SUPPORTED CONDITIONS

The following list identifies conditions from the Orphan Drug Act of 1983, as a disease or condition that affects less than 200,000 people in the United States. The follow conditions are excluded as covered under your plan.

A

- Aarskog Syndrome
- Abetalipoproteinemia
- Ablepharon-Macrostomia Syndrome
- Acanthocheilonemiasis
- Acanthosis Nigricans
- Aceruloplasminemia
- Achalasia
- Achard Thiers Syndrome
- Achondrogenesis
- Achondroplasia
- Acid Sphingomyelinase Deficiency
- Acidemia Isovaleric
- Acidemia, Methylmalonic
- Acoustic Neuroma
- Acquired Aplastic Anemia
- Acquired Hemophilia
- Acquired Lipodystrophy
- Acquired Neuromyotonia
- Acrocallosal Syndrome, Schinzel Type
- Acrodermatitis Enteropathica
- Acrodysostosis
- Acromegaly
- Acromesomelic Dysplasia
- Acromicric Dysplasia
- ACTH Deficiency
- Acute Cholecystitis
- Acute Disseminated Encephalomyelitis
- Acute Eosinophilic Pneumonia
- Acute Intermittent Porphyria
- Acute Myeloid Leukemia
- Acute Promyelocytic Leukemia
- Acute Respiratory Distress Syndrome
- Adams Oliver Syndrome
- ADCY5-Related Dyskinesia
- Addison's Disease
- Adenoid Cystic Carcinoma

- Adenylosuccinate Lyase Deficiency
- Adie Syndrome
- ADNP Syndrome
- Adult Neuronal Ceroid Lipofuscinosis
- Adult Onset Still's Disease
- Adult Polyglucosan Body Disease
- AEC Syndrome
- African Iron Overload
- Agammaglobulinemia
- Agenesis of Corpus Callosum
- Agranulocytosis, Acquired
- Ahumada-Del Castillo Syndrome
- Aicardi Syndrome
- AIDS Dysmorphic Syndrome
- ALAD Porphyria
- Alagille Syndrome
- Alexander Disease
- Alkaptonuria
- Alopecia Areata
- Alpers Disease
- Alpha Thalassemia
- Alpha Thalassemia X-linked Intellectual Disability Syndrome
- Alpha-1 Antitrypsin Deficiency
- Alpha-Mannosidosis
- Alport Syndrome
- Alström Syndrome
- Alternating Hemiplegia of Childhood
- Alveolar Capillary Dysplasia with Misalignment of Pulmonary Veins
- Alveolar Soft Part Sarcoma
- Alveolitis, Extrinsic Allergic
- Ameloblastic Carcinoma
- Ameloblastoma
- Amelogenesis Imperfecta
- Amniotic Band Syndrome
- Amniotic Fluid Embolism
- Amyloidosis
- Amyotrophic Lateral Sclerosis
- Anaplastic Astrocytoma
- Andersen Disease (GSD IV)
- Andersen-Tawil Syndrome
- Anemia of Chronic Disease
- Anemia, Hemolytic, Acquired Autoimmune
- Anemia, Hemolytic, Cold Antibody
- Anemia, Hereditary Nonspherocytic Hemolytic

- Anemia, Megaloblastic
- Anemia, Pernicious
- Anemias, Sideroblastic
- Anencephaly
- Angelman Syndrome
- Angioimmunoblastic T-Cell Lymphoma
- Aniridia
- Aniridia Cerebellar Ataxia Mental Deficiency
- Anthrax
- Antiphospholipid Syndrome
- Antisynthetase Syndrome
- Antithrombin Deficiency
- Antley-Bixler Syndrome
- AP-4-Associated Hereditary Spastic Paraplegia (AP-4-HSP)
- Apert Syndrome
- Aplasia Cutis Congenita
- Apnea, Infantile
- Appendiceal Cancer and Tumors
- Apraxia
- Arachnoid Cysts
- Arginase-1 Deficiency
- Argininie: Glycine Amidinotransferase Deficiency
- Argininosuccinic Aciduria
- Aromatic L-Amino Acid Decarboxylase Deficiency
- Arterial Tortuosity Syndrome
- Arteriovenous Malformation
- Arteritis, Takayasu
- Arthritis, Infectious
- Arthritis, Psoriatic
- Arthrogryposis Multiplex Congenita
- ASAH1-Related Disorders
- Asherman's Syndrome
- Asherson's Syndrome
- Aspartylglycosaminuria
- Aspergillosis
- Asphyxiating Thoracic Dystrophy
- Astrocytoma
- Ataxia Telangiectasia
- Ataxia with Vitamin E Deficiency
- ATR-16 Syndrome
- Atransferrinemia
- Atrial Septal Defects
- Atrioventricular Septal Defect
- Atypical Hemolytic Uremic Syndrome

- Autoimmune Blistering Diseases
- Autoimmune Hepatitis
- Autoimmune Polyendocrine Syndrome Type II
- Autoimmune Polyglandular Syndrome Type 1
- Autoinflammation with Infantile Enterocolitis
- Autosomal Dominant Hereditary Ataxia
- Autosomal Dominant Hyper IgE Syndrome
- Autosomal Dominant Polycystic Kidney Disease
- Autosomal Dominant Porencephaly Type I
- Autosomal Dominant Tubulo-Interstitial Kidney Disease
- Autosomal Recessive Hyper IgE Syndrome
- Autosomal Recessive Hypophosphatemic Rickets Type 2
- Autosomal Recessive Polycystic Kidney Disease

B

- Babesiosis
- Balantidiasis
- Baller-Gerold Syndrome
- Balo Disease
- Banti's Syndrome
- Barakat Syndrome
- Bardet-Biedl Syndrome
- Barth Syndrome
- Bartonellosis
- Bartter Syndrome
- Beckwith-Wiedemann Syndrome
- Behçet's Syndrome
- Bejel
- Bell's Palsy
- Benign Essential Blepharospasm
- Benign Paroxysmal Positional Vertigo
- Bernard-Soulier Syndrome
- Berylliosis
- Best Vitelliform Macular Dystrophy
- Beta Thalassemia
- Bile Acid Synthesis Disorders
- Biliary Atresia
- Binder Type Nasomaxillary Dysplasia
- Binswanger Disease
- Biotinidase Deficiency
- Birt-Hogg-Dubé Syndrome
- Björnstad Syndrome
- Bladder Exstrophy-Epispadias-Cloacal Exstrophy Complex

