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A

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[Nicolaiides-Baraitser Syndrome](#)

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[DNAJC6 Parkinson Disease](#)
[KMT2B-Related Dystonia](#)

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[Congenital Myasthenic Syndromes Overview](#)

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[RPS6KA3-Related Intellectual Disability](#)

Aboussouan, Loutfi S, MD; Cleveland Clinic
[Alpha-1 Antitrypsin Deficiency](#)

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[GJB1 Disorders: Charcot-Marie-Tooth Neuropathy \(CMT1X\) and Central Nervous System Phenotypes](#)

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[Dent Disease](#)

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[Primary Congenital Glaucoma](#)

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[Woodhouse-Sakati Syndrome](#)

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[NR0B1-Related Adrenal Hypoplasia Congenita](#)

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[Congenital Diaphragmatic Hernia Overview](#)

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[Kabuki Syndrome](#)

[Mowat-Wilson Syndrome](#)

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[Myotonic Dystrophy Type 1](#)

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[Chediak-Higashi Syndrome](#)

[Disorders of Intracellular Cobalamin Metabolism](#)

[Free Sialic Acid Storage Disorders](#)

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[Autosomal Dominant Sleep-Related Hypermotor \(Hyperkinetic\) Epilepsy](#)

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[Familial Paroxysmal Nonkinesigenic Dyskinesia](#)

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[Multiple Sulfatase Deficiency](#)

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[Muenke Syndrome](#)

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[Danon Disease](#)

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[COL1A1/2 Osteogenesis Imperfecta](#)

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[ROR2-Related Robinow Syndrome](#)

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[Muenke Syndrome](#)

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[Primary Coenzyme Q10 Deficiency Overview](#)

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[Ornithine Transcarbamylase Deficiency](#)

[Urea Cycle Disorders Overview](#)

- Ahearn, Mary Ellen, MS; Translational Genomics Research Institute
[Spinal Muscular Atrophy, X-Linked Infantile](#)
- Ahmad, Ayesha, MD; University of Michigan School of Medicine
[Pyruvate Carboxylase Deficiency](#)
- Ahmad, Saya, BSc; University Medical Center Utrecht
[Von Hippel-Lindau Syndrome](#)
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[MAPT-Related Frontotemporal Dementia](#)
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[APC-Associated Polyposis Conditions](#)
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[SCARB2-Related Action Myoclonus – Renal Failure Syndrome](#)
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[Multiple Sulfatase Deficiency](#)
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[Aicardi-Goutieres Syndrome](#)
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[DCX-Related Disorders](#)
- Ait-El-Mkadem Saadi, Samira, PhD; Nice Hospital
[CHCHD10-Related Disorders](#)
- Akers, Amy, PhD; Angioma Alliance
[Familial Cerebral Cavernous Malformation](#)
- Akesson, Lauren S, MBBS (Hons), PhD; University of Melbourne
[Fibrodysplasia Ossificans Progressiva](#)
- Akman, H Orhan, PhD; Columbia University Medical Center
[GBE1 Adult Polyglucosan Body Disease](#)
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[Adenosine Deaminase 2 Deficiency](#)
[TNF Receptor-Associated Periodic Fever Syndrome](#)
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[PLPBP Deficiency](#)

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[Stickler Syndrome](#)

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[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

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[Snyder-Robinson Syndrome](#)

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[Long QT Syndrome Overview](#)

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[Spastic Paraplegia 15](#)

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[Hereditary Distal Renal Tubular Acidosis](#)

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[Asparagine Synthetase Deficiency](#)

[Biotin-Thiamine-Responsive Basal Ganglia Disease](#)

[EMC10-Related Neurodevelopmental Disorder](#)

[SLC25A19-Related Thiamine Metabolism Dysfunction](#)

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[Trichorhinophalangeal Syndrome](#)

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[Warsaw Syndrome](#)

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[ATN1-Related Neurodevelopmental Disorder](#)

[NKX6-2-Related Disorder](#)

[Warsaw Syndrome](#)

[Woodhouse-Sakati Syndrome](#)

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[Noonan Syndrome](#)

[Smith-Magenis Syndrome](#)

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[Smith-Magenis Syndrome](#)

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X-Linked Severe Combined Immunodeficiency

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[El-Hattab-Alkuraya Syndrome](#)

[FARS2 Deficiency](#)

[FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion Syndrome](#)

[MELAS](#)

[MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect](#)

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[Fanconi Anemia](#)

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[Megacystis-Microcolon-Intestinal Hypoperistalsis Syndrome Overview](#)

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[Pediatric Genetic Cholestatic Liver Disease Overview](#)

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[ANKRD17-Related Neurodevelopmental Syndrome](#)

[MN1 C-Terminal Truncation Syndrome](#)

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[Peutz-Jeghers Syndrome](#)

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[SCARB2-Related Action Myoclonus – Renal Failure Syndrome](#)

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[Ayme-Gripp Syndrome](#)

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[Progressive Myoclonus Epilepsy, Lafora Type](#)

[SCARB2-Related Action Myoclonus – Renal Failure Syndrome](#)

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[SCARB2-Related Action Myoclonus – Renal Failure Syndrome](#)

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[Huntington Disease-Like 2](#)

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[Variegate Porphyria](#)

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[ANKRD26-Related Thrombocytopenia](#)

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[Glycogen Storage Disease Type V](#)

- Andrews, Caroline V, MSc; Children's Hospital Boston
[Congenital Fibrosis of the Extraocular Muscles Overview](#)
[Duane Syndrome](#)
- Angelini, Corrado, MD; University of Padova
[Calpainopathy](#)
- Angione, Katie, MS; University of Colorado
[CDKL5 Deficiency Disorder](#)
[SLC6A1-Related Neurodevelopmental Disorder](#)
- Anikster, Yair, MD; Safra Children's Hospital
[Costeff Syndrome](#)
- Antonellis, Anthony, PhD; University of Michigan Medical School
[GARS-Associated Axonal Neuropathy](#)
- Anttonen, Anna-Kaisa, MD, PhD; University of Helsinki
[Marinesco-Sjogren Syndrome](#)
- Anyane-Yeboah, Kwame, MD; Columbia University Irving Medical Center
[GNB1 Encephalopathy](#)
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[Dysferlinopathy](#)
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[SOST-Related Sclerosing Bone Dysplasias](#)
- Applegarth, Derek A, PhD; University of British Columbia
[Nonketotic Hyperglycinemia](#)
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[Polymicrogyria Overview](#)
- Arbour, Laura T, MD; University of British Columbia
[Hyaline Fibromatosis Syndrome](#)
- Ardinger, Holly H, MD; GeneReviews
[Pitt-Hopkins Syndrome](#)
- Arenas, Joaquin, PhD; Hospital 12 de Octubre Research Institute
[Glycogen Storage Disease Type V](#)
- Arias, Manuel, MD, PhD; Complejo Hospitalario Universitario
[Spinocerebellar Ataxia Type 36](#)
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[Maternal 15q Duplication Syndrome](#)
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[Cytochrome P450 Oxidoreductase Deficiency](#)

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[PROP1-Related Combined Pituitary Hormone Deficiency](#)

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[Fructose-1,6-Bisphosphatase Deficiency](#)[Primrose Syndrome](#)

Arriaga, Moises A, MBA, MD; Louisiana State University Health Sciences Center

[Usher Syndrome Type I](#)[Usher Syndrome Type II](#)

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[Cranioectodermal Dysplasia](#)

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[Hypokalemic Periodic Paralysis](#)

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[Friedreich Ataxia](#)[Spinocerebellar Ataxia Type 1](#)[Spinocerebellar Ataxia Type 10](#)

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[White-Sutton Syndrome](#)

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[LZTR1- and SMARCB1-Related Schwannomatosis](#)

Astigarraga, Itziar, MD, PhD; Hospital Universitario Cruces

[Familial Hemophagocytic Lymphohistiocytosis](#)

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[Fabry Disease](#)

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[Molybdenum Cofactor Deficiency](#)

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[Peters Plus Syndrome](#)

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[BSCL2-Related Neurologic Disorders / Seipinopathy](#)

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[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)

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[Au-Kline Syndrome](#)
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[Heritable Pulmonary Arterial Hypertension Overview](#)
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[Glycogen Storage Disease Type I](#)
[Phosphorylase Kinase Deficiency](#)
- Autti, Taina, MD, PhD; University of Helsinki
[Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy](#)
- Avner, Ellis D, MD; Children's Hospital Health System of Wisconsin
[Autosomal Recessive Polycystic Kidney Disease – PKHD1](#)
- Axeen, Erika, MD; University of Virginia
[GNAO1-Related Disorder](#)
- Axell, Lisen, MD; University of Colorado Anschutz Medical Center
[APC-Associated Polyposis Conditions](#)
- Axelrod, Felicia B, MD; NYU Medical Center
[NTRK1 Congenital Insensitivity to Pain with Anhidrosis](#)
- Ayhan, Fatma, BS; University of Florida College of Medicine
[Spinocerebellar Ataxia Type 8](#)
- Aylsworth, Arthur S, MD; GC University of North Carolina
[Hypochondroplasia](#)
- Ayres, Lachlan, MB ChB; University Hospitals Dorset
[Hereditary Fructose Intolerance](#)
- Azaiez, Hela, MS, PhD; University of Iowa Hospitals and Clinics
[GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss](#)
[OTOF-Related Deafness](#)
- Azzedine, Hamid, PhD; Academic Medical Center
[SH3TC2-Related Hereditary Motor and Sensory Neuropathy](#)
- B**
- Baas, Frank, MD, PhD; Leiden University Medical Center
[EXOSC3 Pontocerebellar Hypoplasia](#)
[TSEN54 Pontocerebellar Hypoplasia](#)
- Babcock, Holly E, MS; National Human Genome Research Institute
[Osteoglophonic Dysplasia](#)

Babovic-Vuksanovic, Dusica, MD; Mayo Clinic

[FGFR1-Related Hartsfield Syndrome](#)

[LZTR1- and SMARCB1-Related Schwannomatosis](#)

Bachmann, André Stephan, MS, PhD; Michigan State University

[Bachmann-Bupp Syndrome](#)

Bacino, Carlos A, MD; Baylor College of Medicine

[ROR2-Related Robinow Syndrome](#)

Badens, Catherine, PharmD, PhD; Aix-Marseille University

[Trichohepatoenteric Syndrome](#)

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[VPS13A Disease](#)

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[LPIN2-Related Majeed Syndrome](#)

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[Acute Intermittent Porphyria](#)

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[Tubulinopathies Overview](#)

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[Pompe Disease](#)

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[HNRNPH2-Related Neurodevelopmental Disorder](#)

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[TNXB-Related Classical-Like Ehlers-Danlos Syndrome](#)

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[Peutz-Jeghers Syndrome](#)

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[Hereditary Fructose Intolerance](#)

Balasubramanian, Meena, MBBS, MD; University of Sheffield

[ASXL3-Related Disorder](#)

[HNRNPU-Related Neurodevelopmental Disorder](#)

Balasubramanian, Ravikumar, MD, PhD; Massachusetts General Hospital

[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)

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[DEPDC5-Related Epilepsy](#)

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[MYH9-Related Disease](#)

Bale, Sherri J, PhD; GeneDx, Inc

[Autosomal Recessive Congenital Ichthyosis](#)

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[Glycogen Storage Disease Type I](#)

[Phosphorylase Kinase Deficiency](#)

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[Hereditary Hyperekplexia Overview](#)

Ballhausen, Diana, MD; Lausanne University Hospital

[Achondrogenesis Type 1B](#)

[Atelosteogenesis Type 2](#)

[Diastrophic Dysplasia](#)

[SLC26A2-Related Multiple Epiphyseal Dysplasia](#)

Ball, Megan, MD; Murdoch Children's Research Institute

[Mitochondrial DNA-Associated Leigh Syndrome Spectrum](#)

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[Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated](#)

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[TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia](#)

Balwani, Manisha, MD, MS; Icahn School of Medicine at Mount Sinai

[Congenital Erythropoietic Porphyria](#)

[Erythropoietic Protoporphyria, Autosomal Recessive](#)

[X-Linked Protoporphyria](#)

Banka, Siddharth, MD, PhD; University of Manchester

[G6PC3 Deficiency](#)

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[CHCHD10-Related Disorders](#)

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[HYAL2 Deficiency](#)

[PI4KA-Related Disorder](#)

[Troyer Syndrome](#)

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[Familial Dysautonomia](#)

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[Schimke Immunoosseous Dysplasia](#)

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[PURA-Related Neurodevelopmental Disorders](#)

[TRIO-Related Neurodevelopmental Disorder](#)

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[Acute Intermittent Porphyria](#)

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[Hereditary Fibrosing Poikiloderma with Tendon Contractures, Myopathy, and Pulmonary Fibrosis](#)

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[GLYT1 Encephalopathy](#)

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[Alstrom Syndrome](#)

[WFS1 Spectrum Disorder](#)

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[Succinic Semialdehyde Dehydrogenase Deficiency](#)

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[Lysosomal Acid Lipase Deficiency](#)

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[Adenosine Deaminase 2 Deficiency](#)

Barry, Brenda J, MS; Harvard Medical School

[Duane Syndrome](#)

Barth, Peter G, MD, PhD; University of Amsterdam

[EXOSC3 Pontocerebellar Hypoplasia](#)

[TSEN54 Pontocerebellar Hypoplasia](#)

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[HFE-Related Hemochromatosis](#)

Basel-Salmon, Lina, MD, PhD; Rabin Medical Center

[Kaufman Oculocerebrofacial Syndrome](#)

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[COL1A1/2 Osteogenesis Imperfecta](#)

Baskin, Berivan, PhD; Uppsala University Hospital

[Cherubism](#)

Bason, Lynn D, MS; Children's Hospital of Philadelphia

[Alagille Syndrome](#)

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[Holt-Oram Syndrome](#)

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[PRICKLE1-Related Disorders](#)

Bauer, Andrew J, MD; Children's Hospital of Philadelphia

[DICER1 Tumor Predisposition](#)

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[DEPDC5-Related Epilepsy](#)

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[FKBP14 Kyphoscoliotic Ehlers-Danlos Syndrome](#)

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[Spinal Muscular Atrophy, X-Linked Infantile](#)

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[GRIA2-Related Neurodevelopmental Disorder](#)

Bayrak-Toydemir, Pinar, MD, PhD; University of Utah

[Capillary Malformation-Arteriovenous Malformation Syndrome](#)

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[Bardet-Biedl Syndrome Overview](#)

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[Epimerase Deficiency Galactosemia](#)

[Mowat-Wilson Syndrome](#)

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[Dent Disease](#)

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[KCNT1-Related Epilepsy](#)

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[ACTG2 Visceral Myopathy](#)

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[SYNE1 Deficiency](#)

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[X-Linked Congenital Stationary Night Blindness](#)

Becker, Michael A, MD; University of Chicago Medical Center

[Phosphoribosylpyrophosphate Synthetase Superactivity](#)

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[Beckwith-Wiedemann Syndrome](#)

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[Pyruvate Carboxylase Deficiency](#)

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[EPB42-Related Hereditary Spherocytosis](#)

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[AP-4-Associated Hereditary Spastic Paraplegia](#)

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[SOST-Related Sclerosing Bone Dysplasias](#)

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[Androgen Insensitivity Syndrome](#)

Bejjani, Bassem A, MD; Washington State University

[Primary Congenital Glaucoma](#)

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[KCNQ2-Related Disorders](#)

[KCNQ3-Related Disorders](#)

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[Hypochondroplasia](#)

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[CHD7 Disorder](#)

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[INSR-Related Severe Insulin Resistance Syndrome](#)

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[Emery-Dreifuss Muscular Dystrophy](#)

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[MECR-Related Neurologic Disorder](#)

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[TECPR2-Related Hereditary Sensory and Autonomic Neuropathy with Intellectual Disability](#)

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[Hypokalemic Periodic Paralysis](#)

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[Sickle Cell Disease](#)

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[CDKL5 Deficiency Disorder](#)

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[IMAGe Syndrome](#)

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[Carnitine Palmitoyltransferase 1A Deficiency](#)

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[HFE-Related Hemochromatosis](#)

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[ASAH1-Related Disorders](#)

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[Juvenile Hemochromatosis](#)

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[Primary Familial and Congenital Polycythemia](#)

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[Pseudoxanthoma Elasticum](#)

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[X-Linked Agammaglobulinemia](#)

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[Autosomal Dominant Sleep-Related Hypermotor \(Hyperkinetic\) Epilepsy](#)

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[POLR3-Related Leukodystrophy](#)

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[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

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[Congenital Central Hypoventilation Syndrome](#)

[FMRI Disorders](#)

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[Classic Galactosemia and Clinical Variant Galactosemia](#)

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[Trichohepatoenteric Syndrome](#)

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[ALS2-Related Disorder](#)

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[Juvenile Hemochromatosis](#)

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[21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia](#)

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[ACTG2 Visceral Myopathy](#)

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[Isolated Sulfite Oxidase Deficiency](#)

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[Canavan Disease](#)

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[Fructose-1,6-Bisphosphatase Deficiency](#)

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[KCNT1-Related Epilepsy](#)

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[EXOC6B-Related Spondyloepimetaphyseal Dysplasia with Joint Laxity](#)

[Multicentric Osteolysis Nodulosis and Arthropathy](#)

[Progressive Pseudorheumatoid Dysplasia](#)

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[Bryant-Li-Bhoj Neurodevelopmental Syndrome](#)

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[Dopamine Beta-Hydroxylase Deficiency](#)

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[Hypomyelination and Congenital Cataract](#)

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[GLI3-Related Pallister-Hall Syndrome](#)

[Greig Cephalopolysyndactyly Syndrome](#)

[Proteus Syndrome](#)

[SLC25A19-Related Thiamine Metabolism Dysfunction](#)

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[Fructose-1,6-Bisphosphatase Deficiency](#)

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[Long QT Syndrome Overview](#)

[Trichorhinophalangeal Syndrome](#)

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[SHOX Deficiency Disorders](#)

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[Huppke-Brendel Syndrome](#)

[Isolated Sulfite Oxidase Deficiency](#)

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[ADCY5 Dyskinesia](#)

[Alzheimer Disease Overview](#)

[Charcot-Marie-Tooth Hereditary Neuropathy Overview](#)

[GDAP1-Related Hereditary Motor and Sensory Neuropathy](#)

[GJB1 Disorders: Charcot-Marie-Tooth Neuropathy \(CMT1X\) and Central Nervous System Phenotypes](#)

[Hereditary Ataxia Overview](#)

[Hereditary Neuropathy with Liability to Pressure Palsies](#)

[Myotonic Dystrophy Type 1](#)

[SAMD9L Ataxia-Pancytopenia Syndrome](#)

[Spinocerebellar Ataxia Type 14](#)

[Spinocerebellar Ataxia Type 7](#)

Bissell, D Montgomery, MD; University of California San Francisco

[Hereditary Coproporphyrria](#)

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[IRF6-Related Disorders](#)

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[Spastic Paraplegia 15](#)

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[CHD7 Disorder](#)

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[Aromatic L-Amino Acid Decarboxylase Deficiency](#)

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[Autoimmune Lymphoproliferative Syndrome](#)

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[Canavan Disease](#)

Bleyer, Anthony J, MD, MS; Wake Forest School of Medicine

[Autosomal Dominant Tubulointerstitial Kidney Disease -- MUC1](#)

[Autosomal Dominant Tubulointerstitial Kidney Disease -- REN](#)

[Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD](#)

Bloise, Raffaella, MD; ICS Maugeri

[CACNA1C-Related Disorders](#)

[Catecholaminergic Polymorphic Ventricular Tachycardia](#)

Bloomer, Joseph, MD; University of Alabama at Birmingham

[Erythropoietic Protoporphyrria, Autosomal Recessive](#)

[X-Linked Protoporphyrria](#)

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[Chondrodysplasia Punctata 1, X-Linked](#)

[Hypochondroplasia](#)

[Microcephalic Osteodysplastic Primordial Dwarfism Type II](#)

[Rhizomelic Chondrodysplasia Punctata Type 1](#)

[RNU4atac-opathy](#)

Boccuto, Luigi, MD; Greenwood Genetic Center

[Phelan-McDermid Syndrome](#)

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[FLNA Deficiency](#)

[Polymicrogyria Overview](#)

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[X-Linked Adrenoleukodystrophy](#)

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[NSDHL-Related Disorders](#)

[Schimke Immunoosseous Dysplasia](#)

[Snyder-Robinson Syndrome](#)

[Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive](#)

[UNC80 Deficiency](#)

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[ALS2-Related Disorder](#)

[Childhood Ataxia with Central Nervous System Hypomyelination / Vanishing White Matter](#)

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[Woodhouse-Sakati Syndrome](#)

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[Cystic Fibrosis](#)

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[Collagen VI-Related Dystrophies](#)

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[ADAMTSL4-Related Eye Disorders](#)

[Cold-Induced Sweating Syndrome including Crisponi Syndrome](#)

[Congenital Stromal Corneal Dystrophy](#)

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[GAN-Related Neurodegeneration](#)

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[Achondrogenesis Type 1B](#)

[Atelosteogenesis Type 2](#)

[Diastrophic Dysplasia](#)

[SLC26A2-Related Multiple Epiphyseal Dysplasia](#)

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[Familial Porphyria Cutanea Tarda](#)

[Hepatoerythropoietic Porphyria](#)

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[Emery-Dreifuss Muscular Dystrophy](#)

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[SH3TC2-Related Hereditary Motor and Sensory Neuropathy](#)

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[CADASIL](#)

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[ADAMTSL4-Related Eye Disorders](#)

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[GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss](#)

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[Kaufman Oculocerebrofacial Syndrome](#)

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[Retinoblastoma](#)

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[Spinocerebellar Ataxia Type 38](#)

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[CDK13-Related Disorder](#)

[PORCN-Related Developmental Disorders](#)

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[ARSACS](#)

[SYNE1 Deficiency](#)

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[Oculopharyngeal Muscular Dystrophy](#)

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[Rhabdoid Tumor Predisposition Syndrome](#)

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[Trichohepatoenteric Syndrome](#)

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[Thrombocytopenia Absent Radius Syndrome](#)

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[Cherubism](#)

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[PAX2-Related Disorder](#)

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[Fibrous Dysplasia / McCune-Albright Syndrome](#)

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[Mandibulofacial Dysostosis with Microcephaly](#)[Microcephaly-Capillary Malformation Syndrome](#)[THOC6 Intellectual Disability Syndrome](#)[VLDLR Cerebellar Hypoplasia](#)[X-Linked Congenital Stationary Night Blindness](#)

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[Pseudoxanthoma Elasticum](#)

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[Smith-Magenis Syndrome](#)

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[Kaufman Oculocerebrofacial Syndrome](#)

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[BCL11A-Related Intellectual Disability](#)

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[FOXP1 Syndrome](#)

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[ARSACS](#)[GAA-FGF14-Related Ataxia](#)[Oculopharyngeal Muscular Dystrophy](#)

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[UNC80 Deficiency](#)

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[Multiple Endocrine Neoplasia Type 1](#)

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[MUTYH Polyposis](#)

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[MECP2 Disorders](#)

[Spinocerebellar Ataxia Type 1](#)

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[ATP1A3-Related Neurologic Disorders](#)

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[Chondrodysplasia Punctata 1, X-Linked](#)

[Rhizomelic Chondrodysplasia Punctata Type 1](#)

[Zellweger Spectrum Disorder](#)

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[Congenital Stromal Corneal Dystrophy](#)

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[TP63-Related Disorders](#)

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[Smith-Magenis Syndrome](#)

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[Genetic Atypical Hemolytic-Uremic Syndrome](#)

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[DYT1 Early-Onset Isolated Dystonia](#)

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[CADASIL](#)

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[Lowe Syndrome](#)

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[Parkin Type of Early-Onset Parkinson Disease](#)

[Spastic Paraplegia 11](#)

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[Lymphedema-Distichiasis Syndrome](#)

[Milroy Disease](#)

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[GNAO1-Related Disorder](#)

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[COMP-Related Pseudoachondroplasia](#)

[Multiple Epiphyseal Dysplasia, Dominant](#)

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[Oculocutaneous Albinism Type 4](#)

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[Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum](#)

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[RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies](#)

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[Multiple Endocrine Neoplasia Type 4](#)

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[PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia](#)

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[WAS-Related Disorders](#)

Brooks, Brian P, MD, PhD; National Eye Institute

[Oculocutaneous Albinism and Ocular Albinism Overview](#)

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[Warsaw Syndrome](#)

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[Hemophilia A](#)

[Hemophilia B](#)

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[CHMP2B Frontotemporal Dementia](#)

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[Hutchinson-Gilford Progeria Syndrome](#)

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[Epidermolysis Bullosa Simplex](#)

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[Oral-Facial-Digital Syndrome Type I](#)

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[Brugada Syndrome](#)

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[Brugada Syndrome](#)

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[Brugada Syndrome](#)

Brunetti-Pierri, Nicola, MD; University of Naples "Federico II"

[Chondrodysplasia Punctata 1, X-Linked](#)

[Myhre Syndrome](#)

- Brunkow, Mary E, PhD; Institute for Systems Biology
[SOST-Related Sclerosing Bone Dysplasias](#)
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[Autosomal Dominant Robinow Syndrome](#)
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[Hypomyelination and Congenital Cataract](#)
- Brunzell, John D, MD; University of Washington School of Medicine
[Familial Lipoprotein Lipase Deficiency](#)
- Brusco, Alfredo, PhD; University of Turin
[Spinocerebellar Ataxia Type 28](#)
[Spinocerebellar Ataxia Type 38](#)
- Brussino, Alessandro, MD; University of Turin
[Spinocerebellar Ataxia Type 28](#)
- Bryant, Laura, PhD; Children's Hospital of Philadelphia
[Bryant-Li-Bhoj Neurodevelopmental Syndrome](#)
- Bryceson, Yenan, PhD; Karolinska Institutet
[Familial Hemophagocytic Lymphohistiocytosis](#)
- Brzezinski, Jack, MD, PhD; Hospital for Sick Children
[Wilms Tumor Predisposition](#)
- Brüggemann, Norbert, MD; University of Lübeck
[Parkin Type of Early-Onset Parkinson Disease](#)
- Buck, Cassandra, MS; Massachusetts General Hospital
[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)
- Buckley, Michael Francis, MB ChB, PhD; Prince of Wales Hospital
[Hepatic Veno-Occlusive Disease with Immunodeficiency](#)
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[ATP8B1 Deficiency](#)
- Bupp, Caleb, MD; Spectrum Health
[Bachmann-Bupp Syndrome](#)
- Burgmaier, Kathrin, MD; University of Cologne
[Autosomal Recessive Polycystic Kidney Disease – PKHD1](#)
- Burkardt, Deepika, DO; National Human Genome Research Institute
[HIST1H1E Syndrome](#)
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[BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer](#)

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[Abetalipoproteinemia](#)

[APOB-Related Familial Hypobetalipoproteinemia](#)

[Chylomicron Retention Disease](#)

[Familial Combined Hypolipidemia](#)

[Familial Lipoprotein Lipase Deficiency](#)

[Tangier Disease](#)

Burrage, Lindsay, MD, PhD; Baylor College of Medicine

[TANGO2 Deficiency](#)

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[RPE65-Related Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy](#)

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[3q29 Recurrent Deletion](#)

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[APC-Associated Polyposis Conditions](#)

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[Collagen VI-Related Dystrophies](#)

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[WARS2 Deficiency](#)

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[Oculopharyngeal Muscular Dystrophy](#)

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[COL1A1/2 Osteogenesis Imperfecta](#)

[Vascular Ehlers-Danlos Syndrome](#)

Böckenhauer, Detlef, MD, PhD; University Hospitals Leuven

[X-Linked Hypophosphatemia](#)

Bönnemann, Carsten G, MD; National Institute of Neurological Disorders and Stroke

[Collagen VI-Related Dystrophies](#)

C

Cabalza, Jessica L; George Washington University School of Medicine

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

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[CDC73-Related Disorders](#)

Caggana, Michele, ScD; Wadsworth Center - New York State Department of Health

[Krabbe Disease](#)

