K. Meckel Gruber and Joubert Syndrome Diagnosed Prenatally: Allelism between the Two Ciliopathies, Complexities of Mutation Types and Digenic Inheritance. Fetal Pediatr Pathol. 2022 Dec;41(6):1041-1051. doi: 10.1080/15513815.2021.2007434. Epub 2021 Nov 25. PMID: 34821546.	34821546	2021	Sanjay Gandhi post Graduate Institute of Medical Sciences, Lucknow	10	Antenatatal	NA	C	Joubert Syndrome, Meckel Syndrome antenatally diagnosed occipital encephalocele and enlarged kidneys	Exome Sequencing	TMEM138	NA	NA
JS 9	Srivastava S, Manisha R, Dwivedi A, Agarwal H, Saxena D, Agrawal V, Mandal K. Meckel Gruber and Joubert Syndrome Diagnosed Prenatally: Allelism between the Two Ciliopathies, Complexities of Mutation Types and Digenic Inheritance. Fetal Pediatr Pathol. 2022 Dec;41(6):1041-1051. doi: 10.1080/15513815.2021.2007434. Epub 2021 Nov 25. PMID: 34821546.	34821546	2021	Sanjay Gandhi post Graduate Institute of Medical Sciences, Lucknow	10	Antenatatal	NA	C	Joubert Syndrome, Meckel Syndrome antenatally diagnosed occipital encephalocele and enlarged kidneys	Exome Sequencing	SDCCAG8	NA	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21066	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	11	2.5	Female	C	Joubert Syndrome	Clinical Exome Sequencing	NA	NA	5 variants from different patients were novel
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21067	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	12	13	Female	NC	Joubert Syndrome	Clinical Exome Sequencing	NA	NA	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21068	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	13	24	Female	NC	Joubert Syndrome	Clinical Exome Sequencing	MKS1	Deletion	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21069	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	14	5	Female	NC	Joubert Syndrome	Clinical Exome Sequencing	NA	NA	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21070	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	15	12.5	Female	NC	Joubert Syndrome	Clinical Exome Sequencing	PIBF1	Frameshift	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21070	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	15	12.5	Female	NC	Joubert Syndrome	Clinical Exome Sequencing	PIBF1	Missense	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21071	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	16	0.8	Male	C	Joubert Syndrome	Clinical Exome Sequencing	NA	NA	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21071	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	16	0.8	Male	C	Joubert Syndrome	Clinical Exome Sequencing	CPLANE1/C5ORF42	Missense	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21072	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	17	15	Male	NC	Joubert Syndrome	Clinical Exome Sequencing	CPLANE1/C5ORF42	Frameshift	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21073	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	18	13	Male	NC	Joubert Syndrome	Clinical Exome Sequencing	MKS1	Deletion	NA
JS 10	Surisetti BK, Holla VV, Prasad S, Neeraja K, Kamble N, Yadav R, Pal PK. Clinical and Imaging Profile of Patients with Joubert Syndrome. J Mov Disord. 2021 Sep;14(3):231-235. doi: 10.14802/jmd.21074	34592808	2021	National Institute of Mental Health and Neurosciences (NIMHANS), Karnataka, India	19	7	Male	NC	Joubert Syndrome	Clinical Exome Sequencing	NA	NA	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	1 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:20	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CCDC28B	Missense	NA

JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	2 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:20	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CC2D2A	1 Missense	novel
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	2 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:20	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CC2D2A	NA	novel
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	2 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:20	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CC2D2A	NA	novel
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	2 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:20	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CC2D2A	NA	novel
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	3 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:20	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Missense	(2 novel CEP290) patient details not available
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	4 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:21	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Frameshift	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	5 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:22	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Frameshift	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	6 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:23	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	splice-site	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	7 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:24	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	8 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:25	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	9 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:26	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	10 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:27	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Frameshift	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	10 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:27	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Deletion	Novel
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	11 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:28	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	12 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:29	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	13 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:30	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	14 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:31	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Frameshift	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	15 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:32	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Frameshift	NA

JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	16 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:33	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CEP290	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	17 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:34	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	TMEM237	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	18 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:35	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	TCTN3	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	19 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:36	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	TCTN3	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	20 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:37	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	TCTN2	Insertion	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	21 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:38	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	TCTN1	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	22 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:39	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	RPGRIP1L	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	23 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:40	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	RPGRIP1L	Insertion	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	24 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:41	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	RPGRIP1L	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	25 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:42	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	RPGRIP1L	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	26 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:43	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	RPGRIP1L	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	27 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:44	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	NPHP1	Deletion	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	28 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:45	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	INPP5E	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	29 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:46	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	INPP5E	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	30 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:47	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CPLANE1/C5ORF42	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	31 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:48	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CPLANE1/C5ORF42	Missense	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	32 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:49	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	CPLANE1 /C5ORF42	NA	NA
JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	33 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:50	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	AHI1	Missense	NA

JS 11	Radha Rama Devi A, Naushad SM, Lingappa L. Clinical and Molecular Diagnosis of Joubert Syndrome and Related Disorders. Pediatr Neurol. 2020 May;106:43-49. doi: 10.1016/j.pediatrneurol.2020.01.012. Epub 2020 Feb 4. PMID: 32139166.	32139166	2020	34 Department of Genetic Metabolic Unit, Rainbow Children Hospital, Hyderabad, India; Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 2 Department of Biochemical Genetics, Sandor Speciality Diagnostics Pvt Ltd, Hyderabad, India. 3 Department of Pediatric Neurology, Rainbow Children Hospital, Hyderabad, India.	20-78/ 35 families with genetic report	Average Age 2.6 ± 2.3 years	Male: Female Ratio= 36:51	31/55 Subjects were consanguinous	Joubert Syndrome	Exome Sequencing	AHI1	Duplication	NA
JS 12	Karegowda LH, Shenoy PM, Sripathi S, Varman M. Joubert syndrome. BMJ Case Rep. 2014 Mar 20;2014:bcr2014203887. doi: 10.1136/bcr-2014-203887. PMID: 24654256; PMCID: PMC3962959.	24654256	2014	Department of Radiodiagnosis, Kasturba Medical College, Manipal, Manipal, Karnataka, India.	79	10 Year	Male	C	Joubert Syndrome	NA	NA	NA	NA
JS 13	Sampathkumar K, Sooraj YS, Karunakaran N, Ganesh R, Mahaldar AR. Joubert syndrome. Kidney Int. 2008 Nov;74(9):1222. doi: 10.1038/ki.2008.339. PMID: 18854854.	18854854	2008	Department of Nephrology, Meenakshi Mission Hospital and Research Centre, Lake Area, Madurai, Tamil Nadu, India.	80	8 Year	Female	NC	Joubert Syndrome	NA	NA	NA	NA
JS 14	Kumar P, Dey A, Mittal K, Sharma R, Goyal A, Hira P. Joubert syndrome: A classic case. J Family Med Prim Care. 2019 Jan;8(1):311-312. doi: 10.4103/jfmpc.jfmpc_165_18. PMID: 30911530; PMCID: PMC6396587.	30911530	2019	Department of Radiology, Seth GSMC and KEM Hospital, Mumbai, Maharashtra, India.	81	1 Year	Female	NC	Joubert Syndrome	NA	NA	NA	NA
JS 15	Kumar S, Singh D. Joubert Syndrome Mimicking Hypotonic Cerebral Palsy. Indian J Pediatr. 2016 Nov;83(12-13):1505. doi: 10.1007/s12098-016-2196-x. Epub 2016 Jul 9. PMID: 27392618.	27392618	2016	1 Department of Pediatrics, MGM Medical College and LSK Hospital, Kishanganj, Bihar, India. 2 Department of Pediatrics, MGM Medical College and LSK Hospital, Kishanganj, Bihar, India.	82	8 Month	Female	NC	Joubert Syndrome	NA	NA	NA	NA
JS 16	Mugundhan K, Mayan MCV, Nidhin PD, Loganathan G, Balamurugan N. Joubert Syndrome Associated with Seizures. J Assoc Physicians India. 2017 Aug;65(8):96. PMID: 28799313.	28799313	2017	1 Dept. of Neurology, Govt. Mohan Kumaramangalam Medical College and Hospital, Salem, Tamil Nadu 2 G.L. Hospital, Salem, Tamil Nadu 3 SIMS Chellum Hospital, Salem, Tamil Nadu	83	NA	NA	NA	Joubert Syndrome	NA	NA	NA	NA
JS 17	Bhardwaj P, Sharma M, Ahluwalia K. Joubert Syndrome with Orofacial Digital Features. J Neurosci Rural Pract. 2018 Jan-Mar;9(1):152-154. doi: 10.4103/jnrp.jnrp_338_17. PMID: 29456362; PMCID: PMC5812143.	29456362	2018	Department of Pediatrics and Physiology, Indira Gandhi Medical College, Shimla, Himachal Pradesh, India	84	3 Year	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 18	Solomon R, Jana AK, Singh S, Biswas A. Joubert syndrome. Indian Pediatr. 2001 Sep;38(9):1045-9. PMID: 11568384.	11568384	2001	Department of Neonatology, Christian Medical College Hospital, Vellore 632 004, Tamil Nadu, India.	85	NA	NA	NA	Joubert Syndrome	NA	NA	NA	NA
JS 19	Gurjar V, Gurjar M, Pattanshetti C, Sankeshwari B. Lingual Frenectomy in Joubert Syndrome. J Contemp Dent Pract. 2017 Aug 1;18(8):728-731. doi: 10.5005/jp-journals-10024-2115. PMID: 28816198.	28816198	2017	1 Department of Oral and Maxillofacial Surgery, Bharati Vidyapeeth Deemed University Medical College and Hospital Sangli, Maharashtra, India 2 Department of Periodontics, Bharati Vidyapeeth Deemed University Medical College and Hospital, Sangli, Maharashtra India. 4 Department of Prosthodontics, Bharati Vidyapeeth Deemed University Medical College and Hospital, Sangli, Maharashtra India.	86	NA	NA	NA	Joubert Syndrome	NA	NA	NA	NA
JS 20	Raghavan DV, Doshi VV, Nambi S. Joubert syndrome with autism in two siblings: A rare presentation. Indian J Psychiatry. 2016 Jan-Mar;58(1):90-2. doi: 10.4103/0019-5545.174395. PMID: 26985112; PMCID: PMC4776590.	26985112	2016	1 Department of Psychiatry, Institute of Mental Health, Madras Medical College, Chennai, Tamil Nadu, India. 2 Department of Child Psychiatry, Institute of Child Health and Hospital for Children, Madras Medical College, Chennai, Tamil Nadu, India.	87	5 Year	Female	2* C	Joubert Syndrome	NA	NA	NA	NA
JS 20	Raghavan DV, Doshi VV, Nambi S. Joubert syndrome with autism in two siblings: A rare presentation. Indian J Psychiatry. 2016 Jan-Mar;58(1):90-2. doi: 10.4103/0019-5545.174395. PMID: 26985112; PMCID: PMC4776590.	26985112	2016	1 Department of Psychiatry, Institute of Mental Health, Madras Medical College, Chennai, Tamil Nadu, India. 2 Department of Child Psychiatry, Institute of Child Health and Hospital for Children, Madras Medical College, Chennai, Tamil Nadu, India.	88	2 Year 11 months	Male	2* C	Joubert Syndrome	NA	NA	NA	NA
JS 21	Kannauje PK, Pandit V, Wasnik P, Pati SK, Venkatesan N. Diagnosing Joubert Syndrome in Two Adult Siblings: A Very Rare Case Report. Cureus. 2022 Jul 19;14(7):e27042. doi: 10.7759/cureus.27042. PMID: 35989767; PMCID: PMC9388958.	35989767	2022	1 General Medicine, All India Institute of Medical Sciences, Raipur, Raipur, IND. 2 Radiodiagnosis, All India Institute of Medical Sciences, Raipur, Raipur, IND. 3 Internal Medicine, All India Institute of Medical Sciences, Raipur, Raipur, IND.	89	21 Years	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 21	Kannauje PK, Pandit V, Wasnik P, Pati SK, Venkatesan N. Diagnosing Joubert Syndrome in Two Adult Siblings: A Very Rare Case Report. Cureus. 2022 Jul 19;14(7):e27042. doi: 10.7759/cureus.27042. PMID: 35989767; PMCID: PMC9388958.	35989767	2022	1 General Medicine, All India Institute of Medical Sciences, Raipur, Raipur, IND. 2 Radiodiagnosis, All India Institute of Medical Sciences, Raipur, Raipur, IND. 3 Internal Medicine, All India Institute of Medical Sciences, Raipur, Raipur, IND.	90	24 Years	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 22	Koshy B, Oommen SP, Jasper S, Danda S, Surendrababu NR. Development and dysmorphism in Joubert syndrome--short case series from India. J Trop Pediatr. 2010 Jun;56(3):209-12. doi: 10.1093/tropej/fmp084. Epub 2009 Sep 15. PMID: 19755534.	19755534	2010	NA	91	Newborn	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 22	Koshy B, Oommen SP, Jasper S, Danda S, Surendrababu NR. Development and dysmorphism in Joubert syndrome--short case series from India. J Trop Pediatr. 2010 Jun;56(3):209-12. doi: 10.1093/tropej/fmp084. Epub 2009 Sep 15. PMID: 19755534.	19755534	2010	NA	92	3 Years	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 22	Koshy B, Oommen SP, Jasper S, Danda S, Surendrababu NR. Development and dysmorphism in Joubert syndrome--short case series from India. J Trop Pediatr. 2010 Jun;56(3):209-12. doi: 10.1093/tropej/fmp084. Epub 2009 Sep 15. PMID: 19755534.	19755534	2010	NA	93	9 Months	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 22	Koshy B, Oommen SP, Jasper S, Danda S, Surendrababu NR. Development and dysmorphism in Joubert syndrome--short case series from India. J Trop Pediatr. 2010 Jun;56(3):209-12. doi: 10.1093/tropej/fmp084. Epub 2009 Sep 15. PMID: 19755534.	19755534	2010	NA	94	Newborn	Female	NC	Joubert Syndrome	NA	NA	NA	NA
JS 22	Koshy B, Oommen SP, Jasper S, Danda S, Surendrababu NR. Development and dysmorphism in Joubert syndrome--short case series from India. J Trop Pediatr. 2010 Jun;56(3):209-12. doi: 10.1093/tropej/fmp084. Epub 2009 Sep 15. PMID: 19755534.	19755534	2010	NA	95	2.5 Years	Female	NC	Joubert Syndrome	NA	NA	NA	NA