- Blastomycosis
- Blepharophimosis, Ptosis, Epicanthus Inversus Syndrome
- Bloom Syndrome
- Blue Diaper Syndrome
- Blue Rubber Bleb Nevus syndrome
- Bohring-Opitz Syndrome
- Börjeson-Forssman-Lehman Syndrome
- Bosma Arhinia Microphthalmia Syndrome
- Botulism
- Bowen Disease
- Bowen Hutterite Syndrome
- Bowenoid Papulosis
- Branchio Oculo Facial Syndrome
- Branchiootorenal Spectrum Disorders
- Bronchiolitis Obliterans Organizing Pneumonia
- Bronchopulmonary Dysplasia
- Brown Séquard Syndrome
- Brown Syndrome
- Brucellosis
- Brugada Syndrome
- Budd Chiari Syndrome
- Buerger's Disease
- Bullous Pemphigoid

C

- C Syndrome
- C3 Glomerulopathy: Dense Deposit Disease and C3 Glomerulonephritis
- CADASIL
- Campomelic Syndrome
- Camurati-Engelmann Disease
- Canavan Disease
- Candidiasis
- CARASIL
- Carbamoyl Phosphate Synthetase I Deficiency
- Carcinoid Syndrome
- CARD9 Deficiency
- Cardiofaciocutaneous Syndrome
- Carney Complex
- Carnitine Palmitoyltransferase 1A Deficiency
- Carnosinemia
- Caroli Disease
- Carpenter Syndrome
- Castleman Disease

- Cat Eye Syndrome
- Catamenial Pneumothorax
- Catel Manzke Syndrome
- Caudal Regression Syndrome
- Cavernous Malformation
- CDKL5 Deficiency Disorder
- Central Core Disease
- Central Diabetes Insipidus
- Central Pain Syndrome
- Centronuclear Myopathy
- Cerebellar Agenesis
- Cerebellar Degeneration, Subacute
- Cerebral Creatine Deficiency Syndromes
- Cerebral Folate Deficiency
- Cerebral Palsy
- Cerebro Oculo Facio Skeletal Syndrome
- Cerebrocostomandibular Syndrome
- Cerebrotendinous Xanthomatosis
- Cervical Dystonia
- Cervical Teratoma
- Chanarin-Dorfman Syndrome
- Chandler's Syndrome
- Charcot-Marie-Tooth Disease
- CHARGE Syndrome
- Chediak Higashi Syndrome
- Chiari Frommel Syndrome
- Chiari Malformations
- Chikungunya
- Chilaiditi's Syndrome
- Cholangiocarcinoma
- Cholera
- Cholesteryl Ester Storage Disease
- Chondrocalcinosis 2
- Chordoma
- Choroideremia
- Choroiditis, Serpiginous
- Chromosome 10, Distal Trisomy 10q
- Chromosome 10, Monosomy 10p
- Chromosome 11, Partial Monosomy 11q
- Chromosome 11, Partial Trisomy 11q
- Chromosome 13, Partial Monosomy 13q
- Chromosome 14 Ring
- Chromosome 14, Trisomy Mosaic
- Chromosome 15 Ring

- Chromosome 15, Distal Trisomy 15q
- Chromosome 18 Ring
- Chromosome 18, Monosomy 18p
- Chromosome 18, Tetrasomy 18p
- Chromosome 18q- Syndrome
- Chromosome 21 Ring
- Chromosome 22 Ring
- Chromosome 22q11.2 Deletion Syndrome
- Chromosome 3, Monosomy 3p
- Chromosome 3, Trisomy 3q2
- Chromosome 4, Monosomy Distal 4q
- Chromosome 4, Partial Trisomy Distal 4q
- Chromosome 4, Trisomy 4p
- Chromosome 4q Deletion
- Chromosome 5, Trisomy 5p
- Chromosome 6 Ring
- Chromosome 6, Partial Trisomy 6q
- Chromosome 7, Partial Monosomy 7p
- Chromosome 8, Monosomy 8p
- Chromosome 9 Ring
- Chromosome 9, Partial Monosomy 9p
- Chromosome 9, Tetrasomy 9p
- Chronic Eosinophilic Pneumonia
- Chronic Granulomatous Disease
- Chronic Inflammatory Demyelinating Polyneuropathy
- Chronic Intestinal Pseudo-Obstruction
- Chronic Lymphocytic Leukemia
- Chronic Myelogenous Leukemia
- Churg Strauss Syndrome
- Cicatricial Alopecia
- Ciguatera Fish Poisoning
- Citrullinemia Type 1
- Classic Hereditary Hemochromatosis
- Classic Infantile CLN1 Disease
- Cleidocranial Dysplasia
- Clostridial Myonecrosis
- CLOVES Syndrome
- Cluster Headache
- Coats Disease
- Cockayne Syndrome
- Coffin Lowry Syndrome
- Coffin Siris Syndrome
- Cogan Reese Syndrome
- Cohen Syndrome