- Calame, Daniel, MD, PhD; Baylor College of Medicine
[ENTPD1-Related Neurodevelopmental Disorder](#)
- Caldovic, Ljubica, PhD; Children's National Hospital
[Ornithine Transcarbamylase Deficiency](#)
- Cali, Elisa, MD; UCL Queen Square Institute of Neurology
[Riboflavin Transporter Deficiency](#)
- Callewaert, Bert L, MD, PhD; Ghent University Hospital
[Arterial Tortuosity Syndrome](#)
[Congenital Contractural Arachnodactyly](#)
[ELN-Related Cutis Laxa](#)
[LTBP4-Related Cutis Laxa](#)
- Calvert, Guy, DPhil; Dup15q Alliance
[Maternal 15q Duplication Syndrome](#)
- Camacho, Jose A, MD; University of California, Irvine
[Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome](#)
- Camaschella, Clara, MD; Istituto Scientifico San Raffaele
[TFR2-Related Hemochromatosis](#)
- Campeau, Philippe M, MD; Sainte-Justine Hospital
[KAT6B Disorders](#)
[Spondylometaphyseal Dysplasia, Corner Fracture Type](#)
[TBC1D24-Related Disorders](#)
- Campuzano, Oscar, PhD; University of Girona
[Brugada Syndrome](#)
- Cao, Antonio, MD; Consiglio Nazionale delle Ricerche
[Alpha-Thalassemia](#)
[Beta-Thalassemia](#)
- Capece, Tara; University of Pittsburgh
[ZAP70-Related Combined Immunodeficiency](#)
- Capp, Philip K; George Washington University School of Medicine
[Succinic Semialdehyde Dehydrogenase Deficiency](#)
- Caprioli, Jessica, Biol Sci D; IRCCS – Istituto di Ricerche Farmacologiche Mario Negri
[Genetic Atypical Hemolytic-Uremic Syndrome](#)
- Caputi, Caterina, MD; Sapienza Università di Roma
[PRICKLE1-Related Disorders](#)
- Carey, Catherine, MD; Torbay Hospital

[Alstrom Syndrome](#)

Cario, Holger, MD; University Medical Center Ulm

[Primary Familial and Congenital Polycythemia](#)

Carlberg, Katie, MD; University of Washington

[Sickle Cell Disease](#)

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[BAP1 Tumor Predisposition Syndrome](#)

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[Salih Myopathy](#)

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[Huntington Disease](#)

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[DICER1 Tumor Predisposition](#)

Carrillo, Nuria, MD; National Human Genome Research Institute

[Disorders of Intracellular Cobalamin Metabolism](#)[GNE Myopathy](#)[Propionic Acidemia](#)

Carson, Vincent J, MD; Clinic for Special Children

[Maple Syrup Urine Disease](#)

Carter, Melissa T, MD, MSc; The Children's Hospital of Eastern Ontario

[Microcephaly-Capillary Malformation Syndrome](#)[NOTCH3-Related Lateral Meningocele Syndrome](#)

Carvalho, Luciani Renata, MD, PhD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo

[PROP1-Related Combined Pituitary Hormone Deficiency](#)

Carvill, Gemma, PhD; Northwestern University

[CHD2-Related Neurodevelopmental Disorders](#)[KCNT1-Related Epilepsy](#)

Casari, Giorgio, PhD; San Raffaele University

[Spastic Paraplegia 7](#)

Casey, Hannah L, BS; University of Chicago

[Spinocerebellar Ataxia Type 6](#)

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[Prader-Willi Syndrome](#)

Cathey, Sara S, MD; Greenwood Genetic Center

GNPTAB-Related Disorders

Caudill, Marissa; University of Connecticut Health Center

[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)

Cavallin, Mara, MD; Paris Descartes University

[Tubulinopathies Overview](#)

Cebulla, Colleen, MD, PhD; The Ohio State University

[BAP1 Tumor Predisposition Syndrome](#)

Cecchi, Alana C, MS; University of Texas Health Science Center

[Heritable Thoracic Aortic Disease Overview](#)

Cederbaum, Stephen, MD; UCLA Medical Center

[Arginase Deficiency](#)

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[Trichohepatoenteric Syndrome](#)

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[Maternal 15q Duplication Syndrome](#)

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[CTDP1-Related Congenital Cataracts, Facial Dysmorphism, and Neuropathy](#)

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[Xia-Gibbs Syndrome](#)

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[WAS-Related Disorders](#)

Chang, Bernard, MD; Harvard Medical School

[Polymicrogyria Overview](#)

Chang, Irene J, MD; Seattle Children's Hospital Seattle, Washington

[Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

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[TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form](#)

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[Hereditary Multiple Osteochondromas](#)

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[CHKB-Related Muscular Dystrophy](#)

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[RPE65-Related Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy](#)

Chapman, Kimberly A, MD, PhD; Children's National Health System

[Urea Cycle Disorders Overview](#)

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[Smith-Magenis Syndrome](#)

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[CHCHD10-Related Disorders](#)

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[NKX6-2-Related Disorder](#)

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[SAMD9L Ataxia-Pancytopenia Syndrome](#)

[Spinocerebellar Ataxia Type 14](#)

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[Cystic Fibrosis](#)

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[CTNNB1 Neurodevelopmental Disorder](#)

[FAM111A-Related Skeletal Dysplasias](#)

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[Cranio metaphyseal Dysplasia, Autosomal Dominant](#)

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[X-Linked Severe Combined Immunodeficiency](#)

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[Alkaptonuria](#)

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[FLNA Deficiency](#)

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[Aspartylglucosaminuria](#)

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[Glycogen Storage Disease Type I](#)

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[Spinocerebellar Ataxia Type 11](#)

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[Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated](#)

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[Primary Hyperoxaluria Type 1](#)

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[Hereditary Myopathy with Early Respiratory Failure](#)

[Leber Hereditary Optic Neuropathy](#)

[Neuroferritinopathy](#)

[POLG-Related Disorders](#)

[Primary Mitochondrial Disorders Overview](#)

Chiplunkar, Shwetha, MBBS; National Institute of Mental Health & Neurosciences

[Huppke-Brendel Syndrome](#)

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[Nonsyndromic Tooth Agenesis Overview](#)

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[Autosomal Dominant Robinow Syndrome](#)

[PPP1R12A-Related Urogenital and/or Brain Malformation Syndrome](#)

[Warsaw Syndrome](#)

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[Cystic Fibrosis](#)

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[ANKRD17-Related Neurodevelopmental Syndrome](#)

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[GATA1-Related Cytopenia](#)

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[Hereditary Neuropathy with Liability to Pressure Palsies](#)

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[Long QT Syndrome Overview](#)

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[Fumarate Hydratase Deficiency](#)

[MECP2 Disorders](#)

[Phosphoribosylpyrophosphate Synthetase Deficiency](#)

[Phosphoribosylpyrophosphate Synthetase Superactivity](#)

[Squalene Synthase Deficiency](#)

Chrzanowska, Krystyna H, MD, PhD; The Children's Memorial Health Institute

[Nijmegen Breakage Syndrome](#)

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[FREM1 Autosomal Recessive Disorders](#)

[Ritscher-Schinzel Syndrome](#)

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[CTNNB1 Neurodevelopmental Disorder](#)

[MN1 C-Terminal Truncation Syndrome](#)

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[16p11.2 Recurrent Deletion](#)

[Okur-Chung Neurodevelopmental Syndrome](#)

[PACS1 Neurodevelopmental Disorder](#)

[PPP2R5D-Related Neurodevelopmental Disorder](#)

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[DDX41-Associated Familial Myelodysplastic Syndrome and Acute Myeloid Leukemia](#)

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[PLPBP Deficiency](#)

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[Hereditary Coproporphyrria](#)

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[Hypertrophic Cardiomyopathy Overview](#)

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[Cornelia de Lange Syndrome](#)

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[Mucopolysaccharidosis Type I](#)

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[Familial Hypercholesterolemia](#)

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[Hypermanganesemia with Dystonia 1](#)

[SLC39A14 Deficiency](#)

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[Spinocerebellar Ataxia Type 8](#)

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[Poikiloderma with Neutropenia](#)

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[ALS2-Related Disorder](#)

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[Diamond-Blackfan Anemia](#)

Coe, Bradley P, PhD; University of Washington School of Medicine

[DYRK1A Syndrome](#)

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[LAMA2 Muscular Dystrophy](#)

Cogal, Andrea G, BS; Mayo Clinic

[Dent Disease](#)

[Primary Hyperoxaluria Type 1](#)

Cohen, Bruce H, MD; Northeast Ohio Medical University

[POLG-Related Disorders](#)

Cohn, Daniel H, PhD; Cedars-Sinai Medical Center

[COMP-Related Pseudoachondroplasia](#)

Colding-Jørgensen, Eskild, MD; University of Copenhagen

[Myotonia Congenita](#)

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[Sotos Syndrome](#)

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[Von Hippel-Lindau Syndrome](#)

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[Hutchinson-Gilford Progeria Syndrome](#)

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[Fibrous Dysplasia / McCune-Albright Syndrome](#)

[Hyperphosphatemic Familial Tumoral Calcinosis](#)

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[GNB1 Encephalopathy](#)

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[Fumarate Hydratase Deficiency](#)

[Squalene Synthase Deficiency](#)

Conant, Alex, BS; Children's National Health System

[Pelizaeus-Merzbacher-Like Disease 1](#)

[TUBB4A-Related Leukodystrophy](#)

Concannon, Patrick J, PhD; University of Virginia

[Nijmegen Breakage Syndrome](#)

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[Poikiloderma with Neutropenia](#)

Conley, Mary Ellen, MD; University of Tennessee

[X-Linked Agammaglobulinemia](#)

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[Milroy Disease](#)

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[EPB42-Related Hereditary Spherocytosis](#)

[Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

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[Mowat-Wilson Syndrome](#)

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[Perrault Syndrome](#)

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[PIK3CA-Related Overgrowth Spectrum](#)

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[Parkinson Disease Overview](#)

Cook, Edwin H, MD; University of Illinois at Chicago

[3q29 Recurrent Deletion](#)

[Maternal 15q Duplication Syndrome](#)

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[ATP1A3-Related Neurologic Disorders](#)

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[KCNQ2-Related Disorders](#)

[KCNQ3-Related Disorders](#)

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[POLG-Related Disorders](#)

Coppola, Antonietta, MD, PhD; Federico II University of Naples

[DYNC1H1-Related Disorders](#)

Coppola, Giangennaro, MD; University of Salerno

[KCNQ2-Related Disorders](#)

[KCNQ3-Related Disorders](#)

Coppola, Giovanni, MD; University of California Los Angeles

[Primary Familial Brain Calcification](#)

Coppola, Stephanie, BS; Jefferson Medical College

[Krabbe Disease](#)

Corces, Victor, PhD; Emory University School of Medicine

[CTCF-Related Disorder](#)

Cormier-Daire, Valérie, MD, PhD; Paris Cité University

[Geleophysic Dysplasia](#)

Weill-Marchesani Syndrome

Correa, Fernanda Azevedo, MD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo
[PROP1-Related Combined Pituitary Hormone Deficiency](#)

Cortese, Andrea, MD, PhD; University College London
[RFC1 CANVAS / Spectrum Disorder](#)

Coryell, Jason, MS; Oregon Health and Science University
[Pantothenate Kinase-Associated Neurodegeneration](#)

Coucke, Paul, MD, PhD; Ghent University Hospital
[Arterial Tortuosity Syndrome](#)

Coughlin, Curtis, MS; University of Colorado School of Medicine
[Nonketotic Hyperglycinemia](#)

Couillard-Despres, Sebastien, PhD; Paracelsus Medical University Salzburg
[DCX-Related Disorders](#)

Coulter-Mackie, Marion B, PhD; British Columbia's Children's and Women's Hospital
[Primary Hyperoxaluria Type 1](#)

Cowan, Jason, MS; University of Miami Miller School of Medicine
[LMNA-Related Dilated Cardiomyopathy](#)

Cox, Diane W, PhD; University of Alberta
[Alpha-1 Antitrypsin Deficiency](#)
[Wilson Disease](#)

Cragun, Deborah, MS; University of Tampa
[Cytochrome P450 Oxidoreductase Deficiency](#)

Craigen, William J, MD, PhD; Baylor College of Medicine
[Mitochondrial DNA Maintenance Defects Overview](#)
[MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect](#)

Crombez, Eric A, MD; Ultragenyx Pharmaceutical Inc
[Arginase Deficiency](#)

Crosby, Andrew H, BSc, PhD; University of Exeter Medical School
[HYAL2 Deficiency](#)
[PI4KA-Related Disorder](#)
[Troyer Syndrome](#)

Crowley, Leon, MSc; King's College London
[TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia](#)

Crowley, William F, MD; Massachusetts General Hospital
[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)

- Crow, Yanick J, BMedSci, MBBS, PhD; Central Manchester Foundation Trust University Hospitals
[Aicardi-Goutieres Syndrome](#)
- Cruts, Marc, PhD; University of Antwerp
[C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia](#)
- Cruz Corchado, Johnny; University of Iowa
[C3 Glomerulopathy](#)
- Cruz, Vincent, MD, MS; DDC Clinic – Center for Special Needs Children
[GM3 Synthase Deficiency](#)
- Cubells, Joseph F, MD, PhD; Emory University School of Medicine
[3q29 Recurrent Deletion](#)
- Cudrici, Cornelia, MD; National Heart, Lung and Blood Institute
[TNF Receptor-Associated Periodic Fever Syndrome](#)
- Culver, Julie O Bars, MS; Fred Hutchinson Cancer Research Center
[BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer](#)
- Culver, Kathy, MS; Sutter Pacific Medical Foundation/California Pacific Medical Center
[Simpson-Golabi-Behmel Syndrome Type 1](#)
- Cunniff, Christopher M, MD; Weill Cornell Medical College
[Bloom Syndrome](#)
[Smith-Lemli-Opitz Syndrome](#)
- Cunningham, Lea, MD; National Cancer Institute
[RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies](#)
- Cunningham, Michael L, MD, PhD; University of Washington Schools of Medicine and Dentistry
[Saethre-Chotzen Syndrome](#)
- Curiel, Julian, BS; Children's National Health System
[TUBB4A-Related Leukodystrophy](#)
- Cutting, Garry R, MD; Johns Hopkins University
[Cystic Fibrosis](#)
[Thanatophoric Dysplasia](#)
[Treacher Collins Syndrome](#)
- D**
- D'Adamo, Maria Cristina, PhD; University of Malta
[Episodic Ataxia Type 1](#)
- D'Andrea, Alan, MD; Dana Farber Cancer Institute
[Fanconi Anemia](#)

Da Silva, Jorge Diogo, MD, PhD; Santo Antonio University Hospital Center

[Ellis-van Creveld Syndrome](#)

Dafsari, Hormos Salimi, MD; University of Cologne

[DYNC1H1-Related Disorders](#)

[EPG5-Related Disorder](#)

Dagli, Aditi I, MD; Orlando Health Arnold Palmer Hospital

[Angelman Syndrome](#)

[Glycogen Storage Disease Type III](#)

[Glycogen Storage Disease Type VI](#)

Daiger, Stephen P, PhD; University of Texas Health Science Center

[Nonsyndromic Retinitis Pigmentosa Overview](#)

Dai, Hongzheng, PhD; Baylor College of Medicine

[FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion Syndrome](#)

[MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect](#)

Dalal, Ashwin, MD; Centre for DNA Fingerprinting and Diagnostics

[Progressive Pseudorheumatoid Dysplasia](#)

Dale, David C, MD; University of Washington Medical Center

[ELANE-Related Neutropenia](#)

Daley, Shane M, MD; Mayo Clinic

[Jervell and Lange-Nielsen Syndrome](#)

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[Myotonic Dystrophy Type 2](#)

[Spinocerebellar Ataxia Type 8](#)

Daly, Mary B, MD, PhD; Fox Chase Cancer Center

[BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer](#)

Danek, Adrian, MD; Ludwig-Maximilians-Universität

[McLeod Neuroacanthocytosis Syndrome](#)

[VPS13A Disease](#)

Danzi, Matt, PhD; University of Miami Miller School of Medicine

[GAA-FGF14-Related Ataxia](#)

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[Dystrophinopathies](#)

Das, Soma, PhD; University of Chicago

[PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia](#)

[X-Linked Myotubular Myopathy](#)

Davenport, Sandra LH, MD; Sensory Genetics and Neurodevelopment

[CHD7 Disorder](#)

Davies, Alexandra K, PhD; University of Cambridge

[AP-4-Associated Hereditary Spastic Paraplegia](#)

Davis, Joie; NIH

[STAT3 Hyper IgE Syndrome](#)

[X-Linked Severe Combined Immunodeficiency](#)

Day, John W, MD, PhD; Stanford University

[Myotonic Dystrophy Type 2](#)

[Spinocerebellar Ataxia Type 8](#)

Dazzo, Emanuela, PhD; CNR Institute of Neuroscience

[Autosomal Dominant Epilepsy with Auditory Features](#)

De Baere, Elfride, MD, PhD; Ghent University Hospital

[Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome](#)

de Boer, Irene, MD; Leiden University Medical Center

[Retinal Vasculopathy with Cerebral Leukoencephalopathy and Systemic Manifestations](#)

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[CLPB Deficiency](#)

[Feingold Syndrome 1](#)

[Phosphoribosylpyrophosphate Synthetase Deficiency](#)

[Phosphoribosylpyrophosphate Synthetase Superactivity](#)

[SERAC1 Deficiency](#)

De Camilli, Pietro, PhD; Yale University School of Medicine

[VPS13A Disease](#)

de Castro, Luis F, PhD; National Institute of Dental and Craniofacial Research/NIH

[Fibrous Dysplasia / McCune-Albright Syndrome](#)

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[Multiple Sulfatase Deficiency](#)

De Gobbi, Marco, MD, PhD; University of Torino

[TFR2-Related Hemochromatosis](#)

de Graaff, Laura CG, MD, PhD; Erasmus Medical Center

[PROP1-Related Combined Pituitary Hormone Deficiency](#)

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[Serine Deficiency Disorders](#)

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X-Linked Hyper IgM Syndrome

de Leeuw, Nicole, PhD; Radboud University Medical Center

Kleefstra Syndrome

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DYT1 Early-Onset Isolated Dystonia

De León, Diva D, MD; University of Pennsylvania

Permanent Neonatal Diabetes Mellitus

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Arterial Tortuosity Syndrome

Classic Ehlers-Danlos Syndrome

EFEMP2-Related Cutis Laxa

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GNB5-Related Neurodevelopmental Disorder

de Vernejoul, Marie-Christine, MD, PhD; Hopital Lariboisiere

CLCN7-Related Osteopetrosis

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Glucose Transporter Type 1 Deficiency Syndrome

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NTHL1 Tumor Syndrome

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15q13.3 Recurrent Deletion

DYRK1A Syndrome

Gabriele-de Vries Syndrome

Koolen-de Vries Syndrome

TRIO-Related Neurodevelopmental Disorder

WAC-Related Intellectual Disability

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Cornelia de Lange Syndrome

IMAGe Syndrome

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DeFelice, Magee, MD; Nemours Children's Health

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[DICER1 Tumor Predisposition](#)

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[Maturity-Onset Diabetes of the Young Overview](#)

Delatycki, Martin B, MBBS, PhD; Murdoch Childrens Research Institute

[Friedreich Ataxia](#)

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[Canavan Disease](#)

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[Amyotrophic Lateral Sclerosis Overview](#)[Arrhythmogenic Right Ventricular Cardiomyopathy Overview](#)

Dell, Katherine MacRae, MD; Case Western Reserve University School of Medicine

[Autosomal Recessive Polycystic Kidney Disease – PKHD1](#)

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[Perrault Syndrome](#)

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[CDKL5 Deficiency Disorder](#)[SLC6A1-Related Neurodevelopmental Disorder](#)

Demirdas, Serwet, MD, PhD; Erasmus Medical Center

[TNXB-Related Classical-Like Ehlers-Danlos Syndrome](#)

Dempsey, Melissa A, MS; Parkview Health

[Allan-Herndon-Dudley Syndrome](#)[Chondrodysplasia Punctata 2, X-Linked](#)

Demuth, Ilja, PhD; Charité – Universitätsmedizin Berlin

[Nijmegen Breakage Syndrome](#)

Deneubourg, Celine, MSc; King's College London

[EPG5-Related Disorder](#)

Depienne, Christel, PhD; Hôpital Pitié-Salpêtrière

[CLCN2-Related Leukoencephalopathy](#)[Congenital Mirror Movements](#)[Spastic Paraplegia 4](#)

Der Kaloustian, Vazken M, MD; McGill University

[Hidrotic Ectodermal Dysplasia 2](#)

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[Glycogen Storage Disease Type III](#)

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[Familial Cerebral Cavernous Malformation](#)

Desai, Jigar, PhD; Boston Children's Hospital

[Congenital Fibrosis of the Extraocular Muscles Overview](#)

Desnick, Robert J, MD, PhD; Icahn School of Medicine at Mount Sinai

[Congenital Erythropoietic Porphyria](#)[Erythropoietic Protoporphyrin, Autosomal Recessive](#)[Fabry Disease](#)[HEXA Disorders](#)[X-Linked Protoporphyrin](#)

Deutch, Natalie, MS; National Human Genome Research Institute

[RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies](#)[TNF Receptor-Associated Periodic Fever Syndrome](#)

Deuschländer, Angela B, MD; Mayo Clinic

[DYT-GNAL](#)[VPS35-Related Parkinson Disease](#)

Devinsky, Orrin, MD; New York University Langone Medical Center

[Maternal 15q Duplication Syndrome](#)

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[Alpha-Thalassemia](#)[Congenital Dyserythropoietic Anemia Type I](#)

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[FGFR1-Related Hartsfield Syndrome](#)[LZTR1- and SMARCB1-Related Schwannomatosis](#)

Di Donato, Nataliya, MD, PhD; Universitätsklinikum Carl Gustav Carus

[Baraitser-Winter Cerebrofrontofacial Syndrome](#)

Di Giovanni, Giuseppe, PhD; Istituto Euro-Mediterraneo di Scienza e Tecnologia

[Episodic Ataxia Type 1](#)

Di Gregorio, Eleonora, PhD; Città della Salute e della Scienza University Hospital

[Spinocerebellar Ataxia Type 38](#)

Di Meo, Ivano, PhD; IRCCS Foundation Carlo Besta Neurological Institute

[Ethylmalonic Encephalopathy](#)

Di Paola, Jorge, MD; Washington University School of Medicine

[ETV6 Thrombocytopenia and Predisposition to Leukemia](#)

Dias, Cristina, MD, PhD; King's College London

[BCL11A-Related Intellectual Disability](#)

Dias, Renuka, PhD; Birmingham Women's and Children's Hospital
[WFS1 Spectrum Disorder](#)

Diaz, George A, MD, PhD; Icahn School of Medicine at Mount Sinai
[Thiamine-Responsive Megaloblastic Anemia Syndrome](#)

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[SCN9A Neuropathic Pain Syndromes](#)

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[CADASIL](#)

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[Monosomy 7 Predisposition Syndromes Overview](#)

Dietz, Harry C, MD; Johns Hopkins University School of Medicine
[FBN1-Related Marfan Syndrome](#)
[Loeys-Dietz Syndrome](#)

DiGiovanna, John J, MD; National Cancer Institute
[Xeroderma Pigmentosum](#)

Digweed, Martin, PhD; Charité – Universitätsmedizin Berlin
[Nijmegen Breakage Syndrome](#)

DiMauro, Salvatore, MD; Columbia University Medical Center
[MELAS](#)
[MERRF](#)
[Mitochondrial DNA Deletion Syndromes](#)

Dimmock, David P, MD; Children's Hospital of Wisconsin
[Deoxyguanosine Kinase Deficiency](#)

Dindot, Scott, PhD; Texas A&M University
[Maternal 15q Duplication Syndrome](#)

Diop-Bove, Ndeye, PhD; Albert Einstein College of Medicine
[Hereditary Folate Malabsorption](#)

Dirksen, Robert T, PhD; University of Rochester School of Medicine and Dentistry
[Malignant Hyperthermia Susceptibility](#)

DiStasio, Andrew T, PhD; Nationwide Children's Hospital
[ATP7A-Related Copper Transport Disorders](#)

Disteche, Christine M, PhD; University of Washington
[Y Chromosome Infertility](#)

DiStefano, Charlotte, PhD; University of California Los Angeles
[Maternal 15q Duplication Syndrome](#)

Dobson-Stone, Carol, DPhil; University of Sydney

[McLeod Neuroacanthocytosis Syndrome](#)

[VPS13A Disease](#)

Dobyns, William B, MD; University of Minnesota

[ATP6V0A2-Related Cutis Laxa](#)

[Encephalocraniocutaneous Lipomatosis](#)

[PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia](#)

[PIK3CA-Related Overgrowth Spectrum](#)

[Polymicrogyria Overview](#)

Docherty, Louise Esther, BSc (Hons), PhD; Salisbury NHS Foundation Trust

[Diabetes Mellitus, 6q24-Related Transient Neonatal](#)

Doherty, Dan, MD, PhD; University of Washington

[MN1 C-Terminal Truncation Syndrome](#)

Doherty, Emily S, MD; Carilion Clinic

[Muenke Syndrome](#)

Doimo, Mara, PhD; University of Padova

[Primary Coenzyme Q10 Deficiency Overview](#)

Dome, Jeffrey S, MD, PhD; Children's National Health System

[Wilms Tumor Predisposition](#)

Dominik, Natalia, BSc, MSc; UCL Queen Square Institute of Neurology

[Riboflavin Transporter Deficiency](#)

Dong, Hui-Jia; University of Florida College of Medicine

[Angelman Syndrome](#)

Donkervoort, Sandra, MS; National Institute of Neurological Disorders and Stroke

[Amyotrophic Lateral Sclerosis Overview](#)

[Collagen VI-Related Dystrophies](#)

[Inclusion Body Myopathy with Paget Disease of Bone and/or Frontotemporal Dementia](#)

Donnai, Dian, MD; University of Manchester and St Mary's Hospital

[Donnai-Barrow Syndrome](#)

Doros, Leslie, MD; Children's National Medical Center

[DICER1 Tumor Predisposition](#)

Dorsey, Adrienne M; Children's National Medical Center

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Dotti, Maria Teresa, MD; University of Siena

[Cerebrotendinous Xanthomatosis](#)

Douthitt Seibel, Gabrielle, ARNP, MN, MPH; Seattle Children's Hospital

[Sickle Cell Disease](#)

Douzgou, Sofia, MD, PhD; Haukeland University Hospital

[PPP2R1A-Related Neurodevelopmental Disorder](#)

Dowling, James J, MD, PhD; Hospital for Sick Children

[X-Linked Myotubular Myopathy](#)

Downs, Jenny, MSc, PhD; Telethon Kids Institute

[CDKL5 Deficiency Disorder](#)

Drillings, Ian; Children's National Medical Center

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Driscoll, Daniel J, MD, PhD; University of Florida College of Medicine

[Angelman Syndrome](#)

[Prader-Willi Syndrome](#)

Drunat, Séverine, PharmD, PhD; APHP-Robert Debré University Hospital

[ASPM Primary Microcephaly](#)

[Baraitser-Winter Cerebrofrontofacial Syndrome](#)

[WDR62 Primary Microcephaly](#)

du Souich, Christèle, MSc; University of British Columbia

[NSDHL-Related Disorders](#)

[UNC80 Deficiency](#)

Dua Puri, Ratna, MD; Sir Ganga Ram Hospital

[Primrose Syndrome](#)

Dubois, Anna, BSc, MBChB; Royal Victoria Infirmary

[CYLD Cutaneous Syndrome](#)

Dugan, Stefanie N, MS; BloodCenter of Wisconsin

[ANKRD26-Related Thrombocytopenia](#)

Duis, Jessica, MD, MS; Children's Hospital Colorado

[Schinzel-Giedion Syndrome](#)

[SETBP1 Haploinsufficiency Disorder](#)

Duker, Angela, MS; Nemours Children's Health

[Microcephalic Osteodysplastic Primordial Dwarfism Type II](#)

[Rhizomelic Chondrodysplasia Punctata Type 1](#)

[RNU4atac-opathy](#)

Duley, John A, PhD; University of Queensland and Mater Research

[Phosphoribosylpyrophosphate Synthetase Deficiency](#)

[Phosphoribosylpyrophosphate Synthetase Superactivity](#)

Dulski, Jaroslaw, MD, PhD; Medical University of Gdansk

[CSF1R-Related Disorder](#)[DCTN1-Related Neurodegeneration](#)[VPS35-Related Parkinson Disease](#)