JS 23	Holla VV, Stezin A, Chaithra SP, Kamble N, Yadav R, Pal PK. Disabling Myoclonus in a Case of Joubert Syndrome. Mov Disord Clin Pract. 2020 Apr 6;7(4):456-458. doi: 10.1002/mdc3.12933. PMID: 32373664; PMCID: PMC7197299.	32373664	2020	1 Department of Neurology National Institute of Mental Health & Neurosciences Bangalore Karnataka India. 2 Department of Clinical Neurosciences National Institute of Mental Health & Neurosciences Bangalore Karnataka India.	96	13 Year	Male	NA	Joubert Syndrome	Exome Sequencing	CPLANE1 (C5orf42)	Frameshift	NA
JS 23	Holla VV, Stezin A, Chaithra SP, Kamble N, Yadav R, Pal PK. Disabling Myoclonus in a Case of Joubert Syndrome. Mov Disord Clin Pract. 2020 Apr 6;7(4):456-458. doi: 10.1002/mdc3.12933. PMID: 32373664; PMCID: PMC7197299.	32373664	2020	1 Department of Neurology National Institute of Mental Health & Neurosciences Bangalore Karnataka India. 2 Department of Clinical Neurosciences National Institute of Mental Health & Neurosciences Bangalore Karnataka India.	96	13 Year	Male	NA	Joubert Syndrome	Exome Sequencing	CPLANE1 (C5orf42)	Missense	NA
JS 23	Holla VV, Stezin A, Chaithra SP, Kamble N, Yadav R, Pal PK. Disabling Myoclonus in a Case of Joubert Syndrome. Mov Disord Clin Pract. 2020 Apr 6;7(4):456-458. doi: 10.1002/mdc3.12933. PMID: 32373664; PMCID: PMC7197299.	32373664	2020	1 Department of Neurology National Institute of Mental Health & Neurosciences Bangalore Karnataka India. 2 Department of Clinical Neurosciences National Institute of Mental Health & Neurosciences Bangalore Karnataka India.	96	13 Year	Male	NA	Joubert Syndrome	Exome Sequencing	CPLANE1 (C5orf42)	Missense	NA
JS 24	Jain D, Ravishankar V. Ocular Manifestations Leading to a Diagnosis of Joubert Syndrome Related Disorder. Nepal J Ophthalmol. 2022 Jan;14(27):173-177. doi: 10.3126/nepjoph.v14i1.35163. PMID: 35996916.	35996916	2022	Post Graduate Institute of Child Health, Noida, Uttar Pradesh, India.	97	21 Years	Female	NA	Joubert Syndrome	NA	NA	NA	NA
JS 25	Shamsudheen MP, Das U, Taduri G, Guditi S, Karthik R, Thakur R. A Case of Joubert Syndrome with Chronic Kidney Disease. Indian J Nephrol. 2021 Jan-Feb;31(1):61-63. doi: 10.4103/ijn.IJN_287_19. Epub 2021 Jan 27. PMID: 33994691; PMCID: PMC8101675.	33994691	2021	1 Department of Nephrology, Nizam's Institute of Medical Sciences, Punjagutta, Hyderabad, Telangana, India. 2 Department of Radiodiagnosis, Nizam's Institute of Medical Sciences, Punjagutta, Hyderabad, Telangana, India.	98	9 years	Male	4* C	Joubert Syndrome	NA	NA	NA	NA
JS 26	Nag C, Ghosh M, Das K, Ghosh T. Joubert syndrome: the molar tooth sign of the mid-brain. Ann Med Health Sci Res. 2013 Apr;3(2):291-4. doi: 10.4103/2141-9248.113686. PMID: 23919210; PMCID: PMC3728883.	23919210	2013	Department of General Medicine, Burdwan Medical College, Burdwan, West Bengal, India	99	1 Year and 4 Months	Male	C	Joubert Syndrome	NA	NA	NA	NA
JS 27	Choh SA, Choh NA, Bhat SA, Jehangir M. MRI findings in Joubert syndrome. Indian J Pediatr. 2009 Feb;76(2):231-5. doi: 10.1007/s12098-008-0232-1. Epub 2009 Jan 5. PMID: 19129991.	19129991	2009	Department of Pediatrics & Neonatology SKIMS, Srinagar, India	100-102	NA	NA	NA	Joubert Syndrome	NA	NA	NA	NA
JS 28	Arora R. Joubert syndrome: imaging features and illustration of a case. Pol J Radiol. 2014 Oct 27;79:381-3. doi: 10.12659/PJR.890941. PMID: 25360184; PMCID: PMC4213002.	25360184	2014	Department of Radiology, Nizams Institute of Medical Sciences, Hyderabad, India	103	12 Year	Male	NA	Joubert Syndrome	NA	NA	NA	NA
JS 29	Chawla R, Bypareddy R, Vekaria L, Venkatesh P, Ananthashayana VH. Fundus findings in a case of Joubert syndrome. Indian J Ophthalmol. 2017 Apr;65(4):329-330. doi: 10.4103/ijo.IJO_441_16. PMID: 28513504; PMCID: PMC5452592.	28513504	2017	1 Department of Ophthalmology, Dr. Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India. 2 Department of Radio-diagnosis, All India Institute of Medical Sciences, New Delhi, India.	104	9 Month	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 30	Purkait R, Basu R, Das R, Chatterjee U. Association of Joubert syndrome and Hirschsprung disease. Indian Pediatr. 2015 Jan;52(1):61-2. doi: 10.1007/s13312-015-0568-3. PMID: 25638189.	25638189	2015	Department of Paediatric Medicine, NRS Medical College and Hospital, Kolkata; and *Department of Pathology, IPGMER and SSKM Hospital, Kolkata, India.	105	9 Month	Female	C	Joubert Syndrome	NA	NA	NA	NA
JS 31	Luthra A, Singh V. Dexmedetomidine and propofol based total intravenous anesthesia in a case of Joubert syndrome. J Dent Anesth Pain Med. 2020 Apr;20(2):101-103. doi: 10.17245/jdapm.2020.20.2.101. Epub 2020 Apr 27. PMID: 32395616; PMCID: PMC7193062.	32395616	2020	1 Department of Anesthesia and Intensive Care, Post Graduate Institute of Medical Education & Research (PGIMER), Chandigarh, India. 2 Department of Pedodontics and Preventive Dentistry, Oral Health Sciences Centre, Post Graduate Institute of Medical Education & Research (PGIMER), Chandigarh, India.	106	8 Year	Male	NA	Joubert Syndrome	NA	NA	NA	NA
JS 32	Goswami M, Rajwar AS, Verma M. Orocraniofacial findings of a Pediatric Patient with Joubert Syndrome. Int J Clin Pediatr Dent. 2016 Oct-Dec;9(4):379-383. doi: 10.5005/jp-journals-10005-1394. Epub 2016 Dec 5. PMID: 28127172; PMCID: PMC5233707.	28127172	2016	Department of Pedodontics and Preventive Dentistry, Maulana Azad Institute of Dental Sciences, New Delhi, India.	107	7 Year	Female	NA	Joubert Syndrome	NA	NA	NA	NA
JS 33	Singh P, Goraya JS, Saggar K, Ahluwalia A. A report of Joubert syndrome in an infant, with literature review. J Pediatr Neurosci. 2011 Jan;6(1):44-7. doi: 10.4103/1817-1745.84407. PMID: 21977088; PMCID: PMC3173915.	21977088	2011	Department of Radiodiagnosis, Dayanand Medical College and Hospital, Ludhiana, Punjab, India.	108	7 Months	Female	NC	Joubert Syndrome	NA	NA	NA	NA
JS 34	Kaur S, Kulkarni K. Joubert syndrome type I: neuroimaging findings in addition to the 'molar tooth' sign. BMJ Case Rep. 2010 Apr 29;2010:bcr11.2009.2464. doi: 10.1136/bcr.11.2009.2464. PMID: 22736560; PMCID: PMC3047525.	22736560	2010	Indraprastha Apollo Hospital, Pediatrics, Sarita Vihar, New Delhi, Delhi, India.	109	6 Months	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 35	Garg S, Rakesh K, Khattri S, Mishra P, Tikka SK. Bipolar disorder with intellectual disability in Joubert syndrome: A case report. Psychiatry Clin Neurosci. 2019 Dec;73(12):761-762. doi: 10.1111/pcn.12937. Epub 2019 Nov 4. PMID: 31596012.	31596012	2019	1 Department of Psychiatry, Shri Guru Ram Rai Institute of Medical & Health Sciences, Dehradun, India. 2 Department of Psychiatry, All India Institute of Medical Sciences, Raipur, India.	110	27 Year	Female	NA	Joubert Syndrome	NA	NA	NA	NA
JS 36	Baldawa S. An unusual association of classical Joubert syndrome with retrocerebellar arachnoid cyst. Childs Nerv Syst. 2016 Jul;32(7):1181-2. doi: 10.1007/s00381-016-3099-x. Epub 2016 May 2. PMID: 27138555.	27138555	2016	Department of Neurosurgery, Yashodhara Superspeciality Hospital, Solapur, Maharashtra, India	111	NA	NA	NA	Joubert Syndrome	NA	NA	NA	NA
JS 37	Byju N, Jose J, Saifudheen K, Musthafa M. Molar tooth sign with ataxia and see-saw nystagmus (Joubert syndrome). Ann Indian Acad Neurol. 2011 Jan;14(1):62-3. doi: 10.4103/0972-2327.78057. PMID: 21654929; PMCID: PMC3108085.	21654929	2011	Department of Neurology, Medical College, Calicut, Kerala, India.	112	16 Years	Male	C	Joubert Syndrome	NA	NA	NA	NA
JS 37	Mallick D, Thapa R. 'Molar tooth' sign in Joubert syndrome. Pediatr Radiol. 2010 Jan;40(1):131. doi: 10.1007/s00247-009-1338-y. Epub 2009 Jul 7. PMID: 19582441.	19582441	2010	Department of Pediatrics, The Institute of Child Health, 11 Dr. Biresh Guha St, Kolkata, 700 017, India.	113	11 Months	Female	NC	Joubert Syndrome	NA	NA	NA	NA

