Online Resources

Time for disease-specific patient-reported outcome measures: Systematic review on outcomes reported in clinical research on nephronophthisis.
Pediatric Nephrology
Mareike Dahmer-Heath (MD-H)[1*], Sven Optenhövel (SO)[1,2*], Tanja Hechler (TH)[2], Martin Konrad (MK)[1], Jens König (JK)[1] *: authors contributed equally
1 Department of General Pediatrics, University Children's Hospital Münster, Münster, Germany
2 Department of Clinical Psychology and Psychotherapy for Children and Adolescents, University of Münster, Münster, Germany
Correspondence: Jens Christian König, Department of General Pediatrics, Universitätsklinikum Münster, Waldeyerstraße 22, 48149 Münster, Germany. E-mail: jens.koenig@ukmuenster.de

Online Resource 1: PRISMA Checklist

Section and Topic	Item #	Checklist item	Location where item is reported
TITLE			
Title	1	Identify the report as a systematic review.	1
ABSTRACT			
Abstract	2	See the PRISMA 2020 for Abstracts checklist.	
INTRODUCTION			
Rationale	3	Describe the rationale for the review in the context of existing knowledge.	
Objectives	4	Provide an explicit statement of the objective(s) or question(s) the review addresses.	
METHODS			
Eligibility criteria	5	Specify the inclusion and exclusion criteria for the review and how studies were grouped for the syntheses.	4
Information sources	6	Specify all databases, registers, websites, organisations, reference lists and other sources searched or consulted to identify studies. Specify the date when each source was last searched or consulted.	3
Search strategy	7	Present the full search strategies for all databases, registers and websites, including any filters and limits used.	3-4
Selection process	8	Specify the methods used to decide whether a study met the inclusion criteria of the review, including how many reviewers screened each record and each report retrieved, whether they worked independently, and if applicable, details of automation tools used in the process.	4
Data collection process	9	Specify the methods used to collect data from reports, including how many reviewers collected data from each report, whether they worked independently, any processes for obtaining or confirming data from study investigators, and if applicable, details of automation tools used in the process.	4

Section and Topic	Item #	Checklist item	Location where item is reported
Data items	10a	List and define all outcomes for which data were sought. Specify whether all results that were compatible with each outcome domain in each study were sought (e.g. for all measures, time points, analyses), and if not, the methods used to decide which results to collect.	n.a.
	10b	List and define all other variables for which data were sought (e.g. participant and intervention characteristics, funding sources). Describe any assumptions made about any missing or unclear information.	4
Study risk of bias assessment	11	Specify the methods used to assess risk of bias in the included studies, including details of the tool(s) used, how many reviewers assessed each study and whether they worked independently, and if applicable, details of automation tools used in the process.	5
Effect measures	12	Specify for each outcome the effect measure(s) (e.g. risk ratio, mean difference) used in the synthesis or presentation of results.	n.a.
Synthesis methods	13a	Describe the processes used to decide which studies were eligible for each synthesis (e.g. tabulating the study intervention characteristics and comparing against the planned groups for each synthesis (item #5)).	n.a.
	13b	Describe any methods required to prepare the data for presentation or synthesis, such as handling of missing summary statistics, or data conversions.	n.a.
	13c	Describe any methods used to tabulate or visually display results of individual studies and syntheses.	4-5
	13d	Describe any methods used to synthesize results and provide a rationale for the choice(s). If meta-analysis was performed, describe the model(s), method(s) to identify the presence and extent of statistical heterogeneity, and software package(s) used.	5
	13e	Describe any methods used to explore possible causes of heterogeneity among study results (e.g. subgroup analysis, meta-regression).	n.a.
	13f	Describe any sensitivity analyses conducted to assess robustness of the synthesized results.	n.a.
Reporting bias assessment	14	Describe any methods used to assess risk of bias due to missing results in a synthesis (arising from reporting biases).	n.a.
Certainty assessment	15	Describe any methods used to assess certainty (or confidence) in the body of evidence for an outcome.	5
RESULTS			
Study selection	16a	Describe the results of the search and selection process, from the number of records identified in the search to the number of studies included in the review, ideally using a flow diagram.	5-6
	16b	Cite studies that might appear to meet the inclusion criteria, but which were excluded, and explain why they were excluded.	
Study characteristics	17	Cite each included study and present its characteristics.	20-22
Risk of bias in studies	18	Present assessments of risk of bias for each included study.	23
Results of	19	For all outcomes, present, for each study: (a) summary statistics for each group (where appropriate) and (b) an effect estimate and its precision	n.a.

