Supplementary file 2. Rare diseases included in the study

Disease Category	Disease Name	Pubmed ID
Abdominalsurgical	Trisomy 18	37470964
Abdominalsurgical	Laryngotracheoesophageal cleft	37156975
Abdominalsurgical	Caroli disease	35989996
Abdominalsurgical	Johanson-Blizzard Syndrome	34775698
Allergic	Melkersson-Rosenthal syndrome	35218271
Allergic	vibratory angioedema	36130167
Musculoskeletal	Chondrodysplasia with joint dislocations, gPAPP type	34989141
Musculoskeletal	Bruck syndrome	36476632
Musculoskeletal	Fibular aplasia, tibial campomelia, and oligosyndactyly (FATCO syndrome)	35237492
Musculoskeletal	Gollop Wolfgang complex	37094418
Musculoskeletal	Progressive osseous heteroplasia	36575358
Musculoskeletal	Osteochondritis dissecans	37709256
Musculoskeletal	Osteogenesis imperfecta type 1	35600749
Cardiac	Glycogen storage disease due to glycogen debranching enzyme deficiency	35578201
Cardiac	Progressive sensorineural hearing loss-hypertrophic cardiomyopathy syndrome	37001142
Cardiac	Beckwith-Wiedemann syndrome	37749604
Cardiac	Timothy syndrome	36347939
Cardiacmalformation	Congenital mitral stenosis	35799201
Cardiacmalformation	Familial retinal arterial macroaneurysm	35299703
Cardiacmalformation	PHACE Syndrome	37078913
Cardiacmalformation	Tetralogy of Fallot	37734707
Circulatory	Livedoid vasculopathy	35760761
Circulatory	Hereditary hemorrhagic telangiectasia	PMC9452660
Circulatory	Foix-Alajouanine syndrome	37113340
Embryogenesis	Mucopolysaccharidosis type 2	35782619
Embryogenesis	Meckel Syndrome	35360848
Embryogenesis	RNF13-related severe early-onset epileptic encephalopathy	36553410
Embryogenesis	Sotos syndrome	36970544
Embryogenesis	Scimitar Syndrome	33994033
Embryogenesis	Bowen-Conradi syndrome	11310999

Embryogenesis	FATCO syndrome	35713069
Endocrine	X-linked hypophosphatemia	36011303
Endocrine	Von Hippel-Lindau disease	35145668
Endocrine	Turner syndrome	35434097
Endocrine	WAGR syndrome	36011342
Endocrine	Lysosomal acid lipase deficiency	34922935
Gastroenterological	Necrotizing enterocolitis	36058818
Gastroenterological	Gardner syndrome	36875267
Gastroenterological	Bardet-Biedl syndrome	36533229
Gastroenterological	Goblet cell carcinoma	36685551
Genetic	Craniofrontonasal dysplasia	36558986
Genetic	Idiopathic syringomyelia	37490682
Genetic	Familial cold urticaria	36765385
Genetic	Nager syndrome	36530372
Genetic	Aromatase deficiency	36504506
Genetic	Lobar holoprosencephaly	PMC9949831
Genetic	Floating-Harbor syndrome	35664296
Gynaecologicalandobstetric	Phyllodes tumor of the breast	35692391
Gynaecologicalandobstetric	Transverse vaginal septum	36540471
Gynaecologicalandobstetric	Amniotic fluid embolism	35484512
Gynaecologicalandobstetric	Vaginal atresia	34752931
Haematological	Hereditary stomatocytosis	34477311
Haematological	Streptococcus pneumoniae-associated Thrombotic Microangiopathy	36070894
Haematological	Jacobsen Syndrome	35707598
Haematological	Paroxysmal nocturnal hemoglobinuria	37800776
Haematological	Plummer-Vinson Syndrome	37519547
Hepatic	Cholangiocarcinoma	35945590
Hepatic	Primary hepatic neuroendocrine carcinoma	36837619
Hepatic	Pneumonia caused by Pseudomonas aeruginosa infection	37317086
Hepatic	Wilson's Disease	PMC9237335
Hepatic	Cerebrotendinous xanthomatosis	35428606
Immunological	Mendelian susceptibility to mycobacterial disease (MSMD)	37063912
Immunological	Hyper-IgE syndrome	35602476
Immunological	Singleton-Merten dysplasia	35755559

