Lived Experience in Rare Genetic Diseases: A Narrative Synthesis of 317 Qualitative Studies (2004-2024)

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Supplementary Table 2

Disease Categories and Diseases	No of Studies	Countries	HIC	LMIC
Lysosomal Storage Diseases	**53**			
Batten Disease (CLN3 Disease)	3	UK Australia Denmark Germany Sweden Norway Canada USA	8	0
Cystinosis	2	USA	1	0
Fabry Disease	3	Netherlands Spain Canada	3	0
Gaucher Disease (including Type 3)	12	Australia Netherlands Japan Spain Canada USA	6	0
Lysosomal Acid Lipase Deficiency	1	UK	1	0
Metachromatic Leukodystrophy	3	USA Australia	2	0

Mucopolysaccharidosis (MPS) (including MPS I, II, III, IV, VI, Sanfilippo, Morquio)	15	UK Spain Australia Ireland Netherlands Canada France Germany Japan USA Mexico	10	1
Niemann-Pick Disease	4	USA UK Belgium Brazil	3	1
Pompe Disease	8	Australia USA Spain Italy Germany Austria Switzerland Canada	8	0
Sandhoff Disease	1	USA	1	0
Tay-Sachs Disease	1	USA	1	0
Haematological Diseases	**15**			
Aplastic Anaemia	1	China	0	1
Fanconi Anaemia	1	USA	1	
Haemophilia	5	Italy Iran Spain Germany France UK USA China Netherlands	7	2
Paroxysmal Nocturnal Haemoglobinuria	1	Canada UK	2	0

Sickle Cell Disease	4	USA UK Canada	3	0
Thalassemia	3	UK India	1	1
Dermatological Diseases	**10**			
Epidermolysis Bullosa	2	Austria South Africa	1	1
Ichthyosis	2	Norway Philippines India	1	2
Albinism	4	South Africa Uganda	0	2
Primary Lymphoedema (compared to secondary)	1	Spain	1	0
Xeroderma Pigmentosum	1	UK	1	0
Immunological Diseases	**10**			
Ataxia-Telangiectasia	2	UK USA	2	0
Hereditary Angioedema	2	USA Japan	2	0
Mastocytosis	1	UK	1	0
Primary Antibody Deficiency	1	Netherlands	1	0
Immunodeficiencies	2	USA Malaysia	1	1
Activated Phosphoinositide 3-kinase Delta Syndrome (PI3K Delta / APDS)	1	USA UK Spain Australia	4	0
Familial Hemophagocytic Lymphohistiocytosis	1	USA UK	2	0
Neurological Diseases	**54**			
Charcot-Marie-Tooth Disease (including Type 1A)	2	USA Australia	2	0

Familial Amyloid Polyneuropathy (including Hereditary Transthyretin Amyloidosis)	3	Portugal USA Argentina Spain	3	1
Friedreich Ataxia	1	Australia	1	0
Hereditary Spastic Paraplegia	1	Germany	1	0
Huntington's Disease	21	Australia Belgium UK Canada USA Sweden Portugal Slovak Republic	8	0
Machado-Joseph Disease (Spinocerebellar Ataxia Type 3)	1	Portugal	1	0
Myasthenia Gravis	1	USA Germany	2	0
Neurofibromatosis	3	Brazil Iran	0	2
Primary Mitochondrial Myopathy	1	USA	1	
Rett Syndrome	2	China	0	1
Spinocerebellar Ataxia Type 2	1	Japan	1	0
Dravet Syndrome	1	Japan	1	0
Tuberous Sclerosis	1	Japan	1	0
Lennox-Gestaut Syndrome	1	Japan	1	0
Angelman Syndrome	2	UK Argentina Jordan USA Chile Belgium Colombia France	5	3
Fragile X Syndrome	5	Australia UK USA Cameroon	3	1
Adrenoleukodystrophy	1	Australia	1	0

Rare Neurodevelopmental/neurological Diseases (general)	5	Canada Ireland Australia USA UK Egypt France	6	1
Schwannomatosis	1	USA	1	
Muscular Diseases	**23**			
Muscular Dystrophy (various)	1	Sweden	1	0
Duchenne Muscular Dystrophy	**9**	UK USA Japan Jordan Malaysia Puerto Rico Poland	5	2
Facioscapulohumeral Muscular Dystrophy	1	USA	1	0
Limb-Girdle Muscular Dystrophy	1	USA	1	0
Spinal Muscular Atrophy (including Type I, II, Bulbar Muscular Atrophy)	11	USA Australia Germany Finland Canada Malaysia	5	1
Metabolic Diseases	**24**			
Citrin Deficiency	1	China	0	1
Classic Galactosemia	1	USA	1	0
Disorders of Amino Acid Metabolism (AAMDs)	3	Malaysia Iran Philippines	0	3
Familial Chylomicronaemia Syndrome	1	UK Spain	2	0
Familial Hypercholesterolemia	2	Sweden Australia	2	0
Inherited Metabolic Diseases (unspecified)	1	Canada	1	0