Section and Topic	Item #	Checklist item	Location where item is reported
individual studies		(e.g. confidence/credible interval), ideally using structured tables or plots.	
Results of syntheses	20a	For each synthesis, briefly summarise the characteristics and risk of bias among contributing studies.	
	20b	Present results of all statistical syntheses conducted. If meta-analysis was done, present for each the summary estimate and its precision (e.g. confidence/credible interval) and measures of statistical heterogeneity. If comparing groups, describe the direction of the effect.	
	20c	Present results of all investigations of possible causes of heterogeneity among study results.	n.a.
	20d	Present results of all sensitivity analyses conducted to assess the robustness of the synthesized results.	n.a.
Reporting biases	21	Present assessments of risk of bias due to missing results (arising from reporting biases) for each synthesis assessed.	n.a.
Certainty of evidence	22	Present assessments of certainty (or confidence) in the body of evidence for each outcome assessed.	
DISCUSSION			
Discussion	23a	Provide a general interpretation of the results in the context of other evidence.	
	23b	Discuss any limitations of the evidence included in the review.	
	23c	Discuss any limitations of the review processes used.	
	23d	Discuss implications of the results for practice, policy, and future research.	
OTHER INFORMATION			
Registration and protocol	24a	Provide registration information for the review, including register name and registration number, or state that the review was not registered.	
	24b	Indicate where the review protocol can be accessed, or state that a protocol was not prepared.	
	24c	Describe and explain any amendments to information provided at registration or in the protocol.	
Support	25	Describe sources of financial or non-financial support for the review, and the role of the funders or sponsors in the review.	
Competing interests	26	Declare any competing interests of review authors.	
Availability of data, code and other materials	27	Report which of the following are publicly available and where they can be found: template data collection forms; data extracted from included studies; data used for all analyses; analytic code; any other materials used in the review.	

From: Page MJ, McKenzie JE, Bossuyt PM, Boutron I, Hoffmann TC, Mulrow CD, et al. The PRISMA 2020 statement: an updated guideline for reporting systematic reviews. BMJ 2021;372:n71. doi: 10.1136/bmj.n71

Online Resource 2: Normality testing

		Kolmogorov-Smirnov[a]			Shapiro-Wilk		
	Study quality	Statistic	df	Significance	Statistic	df	Significance
SONG Core outome	good	,455	52	<,001	,574	52	<,001
	moderate	,436	28	<,001	,612	28	<,001
	poor	,370	10	<,001	,752	10	,004
SONG Middle tier outcomes	good	,231	52	<,001	,835	50	<,001
	moderate	,331	28	<,001	,717	27	<,001
	poor	,360	10	<,001	,731	10	,002
SONG Outer tier outcomes	good	,332	52	<,001	,739	52	<,001
	moderate	,365	28	<,001	,637	28	<,001
	poor	,381	10	<,001	,640	10	<,001
Clinical outcomes	good	.	50	.	.	50	.
	moderate	.	27	.	.	27	.
	poor	.	10	.	.	10	.
Surrogate outcomes	good	,531	52	<,001	,336	52	<,001
	moderate	,513	28	<,001	,419	28	<,001
	poor	,381	10	<,001	,640	10	<,001
Patient reported outcomes	good	,359	52	<,001	,635	52	<,001
	moderate	,429	28	<,001	,591	28	<,001
	poor	,381	10	<,001	,640	10	<,001

a. Lilliefors' significance correction

Online Resource 3: List of excluded studies

Less than 4 patients

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Alizadeh R, Jamshidi S, Keramatipour M, Moeinian P, Hosseini R, Otukesh H, et al. Whole Exome Sequencing Reveals a XPNPEP3 Novel Mutation Causing Nephronophthisis in a Pediatric Patient. Iranian biomedical journal. 2020;24(6):405-8. https://doi.org/10.29252/ibj.24.6.400.

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AlZabali SM, AlAnazi A, Rahim KA, Faqeehi HY. Clinical improvement of encapsulating peritoneal sclerosis after challenging course and 6 months of total parenteral nutrition in child with nephronophthisis: a case report. Journal of medical case reports. 2021;15(1):366. https://doi.org/10.1186/s13256-021-02905-3.