Immunological	Felty Syndrome	35723962
Immunological	Blau syndrome	37604356
Inbornmetabolism	Hyperornithinemia-hyperammonemia-homocitrullinuria syndrome	35711415
Inbornmetabolism	Hypoxanthine guanine phosphoribosyltransferase partial deficiency	37128514
Inbornmetabolism	mucopolysaccharidosis	34420841
Inbornmetabolism	Propionic acidemia	PMC8607611
Inbornmetabolism	Pyruvate carboxylase deficiency	37484962
Infectious	Ehrlichiosis	35986240
Infectious	Sporotrichosis	37721953
Infectious	Leptospirosis	35145972
Infectious	Ramsay Hunt syndrome	35219691
Infertility	Schaaf-Yang syndrome	36843439
Infertility	Mayer-Rokitansky-Küster-Hauser syndrome	36932787
Infertility	Hyperprolactinemia	37269767
Infertility	Trisomy X	36670650
Neoplastic	Tuberous sclerosis complex	35928867
Neoplastic	Lymphoepithelial-like carcinoma	37193175
Neoplastic	Hepatic Adenomatosis	35355562
Neoplastic	Cowden syndrome	36607858
Neoplastic	Essential thrombocythemia	34895021
Neoplastic	Onychocytic Matricoma	37564690
Neurological	Giant axonal neuropathy	34889507
Neurological	Adult-onset autosomal recessive cerebellar ataxia	35110481
Neurological	Autosomal recessive ataxia, Beauce type	33651373
Neurological	Lennox-Gastaut Syndrome	35795791
Neurological	Borna virus encephalitis	35106511
Neurological	Dravet syndrome	36176564
Neurological	Moebius syndrome	36581828
Odontological	ADULT syndrome	37158316
Odontological	Otodental syndrome	35274545
Odontological	Jalili syndrome	36950920
Odontological	Juvenile hyaline fibromatosis	35726349
Ophthalmic	Cone rod dystrophy	36837600
Ophthalmic	Oculocutaneous albinism	36394576
Ophthalmic	Intermediate uveitis	35907878

Ophthalmic	Klippel-Trenaunay-Weber syndrome	35192386
Ophthalmic	Isolated aniridia	36183072
Ophthalmic	Knobloch syndrome	35693012
Otorhinolaryngological	Orofaciodigital syndrome type 2	3560170
Otorhinolaryngological	Idiopathic bilateral vestibulopathy	37308247
Otorhinolaryngological	MELAS	37576015
Otorhinolaryngological	Kearns-Sayre syndrome	35073857
Otorhinolaryngological	MEGDEL syndrome	37711114
Renal	Idiopathic non-lupus full-house nephropathy	35527024
Renal	Nephroblastoma	36523393
Renal	EEC syndrome	36386837
Renal	Giant cell arteritis	PMC9440460
Renal	Jeune syndrome	35893076
Respiratory	Sprengel deformity	37259666
Respiratory	Nasopharyngeal carcinoma	36345713
Rheumatologicalchildhood	Eosinophilic fasciitis	36606046
Rheumatologicalchildhood	Sweet syndrome	37665047
Rheumatologicalchildhood	Takayasu arteritis	37445428
Rheumatologicalchildhood	PFAPA	35081911
Rhumatological	Kikuchi-Fujimoto disease	30021595
Rhumatological	Overlap myositis	36220202
Rhumatological	Polymyalgia rheumatica	35265557
Rhumatological	Fibroblastic rheumatism	34611730
Rhumatological	Buerger's Disease	36675769
Skin	Linear lichen planus	36590953
Skin	Porokeratotic eccrine ostial and dermal duct nevus	36478599
Skin	Neu-Laxova syndrome	35885441
Skin	Porokeratosis of Mibelli	36451470
Skin	Hennekam syndrome	35626936
Skin	Behçet disease	35661907
Surgicalmaxillo-facial	Fetal alcohol syndrome	37371946
Teratologic	Fetal cytomegalovirus syndrome	36949380
Teratologic	Maternal phenylketonuria	37701331
Teratologic	Fetal valproate syndrome	33455599
Thoracicsurgical	Classical Ehlers-Danlos syndrome	37214418

Thoracicsurgical	De Barsy syndrome	36741656
Thoracicsurgical	Loeys-Dietz syndrome	35668506
Thoracicsurgical	Congenital respiratory-biliary fistula	34890585
Toxiceffects	Fetal valproate spectrum disorder	35942546
Toxiceffects	Cocaine intoxication	35561504
Toxiceffects	Thiocyanate toxicity	35240906
Transplantrelated	Alagille syndrome	36458146
Transplantrelated	Budd-Chiari syndrome	37719537
Transplantrelated	Crigler-Najjar type II	35626936
Transplantrelated	Visceral Myopathy	37168481
Transplantrelated	Pierson syndrome	36829142
Transplantrelated	Nail-patella syndrome	36793587
Urogenital	Congenital primary megaureter, refluxing and obstructed form	35586169
Urogenital	45,X/46,XY Mosaicism，Gonadal dysgenesis	34929697
Urogenital	Aarskog-Scott syndrome	37337880
Urogenital	Campomelic dysplasia	36467484