Dumitrescu, Alexandra M, MD, PhD; University of Chicago Medical Center

[Allan-Herndon-Dudley Syndrome](#)

Dunn, Clinton P, MD; University of Washington

[X-Linked Hyper IgM Syndrome](#)

Dunoyer, Margaux, MD; Assistance Publique-Hôpitaux de Paris

[Congenital Mirror Movements](#)

Dunwoodie, Sally, BSc, PhD; Victor Chang Cardiac Research Institute

[Congenital NAD Deficiency Disorder](#)[Spondylocostal Dysostosis, Autosomal Recessive](#)

Dunø, Morten, PhD; Rigshospitalet

[Myotonia Congenita](#)

Dupré, Nicolas, MD, MSc; Université Laval

[Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum](#)[SYNE1 Deficiency](#)

Dupuis, Lucie, MS, MSc; The Hospital for Sick Children and University of Toronto

[Caffey Disease](#)

Duque Lasio, Maria Laura, MD; Washington University School of Medicine

[Pyruvate Carboxylase Deficiency](#)

Durda, Kate M, MS; University of Iowa

[IRF6-Related Disorders](#)

Durie, Peter R, MD; The Hospital for Sick Children, University of Toronto

[Shwachman-Diamond Syndrome](#)

Durr, Alexandra, MD, PhD; Hôpital de la Pitié-Salpêtrière

[Parkin Type of Early-Onset Parkinson Disease](#)[Spastic Paraplegia 11](#)[Spastic Paraplegia 4](#)[Spinocerebellar Ataxia Type 28](#)

Dye, Thomas J, MD; Cincinnati Children's Hospital Medical Center

[DLG4-Related Synaptopathy](#)

Dykens, Elisabeth, PhD; University of California, Los Angeles

Smith-Magenis Syndrome

Dyment, David A, DPhil, MD; Children's Hospital of Eastern Ontario

[ASAH1-Related Disorders](#)

[SHORT Syndrome](#)

[TRPM3-Related Neurodevelopmental Disorder](#)

Délot, Emmanuèle C, PhD; Children's National Hospital

[Nonsyndromic 46,XX Testicular Disorders/Differences of Sex Development](#)

E

Earl, Rachel, PhD; University of Washington

[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

Ebens, Christen, MD, MPH; University of Minnesota

[Fanconi Anemia](#)

Ebrahimi-Fakhari, Darius, MD, PhD; Harvard Medical School

[AP-4-Associated Hereditary Spastic Paraplegia](#)

[EPG5-Related Disorder](#)

[PRRT2-Associated Paroxysmal Movement Disorders](#)

[Spastic Paraplegia 15](#)

[TECPR2-Related Hereditary Sensory and Autonomic Neuropathy with Intellectual Disability](#)

Eccles, Michael R, PhD; University of Otago

[PAX2-Related Disorder](#)

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[Adenine Phosphoribosyltransferase Deficiency](#)

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[Primary Congenital Glaucoma](#)

Edwards, Corwin Q, MD; University of Utah School of Medicine

[HFE-Related Hemochromatosis](#)

Efthymiou, Stephanie, MSc, PhD; University College London

[GRIA2-Related Neurodevelopmental Disorder](#)

[WARS2 Deficiency](#)

Eggens, Veerle RC, MSc; University of Amsterdam

[EXOSC3 Pontocerebellar Hypoplasia](#)

[TSEN54 Pontocerebellar Hypoplasia](#)

Eichler, Evan E, PhD; University of Washington School of Medicine

[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

[DYRK1A Syndrome](#)

Eichler, Florian S, MD; Massachusetts General Hospital

[Canavan Disease](#)

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[NOTCH3-Related Lateral Meningocele Syndrome](#)

El-Gharbawy, Areeg, MD; Duke University Medical Center

[Glycogen Storage Disease Type I](#)

El-Hattab, Ayman W, MD; University of Sharjah

[Asparagine Synthetase Deficiency](#)

[Deoxyguanosine Kinase Deficiency](#)

[El-Hattab-Alkuraya Syndrome](#)

[FARS2 Deficiency](#)

[FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion Syndrome](#)

[Glycogen Storage Disease Type IV](#)

[MELAS](#)

[Mitochondrial DNA Maintenance Defects Overview](#)

[MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect](#)

[SUCLA2-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria](#)

[SUCLG1-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria](#)

[Systemic Primary Carnitine Deficiency](#)

[TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form](#)

[Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated](#)

Elango, Sonya, MS; Icahn School of Medicine at Mount Sinai / Mount Sinai Beth Israel

[LRRK2 Parkinson Disease](#)

Elizondo, Leah I; Baylor College of Medicine

[Schimke Immunoosseous Dysplasia](#)

Elliott, Alison M, MS, PhD; University of British Columbia

[Ritscher-Schinzel Syndrome](#)

Ellison, David H, MD; Oregon Health & Science University

[Pseudohypoaldosteronism Type II](#)

Elsas, Louis J, MD; University of Miami

[Classic Galactosemia and Clinical Variant Galactosemia](#)

Elsea, Sarah H, PhD; Baylor College of Medicine

[Aromatic L-Amino Acid Decarboxylase Deficiency](#)

[MBD5 Haploinsufficiency](#)

[Smith-Magenis Syndrome](#)

Else, Tobias, MD; University of Michigan

[Hereditary Paraganglioma-Pheochromocytoma Syndromes](#)
[POT1 Tumor Predisposition](#)

Elsharkawi, Ibrahim, MD; Washington University

[Pitt-Hopkins Syndrome](#)

Emanuel, Beverly S, PhD; Children's Hospital of Philadelphia

[22q11.2 Deletion Syndrome](#)

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Eng, Charis, MD, PhD; Cleveland Clinic

[Multiple Endocrine Neoplasia Type 2](#)
[PTEN Hamartoma Tumor Syndrome](#)

Engelborghs, Sebastiaan, MD, PhD; University of Antwerp

[C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia](#)

Engelen, Marc, MD, PhD; Amsterdam University Medical Centers

[Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation](#)

England, Jade, MD, MSc; University of Montreal

[Spondylometaphyseal Dysplasia, Corner Fracture Type](#)

Engle, Elizabeth C, MD; Children's Hospital Boston

[Congenital Fibrosis of the Extraocular Muscles Overview](#)
[Duane Syndrome](#)

Enns, Gregory M, MB ChB; Stanford University

[Carnitine-Acylcarnitine Translocase Deficiency](#)
[MERRF](#)
[NGLY1-Related Congenital Disorder of Deglycosylation](#)

Erez, Ayelet, MD, PhD; Baylor College of Medicine

[Argininosuccinate Lyase Deficiency](#)

Erro, Roberto, MD, PhD; University of Salerno

[Familial Paroxysmal Nonkinesigenic Dyskinesia](#)

Erwin, Angelika, MD, PhD; Cleveland Clinic

[Congenital Erythropoietic Porphyria](#)

Escolar, Maria L, MD, MS; Children's Hospital of Pittsburgh – UPMC

[Krabbe Disease](#)

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[PRICKLE1-Related Disorders](#)

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[Oculopharyngeal Muscular Dystrophy](#)

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[Nevoid Basal Cell Carcinoma Syndrome](#)

[NF2-Related Schwannomatosis](#)

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[Apert Syndrome](#)

[FGFR Craniosynostosis Syndromes Overview](#)

Everett, Lorraine A; National Human Genome Research Institute

[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

Evidente, Virgilio Gerald H, MD; Movement Disorders Center of Arizona

[X-Linked Dystonia-Parkinsonism Syndrome](#)

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[Fumarate Hydratase Deficiency](#)

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[ALS2-Related Disorder](#)

F

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[Trichohepatoenteric Syndrome](#)

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[Nonsyndromic Retinitis Pigmentosa Overview](#)

Fahrner, Jill A, MD, PhD; Johns Hopkins School of Medicine

[TET3-Related Beck-Fahrner Syndrome](#)

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[Multiple Endocrine Neoplasia Type 1](#)

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[INSR-Related Severe Insulin Resistance Syndrome](#)

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[Cohen Syndrome](#)

[FGFR Craniosynostosis Syndromes Overview](#)

[Mitochondrial DNA Deletion Syndromes](#)

[Primary Pyruvate Dehydrogenase Complex Deficiency Overview](#)

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[Rhizomelic Chondrodysplasia Punctata Type 1](#)

Fam, Hok Khim, BSc; University of British Columbia

[Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive](#)

Fanin, Marina, PhD; University of Padova

[Calpainopathy](#)

Fanto, Manolis, PhD; King's College London

[EPG5-Related Disorder](#)

Fan, Yuting, MD; St Luke's Hospital

[Y Chromosome Infertility](#)

Faqeih, Eissa, MD; King Fahad Medical City

[FARS2 Deficiency](#)

Farlow, Janice, BA, BS; Indiana University School of Medicine

[Parkinson Disease Overview](#)

Farndon, Peter A, MD; Birmingham Women's Hospital

[Nevoid Basal Cell Carcinoma Syndrome](#)

Farrer, Matthew, PhD; University of British Columbia

[LRRK2 Parkinson Disease](#)

Fasham, James, BSc, MBChB, PhD; University of Exeter Medical School

[HYAL2 Deficiency](#)

Fatemi, Ali, MD; Kennedy Krieger Institute

[X-Linked Adrenoleukodystrophy](#)

Fauth, Christine, MD; Medical University of Innsbruck

[FKBP14 Kyphoscoliotic Ehlers-Danlos Syndrome](#)

Fecarotta, Simona, MD; Federico II University

[Lysinuric Protein Intolerance](#)

Fechner, Patricia Y, MD; University of Washington

[Nonsyndromic Disorders of Testicular Development Overview](#)

Federico, Antonio, MD; University of Siena

[Cerebrotendinous Xanthomatosis](#)

Feldman, Gerald L, MD, PhD; Wayne State University School of Medicine

[BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer](#)

Feldman, Howard H, MD; University of British Columbia

[GRN Frontotemporal Dementia](#)

Felton, Thomas, MS; McLendon Clinical Laboratories

[DRPLA](#)

Ferreira, Carlos R, MD; National Human Genome Research Institute

[Barth Syndrome](#)

[Generalized Arterial Calcification of Infancy](#)

[Osteoglophonic Dysplasia](#)

[Primrose Syndrome](#)

[Prolidase Deficiency](#)

[Saul-Wilson Syndrome](#)

Fete, Mary, MSN, RN; National Foundation for Ectodermal Dysplasias

[Hypohidrotic Ectodermal Dysplasia](#)

Field, Amanda, MPH; ResourcePath

[DICER1 Tumor Predisposition](#)

Figlewicz, Denise A, PhD; University of Michigan Medical School

[Facioscapulohumeral Muscular Dystrophy](#)

Filipovich, Alexandra H, MD; Cincinnati Children's Hospital

[Familial Hemophagocytic Lymphohistiocytosis](#)

[WAS-Related Disorders](#)

[X-Linked Hyper IgM Syndrome](#)

[X-Linked Lymphoproliferative Disease](#)

Finanger, Erika, MD; Oregon Health and Science University

[Spinal Muscular Atrophy](#)

Fink, John K, MD; University of Michigan

[Hereditary Spastic Paraplegia Overview](#)

Finucane, Brenda M, MS; Geisinger Health System

[17q12 Recurrent Deletion Syndrome](#)

[Maternal 15q Duplication Syndrome](#)

[Smith-Magenis Syndrome](#)

Fishbein, Lauren, MD, PhD; University of Colorado School of Medicine

[Hereditary Paraganglioma-Pheochromocytoma Syndromes](#)

Fisher, Simon E, DPhil; Max Planck Institute for Psycholinguistics

[FOXP2-Related Speech and Language Disorder](#)

Fish, Jennifer, PhD; University of Massachusetts Lowell

[SATB2-Associated Syndrome](#)

Fiskerstrand, Torunn, MD, PhD; University of Bergen

[ADAMTSL4-Related Eye Disorders](#)

Fitzgibbon, Jude, PhD; Queen Mary University of London

[CEBPA-Associated Familial Acute Myeloid Leukemia \(AML\)](#)

FitzPatrick, David R, MD; University of Edinburgh

[SOX2 Disorder](#)

Flanagan, Maeve, BA; Weill Cornell Medical College

[Bloom Syndrome](#)

Flanigan, Kevin M, MD; Nationwide Children's Hospital

[Collagen VI-Related Dystrophies](#)

Fleming, Judith C, PhD

[Thiamine-Responsive Megaloblastic Anemia Syndrome](#)

Flemming, Kelly D, MD; Mayo Clinic

[Familial Cerebral Cavernous Malformation](#)

Florenzano, Pablo, MD; National Institute of Dental and Craniofacial Research

[Fibrous Dysplasia / McCune-Albright Syndrome](#)

Fluharty, Arvan L, PhD; University of California, Los Angeles

[Arylsulfatase A Deficiency](#)

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[Childhood Ataxia with Central Nervous System Hypomyelination / Vanishing White Matter](#)

Foley, A Reghan, MD; National Institute of Neurological Disorders and Stroke

[Collagen VI-Related Dystrophies](#)

Fong, Jamie C, MS; University of California, San Francisco

[Holt-Oram Syndrome](#)

Fontaine, Bertrand, MD, PhD; Assistance Publique - Hôpitaux de Paris

[Hypokalemic Periodic Paralysis](#)

Foroud, Tatiana, PhD; Indiana University School of Medicine

[Parkinson Disease Overview](#)

Forsythe, Elizabeth, BMedSci, MBBS; University College London

[Bardet-Biedl Syndrome Overview](#)

Forsyth, RaeLynn, MD; Johns Hopkins University School of Medicine

[Bardet-Biedl Syndrome Overview](#)

Forwood, Caitlin, BSc (Hons), MBBS (Hons); Sydney Children's Hospitals Network

[CLCN4-Related Neurodevelopmental Disorder](#)

Foss, Kimberly, MS; Seattle Children's Hospital

[PPP2R5D-Related Neurodevelopmental Disorder](#)

Foster, Rebecca, PhD; St Louis Children's Hospital

[Smith-Magenis Syndrome](#)

Fournier, Emmanuel, MD, PhD; Assistance Publique – Hôpitaux de Paris

[Hypokalemic Periodic Paralysis](#)

Fox, Mark H, MD; University of Iowa

[PRICKLE1-Related Disorders](#)

Franco, Brunella, MD; University of Naples Federico II

[Microphthalmia with Linear Skin Defects Syndrome](#)[Oral-Facial-Digital Syndrome Type I](#)

Francomano, Clair A, MD; National Institutes of Health

[Achondroplasia](#)[Hypochondroplasia](#)

Franques, Jérôme, MD; Assistance Publique – Hôpitaux de Marseille

[Hypokalemic Periodic Paralysis](#)

Frantzen, Carlijn, MD; University Medical Center Groningen

[Von Hippel-Lindau Syndrome](#)

Frazier, Marsha L, PhD; UT MD Anderson Cancer Center

[Peutz-Jeghers Syndrome](#)

Freeman, Alexandra F, MD; National Institutes of Health

[STAT3 Hyper IgE Syndrome](#)

French, Tegan; Murdoch Children's Research Institute

[Thanatophoric Dysplasia](#)

Frey, Beat M, MD; Swiss Red Cross

[McLeod Neuroacanthocytosis Syndrome](#)

Fridovich-Keil, Judith L, PhD; Emory University School of Medicine

[Duarte Variant Galactosemia](#)[Epimerase Deficiency Galactosemia](#)

Friedman, Jan M, MD, PhD; University of British Columbia

[Neurofibromatosis 1](#)

Friedman, Jennifer, MD; University of California San Diego

[ADCY5 Dyskinesia](#)[Sepiapterin Reductase Deficiency](#)

Friedman, Thomas B, PhD; National Institute on Deafness and Other Communication Disorders

[Perrault Syndrome](#)

Friez, Michael J, PhD; Greenwood Genetic Center

[GNPTAB-Related Disorders](#)

Frontali, Marina, MD; Italian National Research Council

[DRPLA](#)

Frühwald, Michael C, MD, PhD; University Medical Center Augsburg
[Rhabdoid Tumor Predisposition Syndrome](#)

Fu, Jiao, MD; University of Chicago Medical Center
[Allan-Herndon-Dudley Syndrome](#)

Fujimura, Frank K, PhD; GMP Genetics, Inc
[Hemophilia A](#)
[Hemophilia B](#)

Fukutake, Toshio, MD, PhD; Kameda Medical Center
[HTRA1 Disorder](#)

Fung, Jasmine LF; University of Hong Kong
[MN1 C-Terminal Truncation Syndrome](#)

Furukawa, Yoshiaki, MD, PhD; Juntendo University Faculty of Medicine
[GTP Cyclohydrolase 1-Deficient Dopa-Responsive Dystonia](#)
[Tyrosine Hydroxylase Deficiency](#)

G

Gabriele, Michele, MS, PhD; European Institute of Oncology
[Gabriele-de Vries Syndrome](#)

Gadea, Randi, MS; Children's Mercy Hospitals and Clinics
[Ayme-Gripp Syndrome](#)

Gafni, Rachel I, MD; National Institute of Dental and Craniofacial Research
[Hyperphosphatemic Familial Tumoral Calcinosis](#)

Gahl, William A, MD, PhD; National Human Genome Research Institute
[Alkaptonuria](#)
[Costeff Syndrome](#)
[Cystinosis](#)
[Free Sialic Acid Storage Disorders](#)
[Generalized Arterial Calcification of Infancy](#)
[Hermansky-Pudlak Syndrome](#)

Galanello, Renzo, MD; Ospedale Regionale Microcitemie
[Alpha-Thalassemia](#)
[Beta-Thalassemia](#)

Gallagher, Emily R, MD, MPH; University of Washington
[Saethre-Chotzen Syndrome](#)

Gallie, Brenda L, MD; Hospital for Sick Children
[Retinoblastoma](#)

Gallus, Gian Nicola, Dsci; University of Siena

[Cerebrotendinous Xanthomatosis](#)

Gamache, Pierre-Luc, MD, PhD; CHU de Québec

[SYNE1 Deficiency](#)

Gambello, Michael J, MD, PhD; Emory University School of Medicine

[3q29 Recurrent Deletion](#)

[Duarte Variant Galactosemia](#)

Ganesh, Jaya, MD; Icahn School of Medicine at Mount Sinai

[Lathosterolosis](#)

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[Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency](#)

[Primary Pyruvate Dehydrogenase Complex Deficiency Overview](#)

[TRMU Deficiency](#)

Gangishetti, Prasanna K, MBBS; Mount Sinai School of Medicine

[21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia](#)

Garber, Judy, MD, MPH; Dana Farber Cancer Institute

[Li-Fraumeni Syndrome](#)

Garber, Kathryn, PhD; Emory University School of Medicine

[CTCF-Related Disorder](#)

Garbern, James Y, MD, PhD; University of Rochester Medical Center

[PLP1 Disorders](#)

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[Pulmonary Fibrosis Predisposition Overview](#)

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[Spinocerebellar Ataxia Type 36](#)

Garden, Gwenn A, MD, PhD; University of Washington

[Spinocerebellar Ataxia Type 7](#)

Gardner, Kathy Lou, MD; University of Pittsburgh

[Familial Hemiplegic Migraine](#)

Gardner, RJM, MB ChB; University of Otago

[Spinocerebellar Ataxia Type 20](#)

Garland, Emily M, PhD; Vanderbilt University Medical Center

[Dopamine Beta-Hydroxylase Deficiency](#)

Gasior, Maciej, MD, PhD; George Washington University School of Medicine

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Gassner, Christoph, PhD; Swiss Red Cross

[McLeod Neuroacanthocytosis Syndrome](#)

Gatti, Richard A, MD; David Geffen School of Medicine at UCLA

[Ataxia-Telangiectasia](#)

[Nijmegen Breakage Syndrome](#)

Gaudette, Mara, MS; Northwestern University Medical School

[Amyotrophic Lateral Sclerosis Overview](#)

Gaughan, Sommer, RD; Children's Hospital Colorado

[Hereditary Fructose Intolerance](#)

Gauvreau, Claudie, MD, MSc; Laval University

[Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum](#)

Gazdagh, Gabriella, MD; University Hospital Southampton

[TRIO-Related Neurodevelopmental Disorder](#)

Gazda, Hanna T, MD, PhD; Harvard Medical School

[Diamond-Blackfan Anemia](#)

Gazzerro, Elisabetta, MD; Gaslini Institute

[Hypomyelination and Congenital Cataract](#)

Gear, Russell, MB ChB; Victorian Clinical Genetics Services

[Osteopathia Striata with Cranial Sclerosis](#)

Geberhiwot, Tarekegn, MD; University Hospitals

[Alstrom Syndrome](#)

Gelb, Bruce D, MD; Icahn School of Medicine at Mount Sinai

[Char Syndrome](#)

[Noonan Syndrome with Multiple Lentigines](#)

German, James, MD; Weill Cornell Medical College

[Bloom Syndrome](#)

Gertler, Tracy, MD, PhD; Children's Hospital of Chicago

[KCNT1-Related Epilepsy](#)

Geschwind, Daniel H, MD, PhD; University of California, Los Angeles

[Primary Familial Brain Calcification](#)

Ghali, Neeti, MBChB, MD; London North West University Healthcare NHS Trust

[TNXB-Related Classical-Like Ehlers-Danlos Syndrome](#)

Ghosh, Partha S, MD; Boston Children's Hospital

[Dystrophinopathies](#)

Ghosh, Rajarshi, PhD; National Institute of Allergy and Infectious Diseases

[GARS-Associated Axonal Neuropathy](#)

Gibbs, Richard A, PhD; Baylor College of Medicine

[Xia-Gibbs Syndrome](#)

Gibson, K Michael, PhD; Washington State University Spokane

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Gibson, Ronald L, MD, PhD; University of Washington School of Medicine

[Cystic Fibrosis](#)

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[EED-Related Overgrowth](#)[SETD1B-Related Neurodevelopmental Disorder](#)

Gidaro, Teresa, MD, PhD; Institut de Myologie, CNRS

[Oculopharyngeal Muscular Dystrophy](#)

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[Hereditary Distal Renal Tubular Acidosis](#)

Gilbert, Melissa A, PhD; Children's Hospital of Philadelphia

[Alagille Syndrome](#)

Giles, Rachel H, PhD; University Medical Center Utrecht

[Von Hippel-Lindau Syndrome](#)

Gillis, David, MD; Hadassah Hebrew University Hospital

[Familial Hyperinsulinism](#)

Gimpel, Charlotte, MD; Heidelberg University

[Autosomal Recessive Polycystic Kidney Disease – PKHD1](#)

Ginevic, Ilona, RD; Icahn School of Medicine at Mount Sinai

[Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency / Trifunctional Protein Deficiency](#)

Giovanni, Monica A, MS; Geisinger Health System

[Lysosomal Acid Lipase Deficiency](#)

Girirajan, Santhosh, MBBS, PhD; Pennsylvania State University

[16p12.2 Recurrent Deletion](#)[Smith-Magenis Syndrome](#)

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[EBF3 Neurodevelopmental Disorder](#)[EXOC6B-Related Spondyloepimetaphyseal Dysplasia with Joint Laxity](#)[ISCA1-Related Multiple Mitochondrial Dysfunctions Syndrome](#)[Multicentric Osteolysis Nodulosis and Arthropathy](#)[Progressive Pseudorheumatoid Dysplasia](#)

Giunta, Cecilia, PhD; University Children's Hospital
[FKBP14 Kyphoscoliotic Ehlers-Danlos Syndrome](#)

Giusti, Francesca, MD, PhD; Villa Donatello Hospital
[Multiple Endocrine Neoplasia Type 1](#)

Glaser, Benjamin, MD; Hadassah-Hebrew University Medical Center
[Familial Hyperinsulinism](#)

Glassford, Megan, MMSc; Emory University School of Medicine
[3q29 Recurrent Deletion](#)

Glass, Ian, MBChB, MD; University of Washington
[Joubert Syndrome](#)
[SHOX Deficiency Disorders](#)

Glinton, Kevin, MD, PhD; Baylor College of Medicine
[TANGO2 Deficiency](#)

Gochuico, Bernadette R, MD; National Human Genome Research Institute
[Hermansky-Pudlak Syndrome](#)

Godfrey, Maurice, PhD; University of Nebraska Medical Center
[Congenital Contractural Arachnodactyly](#)

Gogineni, Kishore Sai, MSc; Kasturba Medical College, Manipal
[LPIN2-Related Majeed Syndrome](#)

Golabi, Mahin, MD, MPH; San Francisco General Hospital
[Simpson-Golabi-Behmel Syndrome Type 1](#)

Golas, Gretchen A, MS, RN; National Institutes of Health
[Chediak-Higashi Syndrome](#)

Gold-Von Simson, Gabrielle, BA, MD; NYU Medical Center
[NTRK1 Congenital Insensitivity to Pain with Anhidrosis](#)

Goldberg, Ethan M, MD, PhD; Children's Hospital of Philadelphia
[SCN3A-Related Neurodevelopmental Disorder](#)

Goldberg, Y Paul, MBChB, PhD; Xenon Pharmaceuticals Inc
[Juvenile Hemochromatosis](#)

Goldfarb, Lev G, MD; National Institute of Neurological Disorders and Stroke / NIH
[GARS-Associated Axonal Neuropathy](#)

Goldin, Ehud, PhD; National Institutes of Health
[Mucopolidosis IV](#)

Goldman, I David, MD; Albert Einstein College of Medicine
[FOLR1-Related Cerebral Folate Transport Deficiency](#)

Hereditary Folate Malabsorption

Goldstein, Amy, MD; Children's Hospital of Philadelphia

Mitochondrial DNA Deletion Syndromes

Goldstein, David B, PhD; Columbia University Irving Medical Center

GNB1 Encephalopathy

Goldstein, Jennifer L, MS, PhD; University of North Carolina-Chapel Hill

Glycogen Storage Disease Type I

Phosphorylase Kinase Deficiency

Gomes, Alicia, MS; University of Alabama

LZTR1- and SMARCB1-Related Schwannomatosis

Gomez-Ospina, Natalia, MD, PhD; Stanford University

Arylsulfatase A Deficiency

Gomez, Christopher M, MD, PhD; University of Chicago

Spinocerebellar Ataxia Type 6

Gonzalez-Gandolfi, Christina X; Children's Hospital of Philadelphia

Simpson-Golabi-Behmel Syndrome Type 1

Goodeve, Anne C, PhD; Sheffield Children's NHS Foundation Trust

von Willebrand Disease

Goodman, Stephen I, MD; University of Colorado

Glutaric Acidemia Type I

Goodnight, Scott H, MD; Oregon Health and Science University

Factor V Leiden Thrombophilia

Goodspeed, Kimberly, MD; University of Texas Southwestern Medical Center

Aspartylglucosaminuria

SLC6A1-Related Neurodevelopmental Disorder

Gordillo, Miriam, PhD; Mount Sinai School of Medicine

ESCO2 Spectrum Disorder

Gordon, Christopher T, PhD; Institut Imagine

ANKRD17-Related Neurodevelopmental Syndrome

MN1 C-Terminal Truncation Syndrome

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Milroy Disease

Gordon, Leslie B, MD, PhD; Alpert Medical School of Brown University

Hutchinson-Gilford Progeria Syndrome

Gorell, Emily, DO, MS; University of Cincinnati College of Medicine

[Epidermolysis Bullosa with Pyloric Atresia](#)

Gorman, Gráinne S, MBBCh, PhD; Newcastle University

[RRM2B Mitochondrial DNA Maintenance Defects](#)

Gorospe, J Rafael, MD, PhD; George Washington University School of Medicine

[Alexander Disease](#)

Gosky, Michael, BS, MS; University of Pittsburgh

[LMNB1-Related Autosomal Dominant Leukodystrophy](#)

Gospe, Sidney M, MD, PhD; University of Washington

[Hypermanganesemia with Dystonia 1](#)[Pyridoxine-Dependent Epilepsy - ALDH7A1](#)

Gossye, Helena, MD; University of Antwerp

[C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia](#)

Gottlieb, Bruce, PhD; McGill University

[Androgen Insensitivity Syndrome](#)

Gottlob, Irene, MD; University of Leicester

[FRMD7-Related Infantile Nystagmus](#)

Goudy, Steven, MD; Vanderbilt University

[IRF6-Related Disorders](#)