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Online Resource 5: Study characteristics

Author (year)	Country	Study type	N	N with NPH	Age	Gender (F:M)
Ala-Mello (1996)	Finland	Retrospective	18	6	Mean: 10.4(6.8-13.8)	2:4
Ala-Mello (1998)	Finland	Retrospective	7	7	m	2:5
Ala-Mello (1999)	Finland	Retrospective	59	59	m	m
Antignac (1993)	France	Genetic/linkage analysis	22 families	18 families	m	m
Attanasio (2007)	USA	Genetic/linkage analysis	25 kindreds	m	m	m
Avci (2022)	Turkey	Retrospective	17	17	Mean: 12.6	M
Bakkaloglu (2014)	Turkey	Retrospective	4	4	10-24	2:2
Baris (2006)	USA	Genetic/linkage analysis	7	7	18 m – 12y	3:4
Beal (2024)	England	Retrospective	71	4	m	m
Birtel (2021)	m	Retrospective	16	16	Median: 17(6-53)	3:13
Bizet (2015)	m	Genetic/linkage analysis	8	7	m	m
Blowey (1996)	m	Retrospective	11	11	Mean: 9.5(6-33)	6:5
Braun (2016)	m	Genetic/linkage analysis	103	7	m	m
Caridi (1998)	Italy	Case report	8	8	Mean: 22.8(6-33)	5:3
Caridi (2000)	Italy	Genetic/linkage analysis	68	68	m	34:34
Caridi (2006)	Italy	Genetic/linkage analysis	56	56	m	m
Chaari (2012)	Tunesia	Genetic/linkage analysis	100	100	m	38:62
Chaki (2011)	m	Genetic/linkage analysis	440	m	m	m
Chaki (2012)	m	Genetic/linkage analysis	m	m	m	m
Chen (2022)	China	Genetic/linkage analysis	6	6	Mean: 4.8(1-10)	2:4
Dahmer-Heath (2021)	Germany		75	17	Median: 18(5-75)	4:13
Doreille (2021)	France	Letter to the editors	200	18	m	m
Failler (2014)	m	Genetic/linkage analysis	1255	m	m	m
Geissari (2015)	Iran	Genetic/linkage analysis	16	16	Median: 15(12-22)	10:6
Gjerstad (2023)	Norway	Retrospective	575	16	Median: 8.3(2.7-15.8)	8:8
Green (1990)	Ireland	Retrospective	1020	12	Mean: 31(9-65)	m
Grenda (2007)	m	Prospective	303	19	Mean: 11.9	m
Gretz (1998)	Germany	Retrospective	29	29	Median: 9.6(4-18.5)	13:16
Haider (1998)	Israel	Genetic/linkage analysis	1 family	10	m	3:7
Halbritter (2012)	m	Genetic/linkage analysis	192	91	m	m

Halbritter (2013)	m	Genetic/linkage analysis	1056	447	m	m
Hamiwka (2008)	Canada, USA	Retrospective	224	224	m	112:112
Hildebrandt (1997)	Germany	Retrospective	57	39	m	m
Hoefele (2005)	m	Genetic/linkage analysis	250	190	m	m
Hoefele (2007)	m	Genetic/linkage analysis	20	20	m	m
Hoefele (2011)	Turkey	Case report	4	4	Mean: 19.5(15.6-25.5)	3:1
Hoff (2013)	m	Genetic/linkage analysis	6 families	m	m	m
Hudson (2020)	Australia	Case report	4	4	m	1:3
Hussain (2018)	Pakistan	Genetic/linkage analysis	7	3	m	m
Kang (2015)	South Korea	Retrospective	5	5	Mean: 19.5(15.6-25.5)	1:4
König (2017)	Germany, Austria, the Netherlands	Retrospective	152	77	m	m
König (2022)	m	Genetic/linkage analysis	165	m	m	m
Lee (2015)	South Korea	Genetic/linkage analysis	48	6	Median: 14(1.5-21)	3:3
Li (2023)	China	Genetic/linkage analysis	29	15	m	m
Macia (2017)	France, USA, Norway	Genetic/linkage analysis	8	8	m	m
Mashat (2015)	Saudi-Arabia	Retrospective	55	3	Mean: 9.3	2:1
Medhioub (1994)	France	Genetic/linkage analysis	23 families	m	m	m
Mehrkash (2021)	Iran	Retrospective	30	7	m	m
Mistry (2007)	USA	Genetic/linkage analysis	6	6	Median: 42.5(31-55)	3:3
Murer (2002)	Italy	Immunohistochemistry analysis	17	17	Mean: 8.5	7:10
Nyberg (1995)	Sweden	Retrospective	60	10	Median: 15(10-45)	4:6
Obeidova (2020)	Czech Republic	Genetic/linkage analysis	31	1	m	m
Olbrich (2003)	m	Genetic/linkage analysis	m	38	m	m
Olinger (2003)	m	Genetic/linkage analysis	6	6	m	2:4
Omran (1999)	m	Genetic/linkage analysis	6 families	14	m	m
Omran (2000)	Venezuela	Genetic/linkage analysis	340	29	m	20:9
O´Toole (2010)	m	Genetic/linkage analysis	116 families	m	m	m
Otto (2003)	m	Genetic/linkage analysis	m	9	m	m
Otto (2005)	m	Genetic/linkage analysis	527	435	m	m