- COL4A1/A2-Related Disorders
- Collagen Type VI-Related Disorders
- Colorado Tick Fever
- Common Variable Immune Deficiency
- Complete DiGeorge Syndrome
- Complex Regional Pain Syndrome
- Cone Dystrophy
- Congenital Adrenal Hyperplasia
- Congenital Afibrinogenemia
- Congenital Bilateral Perisylvian Syndrome
- Congenital Central Hypoventilation Syndrome
- Congenital Contractural Arachnodactyly
- Congenital Disorders of Glycosylation
- Congenital Erythropoietic Porphyria
- Congenital Fiber Type Disproportion
- Congenital Fibrosis of the Extraocular Muscles
- Congenital Generalized Lipodystrophy
- Congenital Heart Block
- Congenital Hepatic Fibrosis
- Congenital Hyperinsulinism
- Congenital Lactic Acidosis
- Congenital Muscular Dystrophy
- Congenital Myasthenic Syndromes
- Congenital Myopathy
- Congenital Plasminogen Deficiency
- Congenital Pulmonary Lymphangiectasia
- Congenital Sucrase-Isomaltase Deficiency
- Congenital Syphilis
- Congenital Varicella Syndrome
- Conradi Hünermann Syndrome
- COPA Syndrome
- Cor Triatriatum
- Corneal Dystrophies
- Cornelia de Lange Syndrome
- Corticobasal Degeneration
- Costello Syndrome
- Craniofrontonasal Dysplasia
- Craniometaphyseal Dysplasia
- Craniopharyngioma
- Creatine Transporter Deficiency
- Creutzfeldt Jakob Disease
- Cri du Chat Syndrome
- Crigler Najjar Syndrome
- Cronkhite-Canada Syndrome

- Crouzon Syndrome
- Cryptococcosis
- Cushing Syndrome
- Cutaneous T-Cell Lymphomas
- Cutaneous Vasculitis
- Cutis Laxa
- Cutis Marmorata Telangiectatica Congenita
- Cyclic Neutropenia
- Cyclic Vomiting Syndrome
- Cystic Fibrosis
- Cysticercosis
- Cystinosis
- Cystinuria
- Cytochrome C Oxidase Deficiency
- Cytomegalovirus Infection

D

- Dandy Walker Malformation
- Danon Disease
- De Barsy Syndrome
- De Santis Cacchione Syndrome
- Degos Disease
- Dejerine-Sottas Syndrome
- Dengue Fever
- Dent Disease
- Dentin Dysplasia Type I
- Dentin Dysplasia Type II
- Dentinogenesis Imperfecta Type III
- Denys-Drash Syndrome
- Deoxyhypusine Synthase Disorder
- Depersonalization Disorder
- Dercum's Disease
- Dermatitis Herpetiformis
- Dermatomyositis
- Desmoid Tumor
- Dextrocardia with Situs Inversus
- Diamond Blackfan Anemia
- Diastrophic Dysplasia
- Diencephalic Syndrome
- Diffuse Pulmonary Lymphangiomatosis
- Dilatation of the Pulmonary Artery, Idiopathic
- Distal Myopathy
- Dominant Multiple Epiphyseal Dysplasia

- DOOR Syndrome
- Dracunculosis
- Dravet Syndrome
- Duane syndrome
- Dubin Johnson Syndrome
- Dubowitz Syndrome
- Duchenne Muscular Dystrophy
- Duodenal Atresia or Stenosis
- Dup15q Syndrome
- Dupuytren's Contracture
- Dyggve Melchior Clausen syndrome
- Dysautonomia, Familial
- Dyskeratosis Congenita
- Dysplasia Epiphysealis Hemimelica
- Dystonia

E

- Eales Disease
- Ear, Patella, Short Stature Syndrome
- Ectodermal Dysplasias
- Ectrodactyly Ectodermal Dysplasia Cleft Lip/Palate
- Ehlers Danlos Syndromes
- Eisenmenger Syndrome
- Elephantiasis
- Ellis Van Creveld Syndrome
- Emery Dreifuss Muscular Dystrophy
- Emphysema, Congenital Lobar
- Empty Sella Syndrome
- Encephalitis, Herpes Simplex
- Encephalitis, Japanese
- Encephalocele
- Endocardial Fibroelastosis
- Endocarditis, Infective
- Endomyocardial Fibrosis
- Enterobiasis
- Eosinophilia-Myalgia Syndrome
- Eosinophilic Esophagitis
- Eosinophilic Fasciitis
- Eosinophilic Gastroenteritis
- Epidermal Nevus Syndromes
- Epidermolysis Bullosa
- Epidermolytic Ichthyosis
- Epitheliopathy, Acute Posterior Multifocal Placoid Pigment

- Erdheim Chester Disease
- Erysipelas
- Erythema Multiforme
- Erythrokeratodermia with Ataxia
- Erythromelalgia
- Erythropoietic Protoporphyria and X-Linked Protoporphyria
- Esophageal Atresia and/or Tracheoesophageal Fistula
- Esophageal Cancer
- Essential Iris Atrophy
- Essential Thrombocythemia
- Essential Tremor
- Evans Syndrome
- Ewing Sarcoma

F

- Fabry Disease
- Facioscapulohumeral Muscular Dystrophy
- Factor VII Deficiency
- Factor X Deficiency
- Factor XI Deficiency
- Factor XII Deficiency
- Factor XIII Deficiency
- Familial Adenomatous Polyposis
- Familial Cold Autoinflammatory Syndrome
- Familial Encephalopathy with Neuroserpin Inclusion Bodies
- Familial Eosinophilic Cellulitis
- Familial Hypercholesterolemia
- Familial Hypophosphatemia
- Familial Isolated Hypoparathyroidism
- Familial Lipoprotein Lipase Deficiency
- Familial Mediterranean Fever
- Familial Partial Lipodystrophy
- Familial Platelet Disorder with Associated Myeloid Malignancy
- Fanconi Anemia
- Fascioliasis
- Fatal Familial Insomnia
- Felty Syndrome
- Femoral Facial Syndrome
- Ferroportin Disease
- Fetal Alcohol Syndrome
- Fetal Hydantoin Syndrome
- Fetal Retinoid Syndrome
- Fetal Valproate Syndrome