Goulet, Olivier, MD, PhD; Hôpital Necker-Enfants Malades

[Trichohepatoenteric Syndrome](#)

Gouw, Launce G-C, MD, PhD; University of Utah School of Medicine

[Spinocerebellar Ataxia Type 7](#)

Govindaraj, Periyasamy, PhD; National Institute of Mental Health & Neurosciences

[Huppke-Brendel Syndrome](#)

Graham, Brett, MD, PhD; Indiana University School of Medicine

[TANGO2 Deficiency](#)

Graham, John M, MD, ScD; Cedars-Sinai Medical Center

[Bohring-Opitz Syndrome](#)[KCNK9 Imprinting Syndrome](#)[PIK3CA-Related Overgrowth Spectrum](#)

Graham, Rona K, PhD; University of Sherbrooke

[Huntington Disease](#)

Grand, Kathryn L, MS; Children's Hospital of Philadelphia

[WDR26-Related Intellectual Disability](#)

Grange, Dorothy Katherine, MD; Washington University School of Medicine

[Cantú syndrome](#)

[Hypohidrotic Ectodermal Dysplasia](#)

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[Shprintzen-Goldberg Syndrome](#)

Greenbaum, Larry A, MD, PhD; Emory University

[Hereditary Distal Renal Tubular Acidosis](#)

Greenberg, Samantha, MPH, MS; UT Southwestern Medical Center

[Hereditary Paraganglioma-Pheochromocytoma Syndromes](#)

Greenblatt, Danielle, MB ChB; Guy's and St Thomas' NHS Foundation Trust

[Hidrotic Ectodermal Dysplasia 2](#)

Greene, Carol L, MD; University of Maryland School of Medicine

[Phenylalanine Hydroxylase Deficiency](#)

Green, Eric D, MD, PhD; National Human Genome Research Institute

[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

Greengard, Emily G, MD; University of Minnesota Masonic Children's Hospital

[ALK-Related Neuroblastic Tumor Susceptibility](#)

Green, Glenn Edward, MD; University of Michigan

[Branchiootorenal Spectrum Disorder](#)

[Genetic Hearing Loss Overview](#)

[Jervell and Lange-Nielsen Syndrome](#)

Green, Peter HR, MD; Columbia University Medical Center

[Celiac Disease](#)

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[Type II Collagen Disorders Overview](#)

Gregory, Allison, MS; Oregon Health & Science University

[Beta-Propeller Protein-Associated Neurodegeneration](#)

[Fatty Acid Hydroxylase-Associated Neurodegeneration](#)

[MECR-Related Neurologic Disorder](#)

[Mitochondrial Membrane Protein-Associated Neurodegeneration](#)

[Neurodegeneration with Brain Iron Accumulation Disorders Overview](#)

[Pantothenate Kinase-Associated Neurodegeneration](#)

[PLA2G6-Associated Neurodegeneration](#)

[SLC39A14 Deficiency](#)

Grimaldi, Silvia, MD; Consorzio Siciliano di Riabilitazione – Associazione Italiana Assistenza Spastici (CSR-AIAS)

[DRPLA](#)

Gripp, Karen W, MD; Alfred I duPont Hospital for Children

[Ayme-Gripp Syndrome](#)

[HRAS-Related Costello Syndrome](#)

[NOTCH3-Related Lateral Meningocele Syndrome](#)

Grishchuk, Yulia, PhD; Harvard Medical School

[Mucopolidosis IV](#)

Groffen, Alexander J, PhD; Amsterdam University Medical Center

[Long QT Syndrome Overview](#)

Gropman, Andrea L, MD; Children's National Health System

[Holoprosencephaly Overview](#)

[Smith-Magenis Syndrome](#)

[Urea Cycle Disorders Overview](#)

Gros-Louis, François, PhD; Université Laval

[SYNE1 Deficiency](#)

Gruber, Stephen B, MD, PhD; USC Norris Comprehensive Cancer Center

[Lynch Syndrome](#)

Grzeschik, Karl-Heinz, PhD; Philipps-Universität

[NSDHL-Related Disorders](#)

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[Glycogen Storage Disease Type III](#)

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[LRRK2 Parkinson Disease](#)

Guerin, Andrea, MD, MEd; Queen's University

[Caffey Disease](#)

Gunay-Aygun, Meral, MD; Johns Hopkins University School of Medicine

[Alstrom Syndrome](#)

[Bardet-Biedl Syndrome Overview](#)

[Costeff Syndrome](#)

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[1q21.1 Recurrent Microdeletion](#)

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[Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD](#)

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[WFS1 Spectrum Disorder](#)

Gupta, Sweta, MD; Indiana Hemophilia & Thrombosis Center

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Gurrola, Jose G, MD; University of Iowa

OTOF-Related Deafness

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Hereditary Hemorrhagic Telangiectasia

Günes, Nilay, MD; Istanbul University

Trichorhinophalangeal Syndrome

H

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Beta-Propeller Protein-Associated Neurodegeneration

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Smith-Magenis Syndrome

Hackman, Peter, PhD; University of Helsinki

Salih Myopathy

Udd Distal Myopathy - Tibial Muscular Dystrophy

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CADASIL

Hahn, Angelika F, MD; Western University

Cold-Induced Sweating Syndrome including Crisponi Syndrome

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Huntington Disease

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22q11.2 Deletion Syndrome

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Hypokalemic Periodic Paralysis

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Hypermobile Ehlers-Danlos Syndrome

Hakola, Panu, MD, PhD; Niuvanniemi Hospital

Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy

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1q21.1 Recurrent Microdeletion

Branchiooculofacial Syndrome

[Disorders of GNAS Inactivation](#)

[FGFR Craniosynostosis Syndromes Overview](#)

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[Familial Dysautonomia](#)

[Familial Mediterranean Fever](#)

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[Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy](#)

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[SYNGAP1-Related Intellectual Disability](#)

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[SOST-Related Sclerosing Bone Dysplasias](#)

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[TUBB4A-Related Leukodystrophy](#)

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[SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders](#)

Hamosh, Ada, MD, MPH; Johns Hopkins University School of Medicine

[Nonketotic Hyperglycinemia](#)

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[PTEN Hamartoma Tumor Syndrome](#)

Handley, Mark, PhD; University of Leeds

[RAB18 Deficiency](#)

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[Episodic Ataxia Type 1](#)

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[IPEX Syndrome](#)

[Kabuki Syndrome](#)

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[X-Linked Spondyloepiphyseal Dysplasia Tarda](#)

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[Nonsyndromic Tooth Agenesis Overview](#)

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[Pachyonychia Congenita](#)

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[16p11.2 Recurrent Deletion](#)

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Werner Syndrome

Harbison, Madeleine D, MD; Icahn School of Medicine at Mount Sinai

Silver-Russell Syndrome

Harlalka, Gaurav; St George's, University of London

Troyer Syndrome

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TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia

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DICER1 Tumor Predisposition

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HPRT1 Disorders

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Dent Disease

Polycystic Kidney Disease, Autosomal Dominant

Primary Hyperoxaluria Type 1

Primary Hyperoxaluria Type 3

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Mitochondrial Membrane Protein-Associated Neurodegeneration

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Succinic Semialdehyde Dehydrogenase Deficiency

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Mandibulofacial Dysostosis with Microcephaly

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Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD

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X-Linked Hypophosphatemia

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Rhabdoid Tumor Predisposition Syndrome

Hathaway, Evan R, MS; Children's Hospital of Philadelphia

Simpson-Golabi-Behmel Syndrome Type 1

Haviland, Isabel, MD; Boston Children's Hospital

CDKL5 Deficiency Disorder

Hayashi, Masahiro, MD, PhD; Yamagata University

[Oculocutaneous Albinism Type 4](#)

Hayden, Michael R, ChB, MB, PhD; University of British Columbia

[Huntington Disease](#)

Hayflick, Susan J, MD; Oregon Health & Science University

[Beta-Propeller Protein-Associated Neurodegeneration](#)

[Fatty Acid Hydroxylase-Associated Neurodegeneration](#)

[MECR-Related Neurologic Disorder](#)

[Mitochondrial Membrane Protein-Associated Neurodegeneration](#)

[Neurodegeneration with Brain Iron Accumulation Disorders Overview](#)

[Pantothenate Kinase-Associated Neurodegeneration](#)

[PLA2G6-Associated Neurodegeneration](#)

[SLC39A14 Deficiency](#)

Hedera, Peter, MD, PhD; University of Louisville

[Hereditary Spastic Paraplegia Overview](#)

[Spastic Paraplegia 3A](#)

Hefner, Margaret A, MS; Saint Louis University School of Medicine

[CHD7 Disorder](#)

Hegele, Robert A, MD; Western University

[Abetalipoproteinemia](#)

[APOB-Related Familial Hypobetalipoproteinemia](#)

[Chylomicron Retention Disease](#)

[Familial Combined Hypolipidemia](#)

[Familial Lipoprotein Lipase Deficiency](#)

[Tangier Disease](#)

Hehr, Ute, MD; Center for Human Genetics

[DCX-Related Disorders](#)

Heidlebaugh, Alexis R, ScM; Geisinger Health System

[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

Heiman, Meadow, MS; Indiana Hemophilia & Thrombosis Center

[Complete Plasminogen Activator Inhibitor 1 Deficiency](#)

Heimer, Gali, MD, PhD; Sheba Medical Center, Sackler School of Medicine

[MECR-Related Neurologic Disorder](#)

[TECPR2-Related Hereditary Sensory and Autonomic Neuropathy with Intellectual Disability](#)

Helbig, Ingo, MD; Children's Hospital of Philadelphia

[CHD2-Related Neurodevelopmental Disorders](#)

Helbig, Katherine L, MS; Children's Hospital of Philadelphia

[SCN3A-Related Neurodevelopmental Disorder](#)

Helsmoortel, Céline, MSc; University of Antwerp

[ADNP-Related Disorder](#)

He, Miao, PhD; University of Pennsylvania

[Epimerase Deficiency Galactosemia](#)

Hennekam, Raoul CM, MD, PhD; Academic Medical Center

[Peters Plus Syndrome](#)[TBC1D24-Related Disorders](#)[Trichorhinophalangeal Syndrome](#)

Hennermann, Julia B, MD, PhD; University Medical Center Mainz

[Nonketotic Hyperglycinemia](#)

Henry, Marie-Louise, MS; University of Michigan

[POT1 Tumor Predisposition](#)

Heppner, Jonathan, PhD; University of British Columbia

[Mucopolysaccharidosis Type I](#)

Herbert, Mrudu, MD, MPH; University of Kentucky

[Phosphorylase Kinase Deficiency](#)

Herman, Gail Ellen, MD, PhD; Nationwide Children's Hospital

[Chondrodysplasia Punctata 2, X-Linked](#)[X-Linked Myotubular Myopathy](#)

Herman, Isabella, MD, PhD; Baylor College of Medicine

[ENTPD1-Related Neurodevelopmental Disorder](#)

Hermann, Andreas, MD, PhD; University Medical Center Rostock

[VPS13A Disease](#)

Hershberger, Ray E, MD; The Ohio State University

[Dilated Cardiomyopathy Overview](#)[LMNA-Related Dilated Cardiomyopathy](#)

Hershfield, Michael, MD; Duke University Medical Center

[Adenosine Deaminase Deficiency](#)

HersHKovitz, Tova, MD; The Genetics Institute - Rambam Health Care Campus

[GLYT1 Encephalopathy](#)

Heutink, Peter, PhD; Vrije Universiteit Medical Center

[MAPT-Related Frontotemporal Dementia](#)

Hicks, Debbie, PhD; University of Newcastle upon Tyne

[Collagen VI-Related Dystrophies](#)

High, Frances A, MD, PhD; MassGeneral Hospital for Children
[Congenital Diaphragmatic Hernia Overview](#)

Hildebrand, Michael S, PhD; University of Melbourne
[DFNA2 Nonsyndromic Hearing Loss](#)
[FOXP2-Related Speech and Language Disorder](#)
[Genetic Hearing Loss Overview](#)

Hildebrandt, Friedhelm, MD; Boston Children's Hospital
[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

Hill, D Ashley, MD; George Washington University School of Medicine
[DICER1 Tumor Predisposition](#)

Hines, Melissa, MD; St Jude Children's Research Hospital
[X-Linked Lymphoproliferative Disease](#)

Hing, Anne V, MD; Seattle Children's Hospital
[Apert Syndrome](#)

Hingorani, Melanie, MA, MBBS, MD; Moorfields Eye Hospital
[PAX6-Related Aniridia](#)

Hipp, Heather, MD; Emory University School of Medicine
[FMR1 Disorders](#)

Hirano, Michio, MD; Columbia University Medical Center
[MELAS](#)
[MERRF](#)
[Mitochondrial DNA Deletion Syndromes](#)
[Mitochondrial Neurogastrointestinal Encephalopathy Disease](#)

Hirano, Ryuki, MD, PhD; Kagoshima University Graduate School of Medical and Dental Sciences
[Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive](#)

Hirose, Shinichi, MD, PhD; Fukuoka University
[Autosomal Dominant Sleep-Related Hypermotor \(Hyperkinetic\) Epilepsy](#)

Hirst, Jennifer, PhD; University of Cambridge
[AP-4-Associated Hereditary Spastic Paraplegia](#)

Hisama, Fuki M, MD; University of Washington
[ADCY5 Dyskinesia](#)
[SCN9A Neuropathic Pain Syndromes](#)
[Werner Syndrome](#)

Hisaund, Michael, BSc; University of Leicester
[FRMD7-Related Infantile Nystagmus](#)

Hoang, Stephanie, MSc; University of Alberta Hospital

[Choroideremia](#)

[X-Linked Congenital Retinoschisis](#)

[X-Linked Congenital Stationary Night Blindness](#)

Hobbs, William, MD, PhD; University of Washington

[Sickle Cell Disease](#)

Hobson, Grace M, PhD; Nemours Alfred I duPont Hospital for Children

[Pelizaeus-Merzbacher-Like Disease 1](#)

[PLP1 Disorders](#)

Ho, Carolyn, MD; Brigham and Women's Hospital

[Hypertrophic Cardiomyopathy Overview](#)

Hodge, Jennelle, PhD; Cedars-Sinai Medical Center

[17q12 Recurrent Duplication](#)

Hoffman, Erin P, MS; Littleton Adventist Hospital

[Lysosomal Acid Lipase Deficiency](#)

Hogarth, Penelope, MD; Oregon Health & Science University

[Beta-Propeller Protein-Associated Neurodegeneration](#)

[MECR-Related Neurologic Disorder](#)

[Mitochondrial Membrane Protein-Associated Neurodegeneration](#)

[PLA2G6-Associated Neurodegeneration](#)

Ho, Gladys; The Children's Hospital at Westmead

[MECP2 Disorders](#)

Holder-Espinasse, Muriel, MD, PhD; Guy's and St Thomas' NHS Trust

[Three M Syndrome](#)

Holder, J Lloyd, MD, PhD; Texas Children's Hospital

[SYNGAP1-Related Intellectual Disability](#)

Holland, Steven M, MD; National Institute of Allergy and Infectious Diseases - NIH

[Chronic Granulomatous Disease](#)

[STAT3 Hyper IgE Syndrome](#)

Holm, Ida E, DMSc, MD; Aarhus University

[CHMP2B Frontotemporal Dementia](#)

Hong, Kui, MD, PhD; Heart Institute of Nanchang University

[Brugada Syndrome](#)

Hoogerbrugge, Nicoline, MD, PhD; Radboud University Medical Center

[NTHL1 Tumor Syndrome](#)

Hooper, Amanda J, PhD; University of Western Australia

[Abetalipoproteinemia](#)

[APOB-Related Familial Hypobetalipoproteinemia](#)

[Chylomicron Retention Disease](#)

[Familial Combined Hypolipidemia](#)

[Familial Lipoprotein Lipase Deficiency](#)

[Tangier Disease](#)

Hoover-Fong, Julie E, MD, PhD; Johns Hopkins University

[Autosomal Dominant TRPV4 Disorders](#)

[Nail-Patella Syndrome](#)

Hopfer, Suellen, MS, PhD; Pennsylvania State University

[Primary Familial Brain Calcification](#)

Hopkin, Robert J, MD; University of Cincinnati College of Medicine

[Cytochrome P450 Oxidoreductase Deficiency](#)

Hopkinson, Ian, BSc (Hons), MBChB, PhD; University College London

[Alstrom Syndrome](#)

Hopp, Katharina, PhD; Mayo Clinic - Minnesota

[Dent Disease](#)

Horsthemke, Bernhard, PhD; University of Essen

[Retinoblastoma](#)

Horvath, Anelia, PhD; National Institutes of Health

[Carney Complex](#)

Hosoi, Yasushi, MD; Hamamatsu University School of Medicine

[Aceruloplasminemia](#)

Ho, Stephanie KL, MD; Clinical Genetic Service, Department of Health

[CTNNB1 Neurodevelopmental Disorder](#)

[Strømme Syndrome](#)

Houck, Kimberly, MD; Baylor College of Medicine

[TANGO2 Deficiency](#)

Houge, Gunnar, MD, PhD; Haukeland University Hospital

[PPP2R1A-Related Neurodevelopmental Disorder](#)

Houlden, Henry, MD, PhD; University College London

[DRPLA](#)

[GAA-FGF14-Related Ataxia](#)

[GRIA2-Related Neurodevelopmental Disorder](#)

[NKX6-2-Related Disorder](#)

RFC1 CANVAS / Spectrum Disorder

Riboflavin Transporter Deficiency

Spinocerebellar Ataxia Type 11

WARS2 Deficiency

Howard, Heidi C, PhD; Radboud University Medical Center

Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum

Howard, Vanessa C, MSN, RN; St Jude Children's Research Hospital

X-Linked Agammaglobulinemia

Howe, James R, MD; University of Iowa Hospitals and Clinics

Juvenile Polyposis Syndrome

Hoyme, H Eugene, MD; Sanford Children's Hospital

Hyaline Fibromatosis Syndrome

Hoyos-Martinez, Alfonso, MD; Baylor College of Medicine

TANGO2 Deficiency

Hsiung, Ging-Yuek Robin, MD, MHSc; University of British Columbia and Providence Health Care

GRN Frontotemporal Dementia

Hsu, Amy P; National Institute of Allergy and Infectious Diseases

STAT3 Hyper IgE Syndrome

Hudgins, Louanne, MD; Stanford University

Kabuki Syndrome

Huff, Vicki, PhD; University of Texas MD Anderson Cancer Center

Wilms Tumor Predisposition

Hufnagel, Robert B, MD, PhD; National Eye Institute - NIH

PNPLA6 Disorders

Hughes, Derralynn A, DPhil, MA; University College Medical School

Fabry Disease

Gaucher Disease

Huizing, Marjan, PhD; National Human Genome Research Institute

Costeff Syndrome

GNE Myopathy

Hermansky-Pudlak Syndrome

Hull, Judith L, MS; Memorial Sloan-Kettering Cancer Center

BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Hull, Peter R, MD, PhD; Dalhousie University

Pachyonychia Congenita

- Hulton, Sally-Anne, MD; Birmingham Children's Hospital
[Primary Hyperoxaluria Type 2](#)
- Hume, Stacey, PhD; University of Alberta
[Choroideremia](#)
- Hu, Mimi I, MD; The University of Texas MD Anderson Cancer Center
[CDC73-Related Disorders](#)
- Hundley, Rachel, PhD; Boston Children's Hospital
[16p11.2 Recurrent Deletion](#)
- Hunt, David, MBBS, PhD; Princess Anne Hospital
[PURA-Related Neurodevelopmental Disorders](#)
- Hunter, Alasdair GW, MD; Children's Hospital of Eastern Ontario
[RPS6KA3-Related Intellectual Disability](#)
- Hunter, David G, MD, PhD; Children's Hospital Boston
[Congenital Fibrosis of the Extraocular Muscles Overview](#)
[Duane Syndrome](#)
- Hunter, Elizabeth, BS; UTHealth School of Dentistry at Houston
[Nonsyndromic Tooth Agenesis Overview](#)
- Hunter, Jessica Ezzell, PhD; Kaiser Permanente Northwest
[FMR1 Disorders](#)
- Hunter, JM, PhD; Nationwide Children's Hospital
[Spinal Muscular Atrophy, X-Linked Infantile](#)
- Huntsman, David G, MD; University of British Columbia
[Hereditary Diffuse Gastric Cancer](#)
- Hupertz, Vera, MD; Cleveland Clinic
[Alpha-1 Antitrypsin Deficiency](#)
- Huq, Mahbubul, MD, PhD; University of British Columbia
[Huntington Disease](#)
- Hurley, R Morrison, MD; British Columbia's Children's and Women's Hospital
[Primary Hyperoxaluria Type 1](#)
- Hurst, Anna CE, MD, MS; University of Alabama at Birmingham
[Disorders of GNAS Inactivation](#)
- Huryn, Laryssa A, MD; National Eye Institute
[DICER1 Tumor Predisposition](#)
- Huston, Haley, BS; Bloodworks Northwest
[Hemophilia A](#)

Hemophilia B

Hutchinson, Anne L, BS; Children's Hospital of Philadelphia

Alagille Syndrome

Hyde, Samuel M, MMSc; University of Texas MD Anderson Cancer Center

CDC73-Related Disorders

Häberle, Johannes, MD; University Children's Hospital Zurich

Carbonic Anhydrase VA Deficiency

Iacovazzo, Donato, MD; Queen Mary University of London

X-Linked Acrogigantism

Idkowiak, Jan, MD, PhD; University of Birmingham

Cytochrome P450 Oxidoreductase Deficiency

Idos, Gregory, MD, MS; City of Hope National Medical Center

Lynch Syndrome

Ikeda, Marc D, MD; Children's Hospital of Pittsburgh

ZAP70-Related Combined Immunodeficiency

Ikeda, Shu-ichi, MD, PhD; Shinshu University Hospital

Hereditary Transthyretin Amyloidosis

Ikeda, Yoshio, MD, PhD; Institute of Genetic Medicine

Spinocerebellar Ataxia Type 8

Indo, Yasuhiro, MD, PhD; Kumamoto University Hospital

NTRK1 Congenital Insensitivity to Pain with Anhidrosis

Infante, Elena, MS; University of Pittsburgh

MUTYH Polyposis

Innes, A Micheil, BSc, MD; University of Calgary

Au-Kline Syndrome

SHORT Syndrome

THOC6 Intellectual Disability Syndrome

TRPM3-Related Neurodevelopmental Disorder

Innis, Jeffrey W, MD, PhD; University of Michigan

Hand-Foot-Genital Syndrome

Introne, Wendy J, MD; National Human Genome Research Institute

Alkaptonuria

Chediak-Higashi Syndrome

[Hermansky-Pudlak Syndrome](#)

Irons, Mira, MD; Children's Hospital Boston

[Smith-Lemli-Opitz Syndrome](#)

Irving, Melita, MBBS, MD; Guy's and St Thomas' NHS Trust

[Three M Syndrome](#)

Isaacs, Adrian M, DPhil; University College London

[CHMP2B Frontotemporal Dementia](#)

Ison, Hannah E, MS; Stanford Center for Inherited Cardiovascular Disease

[Familial Hypercholesterolemia](#)

Ito, Daisuke, MD, PhD; Keio University

[BSCL2-Related Neurologic Disorders / Seipinopathy](#)

Iwasa, Yoichiro, MD, PhD; University of Iowa

[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)**J**

Jabs, Ethylin Wang, MD; Icahn School of Medicine at Mount Sinai

[ESCO2 Spectrum Disorder](#)[STAC3 Disorder](#)[Treacher Collins Syndrome](#)

Jackson, Andrew, PhD; University of Edinburgh

[Microcephalic Osteodysplastic Primordial Dwarfism Type II](#)[RNU4atac-opathy](#)

Jackson, Michelle A, MS; University of Texas MD Anderson Cancer Center

[CDC73-Related Disorders](#)

James, Aaron; University of California San Francisco

[Simpson-Golabi-Behmel Syndrome Type 1](#)

James, Paula, MD; Queen's University

[von Willebrand Disease](#)

Jankovic, Joseph, MD; Baylor College of Medicine

[NKX2-1-Related Disorders](#)

Jansen, Anna C, MD, PhD; Universitair Ziekenhuis Brussel

[PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia](#)[Polymicrogyria Overview](#)[Progressive Myoclonus Epilepsy, Lafora Type](#)

Janssens, Veerle, PhD; KU Leuven

[PPP2R1A-Related Neurodevelopmental Disorder](#)

Jasperson, Kory W; University of Utah

[APC-Associated Polyposis Conditions](#)

Jeffery, Steve, PhD; St George's University of London

[Lymphedema-Distichiasis Syndrome](#)

[Milroy Disease](#)

Jen, Joanna C, MD, PhD; Icahn School of Medicine at Mount Sinai

[Familial Hemiplegic Migraine](#)

Jeste, Shafali Spurling, MD; Children's Hospital Los Angeles

[Maternal 15q Duplication Syndrome](#)

Jethva, Reena, MD; St Christopher's Hospital for Children

[Short-Chain Acyl-CoA Dehydrogenase Deficiency](#)

Jewett, Tamison, MD; Wake Forest University School of Medicine

[1q21.1 Recurrent Microdeletion](#)

Jinnah, Hyder A, MD, PhD; Emory University

[HPRT1 Disorders](#)

Jirsa, Milan, MD, PhD; Institute for Clinical and Experimental Medicine

[Rotor Syndrome](#)

Johannesen, Katrine, MD, PhD; Rigshospitalet

[SLC6A1-Related Neurodevelopmental Disorder](#)

Johann, Pascal, MD; German Cancer Consortium (DKTK)

[Rhabdoid Tumor Predisposition Syndrome](#)

Johannsen, Peter, MD, PhD; Rigshospitalet

[CHMP2B Frontotemporal Dementia](#)

Johnson-Kerner, Bethany, MD, PhD; University of California, San Francisco

[DDX3X-Related Neurodevelopmental Disorder](#)

Johnson, Eric W, PhD; Barrow Neurological Institute

[Familial Cerebral Cavernous Malformation](#)

Johnson, Judith, MS; Cincinnati Children's Hospital Medical Center

[Autoimmune Lymphoproliferative Syndrome](#)

[Familial Hemophagocytic Lymphohistiocytosis](#)

[WAS-Related Disorders](#)

[X-Linked Hyper IgM Syndrome](#)

[X-Linked Lymphoproliferative Disease](#)

Johnson, Kyle P, MD; Oregon Health and Science University

Smith-Magenis Syndrome

Johnson, Maribel J, MA, RN; Puget Sound Blood Center

[Hemophilia A](#)

[Hemophilia B](#)

Johnson, Rebecca H, MD; Seattle Children's Hospital

[ALK-Related Neuroblastic Tumor Susceptibility](#)

Johnston, Jennifer J, PhD; National Human Genome Research Institute

[Greig Cephalopolysyndactyly Syndrome](#)

Jones, Mary-Kayt N, BA; University of Iowa Hospitals and Clinics

[GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss](#)

Jordan, Elizabeth, MMSc; The Ohio State University

[Dilated Cardiomyopathy Overview](#)

[LMNA-Related Dilated Cardiomyopathy](#)

Jorgenson, Ronald J, DDS, PhD; Consultant; formerly, President, Applied Genetics, Inc, Austin

[Hypohidrotic Ectodermal Dysplasia](#)

Josephson, Neil C, MD; Seattle Genetics

[Hemophilia A](#)

[Hemophilia B](#)

Joshi, Nishtha; Baylor College of Medicine

[KCNQ2-Related Disorders](#)

[KCNQ3-Related Disorders](#)

Jungbluth, Heinz, MD, PhD; King's College London

[DYNC1H1-Related Disorders](#)

[EPG5-Related Disorder](#)

Jung, Hans H, MD; University Hospital Zurich

[McLeod Neuroacanthocytosis Syndrome](#)

Jurgens, Julie A, PhD; Boston Children's Hospital

[Congenital Fibrosis of the Extraocular Muscles Overview](#)

Jurkat-Rott, Karin, MD, PhD; Ulm University

[Hyperkalemic Periodic Paralysis](#)

Jäggle, Herbert, MD; University of Regensburg

[Achromatopsia](#)

K

Kaback, Michael M, MD; University of California, San Diego

HEXA Disorders

Kacena, Melissa A, PhD; Indiana University School of Medicine

[GATA1-Related Cytopenia](#)

Kahle, Kristopher T, MD, PhD; Massachusetts General Hospital

[Muenke Syndrome](#)