JS 38	Kumar J, Kumar A, Saha S. The molar tooth sign of Joubert syndrome. Arch Neurol. 2007 Apr;64(4):602-3. doi: 10.1001/archneur.64.4.602. PMID: 17420326.	17420326	2007	Minakshi Garden, P.O. Tilak Nagar, New Delhi, India.	114	1 Year	NA	NA	Joubert Syndrome	NA	NA	NA	NA
JS 39	Nagpal T, Pande S. Molar tooth sign: neuroimaging characteristic of Joubert syndrome. Neurol India. 2008 Apr-Jun;56(2):223. doi: 10.4103/0028-3886.42019. PMID: 18688166.	18688166	2008	Jabalpur Hospital and Research Centre and Netaji Subhash Chandra Bose Medical College, Jabalpur, India.	115	2 Years	Male	NC	Joubert Syndrome	NA	NA	NA	NA
JS 40	Subramanian S, Hari S, Santosh Kumar S. Clinics in diagnostic imaging (118). Singapore Med J. 2007 Sep;48(9):869-72; quiz 873. PMID: 17728972.	17728972	2007	Department of Radiodiagnosis, All India Institute of Medical Sciences, New Delhi, India.	116	18 Months	Male	NA	Joubert Syndrome	NA	NA	NA	NA
JS 41	Mistry KA, Bhoil R, Patel A. In response to the article "joubert syndrome imaging features and illustration of a case"; pol j radiol, 2015; 80: 381-83. Pol J Radiol. 2015 Apr 4;80:176-7. doi: 10.12659/PJR.892905. PMID: 25908948; PMCID: PMC4391375.	25908948	2015	1 Department of Radiology, Dr Rajendra Prasad Government Medical College, Tanda, Kangra, Himachal Pradesh, India. 2 Department of Radiology, Indira Gandhi Medical College, Shimla, Himachal Pradesh, India. 3 Department of Physiology, Medical College & SSG Hospital, Vadodara, Gujarat, India.	117	8 Months	Female	NA	Joubert Syndrome	NA	NA	NA	NA
JS 42	Verma SK, Shetty BS, Kanth L, Kumar T. A girl with abnormal head and eye movements: Joubert syndrome (2005:3b). Eur Radiol. 2005 Jun;15(6):1274-6. doi: 10.1007/s00330-004-2568-x. PMID: 15988832.	15988832	2005	Department of Radiology and Imaging, K.G. Hospital and P.G Medical Institute, Coimbatore, India.	118	NA	Female	NA	Joubert Syndrome	NA	NA	NA	NA
JS 43	Chatur C, Balani A, Vadapalli R, Murthy MG. Isolated Unilateral Cerebellar Hemispheric Dysplasia: A Rare Entity. Can J Neurol Sci. 2019 Nov;46(6):760-761. doi: 10.1017/cjn.2019.249. PMID: 31352912.	31352912	2019	1 Department of Radiology, Yashoda Hospital, Hyderabad, India. 2 Department of Radiology, Vijaya Diagnostic Centre, Hyderabad, India.	119	9 Years	Female	NC	Joubert Syndrome	NA	NA	NA	NA
JS 44	Singh J, Dalal P, Rattan KN. Development Delay in a Child with Microcephaly and Birth Asphyxia: Explore Diagnosis beyond Hypotonic Cerebral Palsy. J Pediatr Neurosci. 2021 Oct-Dec;16(4):285-288. doi: 10.4103/jpn.JPN_126_20. Epub 2021 Jul 19. PMID: 36531762; PMCID: PMC9757521.	36531762	2021	1 Department of Pediatrics, Pandit Bhagwat Dayal Sharma Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India. 2 Department of Pediatric Surgery, Pandit Bhagwat Dayal Sharma Post Graduate Institute of Medical Sciences, Rohtak, Haryana, India.	120	2 Years	Female	NC	Joubert Syndrome	NA	NA	NA	NA
JS 45	Kalra V, Sharma S, Garg A. Rhombencephalosynapsis: association with single umbilical artery. Indian J Pediatr. 2008 Nov;75(11):1175-7. doi: 10.1007/s12098-008-0189-0. Epub 2008 Sep 22. PMID: 18810339.	18810339	2008	Department of Pediatrics, All India Institute of Medical Sciences, New Delhi, India.	121	6 Years	Female	NA	Joubert Syndrome	NA	NA	NA	NA
JS 46	Agrawal S. A child with delayed milestones and interesting findings on MR. BMJ Case Rep. 2010 Nov 5;2010:bcr0520103037. doi: 10.1136/bcr.05.2010.3037. PMID: 22791856; PMCID: PMC3029270.	22791856	2010	Ekta Institute of Child Health, Raipur, India.	122	13 Months	Male	2* C	Joubert Syndrome	NA	NA	NA	NA
JS 47	Sriganesh K, Vinay B, Jena S, Sudhir V, Saini J, Umamaheswara Rao GS. Anesthetic management of patients with Joubert syndrome: a retrospective analysis of a single-institutional case series. Paediatr Anaesth. 2014 Nov;24(11):1180-4. doi: 10.1111/pan.12472. Epub 2014 Jul 5. PMID: 25040301.	25040301	2014	Department of Neuroanaesthesia, National Institute of Mental health and Neurosciences (NIMHANS), Bangalore, India.	123-138	6 Months to 21 Years	NA	NA	Joubert Syndrome	NA	NA	NA	NA
BBS													
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	1	11 Years	Male	NC	Polydactyly, Rod cone dystrophy	Clinical Exome	BBS10	Missense	Novel
LCA 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	2	16 years	Male	NC	Bardet-Biedl Syndrome, OBESITY , VISION LOSS	NA	BBS10	Duplication	NA
BBS 1	Dayal D, Seetharaman K, Panigrahi I, Muthuvel B, Agarwal A. Severe Early Onset Obesity due to a Novel Missense Mutation in Exon 3 of the Leptin Gene in an Infant from Northwest India. J Clin Res Pediatr Endocrinol. 2018 Jul 31;10(3):274-278. doi: 10.4274/jcrpe.5501. Epub 2017 Dec 8. PMID: 29217499; PMCID: PMC6083471.	29217499	2018	1 Postgraduate Institute of Medical Education and Research, Department of Pediatrics, Division of Pediatric Endocrinology, Chandigarh, India. 2 Postgraduate Institute of Medical Education and Research, Department of Pediatrics, Division of Genetic-Metabolic, Chandigarh, India	3	10 Months	Female	3* C	Severe Hyperphagia, Dyslipidemia and Early Onset Obesity (EOO)	Sanger Sequencing (Illumina HiSeq 2000 sequencing platform)	LEPTIN	Missense	Novel
BBS 1	Dayal D, Seetharaman K, Panigrahi I, Muthuvel B, Agarwal A. Severe Early Onset Obesity due to a Novel Missense Mutation in Exon 3 of the Leptin Gene in an Infant from Northwest India. J Clin Res Pediatr Endocrinol. 2018 Jul 31;10(3):274-278. doi: 10.4274/jcrpe.5501. Epub 2017 Dec 8. PMID: 29217499; PMCID: PMC6083471.	29217499	2018	1 Postgraduate Institute of Medical Education and Research, Department of Pediatrics, Division of Pediatric Endocrinology, Chandigarh, India. 2 Postgraduate Institute of Medical Education and Research, Department of Pediatrics, Division of Genetic-Metabolic, Chandigarh, India	3	10 Months	Female	3* C	Severe Hyperphagia, Dyslipidemia and Early Onset Obesity (EOO)	Sanger Sequencing (Illumina HiSeq 2000 sequencing platform)	BBS1	Missense	NA
BBS 2	Kaur P, Chaudhry C, Neelam H, Panigrahi I. Bardet-Biedl syndrome presenting with laryngeal web and bifid epiglottis. BMJ Case Rep. 2021 Jan 28;14(1):e236325. doi: 10.1136/bcr-2020-236325. PMID: 33509858; PMCID: PMC7845671.	33509858	2021	1 Pediatrics, Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India. 2 Pediatrics, Postgraduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	4	11 Months	Male	3* C	Bardet-Biedl Syndrome and Simpson-Golabi-Behmel syndrome (SGBS)	Exome Sequencing	BBS10	Frameshift	NA
BBS 3	Ankleshwaria C, Prajapati B, Parmar S, Rathod V, Patel H, Dhorajiya D, Chavda N, Parmar K, Pathan F, Chauhan M. Bardet-Biedl Syndrome Presenting in Adulthood. Indian J Nephrol. 2022 Nov-Dec;32(6):633-636. doi: 10.4103/ijn.ijn_320_21. Epub 2022 Oct 2. PMID: 36704599; PMCID: PMC9872935.	36704599	2022	Department of Internal Medicine, Civil Hospital Ahmedabad, Gujarat, India	5	32 Years	Female	C	Bardet-Biedl Syndrome	Genetic Panel	BBS9	Missense	Novel
BBS 4	Ali G, Sadia, Foo JN, Nasir A, Chang CH, Chew EG, Latif Z, Azeem Z, Ain-Ul-Batool S, Kazmi SAR, Awan NB, Khan AH, Rehman FU, Khalid M, Wali A, Sarwar S, Akhtar W, Ahmed Abbasi A, Nisar R. Identification of a Novel Homozygous Missense (c.443A>T:p.N148I) Mutation in BBS2 in a Kashmiri Family with Bardet-Biedl Syndrome. Biomed Res Int. 2021 Feb 23;2021:6626015. doi:	33688495	2021	1 Department of Biotechnology, University of Azad Jammu and Kashmir, P.O. Box 13100, Muzaffarabad, Pakistan. 2 Lee Kong Chian School of Medicine, Nanyang Technological University Singapore, 11 Mandalay Road, Singapore 308232. 3 Human Genetics, Genome Institute of Singapore, A*STAR, 60 Biopolis Street, Singapore 138672. 4 Molecular Science and Technology, Ajou University, Suwon, Republic of Korea. 5 Department of Zoology, University of Azad Jammu	6	26 Years	Male	C	Bardet-Biedl Syndrome	Whole-exome sequencing on Illumina HiSeq 4000	BBS2	Missense	Novel

BBS 5	Goyal S, Singh IR, Vanita V. Novel mutation in MKKS/BBS6 linked with arRP and polydactyly in a family of North Indian origin. Clin Exp Ophthalmol. 2020 Apr;48(3):343-355. doi: 10.1111/ceo.13719. Epub 2020 Feb 3. PMID: 31989739.	31989739	2020	1 Department of Human Genetics, Guru Nanak Dev University, Amritsar, India. 2 Dr. Daljit Singh Eye Hospital, Amritsar, India.	7	NA	NA	NA	Bardet-Biedl Syndrome	Whole-exome sequencing (WES)	MKKS/BBS6	Missense	Novel
BBS 5	Goyal S, Singh IR, Vanita V. Novel mutation in MKKS/BBS6 linked with arRP and polydactyly in a family of North Indian origin. Clin Exp Ophthalmol. 2020 Apr;48(3):343-355. doi: 10.1111/ceo.13719. Epub 2020 Feb 3. PMID: 31989739.	31989739	2020	1 Department of Human Genetics, Guru Nanak Dev University, Amritsar, India. 2 Dr. Daljit Singh Eye Hospital, Amritsar, India.	8	NA	NA	NA	Bardet-Biedl Syndrome	Whole-exome sequencing (WES)	MKKS/BBS6	Missense	NA
BBS 5	Goyal S, Singh IR, Vanita V. Novel mutation in MKKS/BBS6 linked with arRP and polydactyly in a family of North Indian origin. Clin Exp Ophthalmol. 2020 Apr;48(3):343-355. doi: 10.1111/ceo.13719. Epub 2020 Feb 3. PMID: 31989739.	31989739	2020	1 Department of Human Genetics, Guru Nanak Dev University, Amritsar, India. 2 Dr. Daljit Singh Eye Hospital, Amritsar, India.	9	NA	NA	NA	Bardet-Biedl Syndrome	Whole-exome sequencing (WES)	MKKS/BBS6	Missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	10	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS4	splice site	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	11	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	MKKS/BBS6	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	12	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS10	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	13	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS3	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	14	NA	Male	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS5	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	15	NA	Female	NC	Bardet-Biedl Syndrome	SNP Genotyping	BBS2	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	16	NA	Male	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS1	splice site	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	17	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS10	duplication FS	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	18	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS12	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	19	NA	Male	NC	Bardet-Biedl Syndrome	SNP Genotyping	BBS3	missense	Novel
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	20	NA	Male	NC	Bardet-Biedl Syndrome	SNP Genotyping	BBS9	deletion FS	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	21	NA	Male	NC	Bardet-Biedl Syndrome	SNP Genotyping	BBS1	splice site	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	22	NA	Male	NC	Bardet-Biedl Syndrome	SNP Genotyping	BBS11	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	23	NA	Male	NC	Bardet-Biedl Syndrome	SNP Genotyping	BBS3	missense	NA

BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	24	NA	Male	NC	Bardet-Biedl Syndrome	SNP Genotyping	BBS3	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	25	NA	Male	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS2	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	26	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS9	nonsense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	27	NA	NA	NA	Bardet-Biedl Syndrome	SNP Genotyping	MKKS/BBS6	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	28	NA	Male	C	Bardet-Biedl Syndrome	SNP Genotyping	NA	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	29	NA	Male	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS2	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	30	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	BBS12	deletion FS	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	31	NA	Male	NC	Bardet-Biedl Syndrome	SNP Genotyping	ALMS1	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	32	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	ALMS1	nonsense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	33	NA	NA	NA	Bardet-Biedl Syndrome	SNP Genotyping	MKKS/BBS6	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	34	NA	Female	C	Bardet-Biedl Syndrome	SNP Genotyping	NA	missense	NA
BBS 7	Sathya Priya C, Sen P, Umashankar V, Gupta N, Kabra M, Kumaramanickavel G, Stoetzel C, Dollfus H, Sripriya S. Mutation spectrum in BBS genes guided by homozygosity mapping in an Indian cohort. Clin Genet. 2015 Feb;87(2):161-6. doi: 10.1111/cge.12342. Epub 2014 Feb 18. PMID: 24400638.	24400638	2015	SN ONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India; School of Chemical engineering and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India	35	NA	Female	NC	Bardet-Biedl Syndrome	SNP Genotyping	BBS10	deletion FS	NA
BBS 8	Parameswarappa DC, Das AV, Thakur PS, Takkar B, Multani PK, Padhy SK, Doctor MB, Agarwal K, Jalali S. Retinitis pigmentosa in Laurence-Moon-Bardet-Biedl syndrome in India: Electronic medical records driven big data analytics: Report II. Indian J Ophthalmol. 2022 Jul;70(7):2533-2538. doi: 10.4103/ijo.IJO_2268_21. PMID: 35791150; PMCID: PMC9426086.	35791150	2022	L V Prasad Eye Institute, Hyderabad, Telangana, India.	36-279	15.22 ± 7.56 and 14 Years	244 BBS patients (164 Males, 80	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 9	Sachdev MS, Verma L, Garg SP, Moonis M, Shekar CH, Gupta NK. Bilateral disc oedema in retinitis pigmentosa--an unusual sign. Jpn J Ophthalmol. 1987;31(4):621-6. PMID: 3448326.	3448326	1987	Centre for Ophthalmic Sciences, A.I.I.M.S., Ansari Nagar, New Delhi, India.	280	NA	NA	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 10	Gnanasekaran H, Chandrasekhar SP, Kandeeban S, Periyasamy P, Bhende M, Khetan V, Gupta N, Kabra M, Namboothri S, Sen P, Sripriya S. Mutation profile of Bardet-Biedl syndrome patients from India: Implicative role of multiallelic rare variants and oligogenic inheritance pattern. Clin Genet. 2023 Oct;104(4):443-460. doi: 10.1111/cge.14398. Epub 2023 Jul 11. PMID: 37431782.	37431782	2023	1 SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, Tamilnadu, India. 2 School of Chemical and Biotechnology, SASTRA University, Thanjavur, Tamilnadu, India. 3 Division of Genetics, Department of Pediatrics, AIIMS, New Delhi, India. 4 Shri Bhagwan Mahavir Vitreoretinal Services, Sankara Nethralaya, Chennai, Tamilnadu, India. 5 Department of Pediatric Genetics, Amrita Institute of Medical Sciences and Research Centre,	281-388	NA	108	NA	Bardet-Biedl Syndrome	NA	BBS1 and BBS10 (Number NA)	NA	full text requested or continue without
BBS 11	Chandrasekar SP, Namboothiri S, Sen P, Sarangapani S. Screening for mutation hotspots in Bardet-Biedl syndrome patients from India. Indian J Med Res. 2018 Feb;147(2):177-182. doi: 10.4103/ijmr.IJMR_1822_15. PMID: 29806606; PMCID: PMC5991121.	29806606	2018	1 Department of Paediatric Genetics, Amrita Institute of Medical Sciences & Research Centre, Kochi, India. 2 Department of Vitreo Retina Clinic, Medical Research Foundation, Chennai, India. 3 SN ONGC Department of Genetics & Molecular Biology, Vision Research Foundation, Chennai, India.	389-452	13.4±6.42 yr (range 6-30)	64 BBS patients (44 males, 20	NC	Bardet-Biedl Syndrome	PCR based direct sequencing	ARL6	Missense	NA
BBS 11	Chandrasekar SP, Namboothiri S, Sen P, Sarangapani S. Screening for mutation hotspots in Bardet-Biedl syndrome patients from India. Indian J Med Res. 2018 Feb;147(2):177-182. doi: 10.4103/ijmr.IJMR_1822_15. PMID: 29806606; PMCID: PMC5991121.	29806606	2018	1 Department of Paediatric Genetics, Amrita Institute of Medical Sciences & Research Centre, Kochi, India. 2 Department of Vitreo Retina Clinic, Medical Research Foundation, Chennai, India. 3 SN ONGC Department of Genetics & Molecular Biology, Vision Research Foundation, Chennai, India.	NA	NA	NA	NA	NA	NA	BBS10	Missense	NA

BBS 11	Chandrasekar SP, Namboothiri S, Sen P, Sarangapani S. Screening for mutation hotspots in Bardet-Biedl syndrome patients from India. Indian J Med Res. 2018 Feb;147(2):177-182. doi: 10.4103/ijmr.IJMR_1822_15. PMID: 29806606; PMCID: PMC5991121.	29806606	2018	1 Department of Paediatric Genetics, Amrita Institute of Medical Sciences & Research Centre, Kochi, India. 2 Department of Vitreo Retina Clinic, Medical Research Foundation, Chennai, India. 3 SN ONGC Department of Genetics & Molecular Biology, Vision Research Foundation, Chennai, India.	NA	NA	NA	NA	NA	NA	BBS10	Deletion	NA
BBS 11	Chandrasekar SP, Namboothiri S, Sen P, Sarangapani S. Screening for mutation hotspots in Bardet-Biedl syndrome patients from India. Indian J Med Res. 2018 Feb;147(2):177-182. doi: 10.4103/ijmr.IJMR_1822_15. PMID: 29806606; PMCID: PMC5991121.	29806606	2018	1 Department of Paediatric Genetics, Amrita Institute of Medical Sciences & Research Centre, Kochi, India. 2 Department of Vitreo Retina Clinic, Medical Research Foundation, Chennai, India. 3 SN ONGC Department of Genetics & Molecular Biology, Vision Research Foundation, Chennai, India.	NA	NA	NA	NA	NA	NA	BBS5	Missense	Novel
BBS 12	Hulleman JD, Nguyen A, Ramprasad VL, Murugan S, Gupta R, Mahindrakar A, Angara R, Sankurathri C, Mootha VV. A novel H395R mutation in MKKS/BBS6 causes retinitis pigmentosa and polydactyly without other findings of Bardet-Biedl or McKusick-Kaufman syndrome. Mol Vis. 2016 Jan 24;22:73-81. PMID: 26900326; PMCID: PMC4734152.	26900326	2016	1 Srikiran Institute of Ophthalmology, Kakinada, Andhra Pradesh, India. 2 Department of Ophthalmology, University of Texas Southwestern Medical Center, Dallas, TX. 3 MedGenome, Bangalore, Karnataka, India.	453-454	18 Yrs and 13 Yrs	One Male, One Female	C	Bardet-Biedl Syndrome	Next Generation sequencing	MKKS/BBS6	Missense	NA
BBS 13	Chakrabarty S, Savantre SB, Ramachandra Bhat C, Satyamoorthy K. Multiple genetic mutations implicate spectrum of phenotypes in Bardet-Biedl syndrome. Gene. 2020 Jan 30;725:144164. doi: 10.1016/j.gene.2019.144164. Epub 2019 Oct 19. PMID: 31639430.	31639430	2019	1 Department of Cell and Molecular Biology, Manipal School of Life Sciences, Manipal Academy of Higher Education, Manipal, Karnataka 576104, India. 2 Department of Medicine, K.V.G. Medical College & Hospital, Dakshina Kannada, Sullia 574327, India.	455	23 Year	Male	2* C	Bardet-Biedl Syndrome	Whole Exome Sequencing	BBS10	Missense	Novel
BBS 13	Chakrabarty S, Savantre SB, Ramachandra Bhat C, Satyamoorthy K. Multiple genetic mutations implicate spectrum of phenotypes in Bardet-Biedl syndrome. Gene. 2020 Jan 30;725:144164. doi: 10.1016/j.gene.2019.144164. Epub 2019 Oct 19. PMID: 31639430.	31639430	2019	1 Department of Cell and Molecular Biology, Manipal School of Life Sciences, Manipal Academy of Higher Education, Manipal, Karnataka 576104, India. 2 Department of Medicine, K.V.G. Medical College & Hospital, Dakshina Kannada, Sullia 574327, India.	455	23 Year	Male	2* C	Bardet-Biedl Syndrome	Whole Exome Sequencing	PDE6B	Missense	NA
BBS 13	Chakrabarty S, Savantre SB, Ramachandra Bhat C, Satyamoorthy K. Multiple genetic mutations implicate spectrum of phenotypes in Bardet-Biedl syndrome. Gene. 2020 Jan 30;725:144164. doi: 10.1016/j.gene.2019.144164. Epub 2019 Oct 19. PMID: 31639430.	31639430	2019	1 Department of Cell and Molecular Biology, Manipal School of Life Sciences, Manipal Academy of Higher Education, Manipal, Karnataka 576104, India. 2 Department of Medicine, K.V.G. Medical College & Hospital, Dakshina Kannada, Sullia 574327, India.	455	23 Year	Male	2* C	Bardet-Biedl Syndrome	Whole Exome Sequencing	PDE6B	Missense	NA
BBS 14	Soni NK. Ear, nose and throat manifestations in Laurence-Moon-Biedl-Bardet Syndrome. Indian J Otolaryngol Head Neck Surg. 1997 Mar;49(Suppl 1):61-2. doi: 10.1007/BF03021330. PMID: 23119360; PMCID: PMC3450564.	23119360	1997	Department of Ear, Nose and Throat, Medical College, 334003 Kota, (Rajasthan).	456	5 Day	Female	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 14	Soni NK. Ear, nose and throat manifestations in Laurence-Moon-Biedl-Bardet Syndrome. Indian J Otolaryngol Head Neck Surg. 1997 Mar;49(Suppl 1):61-2. doi: 10.1007/BF03021330. PMID: 23119360; PMCID: PMC3450564.	23119360	1997	Department of Ear, Nose and Throat, Medical College, 334003 Kota, (Rajasthan).	457	8 Years	NA	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 14	Soni NK. Ear, nose and throat manifestations in Laurence-Moon-Biedl-Bardet Syndrome. Indian J Otolaryngol Head Neck Surg. 1997 Mar;49(Suppl 1):61-2. doi: 10.1007/BF03021330. PMID: 23119360; PMCID: PMC3450564.	23119360	1997	Department of Ear, Nose and Throat, Medical College, 334003 Kota, (Rajasthan).	458	11 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 14	Soni NK. Ear, nose and throat manifestations in Laurence-Moon-Biedl-Bardet Syndrome. Indian J Otolaryngol Head Neck Surg. 1997 Mar;49(Suppl 1):61-2. doi: 10.1007/BF03021330. PMID: 23119360; PMCID: PMC3450564.	23119360	1997	Department of Ear, Nose and Throat, Medical College, 334003 Kota, (Rajasthan).	459	12 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 14	Soni NK. Ear, nose and throat manifestations in Laurence-Moon-Biedl-Bardet Syndrome. Indian J Otolaryngol Head Neck Surg. 1997 Mar;49(Suppl 1):61-2. doi: 10.1007/BF03021330. PMID: 23119360; PMCID: PMC3450564.	23119360	1997	Department of Ear, Nose and Throat, Medical College, 334003 Kota, (Rajasthan).	460	8 Years	Female	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 15	Tomar SS, Supekar BB, Chopkar A, Mukhi J, Singh RP. Bardet-Biedl Syndrome with Café-au-Lait Macule: Association or Coincidence? Indian Dermatol Online J. 2020 Mar 9;11(2):246-249. doi: 10.4103/idoj.IDOJ_106_19. PMID: 32477991; PMCID: PMC7247639.	32477991	2020	Department of Dermatology, Venereology and Leprosy, Government Medical College, Nagpur, Maharashtra, India.	461	8 Years	Male	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 16	Sahu JK, Jain V. Laurence-Moon-Bardet-Biedl syndrome. JNMA J Nepal Med Assoc. 2008 Oct-Dec;47(172):235-7. PMID: 19079403.	19079403	2008	Department of Pediatrics, All India Institute of Medical Sciences, New Delhi, India.	462	NA	NA	NA	Nonalcoholic steatohepatitis, Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 17	Y M P, Ashraf M, B M V, Menezes S, Mohan A. A case report on the bardet biedl syndrome with hypokalaemic paralysis. J Clin Diagn Res. 2013 Jun;7(6):1163-4. doi: 10.7860/JCDR/2013/4770.3030. Epub 2013 Jun 1. PMID: 23905129; PMCID: PMC3708224.	23905129	2013	Department of Medicine, Father Muller Medical college , Mangalore, Karnataka, India .	463	22 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 18	Parakh R Md, Nairy DM Md. Bardet-Biedl Syndrome with End Stage Renal Disease. Iran J Med Sci. 2016 Nov;41(6):539-542. PMID: 27853335; PMCID: PMC5106570.	27853335	2016	Department and Institutions, SDM College of Medical Sciences and Hospital, Sattur, Dharwad-580009, State-Karnataka, India.	464	22 Years	Male	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 19	Shrinkhal, Singh A, Agrawal A, Mittal SK, Udenia H, Bandu GH. A rare case of Bardet-Biedl syndrome. Taiwan J Ophthalmol. 2019 Oct 17;10(2):138-140. doi: 10.4103/tjo.tjo_62_19. PMID: 32874845; PMCID: PMC7442099.	32874845	2019	Department of Ophthalmology, All India Institute of Medical Sciences, Rishikesh, Uttarakhand, India.	465	7 Years	Male	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA

BBS 20	Hassan S, Khan QA, Saravanan P, Iram S, Rohail S, Belay NF, Afzal M, Hadi FA, Pande H. Megaloblastic Anemia in Bardet-Biedl Syndrome: A Rare Case Report. Clin Med Insights Case Rep. 2023 Aug 14;16:11795476231193896. doi: 10.1177/11795476231193896. PMID: 37588947; PMCID: PMC10426295.	37588947	2023	Pondicherry Institute of Medical Sciences, Pondicherry, India.	466	16 Years	Female	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 21	Kumar S, Mahajan BB, Mittal J. Bardet-Biedl syndrome: a rare case report from North India. Indian J Dermatol Venereol Leprol. 2012 Mar-Apr;78(2):228. doi: 10.4103/0378-6323.93656. PMID: 22421669.	22421669	2012	Department of Skin and Veneral Diseases, G.G.S. Medical College, Faridkot, Punjab, India.	467	14 Years	Male	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 22	Viswanathan VK, Kanna RM, Shetty AP, Rajasekaran S. Acute flaccid paraparesis (cauda equina syndrome) in a patient with Bardet-Biedl syndrome. Indian J Orthop. 2017 May-Jun;51(3):330-333. doi: 10.4103/0019-5413.205682. PMID: 28566787; PMCID: PMC5439321.	28566787	2017	Department of Spine Surgery, Ganga Hospital, Coimbatore, Tamil Nadu, India.	468	22 Years	Female	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 23	Pal S, Bhattacharyya AR. Laurence-Moon-Bardet-Biedl syndrome. J Indian Med Assoc. 1995 Oct;93(10):391, 393. PMID: 9053416.	9053416	1995	Department of Obstetrics and Gynaecology, Calcutta National Medical College.	NA	NA	NA	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 24	Desai A, Jha O, Iyer V, Dada R, Kumar R, Tandon N. Reversible hypogonadism in Bardet-Biedl syndrome. Fertil Steril. 2009 Jul;92(1):391.e13-5. doi: 10.1016/j.fertnstert.2009.02.023. Epub 2009 Mar 26. PMID: 19327768.	19327768	2009	Department of Endocrinology and Metabolism, All India Institute of Medical Sciences, New Delhi, India.	469	30 Years	Male	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 25	Indraneel KS, V Rajalakshmi, Dayanandan Y, Reddy NM. A Case of Laurence Moon Bardet Biedl Syndrome. J Assoc Physicians India. 2023 Jan;71(1):1. PMID: 37116049.	37116049	2023	Saveetha Medical College & Hospital, Chennai, Tamil Nadu, India.	470	20 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 26	Madireddi J, Acharya V, Suryanarayana J, Hande HM, Shetty R. Bardet-Biedl syndrome: multiple fingers with multiple defects! BMJ Case Rep. 2015 Nov 26;2015:bcr2015211776. doi: 10.1136/bcr-2015-211776. PMID: 26611481; PMCID: PMC4680628.	26611481	2015	Department of Internal Medicine, Kasturba Medical College, Manipal, Manipal, Karnataka, India.	471	45 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 27	Prakash EB. Bardet Biedl Syndrome. J Assoc Physicians India. 2005 Sep;53:781. PMID: 16334622.	16334622	2005	CSI Kalyani Multispeciality Hospital, Mylapore, Chennai.	NA	NA	NA	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 28	Varma C, Bhat RY, Bhatt S. Bardet-Biedl syndrome in two sisters: A rare incidence. J Pediatr Genet. 2013 Mar;2(1):49-51. doi: 10.3233/PGE-13048. PMID: 27625840; PMCID: PMC5020959.	27625840	2013	Department of Pediatrics, Kasturba Medical College, Manipal, Karnataka, India.	472	12 Year	Female	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 28	Varma C, Bhat RY, Bhatt S. Bardet-Biedl syndrome in two sisters: A rare incidence. J Pediatr Genet. 2013 Mar;2(1):49-51. doi: 10.3233/PGE-13048. PMID: 27625840; PMCID: PMC5020959.	27625840	2013	Department of Pediatrics, Kasturba Medical College, Manipal, Karnataka, India.	473	NA	Female	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 29	Chakravarti HN, Ray S, Chakrabarti SK, Biswas D, Ghosh S, Mukhopadhyay S, Chowdhury S. Bardet-Biedl syndrome in two siblings: a rare entity revisited. QJM. 2016 Feb;109(2):123-4. doi: 10.1093/qjmed/hcv114. Epub 2015 May 28. PMID: 26025693.	26025693	2016	Department of Endocrinology and Metabolism, Institute of Post Graduate Medical Education & Research and SSKM Hospital, Kolkata 700020, West Bengal, India	474	12 Year	Male	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 29	Chakravarti HN, Ray S, Chakrabarti SK, Biswas D, Ghosh S, Mukhopadhyay S, Chowdhury S. Bardet-Biedl syndrome in two siblings: a rare entity revisited. QJM. 2016 Feb;109(2):123-4. doi: 10.1093/qjmed/hcv114. Epub 2015 May 28. PMID: 26025693.	26025693	2016	Department of Endocrinology and Metabolism, Institute of Post Graduate Medical Education & Research and SSKM Hospital, Kolkata 700020, West Bengal, India	475	6 Year	Female	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 30	Singh MK, Shrinkhal, Pradhan S, Chakraborty P. Bardet Biedl Syndrome - A Report of Two Cases with Otolaryngologic Symptoms. J Clin Diagn Res. 2017 Mar;11(3):ND01-ND02. doi: 10.7860/JCDR/2017/24499.9466. Epub 2017 Mar 1. PMID: 28511423; PMCID: PMC5427349.	28511423	2017	Institute of Medical Sciences, BHU, Varanasi, Uttar Pradesh, India.	476	8 Years	Female	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 30	Singh MK, Shrinkhal, Pradhan S, Chakraborty P. Bardet Biedl Syndrome - A Report of Two Cases with Otolaryngologic Symptoms. J Clin Diagn Res. 2017 Mar;11(3):ND01-ND02. doi: 10.7860/JCDR/2017/24499.9466. Epub 2017 Mar 1. PMID: 28511423; PMCID: PMC5427349.	28511423	2017	Institute of Medical Sciences, BHU, Varanasi, Uttar Pradesh, India.	477	7 Years	Female	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 31	Singh KK, Kumar R, Prakash J, Krishna A. Bardet-Biedl syndrome presenting with steroid sensitive nephrotic syndrome. Indian J Nephrol. 2015 Sep-Oct;25(5):300-2. doi: 10.4103/0971-4065.151765. PMID: 26628797; PMCID: PMC4588327.	26628797	2015	Indira Gandhi Institute of Medical Sciences, Patna, Bihar, India.	478	10 Years	Male	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 32	Kute VB, Vanikar AV, Gumber MR, Patel HV, Shah PR, Patil SB, Trivedi HL. Bardet-biedl syndrome: a rare cause of chronic kidney disease. Indian J Clin Biochem. 2013 Apr;28(2):201-5. doi: 10.1007/s12291-012-0275-y. Epub 2012 Oct 30. PMID: 24426211; PMCID: PMC3613492.	24426211	2013	1 Department of Nephrology and Clinical Transplantation, Institute of Kidney Diseases and Research Center, Dr. HL Trivedi Institute of Transplantation Sciences IKDRC-ITS, Civil Hospital Campus, Asarwa, Ahmedabad, 380016 Gujarat India. 2 Department of Pathology, Laboratory Medicine, Transfusion Services and Immunohematology, IKDRC-ITS, Ahmedabad, India.	479	17 Years	Female	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 33	Yadav DK, Beniwal MK, Jain A. Bardet-Biedl syndrome a rare cause of cardiomyopathy. Indian Pediatr. 2013 Jun 8;50(6):599-601. PMID: 23942403.	23942403	2013	Division of Pediatric Cardiology, Department of Neonatology and Pediatric Medicine, PGIMER, Dr. RML Hospital, New Delhi, India.	480	13 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA

BBS 34	Majumdar U, Arya G, Singh S, Pillai A, Nair PP. Oro-dental findings in Bardet-Biedl syndrome. BMJ Case Rep. 2012 Apr 23;2012:bcr1220115320. doi: 10.1136/bcr.12.2011.5320. PMID: 22604765; PMCID: PMC3339191.	22604765	2012	Department of Oral and Maxillofacial Surgery, Rishiraj Dental College & Hospital, Bhopal, Madhya Pradesh, India.	481	17 Years	Male	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 35	Hemachandar R. Bardet-Biedl syndrome: A rare cause of end stage renal disease. Int J Appl Basic Med Res. 2015 Jan-Apr;5(1):70-2. doi: 10.4103/2229-516X.149254. PMID: 25664275; PMCID: PMC4318109.	25664275	2015	Department of Nephrology, Mahatma Gandhi Medical College and Research Institute, Pillaiyarkuppam, Puducherry, India.	482	18 Years	Female	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 36	Bedi NK, Grover D. Bardet-Biedl syndrome with urogenital sinus presenting with acute renal failure in a neonate. Indian J Pediatr. 2014 Jul;81(7):719-21. doi: 10.1007/s12098-013-1147-z. Epub 2013 Aug 6. PMID: 23918321.	23918321	2014	Department of Pediatric Surgery, Christian Medical College & Hospital, Brown Road, Ludhiana, Punjab, 141008, India	483	Neonate	Female	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 37	Chatterjee SS, Guha P, Talukdar A, Dasgupta G. Autism: a rare presentation of Bardet-Biedl syndrome. BMJ Case Rep. 2014 Jun 4;2014:bcr2014203882. doi: 10.1136/bcr-2014-203882. PMID: 24899006; PMCID: PMC4054480.	24899006	2014	Medical College Kolkata, Kolkata, West Bengal, India.	484	8 Years	Male	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 38	Garg MK, Madhu SV, Dwarakanath CS, Ammini AC. LAURENCE-MOON-BARDET-BIEDL SYNDROME - PRESENTING WITH ACUTE ONSET OF DIABETES MELLITUS. Med J Armed Forces India. 1998 Apr;54(2):155-156. doi: 10.1016/S0377-1237(17)30511-7. Epub 2017 Jun 26. PMID: 28775455; PMCID: PMC5531385.	28775455	1998	All India Institute of Medical Sciences, New Delhi.	485	22 Years	Male	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 39	Jethani J, Parija S, Shetty S, Vijayalakshmi P. Atypical association of Duane retraction syndrome and Bardet Biedl syndrome. Indian J Ophthalmol. 2007 Mar-Apr;55(2):139-41. doi: 10.4103/0301-4738.30710. PMID: 17322606.	17322606	2007	Department of Pediatric Ophthalmology, Aravind Eye Hospital, Madurai - 625 020, Tamil Nadu, India.	486	7 Years	Male	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 40	Vora KS, Modi MP, Butala BP, Shah VR. Anesthetic management of two cases of Bardet-Biedl syndrome for renal transplantation. Saudi J Kidney Dis Transpl. 2017 Mar-Apr;28(2):384-387. doi: 10.4103/1319-2442.202775. PMID: 28352024.	28352024	2017	Department of Anesthesia and Critical Care, Institute of Kidney Disease and Research Center, Dr. H. L. Trivedi Institute of Transplantation Sciences, Ahmedabad, Gujarat, India.	487	17 Years	Female	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 40	Vora KS, Modi MP, Butala BP, Shah VR. Anesthetic management of two cases of Bardet-Biedl syndrome for renal transplantation. Saudi J Kidney Dis Transpl. 2017 Mar-Apr;28(2):384-387. doi: 10.4103/1319-2442.202775. PMID: 28352024.	28352024	2017	Department of Anesthesia and Critical Care, Institute of Kidney Disease and Research Center, Dr. H. L. Trivedi Institute of Transplantation Sciences, Ahmedabad, Gujarat, India.	488	29 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 41	Chhabria B, Nampoothiri R, Suri V, Jain S. Spot Diagnosis of a Daedalian Genetic Disorder: Bardet-Biedl Syndrome. J Clin Diagn Res. 2016 May;10(5):OL01. doi: 10.7860/JCDR/2016/18321.7754. Epub 2016 May 1. PMID: 27437293; PMCID: PMC4948469.	27437293	2016	Department of Internal Medicine, Post Graduate Institute of Medical Education & Research , Chandigarh, India	489	15 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 42	Alexander V, George T, Devarajan G, Zachariah A. Acute myocardial infarction and haemodynamic stroke in a young patient with Bardet-Biedl syndrome. BMJ Case Rep. 2019 Apr 30;12(4):e229788. doi: 10.1136/bcr-2019-229788. PMID: 31040148; PMCID: PMC6505971.	31040148	2019	Department of Medicine, Christian Medical College and Hospital, Vellore, Tamil Nadu, India.	490	28 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 43	Bhat MT, Santhosh MC, Hegde HV, Rao RP. Anesthetic management of a child with Bardet-Biedl syndrome undergoing post-auricular dermoid excision. J Anaesthesiol Clin Pharmacol. 2014 Jan;30(1):117-8. doi: 10.4103/0970-9185.125732. PMID: 24574615; PMCID: PMC3927278.	24574615	2014	Department of Anaesthesiology, SDM Medical College, Sattur, Dharwad, Karnataka, India.	491	8 Years	Male	NA	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 44	Mahajan R, Kumar Batra Y, Kumar S, Kumar Grover V. Anesthetic management of a patient with Bardet-Biedl syndrome and dilated cardiomyopathy. Minerva Anestesiol. 2007 Mar;73(3):191-4. Epub 2007 Jan 25. PMID: 17242658.	17242658	2007	Post Graduate Institute of Medical Education and Research (Pgimer), Chandigarh, India.	492	32 Years	Female	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 45	Hooda AK, Karan SC, Bishnoi JS, Nandwani A, Sinha T. Renal transplant in a child with Bardet-Biedl syndrome: A rare cause of end-stage renal disease. Indian J Nephrol. 2009 Jul;19(3):112-4. doi: 10.4103/0971-4065.57108. PMID: 20436731; PMCID: PMC2859476.	20436731	2009	Department of Nephrology, Army Hospital (R&R), Delhi Cantt, New Delhi-110 010, India.	493	12 Years	Male	NC	Bardet-Biedl Syndrome	NA	NA	NA	NA
BBS 46	Sowjanya B, Sreenivasulu U, Naidu JN, Sivaranjani N. End stage renal disease, differential diagnosis, a rare genetic disorder: bardet-biedl syndrome: case report and review. Indian J Clin Biochem. 2011 Apr;26(2):214-6. doi: 10.1007/s12291-011-0116-4. Epub 2011 Feb 4. PMID: 22468053; PMCID: PMC3107414.	22468053	2011	Department of Biochemistry, Narayana Medical College, Nellore, AP India.	494	16 Years	Female	C	Bardet-Biedl Syndrome	NA	NA	NA	NA
US													
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	1	4 Years	Male	C	Retinitis Pigmentosa and profound hearing loss	Clinical Exome	MYO7A	Indel	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	2	19 Years	Male	NA	Usher Syndrome	NA	CA4DMXL2	Missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	2	19 Years	Male	NA	Usher Syndrome	NA	HOMER2	Missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	2	19 Years	Male	NA	Usher Syndrome	NA	MYH14	Missense	NA