Otto (2008)	m	Genetic/linkage analysis	470	372	m	m
Otto (2009)	m	Genetic/linkage analysis	9 families	m	m	m
Otto (2010)	m	Genetic/linkage analysis	10 families	m	m	m
Otto (2011)	m	Genetic/linkage analysis	120	46	m	m
Parisi (2004)	USA	Genetic/linkage analysis	25	4	Median: 13(8-17)	2:2
Petzold (2023)	France	Genetic/linkage analysis	600	600	m	m
Qiu (2022)	China	Genetic/linkage analysis	5	m	m	m
Sakakibara (2022)	Japan	Genetic/linkage analysis	574	34	m	m
Sayer (2006)	m	Genetic/linkage analysis	25 kindreds	m	m	m
Schuermann (2002)	m	Genetic/linkage analysis	7 families	14	m	m
Shaheen (2016)	UK, Saudi Arabia	Genetic/linkage analysis	371	8	m	m
Snoek (2018)	m	Genetic/linkage analysis	5606	26	m	14:12
Soliman (2010)	Egypt	Letter to the editors	20	16	m	m
Soliman (2012)	Egypt	Retrospective	20	16	Median: 8.5(5m-12y)	10:6
Soliman (2014)	Egypt	Retrospective	105	30	m	m
Stokman (2018)	The Netherlands	Retrospective	40	18	m	m
Stokman (2019)	The Netherlands	Retrospective	12	6	Median: 13.5(6-21)	4:2
Sugimoto (2016)	Japan	Genetic/linkage analysis	35	35	Median: 14(6-46)	19:16
Sun (2016)	China	Genetic/linkage analysis	18	18	m	6:12
Tang (2019)	China	Genetic/linkage analysis	5	5	Median: 4.6(0.2-13.7)	2:3
Tang (2020)	China	Genetic/linkage analysis	10	10	Mean: 27.9(9-73)	3:7
Tang (2022)	China	Retrospective	60	27	m	m
Taskiran (2014)	Turkey	Genetic/linkage analysis	56	m	m	m
Tayfur (2011)	Turkey	Retrospective	9	9	m	4:5
Tong (2023)	China	Retrospective	4	4	Median: 12,7(12,2-13.8)	1:3
Tory (2009)	m	Genetic/linkage analysis	43 families	25	m	m
Valente (2005)	m	Genetic/linkage analysis	23 families	4	m	m
Wang (2021)	China	Genetic/linkage analysis	69	M	m	m
Wolf (2007)	m	Genetic/linkage analysis	56	0	m	m
Yue (2020)	China	Genetic/linkage analysis	55	55	Median: 7.65(1m-17y)	28:27
Zaki (2011)	Egypt	Retrospective	28	3	Median: 12(10-18)	0:3

Online Resource 6: Quality ratings grouped by quality rating tool and study type.