[Pseudohypoaldosteronism Type II](#)

Kakhlon, Or, PhD; Hadassah-Hebrew University Medical Center

[GBE1 Adult Polyglucosan Body Disease](#)

Kalaydjieva, Luba, MD, PhD; The University of Western Australia

[CTDP1-Related Congenital Cataracts, Facial Dysmorphism, and Neuropathy](#)

Kaler, Stephen G, MD, MPH; The Ohio State University College of Medicine

[ATP7A-Related Copper Transport Disorders](#)

Kalfa, Theodosia A, MD, PhD; Cincinnati Children's Hospital Medical Center and University of Cincinnati Medical School

[EPB42-Related Hereditary Spherocytosis](#)

Kalish, Jennifer M, MD, PhD; Children's Hospital of Philadelphia

[Beckwith-Wiedemann Syndrome](#)

[Simpson-Golabi-Behmel Syndrome Type 1](#)

Kalscheuer, Vera, PhD; Max Planck Institute for Molecular Genetics

[CLCN4-Related Neurodevelopmental Disorder](#)

Kamath, Binita M, MBBChir; Children's Hospital of Philadelphia

[Alagille Syndrome](#)

Kamholz, John, MD, PhD; University of Iowa Hospitals & Clinics

[PLP1 Disorders](#)

Kamihara, Junne, MD, PhD; Dana-Farber/Boston Children's Cancer and Blood Disorders Center

[DICER1 Tumor Predisposition](#)

[FH Tumor Predisposition Syndrome](#)

Kamsteeg, Erik-Jan, MD, PhD; Radboud University Medical Center

[ARSACS](#)

[Ataxia-Telangiectasia](#)

Kang, Jingqiong, PhD; Vanderbilt University Medical Center

[SLC6A1-Related Neurodevelopmental Disorder](#)

Kannu, Peter, MB ChB, PhD; The Hospital for Sick Children

[Cherubism](#)

Kantarci, Sibel, PhD; Quest Diagnostics Nichols Institute

[Donnai-Barrow Syndrome](#)

Kapferer-Seebacher, Ines, DMD; Medical University of Innsbruck

[Periodontal Ehlers-Danlos Syndrome](#)

Kaplan, Julie D, MD; Cleveland Clinic Foundation

[MYRF-Related Cardiac Urogenital Syndrome](#)

Karczeski, Barbara A, MA, MS; Johns Hopkins University School of Medicine

[Cystic Fibrosis](#)[Thanatophoric Dysplasia](#)

Karpati, George, MD; Montreal Neurological Institute

[GNE Myopathy](#)

Kashtan, Clifford E, MD; University of Minnesota

[Alport Syndrome](#)

Kaspar, Roger L, PhD; Transderm, Inc

[Pachyonychia Congenita](#)

Katsanis, Sara Huston, MS; Northwestern University

[Treacher Collins Syndrome](#)

Kaurah, Pardeep, MSc, PhD; University of British Columbia

[Hereditary Diffuse Gastric Cancer](#)

Kaur, Parneet, MSc; Manipal Academy of Higher Education

[ISCA1-Related Multiple Mitochondrial Dysfunctions Syndrome](#)

Kaur, Simranpreet, M Phil, MSc; University of Melbourne

[MECP2 Disorders](#)

Kaya, Namik, PhD; King Faisal Specialist Hospital & Research Center

[ISCA2-Related Mitochondrial Disorder](#)[NKX6-2-Related Disorder](#)

Kaylor, Julie, MS; Arkansas Children's Hospital

[SATB2-Associated Syndrome](#)

Kayser, Michael A, DO; Center for Genetic Testing at Saint Francis Hospital

[Alkaptonuria](#)

Keats, Bronya JB, PhD; Louisiana State University Health Sciences Center

[Usher Syndrome Type I](#)[Usher Syndrome Type II](#)

Keegan, Catherine E, MD, PhD; University of Michigan

[Nonsyndromic Disorders of Testicular Development Overview](#)

Kelkar, Janhawi, MD; Icahn School of Medicine at Mount Sinai

[Lathosterolosis](#)

Kelley, Philip M, PhD; Boys Town National Research Hospital

[OTOF-Related Deafness](#)

Kelly, Edward J, PhD; University of Washington

[Bietti Crystalline Dystrophy](#)

Kelly, Thaddeus E, MD, PhD; University of Virginia Hospital

[Hypochondroplasia](#)

Kepler-Noreuil, Kim, MD; University of Wisconsin School of Medicine and Public Health

[Cranioectodermal Dysplasia](#)[PIK3CA-Related Overgrowth Spectrum](#)

Kerrigan, John F, MD; Phoenix Children's Hospital

[Fumarate Hydratase Deficiency](#)

Khaikin, Yannay, BSc; The Hospital for Sick Children

[STXBP1 Encephalopathy with Epilepsy](#)

Khan, Sadiya S, MD; Northwestern University Feinberg School of Medicine

[Complete Plasminogen Activator Inhibitor 1 Deficiency](#)

Khaytin, Ilya, MD, PhD; Northwestern University

[Congenital Central Hypoventilation Syndrome](#)

Kibrom, Sara, MD; Stanford University School of Medicine

[SCARB2-Related Action Myoclonus – Renal Failure Syndrome](#)

Kidd, Kendrah, MS; Wake Forest University School of Medicine

[Autosomal Dominant Tubulointerstitial Kidney Disease -- MUC1](#)[Autosomal Dominant Tubulointerstitial Kidney Disease -- REN](#)[Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD](#)

Kimberling, William J, PhD; Boys Town National Research Hospital

[Usher Syndrome Type I](#)[Usher Syndrome Type II](#)

Kimonis, Virginia, MD; University of California Irvine Medical Center

[Inclusion Body Myopathy with Paget Disease of Bone and/or Frontotemporal Dementia](#)

Kinsley, Lisa, MS; Northwestern University Feinberg School of Medicine

[Amyotrophic Lateral Sclerosis Overview](#)

Kirk, Jessica, BS; Yale University School of Medicine

[GATA1-Related Cytopenia](#)

Kirmani, Salman, MBBS; Aga Khan University

[Hereditary Paraganglioma-Pheochromocytoma Syndromes](#)

- Kirschner, Lawrence, MD, PhD; The Ohio State University Comprehensive Cancer Center
Multiple Endocrine Neoplasia Type 4
- Kishnani, Priya S, MD; Duke University Medical Center
Glycogen Storage Disease Type I
Phosphorylase Kinase Deficiency
- Kish, Stephen, PhD; University of Toronto
Tyrosine Hydroxylase Deficiency
- Klaiman, Cheryl, PhD; Emory University School of Medicine
3q29 Recurrent Deletion
- Klar, Aharon, MD; Hebrew University
INSR-Related Severe Insulin Resistance Syndrome
- Klasson, Timothy D, BSc; University Medical Center Utrecht
Von Hippel-Lindau Syndrome
- Kleefstra, Tjitske, MD, PhD; Radboud University Medical Center
DDX3X-Related Neurodevelopmental Disorder
Kleefstra Syndrome
- Klein-Tasman, Bonita P, PhD; University of Wisconsin-Milwaukee
7q11.23 Duplication Syndrome
- Klein, Christine, MD; University of Lübeck
Hereditary Dystonia Overview
Parkin Type of Early-Onset Parkinson Disease
PINK1 Type of Young-Onset Parkinson Disease
PRRT2-Associated Paroxysmal Movement Disorders
- Klein, Christopher J, MD; Mayo Clinic
DNMT1-Related Disorder
GBE1 Adult Polyglucosan Body Disease
- Klein, Pierre, MSc; Université Pierre et Marie Curie
Oculopharyngeal Muscular Dystrophy
- Klein, Roger D, JD, MD; Cleveland Clinic
CEBPA-Associated Familial Acute Myeloid Leukemia (AML)
Hereditary Paraganglioma-Pheochromocytoma Syndromes
- Klein, Steven D, MD, PhD; Children's Hospital of Philadelphia
Simpson-Golabi-Behmel Syndrome Type 1
- Kleta, Robert, MD, PhD; National Institutes of Health
Cystinosis

Free Sialic Acid Storage Disorders

Kline, Antonie D, MD; Greater Baltimore Medical Center

Au-Kline Syndrome

Klopstock, Thomas, MD; University Hospital, LMU Munich

Mitochondrial Membrane Protein-Associated Neurodegeneration

Kmiec, Tomasz, MD; Children's Memorial Health Institute

Mitochondrial Membrane Protein-Associated Neurodegeneration

Kmoch, Stanislav, PhD; Charles University

Autosomal Dominant Tubulointerstitial Kidney Disease -- MUC1

Autosomal Dominant Tubulointerstitial Kidney Disease -- REN

Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD

Rotor Syndrome

Knappskog, Per Morten, PhD; Haukeland University Hospital

ADAMTSL4-Related Eye Disorders

Cold-Induced Sweating Syndrome including Crisponi Syndrome

Congenital Stromal Corneal Dystrophy

Knight Johnson, Amy, MS; University of Chicago

Maturity-Onset Diabetes of the Young Overview

Knisely, AS, MD; Medizinische Universität Graz

ATP8B1 Deficiency

Rotor Syndrome

Knoers, Nine, MD, PhD; University Medical Center Groningen

Cranioectodermal Dysplasia

Hereditary Nephrogenic Diabetes Insipidus

Nephronophthisis-Related Ciliopathies

Knowles, Joshua W, MD, PhD; Stanford Center for Inherited Cardiovascular Disease

Familial Hypercholesterolemia

Knowles, Michael R, MD; University of North Carolina at Chapel Hill

Primary Ciliary Dyskinesia

Kobayashi, Keiko, PhD; Kagoshima University Graduate School of Medical and Dental Sciences

Citrin Deficiency

Koenekoop, Robert K, MD, PhD; McGill University Health Center

Usher Syndrome Type I

Usher Syndrome Type II

Koene, Saskia, MD, PhD; Leiden University Medical Center

FOXP1 Syndrome

Koenig, Mary Kay, MD; McGovern Medical School

[Tuberous Sclerosis Complex](#)

Koenig, Michel, MD, PhD; Université de Montpellier

[Ataxia with Oculomotor Apraxia Type 2](#)

Kohlhase, Jürgen, MD; SYNLAB Center for Human Genetics

[SALL4-Related Disorders](#)

[Townes-Brocks Syndrome](#)

Kohlmann, Wendy, MS; University of Michigan Health System

[Lynch Syndrome](#)

Kohl, Susanne, MSc, PhD; Institute for Ophthalmic Research

[Achromatopsia](#)

Kolevzon, Alexander, MD; Icahn School of Medicine at Mount Sinai

[FOXP1 Syndrome](#)

Konkle, Barbara A, MD; University of Washington

[Hemophilia A](#)

[Hemophilia B](#)

Konno, Takuya, MD, PhD; Niigata University

[DCTN1-Related Neurodegeneration](#)

Koolen, David A, MD, PhD; Radboud University Medical Center

[Koolen-de Vries Syndrome](#)

Kool, Marcel, PhD; German Cancer Consortium (DKTK)

[Rhabdoid Tumor Predisposition Syndrome](#)

Kooy, Frank, PhD; University of Antwerp

[ADNP-Related Disorder](#)

Korbonits, Márta, MD, PhD; Queen Mary University of London

[AIP Familial Isolated Pituitary Adenomas](#)

[X-Linked Acrogigantism](#)

Kordes, Uwe, MD; University Medical Center Hamburg – Eppendorf

[Rhabdoid Tumor Predisposition Syndrome](#)

Korf, Bruce R, MD, PhD; University of Alabama at Birmingham

[Dystrophinopathies](#)

Kornak, Uwe, MD, PhD; Universitätsmedizin Göttingen

[ATP6V0A2-Related Cutis Laxa](#)

[CLCN7-Related Osteopetrosis](#)

Koskiniemi, Marja-Leena, MD, PhD; University of Helsinki

[Progressive Myoclonic Epilepsy Type 1](#)

Kostic, Ana, PhD; Icahn School of Medicine at Mount Sinai

[FOXP1 Syndrome](#)

Kowdley, Kris V, MD; Virginia Mason Medical Center

[HFE-Related Hemochromatosis](#)

Koy, Anne, MD; University of Cologne

[GNAO1-Related Disorder](#)

Kraemer, Kenneth H, MD; National Cancer Institute

[Xeroderma Pigmentosum](#)

Krahn, Katherine Nash, MS; University of Iowa

[IRF6-Related Disorders](#)

Krajewski, Karen, MS; Wayne State University School of Medicine

[PLP1 Disorders](#)

Kranc, Kamil R, DPhil, MD; Queen Mary University of London

[Fumarate Hydratase Deficiency](#)

Krantz, Ian D, MD; The Children's Hospital of Philadelphia

[Alagille Syndrome](#)

[Cornelia de Lange Syndrome](#)

Krasnewich, Donna M, MD, PhD; National Institutes of Health

[Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview](#)

[PMM2-CDG](#)

Krause, Amanda, MBBCh, PhD; University of the Witwatersrand

[Huntington Disease-Like 2](#)

Krey, Ilona, MD; University of Leipzig Medical Center

[GRIN2D-Related Developmental and Epileptic Encephalopathy](#)

Kriek, Marjolein, MD, PhD; Leiden University Medical Center

[Peters Plus Syndrome](#)

Kronn, David F, MD; New York Medical College

[Hereditary Folate Malabsorption](#)

Kruer, Michael C, MD; University of South Dakota Sanford School of Medicine

[Fatty Acid Hydroxylase-Associated Neurodegeneration](#)

Kruszka, Paul, MD, MPH; GeneDx

[Holoprosencephaly Overview](#)

[Muenke Syndrome](#)

[Weiss-Kruszka Syndrome](#)

Kucine, Nicole, MD, MS; Weill Cornell Medicine

[Bloom Syndrome](#)

Kuhlenbäumer, Gregor, MD, PhD; University of Kiel

[GAN-Related Neurodegeneration](#)

Kuiper, Roland P, PhD; Princess Máxima Center for Pediatric Oncology

[NTHL1 Tumor Syndrome](#)

Kujovich, Jody L, MD; Oregon Health and Science University

[Factor V Leiden Thrombophilia](#)[Prothrombin Thrombophilia](#)

Kumar, Ajith V, MD; Great Ormond Street Hospital

[AIP Familial Isolated Pituitary Adenomas](#)

Kumar, Anil; University of Leicester

[FRMD7-Related Infantile Nystagmus](#)

Kumaran, Neruban, BSc, MBBS; University College London

[Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview](#)

Kumble, Smitha, MBBS; Murdoch Children's Research Institute

[Chondrodysplasia Punctata 2, X-Linked](#)

Kupfer, Gary, MD; Yale University School of Medicine

[Fanconi Anemia](#)

Kurahashi, Hirokazu, MD, PhD; Aichi Medical University

[Autosomal Dominant Sleep-Related Hypermotor \(Hyperkinetic\) Epilepsy](#)

Kurian, Manju Ann, MBBChir, PhD; UCL Great Ormond Street Institute of Child Health

[Aromatic L-Amino Acid Decarboxylase Deficiency](#)[Beta-Propeller Protein-Associated Neurodegeneration](#)[DNAJC6 Parkinson Disease](#)[KMT2B-Related Dystonia](#)[PLA2G6-Associated Neurodegeneration](#)[SLC12A5-Related Epilepsy of Infancy with Migrating Focal Seizures](#)[SLC39A14 Deficiency](#)[SLC6A3-Related Dopamine Transported Deficiency Syndrome](#)

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[GLYT1 Encephalopathy](#)

Kurth, Ingo, MD; Uniklinik RWTH Aachen

[Hereditary Sensory and Autonomic Neuropathy Type II](#)

Kurtz-Nelson, Evangeline C, PhD; University of Washington

[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

Kurtzman, Tracey L; University of Arizona Colleges of Medicine and Science

[Smith-Lemli-Opitz Syndrome](#)

Kushner, Jessica D, MS; Oregon Health and Science University

[Dilated Cardiomyopathy Overview](#)

Kutler, Mary Jo, DO; The Snyder-Robinson Foundation

[Snyder-Robinson Syndrome](#)

Kutsche, Kerstin, PhD; University Medical Center Hamburg-Eppendorf

[CASK Disorders](#)

[EBF3 Neurodevelopmental Disorder](#)

Kälviäinen, Reetta, MD, PhD; Kuopio University Hospital

[Progressive Myoclonic Epilepsy Type 1](#)

Kölker, Stefan, MD; University Hospital Heidelberg

[Classic Isovaleric Acidemia](#)

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[NSDHL-Related Disorders](#)

Küry, Sébastien, DVM, PhD; Service de Génétique médicale – CHU

[Hereditary Fibrosing Poikiloderma with Tendon Contractures, Myopathy, and Pulmonary Fibrosis](#)

L

La Spada, Albert R, MD, PhD; University of California Irvine School of Medicine

[Spinal and Bulbar Muscular Atrophy](#)

[Spinocerebellar Ataxia Type 7](#)

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[Glycogen Storage Disease Type VI](#)

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[Oculopharyngeal Muscular Dystrophy](#)

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[Alpha-1 Antitrypsin Deficiency](#)

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[CHD4 Neurodevelopmental Disorder](#)

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[Hereditary Coproporphyrria](#)

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[Laing Distal Myopathy](#)

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[TANGO2 Deficiency](#)

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[Smith-Magenis Syndrome](#)

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[CHD7 Disorder](#)[TANGO2 Deficiency](#)

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[SLC6A1-Related Neurodevelopmental Disorder](#)

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[NGLY1-Related Congenital Disorder of Deglycosylation](#)[PMM2-CDG](#)

Lamont, Phillipa, PhD; Royal Perth Hospital

[Laing Distal Myopathy](#)

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[Collagen VI-Related Dystrophies](#)

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[Ethylmalonic Encephalopathy](#)

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[Primary Hyperoxaluria Type 1](#)

Lange, Lara M, MD; University of Lübeck

[PINK1 Type of Young-Onset Parkinson Disease](#)

Langer, Arielle L, MD, MPH; Brigham and Women's Hospital

[Beta-Thalassemia](#)

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[Bloom Syndrome](#)

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[Urea Cycle Disorders Overview](#)

Larizza, Lidia, MD; IRCCS Istituto Auxologico Italiano

[Baller-Gerold Syndrome](#)[Poikiloderma with Neutropenia](#)

Larkin, Allyson, MD; Children's Hospital of Pittsburgh

[ZAP70-Related Combined Immunodeficiency](#)

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[Juvenile Polyposis Syndrome](#)

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[Glutaric Acidemia Type I](#)

LaRusch, Jessica, PhD; Ariel Precision Medicine

[Pancreatitis Overview](#)[PRSS1-Related Hereditary Pancreatitis](#)

LaSalle, Janine M, PhD; University of California Davis School of Medicine

[Maternal 15q Duplication Syndrome](#)

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[Cockayne Syndrome](#)

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[X-Linked Hypophosphatemia](#)

Law, Linda, MBA, MD; Advicenne

[Hereditary Distal Renal Tubular Acidosis](#)

Lawlor, Michael W, MD, PhD; Medical College of Wisconsin

[X-Linked Myotubular Myopathy](#)

Lawrence, Jessica, MSc; Alberta Health Services

[Bestrophinopathies](#)

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[PROP1-Related Combined Pituitary Hormone Deficiency](#)

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[Geleophysic Dysplasia](#)

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[Pachyonychia Congenita](#)

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[Spinal Muscular Atrophy](#)

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[Pycnodysostosis](#)

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[Celiac Disease](#)

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[17q12 Recurrent Deletion Syndrome](#)[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

Lee, Bo Hoon, MD; University of Rochester Medical Center

[PURA-Related Neurodevelopmental Disorders](#)

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[Argininosuccinate Lyase Deficiency](#)
[Cleidocranial Dysplasia Spectrum Disorder](#)
[KAT6B Disorders](#)

Lee, Chung U, MD; Stanford University

[Carnitine-Acylcarnitine Translocase Deficiency](#)
[MERRF](#)

Lee, Kristen N, MD; University of Michigan

[Citrullinemia Type I](#)

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[CTNNB1 Neurodevelopmental Disorder](#)
[MN1 C-Terminal Truncation Syndrome](#)

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[McLeod Neuroacanthocytosis Syndrome](#)

Lee, Thomas, MD, MSc; University of Ottawa

[Bestrophinopathies](#)

Legare, Janet M, MD; University of Wisconsin

[Achondroplasia](#)

Legius, Eric H, MD, PhD; University of Leuven

[Legius Syndrome](#)

LeGuern, Eric, MD, PhD; Hôpital de la Pitié-Salpêtrière

[SH3TC2-Related Hereditary Motor and Sensory Neuropathy](#)

Lehesjoki, Anna-Elina, MD, PhD; University of Helsinki

[Marinesco-Sjogren Syndrome](#)
[Progressive Myoclonic Epilepsy Type 1](#)

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[Pyruvate Carboxylase Deficiency](#)

Lehman, Christopher, MS; Geisinger Health System

[16p11.2 Recurrent Deletion](#)

Lehmann-Horn, Frank, MD, MS, PhD; Ulm University

[Hyperkalemic Periodic Paralysis](#)
[Hypokalemic Periodic Paralysis](#)

Lehmberg, Kai, MD; University Medical Center Eppendorf

[Familial Hemophagocytic Lymphohistiocytosis](#)

Leiding, Jennifer W, MD; Johns Hopkins University

Chronic Granulomatous Disease

Leigh, Margaret W, MD; University of North Carolina at Chapel Hill

Primary Ciliary Dyskinesia

Leistritz, Dru F, MS; University of Washington Medical Center

Werner Syndrome

Lemire, Gabrielle, MD; Children's Hospital of Eastern Ontario Research Institute

KAT6B Disorders

THOC6 Intellectual Disability Syndrome

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GRIN1-Related Neurodevelopmental Disorder

GRIN2B-Related Neurodevelopmental Disorder

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Lemmers, Richard JLF, PhD; Leiden University Medical Center

Facioscapulohumeral Muscular Dystrophy

Lemmink, Henny, PhD; University Medical Center

Hereditary Nephrogenic Diabetes Insipidus

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Usher Syndrome Type I

Usher Syndrome Type II

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CDKL5 Deficiency Disorder

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Alagille Syndrome

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Adult Refsum Disease

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GNPTAB-Related Disorders

Leslie, Elizabeth J, PhD; Emory University

IRF6-Related Disorders

Leslie, Nancy D, MD; Cincinnati Children's Hospital Medical Center

Pompe Disease

Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Lesnik Oberstein, Saskia AJ, MD, PhD; Leiden University Medical Center

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Peters Plus Syndrome

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[Nonsyndromic Tooth Agenesis Overview](#)

Leturcq, France, MD; Hôpital Cochin

[Emery-Dreifuss Muscular Dystrophy](#)

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[Simpson-Golabi-Behmel Syndrome Type 1](#)

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[Strømme Syndrome](#)

Leuzzi, Vincenzo, MD; Sapienza Università di Roma

[PRICKLE1-Related Disorders](#)

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[ASAH1-Related Disorders](#)

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[Polymicrogyria Overview](#)

[PURA-Related Neurodevelopmental Disorders](#)

Levine, Michael A, MD; Children's Hospital of Philadelphia

[Disorders of GNAS Inactivation](#)

Levy-Lahad, Ephrat, MD; Sharre Zedek Medical Center

[BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer](#)

Levy, Amanda M, MSc; Rigshospitalet

[DLG4-Related Synaptopathy](#)

Levy, Harvey L, MD; Boston Children's Hospital

[Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency](#)

Levy, Howard P, MD, PhD; Johns Hopkins University School of Medicine

[Hypermobile Ehlers-Danlos Syndrome](#)

Levy, Rebecca V, BM BCh, MSc; Montefiore Medical Center

[17q12 Recurrent Deletion Syndrome](#)

Lewandowska, Magdalena, MD; Indiana Hemophilia & Thrombosis Center

[Complete Plasminogen Activator Inhibitor 1 Deficiency](#)

Lewis, David B, MD; Stanford University School of Medicine

[Schimke Immunoosseous Dysplasia](#)

Lewis, Richard Alan, MD, MS; Baylor College of Medicine

[Lowe Syndrome](#)

Lichter-Konecki, Uta, MD, PhD; UPMC Children's Hospital of Pittsburgh

[Ornithine Transcarbamylase Deficiency](#)

Urea Cycle Disorders Overview

Li, Chumei, MD, PhD; McMaster University

[FREM1 Autosomal Recessive Disorders](#)

Liebau, Max, MD; University of Cologne

[Autosomal Recessive Polycystic Kidney Disease – PKHD1](#)

Lieske, John C, MD; Mayo Clinic

[Dent Disease](#)

[Primary Hyperoxaluria Type 1](#)

[Primary Hyperoxaluria Type 3](#)

Li, Frederick, MD; Dana Farber Cancer Institute

[Li-Fraumeni Syndrome](#)

Li, Hong, MD, PhD; Emory University School of Medicine

[CTCF-Related Disorder](#)

Lilien, Marc, MD, PhD; University Medical Center Utrecht

[Nephronophthisis-Related Ciliopathies](#)

Li, Longchuan, PhD; Emory University School of Medicine

[3q29 Recurrent Deletion](#)

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[RRM2B Mitochondrial DNA Maintenance Defects](#)

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[Branchiooculofacial Syndrome](#)

[Cranioectodermal Dysplasia](#)

[HRAS-Related Costello Syndrome](#)

[MN1 C-Terminal Truncation Syndrome](#)

[Myhre Syndrome](#)

Lindhurst, Marjorie J, PhD; National Institutes of Health

[SLC25A19-Related Thiamine Metabolism Dysfunction](#)

Lindor, Noralane M, MD; Mayo Clinic

[Myhre Syndrome](#)

Lindsay, Mark E, MD, PhD; MassGeneral Hospital for Children

[Myhre Syndrome](#)

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[Mandibulofacial Dysostosis with Microcephaly](#)

[TRPM3-Related Neurodevelopmental Disorder](#)

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[Von Hippel-Lindau Syndrome](#)

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[Spinocerebellar Ataxia Type 1](#)

Lippner, Elizabeth, MD; Ann & Robert H. Lurie Children's Hospital of Chicago

[Schimke Immunoosseous Dysplasia](#)

Lipska-Zietkiewicz, Beata S, MD, PhD; Medical University of Gdansk

[Genetic Steroid-Resistant Nephrotic Syndrome Overview](#)[WT1 Disorder](#)

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[Familial Porphyria Cutanea Tarda](#)[Hepatoerythropoietic Porphyria](#)

Liu, Paul, MD, PhD; National Human Genome Research Institute

[RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies](#)

Lloyd, Ricardo V, MD, PhD; Mayo Clinic

[Hereditary Paraganglioma-Pheochromocytoma Syndromes](#)

Lochmüller, Hanns, MD; Children's Hospital of Eastern Ontario Research Institute

[Congenital Myasthenic Syndromes Overview](#)

Loeys, Bart L, MD, PhD; Antwerp University Hospital

[EFEMP2-Related Cutis Laxa](#)[FBLN5-Related Cutis Laxa](#)[Loeys-Dietz Syndrome](#)

Lohmann, Dietmar R, MD; University Hospital Essen

[Retinoblastoma](#)

Lohmann, Katja, PhD; University of Lübeck

[Hereditary Dystonia Overview](#)

Lohr, Jamie, MD; University of Minnesota Amplatz Children's Hospital

[Autosomal Dominant Robinow Syndrome](#)

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[CTNNB1 Neurodevelopmental Disorder](#)[FAM111A-Related Skeletal Dysplasias](#)[Strømme Syndrome](#)

Longoni, Mauro, MD; Harvard Medical School

[Congenital Diaphragmatic Hernia Overview](#)[Donnai-Barrow Syndrome](#)

Loomes, Kathleen M, MD; University of Pennsylvania

[Alagille Syndrome](#)

Lopez, Christina, BS; California Pacific Medical Center

[Simpson-Golabi-Behmel Syndrome Type 1](#)

LoPiccolo, Mary Kate, MD; Icahn School of Medicine at Mount Sinai

[Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency / Trifunctional Protein Deficiency](#)

Lory, Philippe, PhD; Université de Montpellier 1 et 2

[Hypokalemic Periodic Paralysis](#)

Lossie, Amy C; University of Florida College of Medicine

[Angelman Syndrome](#)