Lipoprotein Lipase Deficiency	1	UK	1	0
Medium Chain Acyl-CoA Dehydrogenase Deficiency	1	UK	1	0
Phenylketonuria	2	Italy UK	2	0
Primary Carnitine Deficiency	1	Netherlands	1	0
Wilson Disease	2	USA UK Germany	3	0
Acute Hepatic Porphyria	1	UK	1	0
Erythropoietic Protoporphyria	2	USA Canada	2	0
Trimethylaminuria	1	Ireland	1	0
AADC Deficiency	1	UK	1	0
Barth Syndrome	2	UK USA	2	0
Mitochondrial Respiratory Chain Disorder	1	Australia	1	0
Genetic Eye Disorders	**12**			
Aniridia	1	Philippines Canada	1	1
Genetic Eye Disorders (general)	8	UK China Italy	2	1
Leber Congenital Amaurosis	1	France Canada USA Germany	4	0
Retinitis Pigmentosa	1	France Canada USA Germany	4	0
Usher Syndrome	1	Australia	1	0
Cardiovascular Diseases	**3**			
Arrhythmogenic Right Ventricular Cardiomyopathy	2	Canada	1	0
Syndromic Heritable Thoracic Aortic Disease	1	Norway	1	0
Skeletal/Connective Tissue Diseases	**12**			

Achondroplasia	1	USA Spain	2	0
Ehlers-Danlos Syndrome	7	Belgium USA Ireland	3	0
Marfan Syndrome	1	Netherlands	1	0
Osteogenesis Imperfecta	3	Canada USA Begium France Germany Italy Netherlands Poland Spain Sweden UK	11	0
Rare Cancers	**6**			
Li-Fraumeni Syndrome	3	USA	1	0
Multiple Endocrine Neoplasia Types 1 and 2	1	Netherlands	1	0
Rare Cancer (unspecified)	1	UK	1	0
Retinoblastoma	1	Kenya		1
Endocrine/Reproductive Disorders	**4**			
Androgen Insensitivity Syndrome	1	Georgia	0	1
Congenital Hypogonadotropic Hypogonadism	1	Switzerland UK	2	0
Leptin Receptor Deficiency	1	Netherlands	1	0
Congenital Adrenal Hyperplasia	1	India	0	1
Congenital Syndromes	**20**			
22q11.2 Deletion Syndrome	1	Ireland	1	0
Apert Syndrome	1	Turkey	0	1
Bardet-Biedl Syndrome	1	USA South Africa	1	1
Cornelia de Lange Syndrome	2	UK Italy	2	0

Costello Syndrome	1	Online only	n/a	n/a
Goldenhar Syndrome	1	UK	1	0
Holt-Oram Syndrome	1	France	1	0
Noonan Syndrome	1	UK	1	0
Rubinstein-Taybi Syndrome	1	Australia	1	0
Treacher Collins Syndrome	1	Canada	1	0
Russell-Silver Syndrome	1	Taiwan, China	1	0
Rare Trisomy Conditions	1	USA	1	0
Prader-Willi Syndrome	2	USA UK	2	0
PIK3CA-related Overgrowth	1	USA Germany	2	0
Smith-Magenis Syndrome	2	Norway UK	2	0
Williams Syndrome	1	Italy	1	0
Cris du Chat	1	UK	1	0
Respiratory Diseases	**16**			
Cystic Fibrosis	16	USA Australia UK Brazil Portugal Canada	5	1
Kidney/Urogenital Disorders	**3**			
Bartter Syndrome	1	Italy	1	0
Distal Renal Tubular Acidosis	1	France	1	0
Mayer-Rokitansky-Küster-Hauser Syndrome (MRKH)	1	Vietnam	0	1
Other Specific Disorders	**4**			
Amyloidosis (unspecified)	1	Germany	1	0
Behçet's Syndrome	1	Italy	1	0
Finnish Disease Heritage (Kashubian Gene)	1	Finland Poland	2	0
Telomere Biology Disorders	1	USA	1	0

Rare Diseases (General)	**72**			
Rare Diseases (disorder of the corpus callossum as symptom)	2	Australia	1	0
Rare Craniofacial Conditions	1	Norway	1	0
Rare Diseases	69	Norway Germany Poland USA Spain Australia Canada South-East Asia Brazil UK Slovenia Italy India China Netherlands Taiwan, China Africa Europe Malaysia Austria Iran Turkey Ireland	14	7
Undiagnosed Diseases	**8**			
Undiagnosed Diseases	8	USA Australia UK	3	0