- FG Syndrome Type 1
- Fibrodysplasia Ossificans Progressiva
- Fibrolamellar Carcinoma
- Fibromuscular Dysplasia
- Fibrosing Mediastinitis
- Fibrous Dysplasia
- Filariasis
- Filippi Syndrome
- Fitz Hugh Curtis Syndrome
- Floating Harbor Syndrome
- Focal Dermal Hypoplasia
- Focal Segmental Glomerulosclerosis
- Follicular Lymphoma
- Food Protein-Induced Enterocolitis Syndrome
- Formaldehyde Poisoning
- Fountain Syndrome
- Fournier Gangrene
- Fox Fordyce Disease
- Fragile X Syndrome
- Fraser Syndrome
- Freeman Sheldon Syndrome
- Frey Syndrome
- Friedreich's Ataxia
- Froelich Syndrome
- Frontofacionasal Dysplasia
- Frontonasal Dysplasia
- Frontotemporal Degeneration
- Fructose Intolerance, Hereditary
- Fryns Syndrome
- Fucosidosis
- Fukuyama Type Congenital Muscular Dystrophy
- Functional Neurological Disorder

G

- Galactosemia
- Galloway-Mowat Syndrome
- Gastritis, Chronic, Erosive
- Gastritis, Giant Hypertrophic
- Gastrointestinal Stromal Tumors
- Gastroparesis
- Gastroschisis
- Gaucher Disease
- General Myoclonus

- Generalized Arterial Calcification of Infancy
- Geographic Tongue
- Gerstmann Syndrome
- Gerstmann-Sträussler-Scheinker Disease
- Gestational Trophoblastic Disease
- Gianotti Crosti Syndrome
- Giant Axonal Neuropathy
- Giant Cell Arteritis
- Giant Cell Myocarditis
- Giant Congenital Melanocytic Nevus
- Gilbert Syndrome
- Gitelman Syndrome
- Glanzmann Thrombasthenia
- Glioblastoma
- Glioma
- Glucose Transporter Type 1 Deficiency Syndrome
- Glucose-6-Phosphate Dehydrogenase Deficiency
- Glucose-Galactose Malabsorption
- Glutaric Aciduria Type I
- Glutaric Aciduria Type II
- Glutathione Synthetase Deficiency
- Glycogen Storage Disease Type I
- Glycogen Storage Disease Type III
- Glycogen Storage Disease Type IX
- Glycogen Storage Disease Type V
- Glycogen Storage Disease Type VII
- GNE Myopathy
- Goblet Cell Carcinoid
- Goodpasture Syndrome
- Gordon Syndrome
- Gorham-Stout Disease
- Gorlin-Chaudhry-Moss Syndrome
- Gottron Syndrome
- Graft versus Host Disease
- Granuloma Annulare
- Granulomatosis with Polyangiitis
- Graves' Disease
- Greig Cephalopolysyndactyly Syndrome
- Grover's Disease
- Growth Hormone Deficiency
- Growth Hormone Insensitivity
- Guanidinoacetate Methyltransferase Deficiency
- Guillain-Barré Syndrome

H

- Hailey-Hailey Disease
- Haim-Munk Syndrome
- Hairy Cell Leukemia
- Hajdu Cheney Syndrome
- Hallermann Streiff Syndrome
- Hanhart Syndrome
- Hantavirus Pulmonary Syndrome
- Harlequin Ichthyosis
- Hartnup Disease
- Hashimoto Encephalopathy
- Heavy Metal Poisoning
- Hemimegalencephaly
- Hemiplegic Migraine
- Hemophagocytic Lymphohistiocytosis
- Hemophilia A
- Hemophilia B
- Henoch-Schönlein Purpura
- Hepatic Encephalopathy
- Hepatocellular Carcinoma
- Hepatoerythropoietic Porphyria
- Hepatopulmonary Syndrome
- Hepatorenal Syndrome
- Hereditary Angioedema
- Hereditary Breast and Ovarian Cancer Syndrome
- Hereditary Coproporphyria
- Hereditary Hemorrhagic Telangiectasia
- Hereditary Hyperphosphatasia
- Hereditary Leiomyomatosis and Renal Cell Carcinoma
- Hereditary Lymphedema
- Hereditary Multiple Osteochondromas
- Hereditary Neuralgic Amyotrophy
- Hereditary Orotic Aciduria
- Hereditary Sensory and Autonomic Neuropathy Type 1E
- Hereditary Sensory and Autonomic Neuropathy Type II
- Hereditary Sensory and Autonomic Neuropathy Type IV
- Hereditary Sensory Neuropathy Type I
- Hereditary Spastic Paraplegia
- Hereditary Spherocytosis
- Hermansky Pudlak Syndrome
- Herpes, Neonatal
- Hers Disease
- Hiccups, Chronic

- Hidradenitis Suppurativa
- Hirschsprung Disease
- Histidinemia
- Hodgkin's Disease
- Holoprosencephaly
- Holt Oram Syndrome
- Homocystinuria due to Cystathionine Beta-Synthase Deficiency
- Horner's Syndrome
- HTLV Type I and Type II
- Human Granulocytic Ehrlichiosis (HGE)
- Human HOXA1 Syndromes
- Human Monocytic Ehrlichiosis (HME)
- Huntington's Disease
- Hutchinson-Gilford Progeria
- Hydranencephaly
- Hydrocephalus
- Hyper IgM Syndromes
- Hyperekplexia
- Hyperemesis Gravidarum
- Hyperferritinemia Cataract Syndrome
- Hyperhidrosis, Primary
- Hyperlipoproteinemia Type III
- Hyperostosis Frontalis Interna
- Hyperprolinemia Type I
- Hyperprolinemia Type II
- Hypochondroplasia
- Hypohidrotic Ectodermal Dysplasia
- Hypokalemia
- Hypomelanosis of Ito
- Hypoparathyroidism
- Hypophosphatasia
- Hypoplastic Left Heart Syndrome
- Hypothalamic Hamartoma

I

- I Cell Disease
- Ichthyosis
- Ichthyosis Hystrix, Curth Macklin Type
- Ichthyosis Vulgaris
- Ichthyosis, CHILD Syndrome
- Ichthyosis, Erythrokeratodermia Variabilis
- Ichthyosis, Erythrokeratolysis Hiemalis
- Ichthyosis, Lamellar