Lossos, Alexander, MD; Hadassah-Hebrew University Medical Center

[GBE1 Adult Polyglucosan Body Disease](#)

Lotze, Timothy, MD; Baylor College of Medicine

[GARS-Associated Axonal Neuropathy](#)

Louie, Raymond J, PhD; Greenwood Genetic Center

[IPEX Syndrome](#)

Lou, Shu, MD; University of British Columbia

[Schimke Immunoosseous Dysplasia](#)

Loyd, James E, MD; Vanderbilt University Medical Center

[Heritable Pulmonary Arterial Hypertension Overview](#)

Lubarr, Naomi, MD; Beth Israel Medical Center

[DYT1 Early-Onset Isolated Dystonia](#)

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[Maternal 15q Duplication Syndrome](#)

Lucia, Alejandro, MD, PhD; European University of Madrid

[Glycogen Storage Disease Type V](#)

Lucking, Christoph, MD; Ludwig-Maximilians-University

[Parkin Type of Early-Onset Parkinson Disease](#)

Lucky, Anne W, MD; Cincinnati Children's Hospital

[Dystrophic Epidermolysis Bullosa](#)

[Epidermolysis Bullosa Simplex](#)

[Epidermolysis Bullosa with Pyloric Atresia](#)

[Junctional Epidermolysis Bullosa](#)

Luk, Ho-ming, MD; Hong Kong Children's Hospital

[CTNNB1 Neurodevelopmental Disorder](#)

[FAM111A-Related Skeletal Dysplasias](#)

Strømme Syndrome

Lupski, James R, MD, PhD; Baylor College of Medicine

Smith-Magenis Syndrome

Lusk, Laina, MMSc; Children's Hospital of Philadelphia

Maternal 15q Duplication Syndrome

PACS1 Neurodevelopmental Disorder

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Cystic Fibrosis

Lynch, Henry, MD; Creighton University Hereditary Cancer Center

MUTYH Polyposis

Lyons, Michael J, MD; Greenwood Genetic Center

MED12-Related Disorders

Lücke, Thomas, MD, PhD; Hannover Medical School

Schimke Immunoosseous Dysplasia

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TXNL4A-Related Craniofacial Disorders

M

Maas, Saskia, MD; University of Amsterdam

Trichorhinophalangeal Syndrome

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NF2-Related Schwannomatosis

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Alstrom Syndrome

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Bestrophinopathies

Choroideremia

Weill-Marchesani Syndrome

X-Linked Congenital Retinoschisis

X-Linked Congenital Stationary Night Blindness

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Mandibulofacial Dysostosis with Microcephaly

VLDLR Cerebellar Hypoplasia

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Juvenile Polyposis Syndrome

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Juvenile Hemochromatosis

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[Cleidocranial Dysplasia Spectrum Disorder](#)

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[Familial Hemophagocytic Lymphohistiocytosis](#)

Mackay, Deborah JG, MA, PhD; Salisbury NHS Foundation Trust

[Diabetes Mellitus, 6q24-Related Transient Neonatal](#)

MacKenzie, Alex, MD; Children's Hospital of Eastern Ontario

[DLG4-Related Synaptopathy](#)

MacLeod, Erin, PhD, RD; Children's National Hospital

[Ornithine Transcarbamylase Deficiency](#)

MacLeod, Heather, MS; University of Chicago

[Arrhythmogenic Right Ventricular Cardiomyopathy Overview](#)

Maconachie, Gail, BSc, PhD; University of Leicester

[FRMD7-Related Infantile Nystagmus](#)

Madhok, Sehajvir, MD; Columbia University Irving Medical Center

[HNRNP2-Related Neurodevelopmental Disorder](#)

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[Smith-Magenis Syndrome](#)

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[Glycogen Storage Disease Type IV](#)

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[Hereditary Folate Malabsorption](#)

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[Bestrophenopathies](#)

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[PLA2G6-Associated Neurodegeneration](#)

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[Molybdenum Cofactor Deficiency](#)

Maillard, Camille, PhD; Université de Paris

[Tubulinopathies Overview](#)

Makaryan, Vahagn, MD; University of Washington Medical Center

[ELANE-Related Neutropenia](#)

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[MN1 C-Terminal Truncation Syndrome](#)

Malfait, Fransiska, MD, PhD; Ghent University Hospital

[Classic Ehlers-Danlos Syndrome](#)

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[Chediak-Higashi Syndrome](#)[GNE Myopathy](#)[Hermansky-Pudlak Syndrome](#)

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[Alpha-Mannosidosis](#)

Mancini, Cecilia, PhD; University of Turin

[Spinocerebellar Ataxia Type 28](#)

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[Fumarate Hydratase Deficiency](#)

Mannens, Marcel MAM, PhD; University of Amsterdam

[Long QT Syndrome Overview](#)

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[TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia](#)

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[Riboflavin Transporter Deficiency](#)

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[Isolated Methylmalonic Acidemia](#)[STAC3 Disorder](#)

Mansour, Sahar; St George's, University of London

[Lymphedema-Distichiasis Syndrome](#)[Milroy Disease](#)

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[Legius Syndrome](#)

Marafi, Dana, MD, MSc; Kuwait University

[El-Hattab-Alkuraya Syndrome](#)

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[Congenital Central Hypoventilation Syndrome](#)

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[Schaaf-Yang Syndrome](#)

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Feingold Syndrome 1

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[Familial Cerebral Cavernous Malformation](#)

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[Spastic Paraplegia 7](#)

Marcucci, Guido, MD; Ohio State University

[CEBPA-Associated Familial Acute Myeloid Leukemia \(AML\)](#)

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[Huntington Disease-Like 2](#)

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[Multiple Endocrine Neoplasia Type 1](#)

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[GARS-Associated Axonal Neuropathy](#)

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[Congenital NAD Deficiency Disorder](#)

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[Multiple Endocrine Neoplasia Type 2](#)

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[SLC39A8-CDG](#)

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[Hereditary Dystonia Overview](#)

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[Alstrom Syndrome](#)

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[Cystic Fibrosis](#)

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[CDKL5 Deficiency Disorder](#)

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[Familial Hemophagocytic Lymphohistiocytosis](#)

[X-Linked Lymphoproliferative Disease](#)

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[17q12 Recurrent Deletion Syndrome](#)

[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

[PACS1 Neurodevelopmental Disorder](#)

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[CHD7 Disorder](#)

Martin, George M, MD; University of Washington

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[CDC73-Related Disorders](#)

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Martín, Bertha, PhD; University of Iowa

[C3 Glomerulopathy](#)

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Marzin, Pauline, MD; Paris Cité University

[Geleophysic Dysplasia](#)[Weill-Marchesani Syndrome](#)

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[PRICKLE1-Related Disorders](#)

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[Genetic Prion Disease](#)

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[Canavan Disease](#)

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[Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

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[Angelman Syndrome](#)

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[ARSACS](#)

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[Spinocerebellar Ataxia Type 37](#)

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[Coffin-Siris Syndrome](#)

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[Spinocerebellar Ataxia Type 10](#)

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[Dent Disease](#)

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[Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome](#)

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[Enlarged Parietal Foramina](#)

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[Catecholaminergic Polymorphic Ventricular Tachycardia](#)

Mazzeu, Juliana, PhD; University of Brasília

[Autosomal Dominant Robinow Syndrome](#)

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[WFS1 Spectrum Disorder](#)

McCormick, Elizabeth M, MS; Children's Hospital of Philadelphia

[Primary Pyruvate Dehydrogenase Complex Deficiency Overview](#)

McCormick, Sally PA, PhD; University of Otago

[Tangier Disease](#)

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[Autosomal Dominant TRPV4 Disorders](#)

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[Spinocerebellar Ataxia Type 3](#)

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[Holt-Oram Syndrome](#)

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[22q11.2 Deletion Syndrome](#)

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[Hereditary Hemorrhagic Telangiectasia](#)

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[Microcephaly-Capillary Malformation Syndrome](#)

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[RRM2B Mitochondrial DNA Maintenance Defects](#)

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[Spondylometaphyseal Dysplasia, Corner Fracture Type](#)

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[Peutz-Jeghers Syndrome](#)

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[Acid Sphingomyelinase Deficiency](#)

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[Nail-Patella Syndrome](#)

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[Pachyonychia Congenita](#)

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[Primary Familial and Congenital Polycythemia](#)

McNally, Elizabeth, MD, PhD; University of Chicago

[Arrhythmogenic Right Ventricular Cardiomyopathy Overview](#)

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[Au-Kline Syndrome](#)

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[Choroideremia](#)

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[SLC12A5-Related Epilepsy of Infancy with Migrating Focal Seizures](#)

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[ASAH1-Related Disorders](#)

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[Emanuel Syndrome](#)

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[15q13.3 Recurrent Deletion](#)[17q12 Recurrent Duplication](#)[CHD2-Related Neurodevelopmental Disorders](#)[SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders](#)

Mehta, Atul, MA, MD; Royal Free Hospital and University College London School of Medicine

[Fabry Disease](#)

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[Fanconi Anemia](#)

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[Spastic Paraplegia 8](#)[VPS13D Movement Disorder](#)

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[SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders](#)

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[Genetic Atypical Hemolytic-Uremic Syndrome](#)

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[ADAMTSL4-Related Eye Disorders](#)

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[Choroideremia](#)

[X-Linked Congenital Retinoschisis](#)

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[PROP1-Related Combined Pituitary Hormone Deficiency](#)

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[Caffey Disease](#)

[Cleidocranial Dysplasia Spectrum Disorder](#)

[MBD5 Haploinsufficiency](#)

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[Nonsyndromic Tooth Agenesis Overview](#)

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[Factor V Leiden Thrombophilia](#)

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[Hereditary Fibrosing Poikiloderma with Tendon Contractures, Myopathy, and Pulmonary Fibrosis](#)

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[Creatine Deficiency Disorders](#)

[STXBP1 Encephalopathy with Epilepsy](#)

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[DICER1 Tumor Predisposition](#)

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[Sitosterolemia](#)

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[GNB5-Related Neurodevelopmental Disorder](#)

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[MID1-Related Opitz G/BBB Syndrome](#)

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[Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

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[7q11.23 Duplication Syndrome](#)

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[INSR-Related Severe Insulin Resistance Syndrome](#)

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[LZTR1- and SMARCB1-Related Schwannomatosis](#)

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[DICER1 Tumor Predisposition](#)

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[SLC39A14 Deficiency](#)

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[X-Linked Lymphoproliferative Disease](#)

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[KCNQ2-Related Disorders](#)[KCNQ3-Related Disorders](#)

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[Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview](#)

Michael, Julianne, MS; Spectrum Health

[Bachmann-Bupp Syndrome](#)

Michals-Matalon, Kimberlee, PhD, RD; University of Houston

[Canavan Disease](#)

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[SYNGAP1-Related Intellectual Disability](#)

Michelucci, Roberto, MD, PhD; Bellaria Hospital

[Autosomal Dominant Epilepsy with Auditory Features](#)

Milewicz, Dianna M, MD, PhD; University of Texas Health Science Center

[Heritable Thoracic Aortic Disease Overview](#)

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[Faciocapulohumeral Muscular Dystrophy](#)[FGFR Craniosynostosis Syndromes Overview](#)

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[16p11.2 Recurrent Deletion](#)[Dystrophinopathies](#)

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[SCN1A-Related Seizure Disorders](#)

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[Prader-Willi Syndrome](#)

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[TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia](#)

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[Mowat-Wilson Syndrome](#)

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[Dent Disease](#)

[Primary Hyperoxaluria Type 1](#)

[Primary Hyperoxaluria Type 3](#)

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[Hypermanganesemia with Dystonia 1](#)

[PNPO Deficiency](#)

[SLC39A14 Deficiency](#)

Milstone, Leonard M, MD; Yale University

[Pachyonychia Congenita](#)

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[VPS13A Disease](#)

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[Branchiooculofacial Syndrome](#)

[Waardenburg Syndrome Type I](#)

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[Hypomyelination and Congenital Cataract](#)

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[CLCN2-Related Leukoencephalopathy](#)

[Megalencephalic Leukoencephalopathy with Subcortical Cysts](#)

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[Hereditary Folate Malabsorption](#)

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[KCNQ2-Related Disorders](#)

[KCNQ3-Related Disorders](#)

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[Microcephaly-Capillary Malformation Syndrome](#)

[MPPH Syndrome](#)

[PIK3CA-Related Overgrowth Spectrum](#)

[Polymicrogyria Overview](#)

[PPP2R5D-Related Neurodevelopmental Disorder](#)

Misko, Albert, MD, PhD; Massachusetts General Hospital / Harvard Medical School

[Molybdenum Cofactor Deficiency](#)

[Mucopolidosis IV](#)

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[Bietti Crystalline Dystrophy](#)

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[17q12 Recurrent Duplication](#)

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[Phenylalanine Hydroxylase Deficiency](#)

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[16p11.2 Recurrent Deletion](#)[17q12 Recurrent Deletion Syndrome](#)[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

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[Achondrogenesis Type 1B](#)[Atelosteogenesis Type 2](#)[Diastrophic Dysplasia](#)[SLC26A2-Related Multiple Epiphyseal Dysplasia](#)

Miyajima, Hiroaki, MD; Hamamatsu University School of Medicine

[Aceruloplasminemia](#)

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[TANGO2 Deficiency](#)

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[16p12.2 Recurrent Deletion](#)

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[Collagen VI-Related Dystrophies](#)

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[Nonsyndromic Disorders of Testicular Development Overview](#)

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[Alpha-Thalassemia](#)

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[VPS13A Disease](#)

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[CASK Disorders](#)[Encephalocraniocutaneous Lipomatosis](#)

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[Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview](#)

[PAX6-Related Aniridia](#)

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[PAX6-Related Aniridia](#)

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[Carnitine-Acylcarnitine Translocase Deficiency](#)

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[Dilated Cardiomyopathy Overview](#)[LMNA-Related Dilated Cardiomyopathy](#)

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[Stickler Syndrome](#)

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[SERAC1 Deficiency](#)

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[Fanconi Anemia](#)

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[PROP1-Related Combined Pituitary Hormone Deficiency](#)

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[Ataxia with Oculomotor Apraxia Type 2](#)

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[KBG Syndrome](#)

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[17q12 Recurrent Deletion Syndrome](#)

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[Oral-Facial-Digital Syndrome Type I](#)

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[FOXP1 Syndrome](#)[FOXP2-Related Speech and Language Disorder](#)[Koolen-de Vries Syndrome](#)[SETBP1 Haploinsufficiency Disorder](#)

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[Chediak-Higashi Syndrome](#)[Schimke Immunoosseous Dysplasia](#)

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[Ornithine Transcarbamylase Deficiency](#)

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[Microphthalmia with Linear Skin Defects Syndrome](#)

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[Hypophosphatasia](#)

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[ATP8B1 Deficiency](#)

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[7q11.23 Duplication Syndrome](#)[Williams Syndrome](#)

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[Familial Cerebral Cavernous Malformation](#)

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[Christianson Syndrome](#)

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[Multiple Epiphyseal Dysplasia, Dominant](#)[Stickler Syndrome](#)[X-Linked Hypophosphatemia](#)

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[Lymphedema-Distichiasis Syndrome](#)[Milroy Disease](#)

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[Maple Syrup Urine Disease](#)

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[Rhizomelic Chondrodysplasia Punctata Type 1](#)[X-Linked Adrenoleukodystrophy](#)[Zellweger Spectrum Disorder](#)

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[X-Linked Adrenoleukodystrophy](#)[Zellweger Spectrum Disorder](#)

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[Cystic Fibrosis](#)

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[HFE-Related Hemochromatosis](#)

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[PRRT2-Associated Paroxysmal Movement Disorders](#)

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[WARS2 Deficiency](#)

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[TBC1D24-Related Disorders](#)

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[Holoprosencephaly Overview](#)[Muenke Syndrome](#)

Mullegama, Sureni V, PhD; Sam Houston State University

[MBD5 Haploinsufficiency](#)

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[3q29 Recurrent Deletion](#)

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[SHOX Deficiency Disorders](#)

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[Legius Syndrome](#)

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[TRMU Deficiency](#)

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[Xia-Gibbs Syndrome](#)

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[3q29 Recurrent Deletion](#)

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[IRF6-Related Disorders](#)

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[Lysosomal Acid Lipase Deficiency](#)

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[Vascular Ehlers-Danlos Syndrome](#)

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[Shwachman-Diamond Syndrome](#)

Myers, Kenneth A, MD, PhD; University of Melbourne

[GRIN2A-Related Speech Disorders and Epilepsy](#)

Myers, Scott M, MD; Geisinger Health System

[17q12 Recurrent Deletion Syndrome](#)[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

Mymin, David, MBBCh; University of Manitoba

[Sitosterolemia](#)

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[Sitosterolemia](#)

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[Cartilage-Hair Hypoplasia - Anauxetic Dysplasia Spectrum Disorders](#)

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[Congenital Mirror Movements](#)

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[DYNC1H1-Related Disorders](#)

Müller, Juliane S, PhD; University of Cambridge

[Congenital Myasthenic Syndromes Overview](#)

Münchau, Alexander, MD; University of Lübeck

[Hereditary Dystonia Overview](#)

Mütze, Ulrike, MD; University Hospital Heidelberg

[Classic Isovaleric Acidemia](#)

N

Nabais Sá, Maria J, MD, MPH, PhD; Radboud University Medical Center

[Gabriele-de Vries Syndrome](#)

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[Argininosuccinate Lyase Deficiency](#)

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[Huppke-Brendel Syndrome](#)

[Isolated Sulfite Oxidase Deficiency](#)

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[Autoimmune Lymphoproliferative Syndrome](#)

[WAS-Related Disorders](#)

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[Canavan Disease](#)

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[WARS2 Deficiency](#)

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[LMNB1-Related Autosomal Dominant Leukodystrophy](#)

[Pelizaeus-Merzbacher-Like Disease 1](#)

[TUBB4A-Related Leukodystrophy](#)

Naidu, Sakkubai, MD; Kennedy Krieger Institute

[Alexander Disease](#)

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[Hereditary Transthyretin Amyloidosis](#)

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[Monosomy 7 Predisposition Syndromes Overview](#)

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[Hemophilia A](#)[Hemophilia B](#)

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[TSEN54 Pontocerebellar Hypoplasia](#)

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[Cockayne Syndrome](#)

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[CACNA1C-Related Disorders](#)[Catecholaminergic Polymorphic Ventricular Tachycardia](#)

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[EBF3 Neurodevelopmental Disorder](#)[ISCA1-Related Multiple Mitochondrial Dysfunctions Syndrome](#)[LPIN2-Related Majeed Syndrome](#)

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[Carnitine Palmitoyltransferase 1A Deficiency](#)

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[MIRAGE Syndrome](#)

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[ZAP70-Related Combined Immunodeficiency](#)

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[16p11.2 Recurrent Deletion](#)

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[PPP2R5D-Related Neurodevelopmental Disorder](#)

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[Primary Coenzyme Q10 Deficiency Overview](#)

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[Maturity-Onset Diabetes of the Young Overview](#)

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[NGLY1-Related Congenital Disorder of Deglycosylation](#)

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[Cockayne Syndrome](#)

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[VPS13A Disease](#)

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[Potocki-Lupski Syndrome](#)

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[Shwachman-Diamond Syndrome](#)

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[Incontinentia Pigmenti](#)

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[Rhabdoid Tumor Predisposition Syndrome](#)

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[Hereditary Dystonia Overview](#)

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[Cystinosis](#)

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[Silver-Russell Syndrome](#)

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[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

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[TECPR2-Related Hereditary Sensory and Autonomic Neuropathy with Intellectual Disability](#)

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[Perrault Syndrome](#)

[Urofacial Syndrome](#)

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[21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia](#)

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[CLCN4-Related Neurodevelopmental Disorder](#)

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[Cantú syndrome](#)

Nichols, Kim E, MD; St Jude Children's Research Hospital

[Familial Hemophagocytic Lymphohistiocytosis](#)

[Li-Fraumeni Syndrome](#)

[X-Linked Lymphoproliferative Disease](#)

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[SPTLC1-Related Hereditary Sensory Neuropathy](#)

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[HPRT1 Disorders](#)

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[CHMP2B Frontotemporal Dementia](#)

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[MUTYH Polyposis](#)

[NTHL1 Tumor Syndrome](#)

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[Dyskeratosis Congenita and Related Telomere Biology Disorders](#)

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[Lysinuric Protein Intolerance](#)

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[Floating-Harbor Syndrome](#)

[Hypochondroplasia](#)

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[Kleefstra Syndrome](#)

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[Alpha-Mannosidosis](#)

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[21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia](#)

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[Simpson-Golabi-Behmel Syndrome Type 1](#)

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[PROP1-Related Combined Pituitary Hormone Deficiency](#)

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CHKB-Related Muscular Dystrophy

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Nonsyndromic Hearing Loss and Deafness, Mitochondrial

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Autosomal Dominant Epilepsy with Auditory Features

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Donnai-Barrow Syndrome

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Cornelia de Lange Syndrome

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SYNE1 Deficiency

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Genetic Atypical Hemolytic-Uremic Syndrome

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Mucopolysaccharidosis Type III

Tuberous Sclerosis Complex

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Floating-Harbor Syndrome

Smith-Lemli-Opitz Syndrome

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HTRA1 Disorder

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Oral-Facial-Digital Syndrome Type I

Nunes, Mark E, MD; Valley Children's Hospital

Hypophosphatasia

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Lysinuric Protein Intolerance

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Lowe Syndrome

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HPRT1 Disorders



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[KMT2E-Related Neurodevelopmental Disorder](#)

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[GNE Myopathy](#)

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[HPRT1 Disorders](#)

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[Pachyonychia Congenita](#)

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[NTRK1 Congenital Insensitivity to Pain with Anhidrosis](#)

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[Mucopolysaccharidosis Type IVA](#)

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[Short-Chain Acyl-CoA Dehydrogenase Deficiency](#)

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[Thiamine-Responsive Megaloblastic Anemia Syndrome](#)

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[Bietti Crystalline Dystrophy](#)

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[Okur-Chung Neurodevelopmental Syndrome](#)

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[Primary Familial Brain Calcification](#)

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[LAMA2 Muscular Dystrophy](#)

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[Allan-Herndon-Dudley Syndrome](#)

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[SLC25A24 Fontaine Progeroid Syndrome](#)

Olson, Heather, MD, MS; Boston Children's Hospital
[CDKL5 Deficiency Disorder](#)

Olson, Timothy S, MD, PhD; Children's Hospital of Philadelphia
[MIRAGE Syndrome](#)
[Monosomy 7 Predisposition Syndromes Overview](#)

Ombrello, Amanda, MD; National Human Genome Research Institute

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[Cystic Fibrosis](#)

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[HTRA1 Disorder](#)

[Spinocerebellar Ataxia Type 17](#)

Opal, Puneet, MD, PhD; Northwestern University Feinberg School of Medicine

[GAN-Related Neurodegeneration](#)

[Spinocerebellar Ataxia Type 1](#)

Ordonez, Jessica L, MS; Miami Cancer Institute - Baptist Health South Florida

[Congenital Deafness with Labyrinthine Aplasia, Microtia, and Microdontia](#)

[Deafness and Myopia Syndrome](#)

Origa, Raffaella, MD; Ospedale Pediatrico Microcitemico Antonio Cao

[Alpha-Thalassemia](#)

[Beta-Thalassemia](#)

Orrell, Richard W, BSc, MD; University College London

[ALS2-Related Disorder](#)

Orsini, Joseph J, PhD; Wadsworth Center - New York State Department of Health

[Krabbe Disease](#)

Orten, Dana J, PhD; Boys Town National Research Hospital

[Usher Syndrome Type II](#)

Orthmann-Murphy, Jennifer, MD, PhD; Johns Hopkins University

[Pelizaeus-Merzbacher-Like Disease 1](#)

Ortiz-Gonzalez, Xilma, MD, PhD; Children's Hospital of Philadelphia

[SPTBN4 Disorder](#)

Osborne, Jenae, MS; University of Michigan

[POT1 Tumor Predisposition](#)

Osborne, Lucy R, PhD; University of Toronto

[7q11.23 Duplication Syndrome](#)

Oshima, Junko, MD, PhD; University of Washington

[Werner Syndrome](#)

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[SUCLA2-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria](#)

Ostergaard, Pia; St George's, University of London

[Milroy Disease](#)

Ostrer, Harry, MD; New York University School of Medicine

[Nonsyndromic Disorders of Testicular Development Overview](#)

Ostrowski, Philip J, MD; St George's University Hospitals NHS Foundation Trust

[Tatton-Brown-Rahman Syndrome](#)

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[Chondrodysplasia Punctata 1, X-Linked](#)

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[Osteoglophonic Dysplasia](#)

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[Autosomal Dominant Epilepsy with Auditory Features](#)

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[ATP1A3-Related Neurologic Disorders](#)

[DYT1 Early-Onset Isolated Dystonia](#)

[SGCE Myoclonus-Dystonia](#)

P

Paciorkowski, Alex R, MD; University of Rochester Medical Center

[PURA-Related Neurodevelopmental Disorders](#)

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[LMNB1-Related Autosomal Dominant Leukodystrophy](#)

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[Nonsyndromic Retinitis Pigmentosa Overview](#)

[Spinocerebellar Ataxia Type 7](#)

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[Alstrom Syndrome](#)

Pais, Lynn, MS, MSc; Boston Children's Hospital

[KMT2E-Related Neurodevelopmental Disorder](#)

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[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)

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[ATN1-Related Neurodevelopmental Disorder](#)

[CLCN4-Related Neurodevelopmental Disorder](#)

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[ANO5 Muscle Disease](#)

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[Adenine Phosphoribosyltransferase Deficiency](#)

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[BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer](#)

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[Nonsyndromic Hearing Loss and Deafness, Mitochondrial](#)

Pankratz, Nathan D, PhD; Indiana University School of Medicine

[Parkinson Disease Overview](#)

Papanikalaou, George, MD, PhD; National and Kapodistrian University of Athens

[Juvenile Hemochromatosis](#)

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[SOST-Related Sclerosing Bone Dysplasias](#)

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[SETD2 Neurodevelopmental Disorders](#)

Paquis-Flucklinger, Véronique, MD, PhD; Nice Hospital

[CHCHD10-Related Disorders](#)

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[VLDLR Cerebellar Hypoplasia](#)

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[LAMA2 Muscular Dystrophy](#)

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[Familial Hypercholesterolemia](#)

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[Joubert Syndrome](#)

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[Congenital Insensitivity to Pain Overview](#)

Parker, Charles J, MD; University of Utah School of Medicine

[HFE-Related Hemochromatosis](#)

Parkin, Kimberly; Massachusetts General Hospital

[Pitt-Hopkins Syndrome](#)

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[SLC39A8-CDG](#)

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[ALK-Related Neuroblastic Tumor Susceptibility](#)

Parks, Sharie B, PhD; Oregon Health and Science University

[Dilated Cardiomyopathy Overview](#)

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[Spastic Paraplegia 4](#)

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[KCNQ2-Related Disorders](#)

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[Glucose Transporter Type 1 Deficiency Syndrome](#)

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[Autosomal Dominant Epilepsy with Auditory Features](#)

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[Retinoblastoma](#)

Passemard, Sandrine, MD, PhD; APHP-Robert Debré University Hospital

[ASPM Primary Microcephaly](#)

[WDR62 Primary Microcephaly](#)

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[Gaucher Disease](#)

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[Factor V Leiden Thrombophilia](#)

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[Troyer Syndrome](#)

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[Friedreich Ataxia](#)

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Patterson, Marc, MD; Mayo Clinic

[Niemann-Pick Disease Type C](#)

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[Congenital Central Hypoventilation Syndrome](#)

Pauli, Richard M, MD, PhD; University of Wisconsin

[Achondroplasia](#)

Paulson, Henry, MD, PhD; University of Michigan Medical School

[Spinocerebellar Ataxia Type 3](#)

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[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Pearson, Deborah A, PhD; McGovern Medical School

[Tuberous Sclerosis Complex](#)

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[Aromatic L-Amino Acid Decarboxylase Deficiency](#)

Pecci, Alessandro, MD, PhD; IRCCS Policlinico San Matteo Foundation

[MYH9-Related Disease](#)

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[McLeod Neuroacanthocytosis Syndrome](#)[VPS13A Disease](#)

Pellerin, David, MD, MSc; University College London

[GAA-FGF14-Related Ataxia](#)

