Rare Disease Names

Brugada syndrome
Protoporphyria, erythropoietic
Guillain-Barre syndrome
Melanoma, familial
Autism, genetic types
Tetralogy of Fallot
Scleroderma
Great vessels transposition
Focal dystonia
Marfan syndrome
Non-Hodgkin malignant lymphoma
Retinitis pigmentosa
Gelineau disease
Myeloma, multiple
Alpha-1 antitrypsin deficiency
Diaphragmatic hernia, congenital
Juvenile arthritis, idiopathic
Neurofibromatosis type 1
Oesophageal atresia
Polycythemia vera
Charcot-Marie-Tooth disease
Polycystic kidney disease, recessive type
VATER association
Coffin-Lowry syndrome
Rendu-Osler-Weber disease
Dermatitis herpetiformis
Atresia of small intestin
Duodenal atresia
Ehlers-Danlos syndrome, classic type
Hirschsprung disease
Microdeletion 22q11
Spherocytosis hereditary
Turner syndrome
Cardiomyopathy, familial dilated
Breast cancer, familial
MELAS syndrome
Leucinosis
Acyl-CoA dehydrogenase, medium chain, deficiency of
Lennox-Gastaut syndrome
Fragile X syndrome
Primary biliary cirrhosis

Stickler syndrome
Williams syndrome
Willebrand disease
Gastroschisis
Microphthalmia
Omphalocele
Sarcoidosis
MURCS association
Stargardt disease
Glioblastoma
Multiple endocrine neoplasia type 1
Prader-Willi syndrome
Alopecia totalis
Nephroblastoma
Cystic fibrosis
Duane syndrome
Neuroblastoma
Hodgkin disease
Dermatomyositis
Polymyositis
Tuberous sclerosis
Congenital adrenal hyperplasia
Rett syndrome
Angelman syndrome
Cataract, total congenital
Hyperlipidemia type 3
Hemophilia
Trisomy 18
Behcet disease
Immunodeficiency, common variable
Microscopic polyangiitis
Idiopathic torsion dystonia
Oculocutaneous albinism
Facioscapulohumeral muscular dystrophy
Holoprosencephaly
Sclerosing cholangitis
Sotos syndrome
Galactosemia
Optic atrophy, Leber type
Osteogenesis imperfecta
Smith-Lemli-Opitz syndrome
Amyotrophic lateral sclerosis
Treacher-Collins syndrome

Tay-Sachs disease
Christ-Siemens-Touraine syndrome
Pheochromocytoma
Retinoblastoma
Rubinstein-Taybi syndrome
Alzheimer disease, familial
Zollinger-Ellison syndrome
Cornelia de Lange syndrome
Familial adenomatous polyposis
Huntington disease
Acromegaly
Fructose intolerance
Primary ciliary dyskinesia
Supranuclear palsy, progressive
Porphyria, acute intermittent
Sickle cell anemia
Deletion 5p
Myasthenia gravis
Achondroplasia
Steinert myotonic dystrophy
Ceroid lipofuscinosis, neuronal
Phenylketonuria
Smith-Magenis syndrome
Wilson disease
Muscular dystrophy limb girdle type 2A, Erb type
CDG syndrome
Niemann-Pick A disease
Propionic acidemia
Waardenburg syndrome type 1, type2 and type 3
Beckwith-Wiedemann syndrome
Adrenoleukodystrophy, X-linked
Goldenhar syndrome
Usher syndrome
Muscular dystrophy, Duchenne and Becker type
Multiple endocrine neoplasia, type 2
Systemic mastocytosis
Von Hippel-Lindau disease
Polyarteritis nodosa
Friedreich ataxia
Poland anomaly
Proximal spinal muscular atrophy
Saethre-Chotzen syndrome
Wegener granulomatosis

Kennedy disease
Cystinosis
Amaurosis congenita of Leber
BOR syndrome
Bullous pemphigoid
Kartagener syndrome
Niemann-Pick B disease
Pseudoxanthoma elasticum
Leigh disease
Peutz-Jeghers syndrome
Autosomal dominant spinocerebellar ataxia
Albinism ocular
Alport syndrome
Crouzon disease
Deletion 4p
Klippel feil syndrome
Langerhans cell histiocytosis
Nail-patella syndrome
Persistent hyperinsulinemic hypoglycemia of infancy
Aniridia, sporadic
Fabry disease
Variegata porphyria
Budd-Chiari syndrome
Darier disease
X-linked severe combined immunodeficiency, T- B+
Bile ducts paucity, syndromic form
Cat-eye syndrome
Apert syndrome
Spastic paraplegia, familial
Adult Onset Still's disease
Pierre Robin syndrome
Glycogen storage disease type 2
Mucopolysaccharidosis type 3
Zellweger syndrome
Nephronophtisis
3-hydroxyacyl-CoA dehydrogenase, long chain, deficiency of
Albers-Schonberg disease
Angioneurotic edema
Ataxia telangiectasia
Chondrodysplasia punctata, rhizomelic type
Coloboma, ocular
Emery-Dreifuss muscular dystrophy, X-linked
Fanconi anemia

Gaucher disease
Gorlin syndrome
Holt-Oram syndrome
Hypokaliemic periodic paralysis
Isovaleric acidemia
Mucopolysaccharidosis type 1
Nemaline myopathy
Neuroendocrine tumor
Thomsen and Becker disease
Churg-Strauss syndrome
Ellis Van Creveld syndrome
Joubert-Boltshauser syndrome
Bardet-Biedl syndrome
Ebstein anomaly
Hyperkaliemic periodic paralysis
Krabbe disease
Mucolipidosis type 2
Albright hereditary osteodystrophy
Menkes syndrome
Niemann-Pick C disease
Glycogen storage disease type 4
Alpha-sarcoglycanopathy
Beta-sarcoglycanopathy
Delta-sarcoglycanopathy
Gamma-sarcoglycanopathy
Tetrasomy 18p
Neurofibromatosis type 2
Xeroderma pigmentosum
Agammaglobulinemia X-linked
Cowden syndrome
Werner syndrome
Glutaryl-CoA dehydrogenase deficiency
Homocystinuria due to cystathionine beta-synthase deficiency
Mucopolysaccharidosis type 4
Lesch-Nyhan syndrome
Pfeiffer syndrome
Severe combined immunodeficiency T- B-
Anemia congenital hypoplastic, Blackfan-Diamond type
Alkaptonuria
Lissencephaly, type 1, due to LIS 1 anomalies
Lipodystrophy, Berardinelli type
Progeria
Granulomatous disease, chronic

Jeune syndrome
Nanism due to growth hormone resistance
Neurodegeneration with brain iron accumulation (NBIA)
Creutzfeldt-Jakob disease
Lowe syndrome
Mucopolysaccharidosis type 6
CHARGE association
Metachromatic leukodystrophy
Bartter syndrome
Muscular dystrophy fukuyama type, Walker-warburg syndrome, Muscle eye brain disease
Ewing sarcoma
Hypercholesterolemia, familial (homozygous form)
Fibrodysplasia ossificans progressiva
Dopa-responsive dystonia
Tyrosinemia type 1
Factor XIII deficiency, congenital
Perinatal hypophosphatasia