- Ichthyosis, Netherton Syndrome
- Ichthyosis, X Linked
- Idiopathic Intracranial Hypertension
- Idiopathic Pulmonary Fibrosis
- Idiopathic Subglottic Stenosis
- IgA Nephropathy
- Immune Thrombocytopenia
- Imperforate Anus
- Incontinentia Pigmenti
- Infantile Myofibromatosis
- IRF6-Related Disorders
- Ivemark Syndrome

J

- Jackson-Weiss Syndrome
- Jansen Type Metaphyseal Chondrodysplasia
- Jejunal Atresia
- Jervell and Lange-Nielsen Syndrome
- Johanson-Blizzard Syndrome
- Joubert Syndrome
- Juberg-Marsidi Syndrome
- Jumping Frenchmen of Maine
- Juvenile CLN3 Disease
- Juvenile Hemochromatosis
- Juvenile Myelomonocytic Leukemia
- Juvenile Pilocytic Astrocytoma

K

- Kabuki Syndrome
- Kallmann Syndrome
- Kasabach-Merritt Phenomenon
- KAT6A Syndrome
- Kawasaki Disease
- KBG Syndrome
- KCNB1 Encephalopathy
- KCNK9 Imprinting Syndrome
- KCNQ2 Encephalopathy
- Kearns Sayre Syndrome
- Kennedy Disease
- Kenny-Caffey Syndrome
- Keratitis Ichthyosis Deafness Syndrome
- Keratoconus

- Keratomalacia
- Keratosis Follicularis
- Keratosis Follicularis Spinulosa Decalvans
- Keratosis, Seborrheic
- Kernicterus
- Kienböck Disease
- KIF1A-Related Disorder
- Kikuchi's Disease
- Kleine-Levin Syndrome
- Klinefelter Syndrome
- Klippel-Feil Syndrome
- Klippel-Trenaunay Syndrome
- Klüver-Bucy Syndrome
- Kniest Dysplasia
- Kohler Disease
- Kufor Rakeb Syndrome
- Kugelberg Welander Syndrome

L

- L1 Syndrome
- Laband Syndrome
- LADD syndrome
- Lambert-Eaton Myasthenic Syndrome
- Landau Kleffner Syndrome
- Langerhans Cell Histiocytosis
- Larsen Syndrome
- Laryngeal Dystonia
- Laurence-Moon Syndrome
- Leber Congenital Amaurosis
- Leber Hereditary Optic Neuropathy
- Legg Calvé Perthes Disease
- Legionnaires' Disease
- Leigh Syndrome
- Leiomyosarcoma
- Leiomyosarcoma, Inferior Vena Cava
- Leishmaniasis
- Lennox-Gastaut Syndrome
- Lenz Microphthalmia Syndrome
- Leprechaunism
- Leprosy
- Leptospirosis
- Leri Pleonosteosis
- Leri-Weill Dyschondrosteosis

- Lesch Nyhan Syndrome
- Leukocyte Adhesion Deficiency Syndromes
- Leukodystrophy
- Leukodystrophy, Krabbe's
- Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation
- Levy-Yeboa Syndrome
- Li-Fraumeni Syndrome
- Lichen Planus
- Lichen Sclerosus
- Limb-Girdle Muscular Dystrophies
- Liposarcoma
- Lissencephaly
- Listeriosis
- Locked In Syndrome
- Long QT Syndrome
- Low Gamma-GT Familial Intrahepatic Cholestasis
- Lowe syndrome
- Lymphangioleiomyomatosis
- Lymphatic Malformations
- Lymphedema-Distichiasis Syndrome
- Lymphocytic Infiltrate of Jessner
- Lymphomatoid Granulomatosis
- Lysosomal Free Sialic Acid Storage Disorders
- Lysosomal Storage Disorders

M

- Machado-Joseph Disease
- Macroglossia
- Madelung's Disease
- Maffucci Syndrome
- Mal de Debarquement
- Malaria
- Malignant Hyperthermia
- Mallory Weiss Syndrome
- Mandibuloacral Dysplasia
- Mantle Cell Lymphoma
- Maple Syrup Urine Disease
- Marcus Gunn Phenomenon
- Marden Walker Syndrome
- Marfan Syndrome
- Marinesco-Sjögren Syndrome
- Maroteaux Lamy Syndrome

- Marshall Smith Syndrome
- Marshall Syndrome
- Mastocytosis
- Maternally Inherited Leigh Syndrome and NARP Syndrome
- Maxillofacial Dysostosis
- May Hegglin Anomaly
- Mayer-Rokitansky-Küster-Hauser Syndrome
- McCune Albright Syndrome
- McKusick Type Metaphyseal Chondrodysplasia
- MCT8-Specific Thyroid Hormone Cell Transporter Deficiency
- MDR3 Deficiency
- Measles
- Meckel Syndrome
- MECP2 Duplication Syndrome
- Median Arcuate Ligament Syndrome
- Medium Chain Acyl CoA Dehydrogenase Deficiency
- Medullary Sponge Kidney
- Medulloblastoma
- Megalencephaly-Capillary Malformation
- Megalocornea Intellectual Disability Syndrome
- Meige Syndrome
- Melanoma, Malignant
- MELAS Syndrome
- Meleda Disease
- Melkersson Rosenthal Syndrome
- Melnick Needles Syndrome
- Melorheostosis
- Menetrier Disease
- Ménière's Disease
- Meningioma
- Meningitis
- Meningitis, Bacterial
- Meningitis, Tuberculous
- Meningococcal Meningitis
- Meningococcemia
- Menkes Disease
- Merkel Cell Carcinoma
- MERRF Syndrome
- Mesenchymal Chondrosarcoma
- Mesenteric Panniculitis
- Mesothelioma
- Metachromatic Leukodystrophy
- Metaphyseal Chondrodysplasia, Schmid Type
- Metatropic Dysplasia I