Pelzer, Nadine, MD, PhD; Leiden University Medical Center

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[ETV6 Thrombocytopenia and Predisposition to Leukemia](#)

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Pennesi, Mark E, MD, PhD; Casey Eye Institute - Oregon Health & Science University

[Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview](#)[RPE65-Related Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy](#)

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[Ataxia-Telangiectasia](#)

[Hereditary Ataxia Overview](#)

Peron, Angela, MD; San Paolo Hospital, Università degli Studi di Milano

[BCL11A-Related Intellectual Disability](#)

[Snyder-Robinson Syndrome](#)

Perrier, Nancy D, MD; University of Texas MD Anderson Cancer Center

[CDC73-Related Disorders](#)

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[ALK-Related Neuroblastic Tumor Susceptibility](#)

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Petrovic, Aleksandra, MD; University of Washington

[X-Linked Severe Combined Immunodeficiency](#)

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Pfautsch, Miranda; Dr Kiran C Patel College of Medicine - Tampa Bay Campus

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[Hereditary Myopathy with Early Respiratory Failure](#)

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[Dystrophic Epidermolysis Bullosa](#)

[Epidermolysis Bullosa Simplex](#)

[Epidermolysis Bullosa with Pyloric Atresia](#)

[Junctional Epidermolysis Bullosa](#)

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[Phelan-McDermid Syndrome](#)

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[Primary Trimethylaminuria](#)

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[Familial Porphyria Cutanea Tarda](#)[Hepatoerythropoietic Porphyria](#)

Phillips, John A, MD; Vanderbilt University Medical Center

[Heritable Pulmonary Arterial Hypertension Overview](#)[PROP1-Related Combined Pituitary Hormone Deficiency](#)

Phillips, Lindsay, PhD; University of Calgary

[Au-Kline Syndrome](#)

Phornphutkul, Chanika, MD; Brown University

[Alkaptonuria](#)

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[SAMD9L Ataxia-Pancytopenia Syndrome](#)

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Pieke-Dahl, Sandra, PhD; Ohio State University, Department of Psychology

[Usher Syndrome Type I](#)[Usher Syndrome Type II](#)

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[X-Linked Myotubular Myopathy](#)

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[Factor V Leiden Thrombophilia](#)

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[Birt-Hogg-Dube Syndrome](#)

[FH Tumor Predisposition Syndrome](#)

Pitteloud, Nelly, MD; Massachusetts General Hospital

[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)

Pitt, James, PhD; Murdoch Children's Research Institute

[Squalene Synthase Deficiency](#)

Pizzo, Lucilla, MS; Pennsylvania State University

[16p12.2 Recurrent Deletion](#)

Plaisier, Emmanuelle, MD, PhD; Hôpital Tenon / Université Pierre et Marie Curie

[COL4A1-Related Disorders](#)

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[GRIN1-Related Neurodevelopmental Disorder](#)

[GRIN2B-Related Neurodevelopmental Disorder](#)

[GRIN2D-Related Developmental and Epileptic Encephalopathy](#)

Plecko, Barbara, MD; Medical University of Graz

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Plitt, Gilman, MD; Cleveland Clinic

[Multiple Endocrine Neoplasia Type 2](#)

Plon, Sharon E, MD, PhD; Baylor College of Medicine

[Rothmund-Thomson Syndrome](#)

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[LZTR1- and SMARCB1-Related Schwannomatosis](#)

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[Congenital Diaphragmatic Hernia Overview](#)

[Donnai-Barrow Syndrome](#)

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[GNB5-Related Neurodevelopmental Disorder](#)

Porter, Christopher C, MD; Emory University School of Medicine

[ETV6 Thrombocytopenia and Predisposition to Leukemia](#)

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[Mucopolysaccharidosis Type I](#)

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Potocki-Lupski Syndrome
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Prada, Carlos, MD; Ann & Robert H Lurie Children's Hospital of Chicago
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Prades, Silvia, PhD; Ataxia UK
DRPLA

Prasad, Rathi, MBBS, PhD; Queen Mary University of London
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Branchiootorenal Spectrum Disorder

Prasov, Lev, MD, PhD; University of Michigan Medical School
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Prasun, Pankaj, MD; Icahn School of Medicine at Mount Sinai
Lathosterolosis
Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency / Trifunctional Protein Deficiency
Multiple Acyl-CoA Dehydrogenase Deficiency

Preston, Matthew K, MD; University of Washington
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Priori, Silvia G, MD, PhD; ICS Maugeri
CACNA1C-Related Disorders
Catecholaminergic Polymorphic Ventricular Tachycardia

Prior, Thomas W, PhD; Case Western Reserve University
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Prokisch, Holger, PhD; Technische Universität München
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Pronk, JC, PhD; Vrije Universiteit Medical Center
Megalencephalic Leukoencephalopathy with Subcortical Cysts

Proudlock, Frank A, BSc, MSc, PhD; University of Leicester
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[Spinocerebellar Ataxia Type 7](#)

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[STAT3 Hyper IgE Syndrome](#)

[X-Linked Severe Combined Immunodeficiency](#)

Puffenberger, Erik G, PhD; Clinic for Special Children

[Maple Syrup Urine Disease](#)

Pulst, Stefan M, MD; University of Utah

[Spinocerebellar Ataxia Type 13](#)

[Spinocerebellar Ataxia Type 2](#)

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[21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia](#)

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[Spinocerebellar Ataxia Type 11](#)

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[Hereditary Hemorrhagic Telangiectasia](#)

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[MYRF-Related Cardiac Urogenital Syndrome](#)

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[Oculopharyngeal Muscular Dystrophy](#)

Q

Quijano-Roy, Susana, MD, PhD; University of Versailles Saint Quentin-en-Yvelines

[LAMA2 Muscular Dystrophy](#)

Quinonez, Shane C, MD; University of Michigan

[Citrullinemia Type I](#)

[Dihydroipoamide Dehydrogenase Deficiency](#)

Quintero-Rivera, Fabiola, MD; University of California, Irvine

[NFIA-Related Disorder](#)

[Wiedemann-Steiner Syndrome](#)

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[Spinocerebellar Ataxia Type 36](#)

R

Raas-Rothschild, Annick, MD; Sheba Medical Center, Tel HaShomer

[Mucopolipidosis III Gamma](#)

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[SETD2 Neurodevelopmental Disorders](#)
- Rahman, Joyeeta, BSc; UCL Great Ormond Street Institute of Child Health
[Mitochondrial DNA-Associated Leigh Syndrome Spectrum](#)
- Rahman, Nazneen, BM BCh, PhD; Institute of Cancer Research
[EZH2-Related Overgrowth](#)
[Sotos Syndrome](#)
- Rahman, Shamima, PhD; UCL Great Ormond Street Institute of Child Health
[Mitochondrial DNA-Associated Leigh Syndrome Spectrum](#)
[Nuclear Gene-Encoded Leigh Syndrome Spectrum Overview](#)
- Rai, Karan, BS; The Ohio State University
[BAP1 Tumor Predisposition Syndrome](#)
- Raininko, Raili, MD, PhD; Uppsala University
[LMNB1-Related Autosomal Dominant Leukodystrophy](#)
- Rajan, Neil, MBBS, PhD; Royal Victoria Infirmery
[CYLD Cutaneous Syndrome](#)
- Rajasekaran, Surender, MD, MPH; Spectrum Health
[Bachmann-Bupp Syndrome](#)
- Ramnitz, Mary Scott, MD; National Institute of Dental and Craniofacial Research
[Hyperphosphatemic Familial Tumoral Calcinosis](#)
- Ramos, Eliana Marisa, PhD; University of California Los Angeles
[Primary Familial Brain Calcification](#)
- Rampoldi, Luca, PhD; Ospedale San Raffaele
[VPS13A Disease](#)
- Rana, Huma Q, MD; Dana-Farber Cancer Institute
[FH Tumor Predisposition Syndrome](#)
- Rand, Casey M, BS; Ann & Robert H Lurie Children's Hospital of Chicago
[Congenital Central Hypoventilation Syndrome](#)
- Ranum, Laura PW, PhD; University of Florida
[Myotonic Dystrophy Type 2](#)
[Spinocerebellar Ataxia Type 8](#)
- Rappold, Gudrun A, PhD; Heidelberg University
[FOXP1 Syndrome](#)
[SHOX Deficiency Disorders](#)
- Raskind, Wendy H, MD, PhD; University of Washington Medical Center

[ADCY5 Dyskinesia](#)

[GATA1-Related Cytopenia](#)

[Hereditary Multiple Osteochondromas](#)

[SAMD9L Ataxia-Pancytopenia Syndrome](#)

[Spinocerebellar Ataxia Type 14](#)

Ratisoontorn, Chootima, DDS, PhD; Chulalongkorn University

[Saethre-Chotzen Syndrome](#)

Rauen, Katherine A, MD, PhD; University of California, Davis

[Cardiofaciocutaneous Syndrome](#)

[HRAS-Related Costello Syndrome](#)

Rawlings, David J, MD; University of Washington

[X-Linked Severe Combined Immunodeficiency](#)

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[TANGO2 Deficiency](#)

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[Carney Complex](#)

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[DYT1 Early-Onset Isolated Dystonia](#)

[LRRK2 Parkinson Disease](#)

[SGCE Myoclonus-Dystonia](#)

Raymond, F Lucy, MD, PhD; University of Cambridge

[NSDHL-Related Disorders](#)

Raymond, Gerald V, MD; Johns Hopkins Hospital

[X-Linked Adrenoleukodystrophy](#)

[Zellweger Spectrum Disorder](#)

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[Cherubism](#)

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[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

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[STRC-Related Autosomal Recessive Hearing Loss](#)

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[NDP-Related Retinopathies](#)

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[McLeod Neuroacanthocytosis Syndrome](#)

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[Succinic Semialdehyde Dehydrogenase Deficiency](#)

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[Hereditary Hyperekplexia Overview](#)

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[Allan-Herndon-Dudley Syndrome](#)

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[Heritable Thoracic Aortic Disease Overview](#)

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[GLB1-Related Disorders](#)

[Mucopolysaccharidosis Type IVA](#)

[Phenylalanine Hydroxylase Deficiency](#)

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[Cranio metaphyseal Dysplasia, Autosomal Dominant](#)

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[PURA-Related Neurodevelopmental Disorders](#)

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[RFC1 CANVAS / Spectrum Disorder](#)

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[TRMU Deficiency](#)

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[Classic Isovaleric Acidemia](#)

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[Maternal 15q Duplication Syndrome](#)

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[Genetic Atypical Hemolytic-Uremic Syndrome](#)

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[GAA-FGF14-Related Ataxia](#)

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[Malignant Hyperthermia Susceptibility](#)

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[Autosomal Recessive Congenital Ichthyosis](#)

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[Oculopharyngeal Muscular Dystrophy](#)

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[Schmid Metaphyseal Chondrodysplasia](#)

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[CDC73-Related Disorders](#)

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[Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

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[Succinic Semialdehyde Dehydrogenase Deficiency](#)

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[Noonan Syndrome](#)

Roberts, Eve, MD; University of Toronto

[Wilson Disease](#)

Robertson, David, MD; Vanderbilt University

[Dopamine Beta-Hydroxylase Deficiency](#)

Robertson, Nic, PhD; University of Edinburgh

[RNU4atac-opathy](#)

Robertson, Stephen, DPhil; University of Otago

[FLNB Disorders](#)[X-Linked Otopalatodigital Spectrum Disorders](#)

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[FGFR Craniosynostosis Syndromes Overview](#)[Stickler Syndrome](#)

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[KMT2E-Related Neurodevelopmental Disorder](#)

Roetto, Antonella, PhD; University of Torino
[TFR2-Related Hemochromatosis](#)

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[Phelan-McDermid Syndrome](#)
[RPS6KA3-Related Intellectual Disability](#)

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[FKBP14 Kyphoscoliotic Ehlers-Danlos Syndrome](#)

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[MAPT-Related Frontotemporal Dementia](#)

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[Autosomal Dominant Robinow Syndrome](#)

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[Muenke Syndrome](#)

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[Shwachman-Diamond Syndrome](#)

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[COL4A1-Related Disorders](#)

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[Hepatic Veno-Occlusive Disease with Immunodeficiency](#)

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[Malignant Hyperthermia Susceptibility](#)

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[ZAP70-Related Combined Immunodeficiency](#)

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[16p12.2 Recurrent Deletion](#)

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[Bardet-Biedl Syndrome Overview](#)

Rossetti, Sandro, MD; Mayo Clinic
[Dent Disease](#)

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[Prolidase Deficiency](#)

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[VPS35-Related Parkinson Disease](#)

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[SETD1B-Related Neurodevelopmental Disorder](#)

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[GLB1-Related Disorders](#)

Roth, Fanny, PhD; Centre de Recherche en Myologie

[Oculopharyngeal Muscular Dystrophy](#)

Rouleau, Guy A, MD, PhD; McGill University

[Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum](#)

[Oculopharyngeal Muscular Dystrophy](#)

[Spastic Paraplegia 8](#)

[SYNE1 Deficiency](#)

Roulet, Jean-Baptiste, PhD; Washington State University

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Rouzier, Cécile, MD, PhD; Nice Hospital

[CHCHD10-Related Disorders](#)

Roze, Emmanuel, MD, PhD; Assistance Publique-Hôpitaux de Paris

[Congenital Mirror Movements](#)

Ruaud, Lyse, MD, PhD; APHP-Robert Debré University Hospital

[WDR62 Primary Microcephaly](#)

Rudnick, Sean, MD; Wake Forest University School of Medicine

[Familial Porphyria Cutanea Tarda](#)

[Hepatoerythropoietic Porphyria](#)

Ruivenkamp, Claudia, PhD; Leiden University Medical Center

[Peters Plus Syndrome](#)

Rumbos Siurana, Elisa, MSc; University College London
[GRIA2-Related Neurodevelopmental Disorder](#)

Rumsby, Gill, PhD; University College London Hospitals
[Primary Hyperoxaluria Type 2](#)

Ruppe, Mary D, MD; The Methodist Hospital
[X-Linked Hypophosphatemia](#)

Russell, Bianca, MD; Cincinnati Children's Hospital
[Bohring-Opitz Syndrome](#)

Russell, Meaghan K, MPH; MassGeneral Hospital for Children
[Congenital Diaphragmatic Hernia Overview](#)

Russman, Barry S, MD; Oregon Health and Science University
[Spinal Muscular Atrophy](#)

Rutkowski, Anne, MD; Cure CMD
[LAMA2 Muscular Dystrophy](#)

Rutkowski, Timothy P, PhD; Emory University School of Medicine
[3q29 Recurrent Deletion](#)

Rutledge, S Lane, MD; University of Alabama at Birmingham
[Nonketotic Hyperglycinemia](#)

Rutten, Julie, MD, PhD; Leiden University Medical Center
[CADASIL](#)

Ryan, Brigid, PhD; University of Auckland
[MAPT-Related Frontotemporal Dementia](#)

Ryan, Shannon, MSc; Montreal Children's Hospital
[Phenylalanine Hydroxylase Deficiency](#)

Rydning, Siri Lynne, MD; University of Oslo
[Spastic Paraplegia 4](#)

Récan-Budiartha, Dominique, MD; Hopital Cochin
[Emery-Dreifuss Muscular Dystrophy](#)

Rødahl, Eyvind, MD, PhD; University of Bergen
[ADAMTSL4-Related Eye Disorders](#)
[Congenital Stromal Corneal Dystrophy](#)

S

Saal, Howard M, MD; Cincinnati Children's Hospital Medical Center
[IRF6-Related Disorders](#)

Silver-Russell Syndrome

Saba, Julie, MD, PhD; UCSF / Benioff Children's Hospital

Sphingosine Phosphate Lyase Insufficiency Syndrome

Sabet Rasekh, Parisa, MD; Children's National Health System

LMNB1-Related Autosomal Dominant Leukodystrophy

Sacharow, Stephanie J, MD; Boston Children's Hospital

Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency

Spinal Muscular Atrophy, X-Linked Infantile

Sadleir, Lynette Grant, MB ChB; University of Otago

GNB5-Related Neurodevelopmental Disorder

Saenz-Ayala, Sofia, MD; Cincinnati Children's Hospital Medical Center

Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Saffari, Afshin, MD; Harvard Medical School

EPG5-Related Disorder

Saheki, Takeyori, MD, PhD; Kagoshima University Graduate School of Medical and Dental Sciences

Citrin Deficiency

Sahota, Amrik, PhD; Rutgers University

Adenine Phosphoribosyltransferase Deficiency

Saito, Kayoko, MD, PhD; Tokyo Women's Medical University School of Medicine

Fukuyama Congenital Muscular Dystrophy

Sajorda, Brian J, BA; Children's Hospital of Philadelphia

Simpson-Golabi-Behmel Syndrome Type 1

Sako, Shuhei, MD; Jikei University School of Medicine

Thiamine-Responsive Megaloblastic Anemia Syndrome

Saldaris, Jacinta, PhD; Telethon Kids Institute

CDKL5 Deficiency Disorder

Salgado, Carlos, MD; Stanford University School of Medicine

Schimke Immunoosseous Dysplasia

Salih, Mustafa A, Dr Med Sci, MBBS, MD; King Saud University

Salih Myopathy

SH3TC2-Related Hereditary Motor and Sensory Neuropathy

Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive

Salomons, Gajja S, PhD; Amsterdam University Medical Centers

Creatine Deficiency Disorders

Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation

Salpea, Paraskevi, PhD; National Institutes of Health
[Carney Complex](#)

Salpietro, Vincenzo, MD; University College London
[GRIA2-Related Neurodevelopmental Disorder](#)

Salter, Claire, BMedSci, MBBS; University of Exeter Medical School
[PI4KA-Related Disorder](#)

Salviati, Leonardo, MD, PhD; University of Padova
[Primary Coenzyme Q10 Deficiency Overview](#)

Sambuughin, Nyamkhishig, PhD; Uniformed Services University
[Malignant Hyperthermia Susceptibility](#)

Sampaio Moura, Natalia, BS; National Human Genome Research Institute
[Adenosine Deaminase 2 Deficiency](#)

Samson, Ricardo A, MD; Children's Heart Center Nevada
[Jervell and Lange-Nielsen Syndrome](#)

Sanchez Russo, Rossana, MD; Emory University School of Medicine
[3q29 Recurrent Deletion](#)

Sandoval, Claudio, MD; New York Medical College
[Hereditary Folate Malabsorption](#)

Sands, Tristan T, MD, PhD; Columbia University Irving Medical Center
[GNB1 Encephalopathy](#)

Santani, Avni B, PhD; Children's Hospital of Philadelphia
[Carnitine Palmitoyltransferase 1A Deficiency](#)

Santen, Gijs, MD, PhD; Leiden University Medical Center
[ARID1B-Related Disorder](#)
[Coffin-Siris Syndrome](#)

Santos, Fernando, MD, PhD; University of Oviedo
[Hereditary Distal Renal Tubular Acidosis](#)

Santos, Manuela, MD; Centro Hospitalar Universitário do Porto
[LAMA2 Muscular Dystrophy](#)

Sanz, Maureen M, PhD; Molloy College
[Bloom Syndrome](#)

Sapp, Julie C, ScM; National Institutes of Health
[Proteus Syndrome](#)

Sardh, Eliane, MD, PhD; Karolinska University Hospital
[Acute Intermittent Porphyria](#)

Sarquella-Brugada, Georgia, MD, PhD; Hospital Sant Joan de Deu

[Brugada Syndrome](#)

Sarret, Catherine, MD, PhD; Centre Hospitalier Universitaire de Clermont-Ferrand

[Allan-Herndon-Dudley Syndrome](#)

Sas, David J, DO; Mayo Clinic

[Primary Hyperoxaluria Type 1](#)

[Primary Hyperoxaluria Type 3](#)

Sassa, Shigeru, MD, PhD; The Rockefeller University

[Acute Intermittent Porphyria](#)

Sattler, Elke C, MD; Ludwig Maximilian University of Munich

[Birt-Hogg-Dube Syndrome](#)

Saulnier, Celine A, PhD; Neurodevelopmental Assessment & Consulting Services

[3q29 Recurrent Deletion](#)

Saul, Robert A, MD; Greenwood Genetics Center

[FMR1 Disorders](#)

Saunders-Pullman, Rachel, MD, MPH; Icahn School of Medicine at Mount Sinai / Mount Sinai Beth Israel

[LRRK2 Parkinson Disease](#)

[SGCE Myoclonus-Dystonia](#)

Saunders, Carol J, PhD; University of Missouri

[Pitt-Hopkins Syndrome](#)

Sauvé, Yves, PhD; University of Alberta

[X-Linked Congenital Stationary Night Blindness](#)

Savage, Sharon A, MD; National Cancer Institute, NIH

[Dyskeratosis Congenita and Related Telomere Biology Disorders](#)

Savant, Adrienne, MD, MS; Tulane University School of Medicine

[Cystic Fibrosis](#)

Savarese, Marco, PhD; University of Helsinki

[Salih Myopathy](#)

Savarirayan, Ravi, MBBS, MD; University of Melbourne

[Chondrodysplasia Punctata 2, X-Linked](#)

[Fibrodysplasia Ossificans Progressiva](#)

[Osteopathia Striata with Cranial Sclerosis](#)

[Pycnodysostosis](#)

[Schmid Metaphyseal Chondrodysplasia](#)

[Thanatophoric Dysplasia](#)

Type II Collagen Disorders Overview

Savoia, Anna, PhD; Institute for Maternal and Child Health - IRCCS Burlo Garofolo

[MYH9-Related Disease](#)

Scaglia, Fernando, MD; Baylor College of Medicine

[Deoxyguanosine Kinase Deficiency](#)

[MELAS](#)

[Mitochondrial DNA Maintenance Defects Overview](#)

[MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect](#)

[SUCLA2-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria](#)

[SUCLG1-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria](#)

[TANGO2 Deficiency](#)

[TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form](#)

Scarpa, Maurizio, MD, PhD; Horst Schmidt Klinik

[Mucopolysaccharidosis Type II](#)

Schaaf, Christian P, MD, PhD; Heidelberg University

[15q13.3 Recurrent Deletion](#)

[NR2F1-Related Neurodevelopmental Disorder](#)

[Schaaf-Yang Syndrome](#)

[Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated](#)

Schaefer, Amanda M, MS; University of Iowa Hospitals and Clinics

[Genetic Hearing Loss Overview](#)

[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

Schaefer, Franz, MD; Heidelberg University

[Autosomal Recessive Polycystic Kidney Disease – PKHD1](#)

Schanen, N Carolyn, MD, PhD; Dup15q Alliance

[Maternal 15q Duplication Syndrome](#)

Scharenberg, Andrew M, MD; University of Washington

[X-Linked Severe Combined Immunodeficiency](#)

Scharer, Gunter, MD, PhD; Denver Genetic Laboratories, University of Colorado

[Nonketotic Hyperglycinemia](#)

Scheffer, Ingrid E, MBBS, PhD; University of Melbourne

[Autosomal Dominant Sleep-Related Hypermotor \(Hyperkinetic\) Epilepsy](#)

[FOXP2-Related Speech and Language Disorder](#)

[GRIN2A-Related Speech Disorders and Epilepsy](#)

Scheper, Gert C, PhD; Vrije Universiteit Medical Center

[Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation](#)
[Megalencephalic Leukoencephalopathy with Subcortical Cysts](#)

Scherer, Gerd, PhD; University of Freiburg

[Campomelic Dysplasia](#)

Scheuerle, Angela E, MD; University of Texas Southwestern Medical Center

[Incontinentia Pigmenti](#)

Schiffmann, Raphael, MD, MHSc; Baylor Scott & White Research Institute

[Childhood Ataxia with Central Nervous System Hypomyelination / Vanishing White Matter](#)
[Mucopolysaccharidosis IV](#)

Schilsky, Michael, MD; Yale University School of Medicine

[Wilson Disease](#)

Schimke, R Neil, MD; University of Kansas Medical Center

[Von Hippel-Lindau Syndrome](#)

Schimmenti, Lisa A, MD; Mayo Clinic

[Myhre Syndrome](#)

[NDP-Related Retinopathies](#)

[PAX2-Related Disorder](#)

Schindler, Alice, MS; National Institute of Neurological Disorders and Stroke

[Autosomal Dominant TRPV4 Disorders](#)

Schinkel, Alfred, PhD; Netherlands Cancer Institute

[Rotor Syndrome](#)

Schirwani, Schaida, MB ChB, MSc; Sheffield Children's NHS Foundation Trust

[ASXL3-Related Disorder](#)

Schlade-Bartusiak, Kamilla, PhD; University of Alberta

[Alpha-1 Antitrypsin Deficiency](#)

Schlechter, Catherine, MS; Casey Eye Institute - Oregon Health Sciences University

[Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview](#)

Schlis, Krysta, MD; Children's Hospital & Research Center Oakland

[Sickle Cell Disease](#)

Schlotawa, Lars, MD; University Medical Center Göttingen

[Multiple Sulfatase Deficiency](#)

Schmale, Gregory A, MD; University of Washington

[Hereditary Multiple Osteochondromas](#)

Schmitz, Matthias, MD; University Medical School

[Genetic Prion Disease](#)

Schneider, Katherine A, MPH; Dana Farber Cancer Institute

[Li-Fraumeni Syndrome](#)

Schneider, Susanne A, MD, PhD; Ludwig Maximilian University

[PINK1 Type of Young-Onset Parkinson Disease](#)

Schneppenheim, Reinhard, MD, PhD; University Medical Center Hamburg – Eppendorf

[Rhabdoid Tumor Predisposition Syndrome](#)

Schon, Katherine Rose, MA, MB ChB; Addenbrooke's Hospital

[Congenital Insensitivity to Pain Overview](#)

Schoser, Benedikt, MD; Ludwig-Maximilians University

[Myotonic Dystrophy Type 2](#)

Schreiber, John M, MD; Children's National Hospital

[SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders](#)

Schreuder, Andrea B, MD, PhD; University Medical Center Groningen

[Glycogen Storage Disease Type III](#)

Schrier Vergano, Samantha A, MD; Children's Hospital of the King's Daughters

[ARID1B-Related Disorder](#)[Coffin-Siris Syndrome](#)[IMAGe Syndrome](#)

Schroer, Richard, MD; Greenwood Genetic Center

[Epimerase Deficiency Galactosemia](#)

Schuchman, Edward H, PhD; Icahn School of Medicine at Mount Sinai

[Acid Sphingomyelinase Deficiency](#)

Schuelke, Markus, MD; Charité - Universitätsmedizin Berlin

[Ataxia with Vitamin E Deficiency](#)

Schultz, Kris Ann P, MD; Children's Minnesota

[DICER1 Tumor Predisposition](#)[FH Tumor Predisposition Syndrome](#)

Schulz, Ansgar, MD; Universitätsklinik für Kinder- und Jugendmedizin

[CLCN7-Related Osteopetrosis](#)

Schulze, Jeanine, MS; Indiana University School of Medicine

[Parkinson Disease Overview](#)

Schutte, Brian C, PhD; Michigan State University

[IRF6-Related Disorders](#)

Schwartz, Charles E, MS, PhD; Greenwood Genetic Center

[RPS6KA3-Related Intellectual Disability](#)

[Snyder-Robinson Syndrome](#)

Schwartz, David A, MD; University of Colorado Denver

[Pulmonary Fibrosis Predisposition Overview](#)

Schwartz, Mary E; PC Project

[Pachyonychia Congenita](#)

Schwartz, Stuart, PhD; Laboratory Corporation of America

[Prader-Willi Syndrome](#)

Schwarz, Guenter; Cologne University

[Molybdenum Cofactor Deficiency](#)

Scott, C Ronald, MD; University of Washington

[Tyrosinemia Type I](#)

Scott, Daryl A, MD, PhD; University of Utah

[GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss](#)

[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

[RERE-Related Disorders](#)

Scriver, Charles R, MD; McGill University

[Phenylalanine Hydroxylase Deficiency](#)

Scruggs, Brittni A, MD, PhD; Mayo Clinic

[NDP-Related Retinopathies](#)

Seabra, Miguel C, MD, PhD; Imperial College School of Medicine

[Choroideremia](#)

Sebastio, Gianfranco, MD; Federico II University of Naples

[Lysinuric Protein Intolerance](#)

Seckington, Rebecca; University of Queensland School of Medicine

[HFE-Related Hemochromatosis](#)