US 1	Kumar A, Babu M, Kimberling WJ, Venkatesh CP. Genetic analysis of a four generation Indian family with Usher syndrome: a novel insertion mutation in MYO7A. Mol Vis. 2004 Nov 24;10:910-6. PMID: 15592175.	15592175	2004	Department of Molecular Reproduction, Development, and Genetics, Indian Institute of Science, Bangalore, India	3	Variable	Male and Female Both	C	Usher Syndrome	Microsatellite Marker Based PCR and Sequencing (PCR-SSCP)	MYO7A	Insertion Frameshift	Novel
US 2	Jain PK, Lalwani AK, Li XC, Singleton TL, Smith TN, Chen A, Deshmukh D, Verma IC, Smith RJ, Wilcox ER. A gene for recessive nonsyndromic sensorineural deafness (DFNB18) maps to the chromosomal region 11p14-p15.1 containing the Usher syndrome type 1C gene. Genomics. 1998 Jun 1;50(2):290-2. doi: [illegible]	9653658	1998	National Institute on Deafness and Other Communication Disorders, National Institutes of Health, Rockville, Maryland 20850-3227, USA.	4	NA	NA	C	Usher Syndrome	Haplotype analysis	DFNB18 on chromosome 11p14-p15.1	NA	NA
US 3	Singanamalla B, Bhagwat C, Madaan P, Nayak G, Saini L. An Infant with Motor Delay and Deafness Due to Usher Syndrome. Indian J Pediatr. 2021 Aug;88(8):827. doi: 10.1007/s12098-021-03799-1. Epub 2021 May 31. PMID: 34057602.	34057602	2021	Post Graduate Institute of Medical Education & Research (PGIMER), Chandigarh, 160012, India	5	11 month	Female	NA	Usher Syndrome	Next-generation sequencing	MYO7A	Missense	NA
US 4	Parameswarappa DC, Das AV, Doctor MB, Natarajan R, Agarwal K, Jalali S. Retinitis pigmentosa in Usher syndrome in India: Electronic medical records driven big data analytics: Report III. Indian J Ophthalmol. 2022 Jul;70(7):2540-2545. doi: 10.4103/ijo.IJO_2272_21. PMID: 35791152; PMCID: PMC9426063.	35791152	2022	1 Srimati Kanuri Santamma Centre for Vitreoretinal Diseases, Hyderabad, Telangana, India. 2 Department of EyeSmart EMR & Aeye, L V Prasad Eye Institute, Hyderabad, Telangana, India. 3 Standard Charted Eye Care Education, Hyderabad, Telangana, India. 4 Department of Ophthalmic Biophysics, L V Prasad Eye Institute, Hyderabad, Telangana, India.	6-406	27 Years (Median Age)	246 Males and 155 Females	NC	Usher Syndrome	NA	NA	NA	NA
US 5	Pehere NK, Khanna RC, Marlapati R, Sannapaneni K. Prevalence of ophthalmic disorders among hearing-impaired school children in Guntur district of Andhra Pradesh. Indian J Ophthalmol. 2019 Apr;67(4):530-535. doi: 10.4103/ijo.IJO_995_18. PMID: 30900588; PMCID: PMC6446638.	30900588	2019	1 The David Brown Children's Eye Care Centre, L V Prasad Eye Institute, Vijayawada, Andhra Pradesh, India. 2 Gullapalli Pratibha Rao International Centre for Advancement in Rural Eye Care (GPR-ICARE), L V Prasad Eye Institute, Hyderabad, Andhra Pradesh, India. 3 Paramedical Ophthalmic Officer, NPCB Area Hospital, Narsaraopeta, Guntur, Andhra Pradesh, India.	407-808	6 Years to 16 Years	239 Males, 163 Females	Mix of Consanguinous and Non [illegible]	Usher Syndrome	NA	NA	NA	NA
US 6	Nair G, Dham R, Sekhar A, Kumar RS, Kameswaran M. Cochlear Implantation in Children with Usher's Syndrome: A South Asian Experience. Indian J Otolaryngol Head Neck Surg. 2020 Mar;72(1):140-144. doi: 10.1007/s12070-019-01759-y. Epub 2019 Nov 7. PMID: 32158671; PMCID: PMC7040150.	32158671	2020	1 Government Medical College, Ernakulam, Kochi, Kerala India. 2 Madras ENT Research Foundation, Chennai, Tamil Nadu India.	809-835	1-6 Years	18 Males and 9 Females	NA	Usher Syndrome	NA	NA	NA	NA
US 7	Kulkarni SS, Karkhanis VS, Joshi JM. Usher's syndrome: Can primarily be a primary ciliary disorder? Lung India. 2014 Jul;31(3):301-2. doi: 10.4103/0970-2113.135791. PMID: 25125827; PMCID: PMC4129612.	25125827	2014	Department of Pulmonary Medicine, T. N. Medical College, B.Y.L. Nair Hospital, Mumbai, Maharashtra, India.	836	20 Years	Male	NC	Usher Syndrome	NA	NA	NA	NA
US 8	Praharaj SK, Acharya M, Sarvanan A, Kongasseri S, Behere RV, Sharma PS. Tip II Usher sendromu ile ilişkili mani [Mania associated with Usher syndrome type II]. Turk Psikiyatri Derg. 2012 Fall;23(3):219-21. Turkish. PMID: 22949292.	22949292	2012	Department of Psychiatry, Kasturba Medical College, Manipal, Karnataka, India	837	30 Years	Male	NA	Usher Syndrome	NA	NA	NA	NA
US 9	Puthalath A, Samanta R, Saraswat N, Agrawal A, Singh A, Jamil M. A rare case of type 3 usher syndrome with bilateral cystoid macular edema treated with topical dorzolamide. Taiwan J Ophthalmol. 2020 Apr 21;11(2):183-186. doi: 10.4103/tjo.tjo_6_20. PMID: 34295626; PMCID: PMC8259521.	34295626	2020	Department of Ophthalmology, All India Institute of Medical Sciences (AIIMS), Rishikesh, Uttarakhand, India.	838	30 Years	Female	NC	Usher Syndrome	NA	NA	NA	NA
US 10	Prakash V, Hazra DK, Srivastava SK. Usher's syndrome (a case report). J Assoc Physicians India. 1974 Mar;22(3):285-7. PMID: 4844709.	4844709	1974	NA	839	NA	NA	NA	Usher Syndrome	NA	NA	NA	NA
US 11	Rani A, Pal N, Azad RV, Sharma YR, Chandra P, Vikram Singh D. Tractional retinal detachment in Usher syndrome type II. Clin Exp Ophthalmol. 2005 Aug;33(4):436-7. doi: 10.1111/j.1442-9071.2005.01014.x. PMID: 16033369.	16033369	2005	Vitreo-Retina Services, Dr Rajendra Prasad Centre for Ophthalmic Sciences, All India Institute of Medical Sciences, New Delhi, India.	840	50 Years	Male	NA	Usher Syndrome	NA	NA	NA	NA
US 12	Murthy R, Honavar SG. Secondary vasoproliferative retinal tumor associated with Usher syndrome type 1. J AAPOS. 2009 Feb;13(1):97-8. doi: 10.1016/j.jaapos.2008.07.012. Epub 2008 Nov 20. PMID: 19022692.	19022692	2009	Ocular Oncology, Oculoplasty and Orbital Diseases, LV Prasad Eye Institute, Hyderabad, India.	841	7 Years	Male	C	Usher Syndrome	NA	NA	NA	NA
AS													
AS 2	Das Bhowmik A, Gupta N, Dalal A, Kabra M. Whole exome sequencing identifies a homozygous nonsense variation in ALMS1 gene in a patient with syndromic obesity. Obes Res Clin Pract. 2017 Mar-Apr;11(2):241-246. doi: 10.1016/j.orcp.2016.09.004. Epub 2016 Sep 21. PMID: 27665122.	27665122	2017	Diagnostics Division, Centre for DNA Fingerprinting and Diagnostics, Hyderabad, India	1	4 year 9 month	Male	C	Alstrom Syndrome	Whole Exome Sequencing	ALMS1	Nonsense	NA
AS 3	Nerakh G, Ranganath P. Alström Syndrome Presenting as Isolated Dilated Cardiomyopathy. Indian J Pediatr. 2019 Mar;86(3):296-298. doi: 10.1007/s12098-018-2807-9. Epub 2018 Nov 28. PMID: 30484169.	30484169	2019	Diagnostics Division, Centre for DNA Fingerprinting and Diagnostics, Hyderabad, Telangana, India	2	2 month	Male	7* C	Alstrom Syndrome	Illumina sequencing platform (Illumina Inc., San Diego, California, United States)	ALMS1	Nonsense	NA
AS 4	Rajan SK, Ameen KH, Hariharan C, Natarajhan VS. Alstrom syndrome. J Assoc Physicians India. 2002 Feb;50:278. PMID: 12038666.	12038666	2002	Department of Medicine, Stanley Medical College and Hospital, Chennai.	NA	NA	NA	NA	Alstrom Syndrome	NA	NA	NA	NA
AS 5	Garg RA, Singh J, Mathur BB. Alstrom syndrome. Indian Pediatr. 1991 Jul;28(7):799-801. PMID: 1800358.port-	1800358	1991	Department of Pediatrics and General Medicine, Holy Family Hospital, New Delhi.	NA	NA	NA	NA	Alstrom Syndrome	NA	NA	NA	NA
AS 6	Joshi AS, Mohite AR, Varthakavi PK, Rathi PM. Alström Syndrome with Portal Hypertension. J Assoc Physicians India. 2016 Oct;64(10):92-93. PMID: 27766814.	27766814	2016	1. Department of Endocrinology. 2 Department of Gastroenterology. 3 Department of Endocrinology. 4 Department of Gastroenterology, Topiwala National Medical College and BYL Nair Ch Hospital, Mumbai, Maharashtra.	3	NA	NA	NA	Type II diabetes mellitus and portal hypertension, Unilateral anorchia with hypergonadotropic hypogonadism	NA	NA	NA	NA
AS 7	Wani IY, Ganie A, Mehraj I, Wani YY, Naik M. Alström syndrome: A rare association of retinitis pigmentosa with insulin resistance syndrome. Indian J Endocrinol Metab. 2013 Sep;17(5):949-50. doi: 10.4103/2230-8210.117211. PMID: 24083195; PMCID: PMC3784897.	24083195	2013	Department of Internal Medicine, SKIMS MCH, Bemina, Srinagar, J and K, India.	4	15 Year	Female	NA	Type 2 Diabetes Mellitus, Abnormal hormonal profile, RP with Alstrom	NA	NA	NA	NA
AS 8	Ahmad A, D'Souza B, Yadav C, Agarwal A, Kumar A, Nandini M, D'Souza V, Poornima AM, Kamath N. Metabolic Syndrome in Childhood: Rare Case of Alstrom Syndrome with Blindness. Indian J Clin Biochem. 2016 Oct;31(4):480-2. doi: 10.1007/s12291-015-0543-8. Epub 2015 Dec 28. PMID: 27605748; PMCID: PMC4992483.	27605748	2016	1 Department of Biochemistry, Center for Basic Sciences, Kasturba Medical College, Manipal University, Bejai, Mangalore, Karnataka 575004 India. 2 Kanachur Institute of Medical Sciences, Deralakatte, Karnataka India. 3 Department of Pediatrics, Kasturba Medical College, Manipal University, Mangalore, India.	5	9 Year	Male	C	Polyphagia, polyuria, polydipsia, generalized weakness, Type 2 Diabetes Mellitus	NA	NA	NA	NA
AS 9	Tiwari A, Awasthi D, Tayal S, Ganguly S. Alstrom syndrome: A rare genetic disorder and its anaesthetic significance. Indian J Anaesth. 2010 Mar;54(2):154-6. doi: 10.4103/0019-5049.63628. PMID: 20661355; PMCID: PMC2900742.	20661355	2010	Department of Anaesthesiology and Critical Care, St. Stephens Hospital, Delhi - 110 054, India.	6	12 Year	Male	NA	Alstrom Syndrome with Gastro-oesophageal reflux disorder (GERD) grade III with gastropathy, hiatus hernia, hypothyroidism, acanthosis	NA	NA	NA	NA