- Mevalonate Kinase Deficiency
- Microvillus Inclusion Disease
- Mikulicz Syndrome
- Miller Fisher Syndrome
- Miller Syndrome
- Mitochondrial Neurogastrointestinal Encephalopathy
- Mitral Valve Prolapse Syndrome
- Mixed Connective Tissue Disease (MCTD)
- Mixed Cryoglobulinemia
- Moebius Syndrome
- Monilethrix
- Mosaic Trisomy 22
- Mosaic Trisomy 9
- Mowat-Wilson Syndrome
- Moyamoya Disease
- Mucha Habermann Disease
- Muckle-Wells Syndrome
- Mucolipidosis IV
- Mucopolysaccharidoses
- Mucopolysaccharidosis IV
- Mucopolysaccharidosis Type I
- Mucopolysaccharidosis Type II
- Mucopolysaccharidosis Type III
- Mucopolysaccharidosis Type VII
- Mucormycosis
- Mucous Membrane Pemphigoid
- Mulibrey Nanism
- Multifocal Motor Neuropathy
- Multiple Endocrine Neoplasia Type 1
- Multiple Endocrine Neoplasia Type 2
- Multiple Myeloma
- Multiple Sclerosis
- Multiple Sulfatase Deficiency
- Multiple System Atrophy
- Mulvihill Smith Syndrome
- Mumps
- Muscular Dystrophy, Becker
- Mutism, Selective
- Myasthenia Gravis
- Mycosis Fungoides
- Myelodysplastic Syndromes
- Myhre Syndrome
- Myocarditis
- Myopathy, Myofibrillar

- Myopathy, Scapuloperoneal
- Myotonia Congenita
- Myotonic Dystrophy

N

- N-Acetylglutamate Synthetase Deficiency
- Nager Syndrome
- Nail Patella Syndrome
- Nance-Horan Syndrome
- Narcolepsy
- Necrotizing Enterocolitis
- Necrotizing Fasciitis
- Nelson Syndrome
- Nemaline Myopathy
- Neonatal Cholestasis
- Neonatal Hemochromatosis
- Neonatal Lupus
- Neonatal-Onset Multisystem Inflammatory Disease
- Nephrogenic Diabetes Insipidus
- Nephrogenic Systemic Fibrosis
- Neu Laxova Syndrome
- Neuroacanthocytosis
- Neurofibromatosis 1
- Neurofibromatosis 2
- Neuroleptic Malignant Syndrome
- Neuromyelitis Optica Spectrum Disorder
- Neuropathy, Congenital Hypomyelination
- Neurotrophic Keratitis
- Nevoid Basal Cell Carcinoma Syndrome
- Nevus Sebaceus Syndrome
- New-Onset Refractory Status Epilepticus (NORSE) and Febrile Infection-Related Epilepsy Syndrome (FIRES)
- NGLY1 Deficiency
- Niemann Pick Disease Type C
- Nocardiosis
- Non-24-Hour Sleep-Wake Disorder
- Nonketotic Hyperglycinemia
- Nontuberculous Mycobacterial Lung Disease
- Noonan Syndrome
- Noonan Syndrome with Multiple Lentigines
- Norrie Disease

O

- Ocular Albinism
- Ocular Melanoma
- Ocular Motor Apraxia, Cogan Type
- Oculo-Auriculo-Vertebral Spectrum
- Oculo-Dento-Digital Dysplasia
- Oculocerebral Syndrome with Hypopigmentation
- Oculocerebrocutaneous Syndrome
- Oculocutaneous Albinism
- Oculopharyngeal Muscular Dystrophy
- Ogilvie syndrome
- Olivopontocerebellar Atrophy
- Ollier Disease
- Opsoclonus-Myoclonus Syndrome
- Optic Nerve Hypoplasia
- Oral-Facial-Digital Syndrome
- Ornithine Transcarbamylase Deficiency
- Orocraniodigital Syndrome
- Orthostatic Hypotension
- OSMED, Heterozygous
- OSMED, Homozygous
- Osteogenesis Imperfecta
- Osteomyelitis
- Osteonecrosis
- Osteopetrosis
- Osteosarcoma
- Otopalatodigital Syndrome Type I and II
- Ovarian Cancer
- Ovotesticular Disorder of Sex Development

P

- Pachydermoperiostosis
- Pachyonychia Congenita
- Paget's Disease
- Paget's Disease of the Breast
- Pallister Killian Mosaic Syndrome
- Pallister W Syndrome
- Pallister-Hall Syndrome
- Palmoplantar Pustulosis
- Pancreatic Neuroendocrine Neoplasms (pNENs)
- Panniculitis, Idiopathic Nodular
- Pantothenate Kinase-Associated Neurodegeneration
- Papillitis
- Papillon Lefèvre Syndrome

- Paracoccidioidomycosis
- Paramyotonia Congenita
- Paraneoplastic Neurologic Syndromes
- Paroxysmal Cold Hemoglobinuria
- Paroxysmal Nocturnal Hemoglobinuria
- Parry Romberg Syndrome
- Pars Planitis
- Parsonage Turner Syndrome
- Partial Androgen Insensitivity Syndrome
- Pediatric Cardiomyopathy
- Pediatric Crohn's Disease
- Pediatric Non-Small Cell Lung Cancer
- Peeling Skin Syndrome
- Pelizaeus Merzbacher disease
- Pemphigus and Pemphigoid
- Penta X Syndrome
- Pentalogy of Cantrell
- PEPCK Deficiency
- Perniosis
- Pertussis
- Peutz Jeghers Syndrome
- Pfeiffer Syndrome
- PHACE Syndrome
- Phelan-McDermid Syndrome
- Phenylketonuria
- Pheochromocytoma
- Phosphoglycerate Kinase Deficiency
- Pierre Robin Sequence
- Pinta
- Pitt-Hopkins Syndrome
- Pityriasis Rosea
- Pityriasis Rubra Pilaris
- PLA2G6-Associated Neurodegeneration
- Plague
- Pleuropulmonary Blastoma
- PMM2-CDG
- Pneumocystis Pneumonia
- POEMS Syndrome
- Poland Syndrome
- Polyarteritis Nodosa
- Polycystic Liver Disease
- Polycythemia Vera
- Polymorphous Low-Grade Adenocarcinoma
- Polymyalgia Rheumatica