Study	Quality rating		
	Good	Moderate	Poor
Retrospective studies evaluated with the CASP Checklist for cohort studies			
Ala-Mello (1996)	x		
Ala-Mello (1998)		x	
Ala-Mello (1990)	x		
Avci (2022)		x	
Birtel (2021)	x		
Blowey (1996)			x
Gjerstad (2023)	x		
Green (1990)		x	
Gretz (1998)		x	
Hamiwka (2008)		x	
Soliman (2012)	x		
Soliman (2014)		x	
Stokman (2018)	x		
Stokman (2019)	x		
Tang (2022)	x		
Tayfur (2011)	x		
Zaki (2011)	x		
Retrospective studies evaluated with the JBL Checklist for case studies			
Bakkaloglu (2014)	x		
Tong (2023)		x	
Prospective studies evaluated with the CASP Checklist for cohort studies			
Dahmer-Heath (2021)	x		
Grenda (2007)	x		
Genetic studies evaluated with the CASP Checklist for cohort studies			
Antignac (1993)	x		
Attanasio (2007)			x
Bizet (2015)		x	
Braun (2016)	x		
Caridi (2000)		x	
Caridi (2006)	x		
Chaki (2011)		x	
Chaki (2012)		x	
Chaari (2012)	x		
Chen (2022)		x	
Geissari (2015)	x		
Haider (1998)	x		
Halbritter (2012)		x	
Halbritter (2013)		x	
Hoefele (2005)	x		
Hoefele (2007)		x	
Hoff (2013)		x	
König (2022)	x		
Lee (2015)		x	
Li (2023)	x		
Macia (2017)		x	
Medhioub (1994)		x	
Obeidova (2020)	x		
Olbrich (2003)		x	
Olinger (2003)			x
Omran (1999)		x	
Omran (2000)	x		
Otto (2003)		x	

Otto (2005)		x	
Otto (2008)	x		
Otto (2009)	x		
Otto (2010)		x	
Otto (2011)			x
Parisi (2004)	x		
Petzold (2023)	x		
Qiu (2022)	x		
Sakakibara (2022)	x		
Sayer (2006)		x	
Schuermann (2002)		x	
Shaheen (2016)	x		
Snoek (2018)	x		
Sugimoto (2016)		x	
Sun (2016)	x		
Tang (2019)		x	
Tang (2020)	x		
Taskiran (2014)			x
Tory (2009)	x		
Valente (2005)		x	
Wang (2021)	x		
Wolf (2007)	x		
Yue (2020)	x		
Genetic studies evaluated with the JBL Checklist for case studies			
Baris (2006)	x		
Failler (2014)	x		
Hussain (2018)	x		
Mistry (2007)	x		
O´Toole (2010)		x	
Case reports evaluated with the JBL Checklist for case studies			
Caridi (1998)		x	
Hoefele (2011)	x		
Hudson (2020)	x		
Letters to the editor evaluated with the CASP Checklist for cohort studies			
Doreille (2021)	x		
Soliman (2010)	x		
Immunohistochemistry studies evaluated with the CASP Checklist for cohort studies			
Murer (2002)		x	

Online Resource 7: Definition of clinical outcomes, surrogate parameter and patient-reported outcomes with reporting frequency

	Clinical outcomes			Surrogate parameter			Patient-reported outcomes		
General	Failure to thrive	13/90	(14%)				Fatigue	4/90	(5%)
	Laterality defects	12/90	(13%)				Pruritus	2/90	(2%)
Cardiac	Aortic aneurysm/ insufficiency	2/90	(2%)	Bradycardia	1/90	(1%)			
	Aortic coarctation	3/90	(3%)						
	Cardiomyopathy	11/90	(12%)						
	Congenital structural heart defect	21/90	(23%)						
	Dextocardia	1/90	(1%)						
Endocrinology	Delayed puberty	1/90	(1%)						
	Diabetes mellitus	3/90	(3%)						
	Growth hormon deficiency	1/90	(1%)						
	Hypogenitalism	5/90	(6%)						
	Hypogonadism	3/90	(3%)						
	Inverted nipples	1/90	(1%)						
	Polycystic ovary syndrome	2/90	(2%)						
Gastrointestinal	Gastrointestinal manifestation	2/90	(2%)				Nausea	1/90	(1%)
	Pancreatic cysts	4/90	(4%)						
Kidney	CAKUT disorders	12/90	(13%)	Delayed tooth eruption	2/90	(2%)	Enuresis	12/90	(13%)
	Edema	2/90	(2%)	GFR	64/90	(71%)	Nocturia	3/90	(3%)
	End stage kidney disease	75/90	(83%)				Polydipsia/polyuria	35/90	(39%)
	Hematuria	4/90	(4%)				Salt craving	1/90	(1%)
	Histology	49/90	(54%)						
	Isosthenuria	1/90	(1%)						
	Kidney cysts	47/90	(52%)						
	Kidney echogenicity	39/90	(43%)						
	Kidney size	30/90	(33%)						
	Kidney volume	1/90	(1%)						