AS 10	Bronson SC, Anand Moses CR, Periyandavar I, Dharmarajan P, Suresh E, Shanmugam A, Vasuki R, Venkatesh D, Amudha J. Diabetes in the young - a case of Alström syndrome with myopathy. J R Coll Physicians Edinb. 2015 Mar;45(1):33-7. doi: 10.4997/JRCPE.2015.108. PMID: 25874828.	25874828	2015	SC Bronson, Institute of Diabetology, Madras Medical College and Rajiv Gandhi Government General Hospital, Chennai 600003, Tamil Nadu, India. Email dr.s.charlesb@gmail.com.	7	NA	NA	NA	Recurrent, severe, steroid responsive myopathy, Alstrom	NA	NA	NA	NA
Unclassified Clinical diagnosis													
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	1	36 Years	M	No	Retinitis Pigmentosa, atrophic macular changes.	Clinical Exome	PROM1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	2	22 Years	M	No	Cone dystrophy?	Clinical Exome	GUCY2D	Substitution	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	3	6 Years	F	Yes	Leber congenital amaurosis or severe early childhood onset retinal dystrophy	Clinical Exome	CRB1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	4	6 Years	M	Yes	Suspected Cone dystrophy?	Clinical Exome	CNGA3	Indel FS	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	5	6 Years	M	Yes	Leber congenital amaurosis?	Clinical Exome	GUCY2D	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	6	24 Years	M	No	Cone rod dystrophy or Stargardt disease	Clinical Exome	PROM1	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	7	59 Years	M	No	Retinitis pigmentosa? or Choroideremia	Clinical Exome	CHM	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	8	2 Years	M	Yes	Rod cone dystrophy/ Leber congenital amaurosis	Clinical Exome	RPGRIP1	Nonsense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	9	3 Years	F	No	Rod-cone dystrophy?	Clinical Exome	LRAT	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	10	3 Years	M	No	Rod-cone dystrophy or Familial exudative vitreoretinopathy	Clinical Exome	NR2E3	Indel	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	11	4 Years	F	Yes	Unspecified hereditary retinal dystrophy	Clinical Exome	GUCY2D	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	12	47 Years	M	Yes	Cone rod dystrophy or Retinitis pigmentosa or Stargardt disease	Clinical Exome	C8orf37	3' splice site	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	13	15 Years	M	Yes	Leber congenital amaurosis / Retinitis pigmentosa	Clinical Exome	AIPL1	Deletion	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	14	10 Years	F	Yes	Unspecified hereditary retinal dystrophy	Clinical Exome	LCA5	Nonsense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	15	9 Years	F	Yes	Unspecified hereditary retinal dystrophy	Clinical Exome	AIPL1	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	16	2 Years	F	No	Cone-rod dystrophy or Leber congenital amaurosis	Clinical Exome	RPGRIP1	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	16	2 Years	F	No	Cone-rod dystrophy or Leber congenital amaurosis	Clinical Exome	RPGRIP1	Non-sense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	17	36 Years	M	Yes	Atypical retinitis pigmentosa or cone-rod dystrophy	Clinical Exome	MFSD8	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	18	2 Years	F	Yes	Unspecified Hereditary retinal dystrophy	Clinical Exome	ALMS1	Deletion FS	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	19	6 Months	M	Yes	Retinal dystrophy	Clinical Exome	CRX	Duplication	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	20	12 Years	F	No	Unspecified hereditary retinal dystrophy	Clinical Exome	ABCA4	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	20	12 Years	F	No	Unspecified hereditary retinal dystrophy	Clinical Exome	ABCA4	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	21	3 Years	F	Yes	Cone rod dystrophy or Rod cone dystrophy	Clinical Exome	PROM1	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	21	3 Years	F	Yes	Cone rod dystrophy or Rod cone dystrophy	Clinical Exome	PRPH2	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	21	3 Years	F	Yes	Cone rod dystrophy or Rod cone dystrophy	Clinical Exome	CA4	3' UTR variation	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	22	23 Years	F	No	Occult macular dystrophy?	Clinical Exome	NRL	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	23	20 Years	M	No	Cone-rod dystrophy or Atypical RP	Clinical Exome	RPGRIP1	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	24	11 Years	F	No	Macular dystrophy	Clinical Exome	PDE6A	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	24	11 Years	F	No	Macular dystrophy	Clinical Exome	ABCA4	5' splice site	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	25	25 Years	M	No	Retinitis pigmentosa or choroideremia	Clinical Exome	PITPNM3	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	26	7 Years	M	No	Rod-cone dystrophy or congenital stationary night blindness	Clinical Exome	TTC8	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	27	8 Years	F	Yes	Rod cone dysfunction or Familial exudative vitreoretinopathy	Clinical Exome	TTC8	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	28	17 years	M	Yes	Rod-cone dystrophy or macular dystrophy	Clinical Exome	PCDH15	Missense	NA

IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	29	4 Years	F	Yes	Advanced retinal dystrophy/ Rod cone dystrophy	Clinical Exome	RPGR	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	30	4 Years	F	Yes	Stargardt disease or Rod-cone dystrophy?	Clinical Exome	IFT140	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	31	9 Years	F	Yes	Pigmentary retinal dystrophy or cone dystrophy	Clinical Exome	CNGA3	Missense	Novel
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	31	9 Years	F	Yes	Pigmentary retinal dystrophy or cone dystrophy	Clinical Exome	CABP4	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	32	8 years	F	Yes	CSNB/ESCS	Clinical Exome	GRM6	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	33	5 years	M	No	Cone dystrophy?	Clinical Exome	No varients identified	NA	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	34	1 year	F	Yes	CRD?LCA?	Clinical Exome	CEP290	Missense	NA
IRD 1	Inherited retinal disorders: a genotype-phenotype correlation in an Indian cohort and the importance of genetic testing and genetic counselling.	36648511	2022	Narayana Netralaya Foundataion	35	1 year	F	No	Inherited retinal dystrophy, Rods and cones affected		No varients identified	NA	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	36	3	M	NA	?LCA	TP	*GUCY2D*	splice site	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	37	24	F	NA	?Occult macular dystrophy	TP	*NRL*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	38	51	M	NA	?Retinitis pigmentosa ?Choroideremia	TP	*PDE6B*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	39	28	F	NA	?Familial exudative vitreoretinopathy ?Noorie disease	TP	*FZD4*	deletion FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	40	15	F	NA	?Stargardt disease	TP	*ABCA4*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	40	15	F	NA			*ABCA4*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	41	8	M	NA	?Cone dystrophy ?Stargardt disease	TP	*ABCA4*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	42	8	M	NA			*ABCA4*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	42	8	M	NA			*ABCA4*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	42	8	M	NA			*ABCA4*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	43	11	M	NA	?BBS	TP	*BBS10*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	43	11	M	NA			*CEP290*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	44	6	M	NA	?LCA	TP	*SPATA7*	insertion FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	45	7	M	NA	?LCA	TP	*GUCY2D*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	46	26	M	NA	?retinitis pigmentosa ?choroideremia	TP	*CHM*	gene deletion	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	46	26	M	NA	?retinitis pigmentosa ?choroideremia	TP	*PITPNM3*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	47	9	F	NA	?rod cone dysfunction ?familial exudative vitreoretinopathy	TP	*TTC8*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	48	6	F	NA	?LCA ?Early onset retinal dystrophy	TP	*CRB1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	49	59	M	NA	?retinitis pigmentosa ?choroideremia	TP	*CHM*	tri-exon deletion	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	50	22	M	NA	?Rod cone dystrophy	TP	*CRB1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	50	22	M	NA	?Rod cone dystrophy	TP	*CRB1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	51	0.83	F	NA	?LCA	TP	*USH2A*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	51	0.83	F	NA	?LCA	TP	*USH2A*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	51	0.83	F	NA	?LCA	TP	*CEP290*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	55	2	M	NA	?LCA	TP	*RPGRIP1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	55	2	M	NA	?LCA	TP	*CRB1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	57	7	M	NA	?Rod cone dystrophy ?CSNB	TP	*TTC8*	missense	NA

IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	59	61	M	NA	Gradual blindness, "FEVR/Noories"	TP	*FZD4*	deletion FS	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	61	25	F	NA	?Cone rod dystrophy ?RP	TP	*PROM1*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	62	9	F	NA	?Cone dystrophy	TP	*CRX*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	63	24	M	NA	?Stargardt/Cone dystrophy	WES	*GUCY2D*	missense	NA
IRD 2	Prevalence of mutations in inherited retinal diseases: A comparison between the United States and India.	31816670	2020	Narayana Nethralaya /Medgenome	64	13	F	NA	?Stargardt disease	WES	*CDH3*	missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	65	9 months/male	M	NA	Congenital stationary night blindness/LCA	exome sequencing/panel-based sequencing/chromosomal microarray	GUCY2D	missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	66	9 months/male	M	NA	Congenital muscle dystrophy? Walker Warburg syndrome	exome sequencing/panel-based sequencing/chromosomal microarray	POMT2	missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	67	10 years/male	M	NA	?Neurometabolic disorder ?Ciliopathy ?Mitochondrial disorder B/L retinitis pigmentosa a/w failure to thrive, chronic kidney disease, GH deficiency and hypothyroidism, leukoencephalopathy with basal	exome sequencing/panel-based sequencing/chromosomal microarray	IFT 172 PRPF8	missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	68	10 years/female	F	NA	Single gene/chromosomal deletion/duplication syndrome	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	69	5 months/female	F	NA	Single gene/chromosomal deletion/duplication syndrome	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	70	6 months/male	M	NA	Chromosomal deletion/duplication syndrome or ?Goldenhar syndrome	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	71	2 months/female	F	NA	Chromosomal deletion/duplication	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	72	10 months/female	F	NA	Single gene disorder ? Kenny-Caffey syndrome type 2	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	73	6 weeks/female	F	NA	Galactosemia, ?TORCH infection	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	74	2 days/female	F	NA	Binders phenotype (?genetic/?nongenetic)	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	75	6 weeks/male	M	NA	Chromosome deletion/duplication/single-gene disorder	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	76	1 year/female	F	NA	Ciliopathy	exome sequencing/panel-based sequencing/chromosomal microarray	IFT 140	missense	NA

IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	77	3 weeks/female	F	NA	Single-gene disorder ?Cerebro-oculo-facio-skeletal syndrome	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	78	2 years/male	M	NA	Single-gene disorder/chromosomal deletion/duplication syndrome	exome sequencing/panel-based sequencing/chromosomal microarray	Chromosome 6 microdeletion /Chromosome 17 duplication		NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	79	2 years/female	F	NA	Isolated eye abnormality	exome sequencing/panel-based sequencing/chromosomal microarray	CYP1B1	missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	80	10 years/male	M	NA	Isolated eye anomaly	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	81	6 weeks/male	M	NA	Single-gene disorder ?Galactosemia ?Mitochondrial disorder	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	82	11 years/male	M	NA	Single-gene disorder ?Galactosemia	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	83	8 months/female	F	NA	Familial microphthalmia (?isolated/syndromic)	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	84	9 years/male	M	NA	Single-gene disorder	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	85	26 years/male	M	NA	Isolated eye abnormality	exome sequencing/panel-based sequencing/chromosomal microarray	PITX2	duplication	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	86	4 years/male	M	NA	Single-gene disorders ?Stickler syndrome	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	87	1 month/male	M	NA	Isolated eye abnormality	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	88	7 years/male	M	NA	Isolated eye abnormality	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	89	7 months/male	M	NA	Isolated eye abnormality	exome sequencing/panel-based sequencing/chromosomal microarray	FOXC1	exon deletion	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	90	11 months/female	F	NA	Isolated eye abnormality	exome sequencing/panel-based sequencing/chromosomal microarray	NA	NA	NA

IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	91	2 years/male	M	NA	Isolated eye abnormality	exome sequencing/panel-based sequencing/chromosomal microarray	FOXE3	missense	NA
IRD 3	Spectrum of congenital and inherited ocular disorders seen in a genetic clinic: Experience of a developing ocular genetic service.	36872713	2023	Advanced Pediatrics Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India Advanced Eye Centre, Post Graduate Institute of Medical Education and Research (PGIMER), Chandigarh, India	92	29 years/female	F	NA	Isolated eye abnormality	exome sequencing/panel-based sequencing/chromosomal microarray	RBP4	missense	NA
IRD 4	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis.	34662339	2021	Shiley Eye Institute, University of California San Diego, La Jolla, California, United States of America; School of Biotechnology, REVA University, Bengaluru, Karnataka, India; Retina and Genomics Institute, Yucatán, México; Laboratoire de Diagnostic Moleculaire, Hôpital Maisonneuve Rosemont, Montreal, Quebec, Canada; Ophthalmology, University of California San Francisco, San Francisco, California, United States of America; 6 Institute for Genomic Medicine, University of California, San Diego, La Jolla, California, United States of America; 7 GeneDx, Gaithersburg, Maryland, United States of America; 8 The Wilmer Eye Institute, Johns Hopkins University School of Medicine, Baltimore, Maryland, United States of America; 9 Ophthalmology & Visual Science, University of Michigan Kellogg Eye Center, Ann Arbor, Michigan, United States of America; 10 Ophthalmology, University of Arizona College of Medicine Phoenix, Phoenix, Arizona, United States of America; 11 Byers Eye Institute, Stanford, Palo Alto, California, United States of America; 12 Genetics and Ophthalmology, Genelabor, Goiânia, Brazil; 13 School of Regenerative Medicine, Manipal University, Bengaluru, Karnataka, India; 14 Casey Eye Institute, Oregon Health & Science University, Portland, Oregon, United States of America; 15 Ophthalmic Genetics and Visual Function Branch, National Eye Institute, National Institutes of Health, Bethesda, Maryland, United States of America; 16 National Centre of Excellence in Molecular Biology, University of the Punjab, Lahore, Pakistan; 17 Allama Iqbal Medical College, University of Health Sciences, Lahore, Pakistan; 18 National Eye Institute, Bethesda, Maryland, United States of America; 19 Ophthalmology & Vision Science, UC Davis Medical Center, California, United States of America; 20 Department of Pediatrics, Rady Children's Hospital, Division of Genome Information Sciences, San Diego, California, United States of America; Case Western Reserve University, UNITED STATES	93	NA	F	NC	unclassified	targeted gene(s) or mutation(s) screening, linkage analysis and exome sequencing	PRPH2	missense	known
IRD 4	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis.	34662339	2021	Shiley Eye Institute, University of California San Diego, La Jolla, California, United States of America; School of Biotechnology, REVA University, Bengaluru, Karnataka, India; Retina and Genomics Institute, Yucatán, México; Laboratoire de Diagnostic Moleculaire, Hôpital Maisonneuve Rosemont, Montreal, Quebec, Canada; Ophthalmology, University of California San Francisco, San Francisco, California, United States of America; 6 Institute for Genomic Medicine, University of California, San Diego, La Jolla, California, United States of America; 7 GeneDx, Gaithersburg, Maryland, United States of America; 8 The Wilmer Eye Institute, Johns Hopkins University School of Medicine, Baltimore, Maryland, United States of America; 9 Ophthalmology & Visual Science, University of Michigan Kellogg Eye Center, Ann Arbor, Michigan, United States of America; 10 Ophthalmology, University of Arizona College of Medicine Phoenix, Phoenix, Arizona, United States of America; 11 Byers Eye Institute, Stanford, Palo Alto, California, United States of America; 12 Genetics and Ophthalmology, Genelabor, Goiânia, Brazil; 13 School of Regenerative Medicine, Manipal University, Bengaluru, Karnataka, India; 14 Casey Eye Institute, Oregon Health & Science University, Portland, Oregon, United States of America; 15 Ophthalmic Genetics and Visual Function Branch, National Eye Institute, National Institutes of Health, Bethesda, Maryland, United States of America; 16 National Centre of Excellence in Molecular Biology, University of the Punjab, Lahore, Pakistan; 17 Allama Iqbal Medical College, University of Health Sciences, Lahore, Pakistan; 18 National Eye Institute, Bethesda, Maryland, United States of America; 19 Ophthalmology & Vision Science, UC Davis Medical Center, California, United States of America; 20 Department of Pediatrics, Rady Children's Hospital, Division of Genome Information Sciences, San Diego, California, United States of America; Case Western Reserve University, UNITED STATES	93	NA	F	NC	unclassified	targeted gene(s) or mutation(s) screening, linkage analysis and exome sequencing	ROM1	duplication FS	known
IRD 4	Deciphering the genetic architecture and ethnographic distribution of IRD in three ethnic populations by whole genome sequence analysis.	34662339	2021	Shiley Eye Institute, University of California San Diego, La Jolla, California, United States of America; School of Biotechnology, REVA University, Bengaluru, Karnataka, India; Retina and Genomics Institute, Yucatán, México; Laboratoire de Diagnostic Moleculaire, Hôpital Maisonneuve Rosemont, Montreal, Quebec, Canada; Ophthalmology, University of California San Francisco, San Francisco, California, United States of America; 6 Institute for Genomic Medicine, University of California, San Diego, La Jolla, California, United States of America; 7 GeneDx, Gaithersburg, Maryland, United States of America; 8 The Wilmer Eye Institute, Johns Hopkins University School of Medicine, Baltimore, Maryland, United States of America; 9 Ophthalmology & Visual Science, University of Michigan Kellogg Eye Center, Ann Arbor, Michigan, United States of America; 10 Ophthalmology, University of Arizona College of Medicine Phoenix, Phoenix, Arizona, United States of America; 11 Byers Eye Institute, Stanford, Palo Alto, California, United States of America; 12 Genetics and Ophthalmology, Genelabor, Goiânia, Brazil; 13 School of Regenerative Medicine, Manipal University, Bengaluru, Karnataka, India; 14 Casey Eye Institute, Oregon Health & Science University, Portland, Oregon, United States of America; 15 Ophthalmic Genetics and Visual Function Branch, National Eye Institute, National Institutes of Health, Bethesda, Maryland, United States of America; 16 National Centre of Excellence in Molecular Biology, University of the Punjab, Lahore, Pakistan; 17 Allama Iqbal Medical College, University of Health Sciences, Lahore, Pakistan; 18 National Eye Institute, Bethesda, Maryland, United States of America; 19 Ophthalmology & Vision Science, UC Davis Medical Center, California, United States of America; 20 Department of Pediatrics, Rady Children's Hospital, Division of Genome Information Sciences, San Diego, California, United States of America; Case Western Reserve University, UNITED STATES	94	NA	M	NC	unclassified	targeted gene(s) or mutation(s) screening, linkage analysis and exome sequencing	PROM1	missense	known
IRD 5	Derivation of three induced pluripotent stem cell lines under feeder-free culture conditions from peripheral blood mononuclear cells (PBMC) of Indian patients suffering from inherited retinal diseases carrying different mutations.	32278301	2020	Eyestem Research Private Limited, C-CAMP, GKVK Post, Bangalore 560065, Karnataka, India. Centre for Eye Genetics and Research, Cytecare Hospital, Bangalore 560064, Karnataka, India. Eyestem Research Private Limited, C-CAMP, GKVK Post, Bangalore 560065, Karnataka, India.	95	11	M	NA	LCA/RP	NA	RDH12	nonsense	NA

IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	96	20	M	C	LCA/RP	Homozygosity mapping guided NGS	RDH12	missense	Novel
IRD 6	Homozygosity mapping guided next generation sequencing to identify the causative genetic variation in inherited retinal degenerative diseases.	27383656	2015	SNONGC Department of Genetics and Molecular Biology, Vision Research Foundation, Chennai, India. PhD Scholar, Birla Institute of Technology & Science (BITS), Hyderabad, India. Departments of Paediatric Ophthalmology and Strabismus, Chennai, India. Vitreo-Retinal Services, Medical Research Foundation, Chennai, India. Strand Center for Genomics and Personalized Medicine, Strand Life Sciences, Bengaluru, India.	96	20	M	C	LCA/RP	Homozygosity mapping guided NGS	ABCA4	missense	NA
IRD 7	A frameshift mutation in prominin (mouse)-like 1 causes human retinal degeneration.	10587575	2000	Biochemistry Department, University of Otago, PO Box 56, Dunedin, New Zealand. marion.maw@stonebow.otago.ac.nz	97	NA	F	Y	Retinal degeneration		PROM1	NA	NA
LCA 4	Rare case of dual diagnosis in consanguineous family: a case report.	34016806	2021	Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, India.	98	11	M	Yes	severe achondroplasia or acromesomelic dysplasia with blindmess, suggestive underlying	Clinical exome	NPR2	Missense	NA
LCA 4	Rare case of dual diagnosis in consanguineous family: a case report.	34016806	2021	Department of Medical Genetics, Sanjay Gandhi Postgraduate Institute of Medical Sciences, Lucknow, India.	98	11	M	Yes	severe achondroplasia or acromesomelic dysplasia with blindmess, suggestive underlying disorder causing cortical blindness	Clinical exome	LCA5	deletion FS	NA
LCA 16	Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration.	17186464	2006	From the Departments of Ophthalmology and Visual Sciences (J.S.F.; K.B.; M.O.; E.F.; D.A.T.; J.R.H. A.S.) and Human Genetics (A.S.), W. K. Kellogg Eye Center, University of Michigan, Ann Arbor; the Jackson Laboratory, Bar Harbor, Maine (B.C.; N.L.H.); Kallam Anji Reddy Molecular Genetics Laboratory (C.K.; H.P.S.) and Kannuri Santhamma Retina-Vitreous Services (S.J.), L. V. Prasad Eye Institute, Banjara Hills, Hyderaba, India; Department of Molecular Genetics, Institute of Ophthalmology, University College London (C.C.; A.R.W.; S.S.B.), and Moorfield Eye Hospital (A.R.W.), London; Department of Ophthalmology, University Hospital of Lund, Lund, Sweden (S.A.); and Department of Ophthalmology, Scheie Eye Institute, University of Pennsylvania, Philadelphia (S.G.J.)	99	12	NA	NA	Retinal dystrophy	Mutation screening	RD3	missense	Novel gene
LCA 16	Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration.	17186464	2006	From the Departments of Ophthalmology and Visual Sciences (J.S.F.; K.B.; M.O.; E.F.; D.A.T.; J.R.H. A.S.) and Human Genetics (A.S.), W. K. Kellogg Eye Center, University of Michigan, Ann Arbor; the Jackson Laboratory, Bar Harbor, Maine (B.C.; N.L.H.); Kallam Anji Reddy Molecular Genetics Laboratory (C.K.; H.P.S.) and Kannuri Santhamma Retina-Vitreous Services (S.J.), L. V. Prasad Eye Institute, Banjara Hills, Hyderaba, India; Department of Molecular Genetics, Institute of Ophthalmology, University College London (C.C.; A.R.W.; S.S.B.), and Moorfield Eye Hospital (A.R.W.), London; Department of Ophthalmology, University Hospital of Lund, Lund, Sweden (S.A.); and Department of Ophthalmology, Scheie Eye Institute, University of Pennsylvania, Philadelphia (S.G.J.)	100	13	NA	NA	Retinal dystrophy	Mutation screening	RD3	missense	Novel gene
LCA 16	Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration.	17186464	2006	From the Departments of Ophthalmology and Visual Sciences (J.S.F.; K.B.; M.O.; E.F.; D.A.T.; J.R.H. A.S.) and Human Genetics (A.S.), W. K. Kellogg Eye Center, University of Michigan, Ann Arbor; the Jackson Laboratory, Bar Harbor, Maine (B.C.; N.L.H.); Kallam Anji Reddy Molecular Genetics Laboratory (C.K.; H.P.S.) and Kannuri Santhamma Retina-Vitreous Services (S.J.), L. V. Prasad Eye Institute, Banjara Hills, Hyderaba, India; Department of Molecular Genetics, Institute of Ophthalmology, University College London (C.C.; A.R.W.; S.S.B.), and Moorfield Eye Hospital (A.R.W.), London; Department of Ophthalmology, University Hospital of Lund, Lund, Sweden (S.A.); and Department of Ophthalmology, Scheie Eye Institute, University of Pennsylvania, Philadelphia (S.G.J.)	101	14	NA	NA	Retinal dystrophy	Mutation screening	RD3	missense	Novel gene
LCA 16	Premature truncation of a novel protein, RD3, exhibiting subnuclear localization is associated with retinal degeneration.	17186464	2006	From the Departments of Ophthalmology and Visual Sciences (J.S.F.; K.B.; M.O.; E.F.; D.A.T.; J.R.H. A.S.) and Human Genetics (A.S.), W. K. Kellogg Eye Center, University of Michigan, Ann Arbor; the Jackson Laboratory, Bar Harbor, Maine (B.C.; N.L.H.); Kallam Anji Reddy Molecular Genetics Laboratory (C.K.; H.P.S.) and Kannuri Santhamma Retina-Vitreous Services (S.J.), L. V. Prasad Eye Institute, Banjara Hills, Hyderaba, India; Department of Molecular Genetics, Institute of Ophthalmology, University College London (C.C.; A.R.W.; S.S.B.), and Moorfield Eye Hospital (A.R.W.), London; Department of Ophthalmology, University Hospital of Lund, Lund, Sweden (S.A.); and Department of Ophthalmology, Scheie Eye Institute, University of Pennsylvania, Philadelphia (S.G.J.)	102	15	NA	C	Retinal dystrophy	Mutation screening	RD3	splice site	Novel gene
LCA 17	Electroretinographic assessment and diagnostic reappraisal of children with visual dysfunction: a prospective study.	17322600	2007	Department of Retina-Vitreous Services, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamil Nadu, India. drvasumathy@yahoo.com	103	4.4+/- 3.2	4 M/ 5 F	NA	CSNB	NA	NA	NA	NA
LCA 17	Electroretinographic assessment and diagnostic reappraisal of children with visual dysfunction: a prospective study.	17322600	2007	Department of Retina-Vitreous Services, Aravind Eye Hospital and Postgraduate Institute of Ophthalmology, Madurai, Tamil Nadu, India. drvasumathy@yahoo.com	104	4.4+/- 3.2	19 M/ 20 F	NA	Others	NA	NA	NA	NA

Index	
C	Consanguineous
NC	Non-consanguineous
NA	Not available
ND	Not detected
N	No
Y	Yes
R	Reported
M	Male

F	Female
TP	Targeted panel
WES	Whole exome sequencing
CES	Clinical exome sequencing