- Polymyositis and Necrotizing Myopathy
- Pompe Disease
- Pontocerebellar Hypoplasia
- Porphyria
- Porphyria Cutanea Tarda
- Post Polio Syndrome
- Post-Transplant Lymphoproliferative Disease
- Posterior Uveitis
- Potter Syndrome
- Prader-Willi Syndrome
- Precocious Puberty
- Primary Biliary Cholangitis
- Primary Central Nervous System Lymphoma
- Primary Ciliary Dyskinesia
- Primary Craniosynostosis
- Primary Distal Renal Tubular Acidosis
- Primary Familial Brain Calcification
- Primary Gastric Lymphoma
- Primary Hyperoxaluria
- Primary Hyperparathyroidism
- Primary Intestinal Lymphangiectasia
- Primary Lateral Sclerosis
- Primary Mitochondrial Myopathies
- Primary Myelofibrosis
- Primary Orthostatic Tremor
- Primary Sclerosing Cholangitis
- Primary Visual Agnosia
- Proctitis
- Progressive Multifocal Leukoencephalopathy
- Progressive Myoclonus Epilepsy
- Progressive Osseous Heteroplasia
- Progressive Supranuclear Palsy
- Progressive Symmetric Erythrokeratoderma
- Prolactinoma
- Propionic Acidemia
- Protein C Deficiency
- Protein S Deficiency
- Proteus Syndrome
- Prune Belly Syndrome
- Pseudo Hurler Polydystrophy
- Pseudoachondroplasia
- Pseudocholinesterase Deficiency
- Pseudohypoparathyroidism
- Pseudomyxoma Peritonei

- Pseudoxanthoma Elasticum
- Psittacosis
- PTEN Hamartoma Tumor Syndrome
- Pterygium Syndrome, Multiple
- Pulmonary Alveolar Proteinosis
- Pulmonary Arterial Hypertension
- Pure Autonomic Failure
- Pure Red Cell Aplasia, Acquired
- Pycnodysostosis
- Pyoderma Gangrenosum
- Pyridoxine-Dependent Epilepsy
- Pyruvate Carboxylase Deficiency
- Pyruvate Dehydrogenase Complex Deficiency
- Pyruvate Kinase Deficiency

Q

- Q fever

R

- Rabies
- Rabson-Mendenhall Syndrome
- Radiation Sickness
- Ramsay Hunt Syndrome
- Rapid-onset Obesity with Hypothalamic Dysfunction, Hypoventilation, and Autonomic Dysregulation
- Rasmussen Encephalitis
- Reactive Arthritis
- Recessive Multiple Epiphyseal Dysplasia
- Recurrent Respiratory Papillomatosis
- Refractory Celiac Disease
- Refsum Disease
- Relapsing Polychondritis
- Renal Agenesis, Bilateral
- Renal Cell Carcinoma
- Renal Glycosuria
- Renal Medullary Carcinoma
- Respiratory Distress Syndrome, Infant
- Restless Legs Syndrome
- Retinal Vasculopathy with Cerebral Leukoencephalopathy and Systemic Manifestations
- Retinitis Pigmentosa
- Retinoblastoma

- Retinopathy of Prematurity
- Retinoschisis
- Retroperitoneal Fibrosis
- Rett Syndrome
- Reye Syndrome
- Rheumatic Fever
- Riboflavin Transporter Deficiency
- Rieger Syndrome
- Ring Chromosome 4
- Roberts Syndrome
- Robinow Syndrome
- Rocky Mountain Spotted Fever
- Rosai-Dorfman Disease
- Rosenberg Chutorian Syndrome
- Rothmund-Thomson Syndrome
- Roussy Lévy Syndrome
- Rubella
- Rubella, Congenital
- Rubinstein Taybi Syndrome
- Russell Silver Syndrome
- Ruvalcaba Syndrome
- RYR-1-Related Diseases

S

- Sacrococcygeal Teratoma
- Saethre Chotzen Syndrome
- Sakati Syndrome
- Sandhoff Disease
- Schimke Immuno-Osseous Dysplasia
- Schindler disease
- Schinzel Giedion Syndrome
- Schinzel Syndrome
- Schnitzler Syndrome
- Schwartz Jampel Syndrome
- Scleroderma
- Scott Craniodigital Syndrome
- Seckel Syndrome
- Segawa Syndrome
- Senior Løken Syndrome
- Sennetsu Fever
- Sepiapterin Reductase Deficiency
- SETBP1 Disorder
- Setleis Syndrome

- Severe Chronic Neutropenia
- Severe Combined Immunodeficiency
- Sheehan Syndrome
- Short Bowel Syndrome
- Short Chain Acyl CoA Dehydrogenase Deficiency
- SHORT Syndrome
- Shprintzen Goldberg Syndrome
- Shwachman Diamond Syndrome
- Sialadenitis
- Sialidosis
- Sickle Cell Disease
- Simian B Virus Infection
- Simple Pulmonary Eosinophilia
- Simpson Dysmorphia Syndrome
- Singleton Merten syndrome
- Sinonasal Undifferentiated Carcinoma
- Sirenomelia
- Sitosterolemia
- Sjögren-Larsson Syndrome
- SLC13A5 Epileptic Encephalopathy
- SLC6A1 Epileptic Encephalopathy
- Small Cell Lung Cancer
- Smallpox
- Smith Lemli Opitz Syndrome
- Smith Magenis Syndrome
- Sneddon Syndrome
- Snyder-Robinson Syndrome
- Soft Tissue Sarcoma
- Sotos Syndrome
- Spastic Paraplegia 47
- Spastic Paraplegia 50
- Spastic Paraplegia 51
- Spastic Paraplegia 52
- Spina Bifida
- Spinal Muscular Atrophy
- Spinal Muscular Atrophy with Respiratory Distress
- Spinocerebellar Ataxia with Axonal Neuropathy
- Split Hand/Split Foot Malformation
- Spondylocostal Dysplasia
- Spondyloepiphyseal Dysplasia Tarda
- Spondyloepiphyseal Dysplasia, Congenital
- Spondylothoracic Dysplasia
- Spontaneous Intracranial Hypotension
- Sporadic Inclusion Body Myositis