	Nephrocalcinosis	2/90	(2%)						
	Nephrotic syndrome	1/90	(1%)						
	Proteinuria	22/90	(24%)						
	Renal tubular acidosis	2/90	(2%)						
	Renomegaly	1/90	(1%)						
	Urinary tract infection	3/90	(3%)						
	Urine concentration defect	15/90	(17%)						
Lab/blood	Anemia	23/90	(26%)	Acedosis	1/90	(1%)			
	Dyselectrolytemia	4/90	(4%)	Alkaline phosphatase	2/90	(2%)			
	Hyperlipidosis	1/90	(1%)	Blood urea nitrogen (BUN)	3/90	(3%)			
	Hypertension	27/90	(30%)	Infection	3/90	(3%)			
				Phosporus	3/90	(3%)			
				Serum creatinine	28/90	(31%)			
Liver	Cholestatic hepatopathy	23/90	(26%)	Esophageal varices	2/90	(2%)			
	Fatty liver	1/90	(1%)	Hepatic pruritus	2/90	(2%)			
	Hepatomegaly	13/90	(14%)	Liver disease	26/90	(29%)			
	Liver cysts	8/90	(9%)	Thrombopenia	1/90	(1%)			
	Liver fibrosis	35/90	(39%)						
	Splenomegaly	4/90	(4%)						
Lungs	Pulmonary hypoplasia	2/90	(2%)						
mental development/ neurological disease	Ataxia	5/90	(5%)	IQ	1/90	(1%)	Dizziness	2/90	(2%)
	Cerebrovascular disease	3/90	(3%)						
	Congenital contractures	1/90	(1%)						
	Epilepsy	12/90	(13%)						
	Hearing loss / impairment	13/90	(14%)						
	Hypotonia	13/90	(14%)						

	Infratentorial congenital brain malformation	27/90	(30%)
	MRI brain performed	11/90	(12%)
	Neural tube defects	6/90	(7%)
	Neurodevelopmental disorders	39/90	(43%)
	Peripheral neuropathy	3/90	(3%)
	Sense of smell (anosmia, hyposmia)	2/90	(2%)
	Spasticity/ophistotonus	2/90	(2%)
	Supratentorial congenital brain malformation	12/90	(13%)
	Tremor	2/90	(2%)
Oncology	Neoplasm	7/90	(8%)
Opthalmological	amblyopia	1/90	(1%)
	anisocoria	1/90	(1%)
	cataracts	4/90	(4%)
	color blindness	2/90	(2%)
	eye movement disorders	20/90	(22%)
	LCA leber congenital amaurosis	11/90	(12%)
	night blindness	5/90	(6%)
	nystagmus	19/90	(21%)
	occular involvement	5/90	(6%)
	oculocerebral disease	1/90	(1%)
	optic nerve disorders	5/90	(6%)
	outer eye abnormalities	5/90	(6%)
	reduced vision	35/90	(39%)
	refractory error	4/90	(4%)
	retinal abnormalities	53/90	(59%)
	strabismus	15/90	(17%)
	structural eye malformations	18/90	(20%)

Pain				Abdominal pain	1/90	(1%)
				Back pain	1/90	(1%)
Prenatal	Oligohydramnios	1/90	(1%)			
Respiratory	Apnea	2/90	(2%)			
	Asthma	2/90	(2%)			
	Bronchiectasis	1/90	(1%)			
	Respiratory distress	4/90	(4%)			
	Respiratory infections	11/90	(12%)			
	Sleep apnea	1/90	(1%)			
	Tachypnea	8/90	(9%)			
Skeletal	Chonodrodysplasia	1/90	(1%)			
	Craniofacial abnormalities	15/90	(17%)			
	Dental malformations	2/90	(2%)			
	Hyperextensibility elbows/knees	2/90	(2%)			
	Skeletal malformations	33/90	(37%)			
Skin	Albismus	1/90	(1%)			
	Atopy	1/90	(1%)			
	Cafe au lait macules	1/90	(1%)			
	Cholesteatoma	2/90	(2%)			
	Ichtyosis	1/90	(1%)			
Social				Impact daily life activities	1/90	(1%)