- Sporadic Porencephaly
- Sprengel Deformity
- Staphylococcal Scalded Skin Syndrome
- Status Epilepticus
- STEC Hemolytic Uremic Syndrome
- Stevens-Johnson Syndrome and Toxic Epidermal Necrolysis
- Stickler Syndrome
- Stiff Person Syndrome
- Stomach Cancer
- Sturge Weber Syndrome
- Stuve-Wiedemann Syndrome
- Subacute Sclerosing Panencephalitis
- Succinic Semialdehyde Dehydrogenase Deficiency
- Sudden Infant Death Syndrome
- Sudden Unexplained Death in Childhood
- Superficial Siderosis
- Superior Mesenteric Artery Syndrome
- Superior Semicircular Canal Dehiscence
- Susac Syndrome
- Sutton Disease II
- Sweet Syndrome
- Swyer syndrome
- Sydenham Chorea
- SYNGAP1-related NSID
- Syphilis, Acquired
- Syringobulbia
- Syringomyelia
- Systemic Capillary Leak Syndrome
- Systemic Primary Carnitine Deficiency
- Systemic Scleroderma

T

- Takotsubo Cardiomyopathy
- Tangier Disease
- TANGO2-Related Metabolic Encephalopathy and Arrhythmias
- Tardive Dyskinesia
- Tarlov Cysts
- Tarsal Tunnel Syndrome
- Tay Sachs Disease
- Tenosynovial Giant Cell Tumor
- Testicular Cancer
- Tethered Cord Syndrome
- Tetrahydrobiopterin Deficiency

- Tetralogy of Fallot
- Thoracic Outlet Syndrome
- Three M Syndrome
- Thrombocytopenia Absent Radius Syndrome
- Thrombotic Thrombocytopenic Purpura
- Thyroid Cancer
- Thyroid Eye Disease
- Tietze Syndrome
- Timothy Syndrome
- Tinnitus
- Tolosa Hunt Syndrome
- Tongue Cancer
- Tongue, Hairy
- Tooth Agenesis
- Tooth and Nail Syndrome
- TORCH Syndrome
- Townes Brocks Syndrome
- Toxic Shock Syndrome
- Transverse Myelitis
- Treacher Collins Syndrome
- Tricho Dento Osseous Syndrome
- Trichorhinophalangeal Syndrome Type I
- Trichorhinophalangeal Syndrome Type II
- Trichorhinophalangeal Syndrome Type III
- Trichothiodystrophy
- Trichotillomania
- Trigeminal Neuralgia
- Trimethylaminuria
- Triosephosphate Isomerase Deficiency
- Triploidy
- Trismus-Pseudocamptodactyly Syndrome
- Trisomy 13 Syndrome
- Trisomy 18
- Trisomy 9p (Multiple Variants)
- Trisomy X
- Tropical Sprue
- Truncus Arteriosus
- Tuberculosis
- Tuberous Sclerosis
- Tularemia
- Tumor Necrosis Factor Receptor-Associated Periodic Syndrome
- Turcot Syndrome
- Turner Syndrome
- Twin-Twin Transfusion Syndrome

- Typhoid
- Tyrosine Hydroxylase Deficiency
- Tyrosinemia Type 1

U

- Ulcerative Colitis
- Urachal Cancer
- Urofacial Syndrome
- Urticaria, Cold
- Urticaria, Papular
- Urticaria, Physical
- Usher Syndrome
- USP7-Related Diseases
- Uterine Leiomyosarcoma

V

- VACTERL Association
- VACTERL with Hydrocephalus
- Valinemia
- Variegate Porphyria
- Vascular Malformations of the Brain
- Vasculitis
- Ventricular Septal Defects
- Vernal Keratonconjunctivitis
- Very Long Chain Acyl CoA Dehydrogenase Deficiency (LCAD)
- Visual Snow Syndrome
- Vitamin D Deficiency Rickets
- Vogt-Koyanagi-Harada Disease
- Von Hippel-Lindau Disease
- Von Willebrand Disease

W

- Waardenburg Syndrome
- WAGR Syndrome/11p Deletion Syndrome
- Waldenström's Macroglobulinemia
- Walker Warburg Syndrome
- Wandering Spleen
- Warburg Micro Syndrome
- Warm Antibody Hemolytic Anemia
- WAS Related Disorders
- Weaver Syndrome

- Weil Syndrome
- Weill Marchesani Syndrome
- Weismann Netter Stuhl Syndrome
- Werdnig-Hoffmann Disease
- Werner Syndrome
- Wernicke-Korsakoff Syndrome
- West Nile Encephalitis
- West Syndrome
- WHIM Syndrome
- Whipple Disease
- Wieacker Syndrome
- Wiedemann Rautenstrauch Syndrome
- Wildervanck Syndrome
- Williams Syndrome
- Wilms' Tumor
- Wilson Disease
- Winchester Syndrome
- WNT4 Deficiency
- Wolf-Hirschhorn Syndrome
- Wolff Parkinson White Syndrome
- Wolfram Syndrome
- Wolman Disease
- Wyburn-Mason Syndrome

X

- X linked Lymphoproliferative Syndrome
- X-Linked Adrenoleukodystrophy
- X-Linked Myopathy with Excessive Autophagy
- X-Linked Myotubular Myopathy
- X-linked Opitz G/BBB Syndrome
- X-Linked Protoporphyria
- X-linked Retinoschisis
- Xeroderma Pigmentosum
- XYY Syndrome

Y

- Yaws
- Yellow Fever
- Yellow Nail syndrome
- Yunis Varon Syndrome

Z

- Zellweger Spectrum Disorders
- Zollinger-Ellison Syndrome