Sedel, Frederic, MD, PhD; MedDay Pharmaceuticals

[CLCN2-Related Leukoencephalopathy](#)

Sekijima, Yoshiki, MD, PhD; Shinshu University School of Medicine

[Hereditary Transthyretin Amyloidosis](#)

Selber, Paulo, MD; Royal Children's Hospital Melbourne

[PURA-Related Neurodevelopmental Disorders](#)

Seminara, Stephanie, MD; Massachusetts General Hospital

[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)

Senaratne, T Niroshini, PhD; University of California, Los Angeles

[NFIA-Related Disorder](#)

Sentner, Christiaan P, MD; University Medical Center Groningen

[Glycogen Storage Disease Type III](#)

Sequerra Amram Cohen, Ana, PhD; Icahn School of Medicine at Mount Sinai

[EED-Related Overgrowth](#)

Sereda, Christina, MSc; National Institutes of Health

[Choroideremia](#)

Seront, Emmanuel, MD, PhD; Cliniques universitaires Saint-Luc

[TEK-Related Venous Malformations](#)

Sethi, Sanjeev, MD, PhD; Mayo Clinic

[C3 Glomerulopathy](#)

Shah, Hitesh, MS; Manipal Academy of Higher Education

[Multicentric Osteolysis Nodulosis and Arthropathy](#)[Progressive Pseudorheumatoid Dysplasia](#)

Shakkottai, Vikram, MD, PhD; University of Michigan Medical School

[Spinocerebellar Ataxia Type 3](#)

Shapiro, Amy D, MD; Indiana Hemophilia & Thrombosis Center

[Complete Plasminogen Activator Inhibitor 1 Deficiency](#)

Sharer, J Daniel, PhD; University of Alabama at Birmingham

[Duarte Variant Galactosemia](#)

Sharpe, Lindsay T, MA, PhD; Institute of Ophthalmology

[Achromatopsia](#)

Shashi, Vandana, MD; Duke University

[NGLY1-Related Congenital Disorder of Deglycosylation](#)

Shaw, Adam, BMed, MD; Guy's & Saint Thomas' Hospitals

[Trichorhinophalangeal Syndrome](#)

Shaw, Chris, BS; University of Washington

[ADCY5 Dyskinesia](#)

Shchelochkov, Oleg A, MD; National Institutes of Health

[Propionic Acidemia](#)

Shearer, A Eliot, MD, PhD; Harvard Medical School

[Genetic Hearing Loss Overview](#)[OTOF-Related Deafness](#)[STRC-Related Autosomal Recessive Hearing Loss](#)

Sheen, Volney L, MD, PhD; Harvard Medical School

[FLNA Deficiency](#)

Sheffield, Val C, MD, PhD; University of Iowa College of Medicine

[GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss](#)

[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

Shelton, Celeste, MS, PhD; Ariel Precision Medicine

[Pancreatitis Overview](#)

[PRSS1-Related Hereditary Pancreatitis](#)

Shen, Yiping, PhD; Boston Children's Hospital

[16p11.2 Recurrent Deletion](#)

Shephard, Elizabeth A, MSc, PhD; University College London

[Primary Trimethylaminuria](#)

Sheppard, Sarah E, MD, PhD; Eunice Kennedy Shriver National Institute of Child Health and Human Development

[Wiedemann-Steiner Syndrome](#)

Sheridan, Eamonn, MD; University of Leeds

[RAB18 Deficiency](#)

Sherr, Elliott H, MD, PhD; University of California, San Francisco

[DDX3X-Related Neurodevelopmental Disorder](#)

[RERE-Related Disorders](#)

Shieh, Joseph TC, MD, PhD; University of California San Francisco

[Hyaline Fibromatosis Syndrome](#)

Shih, Vivian E, MD; Massachusetts General Hospital

[Fumarate Hydratase Deficiency](#)

Shiloh-Malawsky, Yael, MD; University of North Carolina at Chapel Hill

[DRPLA](#)

Shimamura, Akiko, MD, PhD; Dana Farber Cancer Institute

[Fanconi Anemia](#)

Shirvan, Leila, BA; National Human Genome Research Institute

[HEXA Disorders](#)

Shneider, Benjamin L, MD; University of Pittsburgh Medical Center

[ATP8B1 Deficiency](#)

Shoffner, John M, MD; Medical Neurogenetics, LLC

[Mitochondrial Neurogastrointestinal Encephalopathy Disease](#)

Shohat, Mordechai, MD; Maccabi Genetic Institute

[Familial Dysautonomia](#)

[Familial Mediterranean Fever](#)

Sholto-Douglas-Vernon, Carolyn, PhD; St George's University of London

[Milroy Disease](#)

Shooner, Kerry, MS; Cincinnati Children's Hospital Medical Center

[Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

Shukla, Anju, MD; Manipal Academy of Higher Education

[ISCA1-Related Multiple Mitochondrial Dysfunctions Syndrome](#)[Multicentric Osteolysis Nodulosis and Arthropathy](#)[Progressive Pseudorheumatoid Dysplasia](#)

Shultz, Sarah, PhD; Emory University School of Medicine

[3q29 Recurrent Deletion](#)

Shuman, Cheryl, MS; University of Toronto

[Beckwith-Wiedemann Syndrome](#)

Siddique, Nailah, MSN, RN; Northwestern University Feinberg School of Medicine

[Amyotrophic Lateral Sclerosis Overview](#)

Siddique, Teepu, DSc (Hon), MD; Northwestern University Feinberg Medical School

[Amyotrophic Lateral Sclerosis Overview](#)

Siebert, Reiner, MD; Ulm University & Ulm University Medical Center

[Rhabdoid Tumor Predisposition Syndrome](#)

Sieff, Colin, MBBCh; Boston Children's Hospital and Dana Farber Cancer Institute

[Diamond-Blackfan Anemia](#)

Sieni, Elena, MD; Meyer Children's University Hospital

[Familial Hemophagocytic Lymphohistiocytosis](#)

Sieving, Paul A, MD, PhD; University of California, Davis

[X-Linked Congenital Retinoschisis](#)

Silber, Sherman J, MD; Saint Luke's Hospital

[Y Chromosome Infertility](#)

Simons, Cas, PhD; University of Queensland

[TUBB4A-Related Leukodystrophy](#)

Simpson, Kara L, MS; Children's National Hospital

[Ornithine Transcarbamylase Deficiency](#)[Urea Cycle Disorders Overview](#)

Sims, Katherine B, MD; Massachusetts General Hospital / Harvard Medical School

[NDP-Related Retinopathies](#)

Singal, Ashwani K, MD, MSc; University of South Dakota Sanford School of Medicine

Variegate Porphyria

Singer, Kaitlyn, MS; Geisinger Health System

[16p11.2 Recurrent Deletion](#)

Singh, Gautam K, MD; Central Michigan University School of Medicine

[Cantú syndrome](#)

Singh, Rani H, PhD, RD; Emory University School of Medicine

[Duarte Variant Galactosemia](#)

Singh, Swati, MSc; Manipal Academy of Higher Education

[EXOC6B-Related Spondyloepimetaphyseal Dysplasia with Joint Laxity](#)

Sinnreich, Michael, MD, PhD; Montreal Neurological Institute

[GNE Myopathy](#)

Siper, Paige, PhD; Icahn School of Medicine at Mount Sinai

[FOXP1 Syndrome](#)

Sisodiya, Sanjay, MD, PhD; University College London

[TBC1D24-Related Disorders](#)

Sivakumar, Kumaraswamy, MD; St Joseph's Hospital and Medical Center

[GARS-Associated Axonal Neuropathy](#)

Skefos, Catherine M, MA, MS; The University of Texas MD Anderson Cancer Center

[CDC73-Related Disorders](#)

Skinner, Jonathan Robert, MB ChB, MD; University of Auckland

[GNB5-Related Neurodevelopmental Disorder](#)

Skraban, Cara M, MD; Children's Hospital of Philadelphia

[WDR26-Related Intellectual Disability](#)

Slattery, Susan M, MD, MS; Northwestern University

[Congenital Central Hypoventilation Syndrome](#)

Slaugenhaupt, Susan A, PhD; MGH Center for Human Genetic Research / Harvard Medical School

[Mucopolipidosis IV](#)

Slavotinek, Anne M, MBBS, PhD; University of California, San Francisco

[FREM1 Autosomal Recessive Disorders](#)

[Fryns Syndrome](#)

[McKusick-Kaufman Syndrome](#)

Sleasman, John W, MD; Duke University Medical Center

[IPEX Syndrome](#)

Sloan, Jennifer L, MS, PhD; National Human Genome Research Institute

[Disorders of Intracellular Cobalamin Metabolism](#)

[Isolated Methylmalonic Acidemia](#)

Sloman, Melissa, BSc; Royal Devon University Healthcare NHS Foundation Trust

[Spondylocostal Dysostosis, Autosomal Recessive](#)

Smaoui, Nizar, MD; GeneDx, Inc

[Choroideremia](#)[X-Linked Congenital Retinoschisis](#)

Smith-Simmer, Kelcy, MMSc; University of Wisconsin Carbone Cancer Center

[DDX41-Associated Familial Myelodysplastic Syndrome and Acute Myeloid Leukemia](#)

Smith, Adam C, MS; University of Toronto

[Beckwith-Wiedemann Syndrome](#)

Smith, Ann CM, DSc (Hon), MA; National Institutes of Health

[Smith-Magenis Syndrome](#)

Smith, CI Edvard, MD, PhD; Karolinska Institutet

[X-Linked Agammaglobulinemia](#)

Smith, Edward R, MD; Boston Children's Hospital

[Familial Cerebral Cavernous Malformation](#)

Smith, Frances JD, PhD; University of Dundee

[Pachyonychia Congenita](#)

Smith, Janine, MD; National Institutes of Health

[Mucopolipidosis IV](#)

Smith, Rebecca, MS; Geisinger Health System

[16p11.2 Recurrent Deletion](#)

Smith, Richard JH, MD; University of Iowa Hospitals and Clinics

[Branchiootorenal Spectrum Disorder](#)[C3 Glomerulopathy](#)[DFNA2 Nonsyndromic Hearing Loss](#)[Genetic Hearing Loss Overview](#)[GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss](#)[OTOF-Related Deafness](#)[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

Smith, Simone, MSc; Geisinger Health System

[PACS1 Neurodevelopmental Disorder](#)

Smith, Stephanie C, MS; University of Mississippi Medical Center

[Spinocerebellar Ataxia Type 3](#)

Sniderman King, Lisa, MSc; Genzyme Corporation

[Tyrosinemia Type I](#)

Snijders Blok, Lot, MD; Radboud University Medical Center

[DDX3X-Related Neurodevelopmental Disorder](#)

Snow-Bailey, Karen, PhD; Auckland City Hospital

[Multiple Endocrine Neoplasia Type 2](#)

Snyder, Cara L, MS; CLS Genetic Consulting, LLC

[Celiac Disease](#)

Soares, Ana Rita, MD; Santo Antonio University Hospital Center

[Ellis-van Creveld Syndrome](#)

Sobacchi, Cristina, MS; Humanitas Clinical and Research Center

[CLCN7-Related Osteopetrosis](#)

Sobeih, Magdi M, MD, PhD; Boston Children's Hospital

[16p11.2 Recurrent Deletion](#)

Sobrido, Maria J, MD, PhD; Fundación Pública Galega de Medicina Xenómica-SERGAS

[Primary Familial Brain Calcification](#)[Spinocerebellar Ataxia Type 36](#)

So, Jodi Y, BA; Stanford School of Medicine

[Epidermolysis Bullosa Simplex](#)

Soldovieri, Maria Virginia, PhD; University of Molise

[KCNQ2-Related Disorders](#)[KCNQ3-Related Disorders](#)

Soler-Alfonso, Claudia, MD; Baylor College of Medicine

[TANGO2 Deficiency](#)

Solomon, Benjamin D, MD; National Institutes of Health

[Holoprosencephaly Overview](#)

Solomon, Beth, MS; National Institutes of Health

[Smith-Magenis Syndrome](#)

Solomon, Cindy, MS; Myriad Genetic Laboratories, Inc

[APC-Associated Polyposis Conditions](#)

Solomon, Sheila, MS; GeneDx, Inc

[Pancreatitis Overview](#)[PRSS1-Related Hereditary Pancreatitis](#)

Song, Yuan-Zong, MD, PhD; Jinan University

[Citrin Deficiency](#)

Sotero de Menezes, Marcio A, MD; Swedish Neuroscience Institute

[SCN1A-Related Seizure Disorders](#)

Spacey, Sian, MD; University of British Columbia

[Familial Paroxysmal Nonkinesigenic Dyskinesia](#)

Sparks, Susan E, MD, PhD; Sanofi Genzyme

[Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview](#)[LAMA2 Muscular Dystrophy](#)[PMM2-CDG](#)

Spence, Sarah J, MD, PhD; Children's Hospital Boston

[Maternal 15q Duplication Syndrome](#)

Sperandio, Maria Pia, PhD; Federico II University

[Lysinuric Protein Intolerance](#)

Spiegel, Ronen, MD; Emek Medical Center

[Mucopolidosis III Gamma](#)

Spinner, Nancy B, PhD; Children's Hospital of Philadelphia

[Alagille Syndrome](#)

Splawski, Igor, PhD; Assistant, Children's Hospital Boston

[CACNA1C-Related Disorders](#)

Sprecher, Eli, MD, PhD; Tel Aviv Medical Center

[Pachyonychia Congenita](#)

Squires, James E, MD, MS; UPMC Children's Hospital of Pittsburgh

[ATP8B1 Deficiency](#)[Pediatric Genetic Cholestatic Liver Disease Overview](#)

Sreedharan, Jemeen, BSc, MBBS, PhD; King's College London

[TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia](#)

Srivastava, Siddharth, MD; Boston Children's Hospital

[Alexander Disease](#)[SETBP1 Haploinsufficiency Disorder](#)

Stanich, Peter P, MD; The Ohio State University Wexner Medical Center

[APC-Associated Polyposis Conditions](#)

Stanley, Charles A, MD; University of Pennsylvania

[Permanent Neonatal Diabetes Mellitus](#)

Starr, Lois J, MD, PhD; University of Nebraska Medical Center

[Myhre Syndrome](#)[SLC25A24 Fontaine Progeroid Syndrome](#)

Statland, Jeffrey M, MD; University of Kansas Medical Center

[Andersen-Tawil Syndrome](#)

Steeds, Rick, MD; University Hospitals

[Alstrom Syndrome](#)

Steeves, Marcie, MS; MassGeneral Hospital for Children

[Pitt-Hopkins Syndrome](#)

Steinberg, Steven J, PhD; ARUP Laboratories

[Rhizomelic Chondrodysplasia Punctata Type 1](#)[X-Linked Adrenoleukodystrophy](#)[Zellweger Spectrum Disorder](#)

Steiner, Robert D, MD; University of Wisconsin School of Medicine and Public Health

[COL1A1/2 Osteogenesis Imperfecta](#)[Sitosterolemia](#)

Stein, Jennifer L, MS; Cleveland Clinic Genomic Medicine Institute

[PTEN Hamartoma Tumor Syndrome](#)

Steinlein, Ortrud K, MD, PhD; Ludwig Maximilian University of Munich

[Birt-Hogg-Dube Syndrome](#)

Steinman, Kyle J, MD; Seattle Children's Hospital

[16p11.2 Recurrent Deletion](#)

Steinmann, Beat, MD; University Children's Hospital

[PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome](#)

Stephens, Karen, PhD; University of Washington Medical Center

[Epidermolysis Bullosa Simplex](#)

Stephenson, John BP; Royal Hospital for Sick Children

[Aicardi-Goutieres Syndrome](#)

Sternberg, Damien, MD, PhD; Assistance Publique - Hôpitaux de Paris

[Hypokalemic Periodic Paralysis](#)

Sternen, Darci L, MS; Children's Hospital and Regional Medical Center

[Cystic Fibrosis](#)

Stevanin, Giovanni, PhD; Hôpital de la Pitié-Salpêtrière

[Spastic Paraplegia 11](#)

Stevens, Cathy A, MD; University of Tennessee College of Medicine

[Rubinstein-Taybi Syndrome](#)

Stevenson, David A, MD; Stanford University

[Capillary Malformation-Arteriovenous Malformation Syndrome](#)[Hereditary Hemorrhagic Telangiectasia](#)

Legius Syndrome

Stevenson, Roger E, MD; Greenwood Genetic Center

[Alpha-Thalassemia X-Linked Intellectual Disability Syndrome](#)

[Snyder-Robinson Syndrome](#)

Stewart, Blythe; University of Edinburgh

[MYRF-Related Cardiac Urogenital Syndrome](#)

Stewart, Douglas R, MD; National Cancer Institute

[DICER1 Tumor Predisposition](#)

Stojinski, Carol, MD, MS; Drexel University College of Medicine

[Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency](#)

Stojkovic, Tanya, MD; Institut de Myologie

[Oculopharyngeal Muscular Dystrophy](#)

Stokman, Marijn, MD, PhD; Radboud University Medical Centre

[Nephronophthisis-Related Ciliopathies](#)

Stolle, Catherine A, PhD; Children's Hospital of Philadelphia

[Von Hippel-Lindau Syndrome](#)

Stoller, James K, MD, MS; Cleveland Clinic

[Alpha-1 Antitrypsin Deficiency](#)

Stone, Jeremy T, BSc; Mayo Clinic

[LRRK2 Parkinson Disease](#)

Storey, Elsdon, DPhil; Monash University

[Spinocerebellar Ataxia Type 20](#)

Stover, Niamh B, MS; Banner Health

[Bietti Crystalline Dystrophy](#)

Stratakis, Constantine A, DSc, MD; Foundation for Research & Technology Hellas

[Carney Complex](#)

Stratton, Pamela, MD; National Institute of Neurological Disorders and Stroke

[DICER1 Tumor Predisposition](#)

Strauss, Arnold W, MD; Cincinnati Children's Hospital Medical Center

[Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

Strauss, Kevin A, MD; Clinic for Special Children

[Maple Syrup Urine Disease](#)

Strober, Jonathan, MD; UCSF / Benioff Children's Hospital

[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

Stuart, Helen M, MD; University of Manchester and Central Manchester University Hospitals NHS Foundation Trust

[Urofacial Syndrome](#)

Stumpel, Connie, MD, PhD; Academic Hospital Maastricht

[L1 Syndrome](#)

Stutterd, Chloe, MBBS; University of Melbourne

[Polymicrogyria Overview](#)

Stöckler-Ipsiroglu, Sylvia, MBA, MD, PhD; British Columbia Children's Hospital

[Creatine Deficiency Disorders](#)

Subramony, SH, MD; University of Florida

[Spinocerebellar Ataxia Type 1](#)

[Spinocerebellar Ataxia Type 3](#)

[Spinocerebellar Ataxia Type 8](#)

Suchy, Sharon F, PhD; GeneDx, Inc

[Chondrodysplasia Punctata 1, X-Linked](#)

Suit, Lindsey; University of California, San Francisco

[DDX3X-Related Neurodevelopmental Disorder](#)

Sullivan, Bonnie, MD; Children's Mercy Kansas City

[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

Sumegi, Janos, MD, PhD; Cincinnati Children's Hospital Medical Center

[X-Linked Lymphoproliferative Disease](#)

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[Urea Cycle Disorders Overview](#)

Sumner, Charlotte J, MD; Johns Hopkins University School of Medicine

[Autosomal Dominant TRPV4 Disorders](#)

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[Arginase Deficiency](#)

[Mucopolysaccharidosis Type VII](#)

Sundal, Christina, MD, PhD; Gothenburg University

[CSF1R-Related Disorder](#)

Suominen, Tiina, MSc; University of Tampere

[Salih Myopathy](#)

[Udd Distal Myopathy - Tibial Muscular Dystrophy](#)

Superti-Furga, Andrea, MD; University of Lausanne

[Achondrogenesis Type 1B](#)

[Atelosteogenesis Type 2](#)

[Campomelic Dysplasia](#)

[CHST3-Related Skeletal Dysplasia](#)

[Diastrophic Dysplasia](#)

[SLC26A2-Related Multiple Epiphyseal Dysplasia](#)

Sutton, V Reid, MD; Baylor College of Medicine

[Aicardi Syndrome](#)

[PORCN-Related Developmental Disorders](#)

[TP63-Related Disorders](#)

[White-Sutton Syndrome](#)

Suwannarat, Pim, MD; Faculty of Medicine, Ramathibodi Hospital

[Alkaptonuria](#)

Suzuki, Tamio, MD, PhD; Yamagata University

[Oculocutaneous Albinism Type 4](#)

Sveden, Abigail, MS; Boston Children's Hospital

[ANKRD17-Related Neurodevelopmental Syndrome](#)

Swanson, Michael, PhD; University of Colorado School of Medicine

[Nonketotic Hyperglycinemia](#)

Swadner, Kathleen J, PhD; Massachusetts General Hospital

[ATP1A3-Related Neurologic Disorders](#)

Sweeney, Elizabeth, MB ChB, MD; Liverpool Women's Hospital

[Nail-Patella Syndrome](#)

Sweeney, William E, MS; Medical College and Children's Health System of Wisconsin

[Autosomal Recessive Polycystic Kidney Disease – PKHD1](#)

Sweetser, David A, MD, PhD; Massachusetts General Hospital

[GNAO1-Related Disorder](#)

[Pitt-Hopkins Syndrome](#)

Swoboda, Kathryn J, MD; Massachusetts General Hospital

[ATP1A3-Related Neurologic Disorders](#)

[Tyrosine Hydroxylase Deficiency](#)

Sybert, Virginia P, MD; University of Washington

[Epidermolysis Bullosa Simplex](#)

Symoens, Sofie, PhD; Ghent University Hospital

[Classic Ehlers-Danlos Syndrome](#)

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GAA-FGF14-Related Ataxia

PNPLA6 Disorders

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Classic Ehlers-Danlos Syndrome

T

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Biotin-Thiamine-Responsive Basal Ganglia Disease

SLC25A19-Related Thiamine Metabolism Dysfunction

Tabti, Nacira, MD, PhD; Assistance Publique - Hôpitaux de Paris

Hypokalemic Periodic Paralysis

Tagliatela, Maurizio, MD, PhD; University of Naples Federico II

KCNQ2-Related Disorders

KCNQ3-Related Disorders

Tait, Jonathan F, MD, PhD; University of Washington School of Medicine

Cystic Fibrosis

HFE-Related Hemochromatosis

Takahashi, Hitoshi, MD, PhD; Niigata University

Spinocerebellar Ataxia Type 17

Takahashi, Toshiaki, MD, PhD; National Hospital Organization – Sendai-Nishitaga Hospital

Dysferlinopathy

Takasaki, Kaoru, MD; Children's Hospital of Philadelphia

GATA1-Related Cytopenia

Takashima, Hiroshi, MD, PhD; Kagoshima University Graduate School of Medical and Dental Sciences

Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive

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Pulmonary Fibrosis Predisposition Overview

Tallaksen, Chantal; Hôpital de la Pitié-Salpêtrière

Spastic Paraplegia 4

Taly, Arun B, MD; National Institute of Mental Health & Neurosciences

Huppke-Brendel Syndrome

Isolated Sulfite Oxidase Deficiency

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Alpha-Thalassemia

Congenital Dyserythropoietic Anemia Type I

Tamburro, Joan, DO; Cleveland Clinic

[Isolated and Classic Cutis Marmorata Telangiectatica Congenita](#)

Tamura, Deborah, RN; National Cancer Institute

[Xeroderma Pigmentosum](#)

Tanase-Nakao, Kanako, MD, PhD; National Research Institute for Child Health and Development

[MIRAGE Syndrome](#)

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[Chondrodysplasia Punctata 2, X-Linked](#)

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[Fanconi Anemia](#)

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[Mucopolysaccharidosis Type IVA](#)

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[IPEX Syndrome](#)

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[Cranioectodermal Dysplasia](#)

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[Bohring-Opitz Syndrome](#)

Tarleton, Jack C, PhD; Fullerton Genetics Center

[FMR1 Disorders](#)

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[Adenosine Deaminase Deficiency](#)

Tartaglia, Marco, PhD; Bambino Gesù Children's Hospital IRCSS

[Noonan Syndrome with Multiple Lentigines](#)

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[EZH2-Related Overgrowth](#)

[HIST1H1E Syndrome](#)

[Sotos Syndrome](#)

[Tatton-Brown-Rahman Syndrome](#)

Tawana, Kiran, MB ChB, PhD; Queen Mary University of London

[CEBPA-Associated Familial Acute Myeloid Leukemia \(AML\)](#)

Tawil, Rabi, MD; University of Rochester Medical Center

[Andersen-Tawil Syndrome](#)

[Facioscapulohumeral Muscular Dystrophy](#)

Taylor, Annette K, MS, PhD; Laboratory Corporation of America Holdings

Celiac Disease

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[16p11.2 Recurrent Deletion](#)

[CHD8-Related Neurodevelopmental Disorder with Overgrowth](#)

[PACS1 Neurodevelopmental Disorder](#)

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[ATP8B1 Deficiency](#)

Taylor, Matthew RG, MD, PhD; University of Colorado Anschutz Medical Campus

[Danon Disease](#)

Taylor, Robert W, PhD; Newcastle University

[RRM2B Mitochondrial DNA Maintenance Defects](#)

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[ROR2-Related Robinow Syndrome](#)

Tchan, Michel, BMedSci, MBBS, PhD; Westmead Hospital

[Aspartylglucosaminuria](#)

Teebi, Ahmad, MD; Education City

[Cherubism](#)

Tekendo-Ngongang, Cedrik, MD; National Human Genome Research Institute

[Holoprosencephaly Overview](#)

Tekin, Mustafa, MD; John P Hussman Institute for Human Genomics

[Congenital Deafness with Labyrinthine Aplasia, Microtia, and Microdontia](#)

[Deafness and Myopia Syndrome](#)

[KBG Syndrome](#)

Temple, Isabel Karen, MD; Southampton University Hospitals NHS Trust

[Diabetes Mellitus, 6q24-Related Transient Neonatal](#)

Teng, Joyce, MD, PhD; Stanford School of Medicine

[Epidermolysis Bullosa Simplex](#)

Terry, Sharon F, MA; PXE International

[Pseudoxanthoma Elasticum](#)

Terwindt, Gisela M, MD, PhD; Leiden University Medical Center

[CADASIL](#)

[Retinal Vasculopathy with Cerebral Leukoencephalopathy and Systemic Manifestations](#)

Testa, Giuseppe, MA, MD, PhD; European Institute of Oncology

[Gabriele-de Vries Syndrome](#)

Thabet, Farah, MD; Fattouma Bourguiba University Hospital

[SLC25A19-Related Thiamine Metabolism Dysfunction](#)

Thauvin-Robinet, Christel, MD, PhD; University of Burgundy

[Oral-Facial-Digital Syndrome Type I](#)

Thibert, Ronald L, DO, MSPH; Massachusetts General Hospital

[Maternal 15q Duplication Syndrome](#)[Pitt-Hopkins Syndrome](#)

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[Cartilage-Hair Hypoplasia - Anauxetic Dysplasia Spectrum Disorders](#)

Thiel, Moritz, MD; University of Cologne

[GNAO1-Related Disorder](#)

Thoene, Jess G, MD; University of Michigan

[Citrullinemia Type I](#)[Dihydrolipoamide Dehydrogenase Deficiency](#)

Thomas, Julian, MD; Children's Hospital Orange County

[DDX3X-Related Neurodevelopmental Disorder](#)

Thomas, Mervyn G, MBChB, PhD; University of Leicester

[FRMD7-Related Infantile Nystagmus](#)[Oculocutaneous Albinism and Ocular Albinism Overview](#)

Thomas, Rhys, PhD; Newcastle University

[Hereditary Hyperekplexia Overview](#)

Thomas, Shery; University of Leicester

[FRMD7-Related Infantile Nystagmus](#)

Thompson, Arthur R, MD, PhD; University of Washington

[Hemophilia A](#)[Hemophilia B](#)

Thompson, Reid, MD; Johns Hopkins University

[Barth Syndrome](#)

Thorburn, David R, PhD; Royal Children's Hospital and University of Melbourne

[Mitochondrial DNA-Associated Leigh Syndrome Spectrum](#)[Nuclear Gene-Encoded Leigh Syndrome Spectrum Overview](#)

Thorpe, Ryan K, MD; University of Iowa

[OTOF-Related Deafness](#)

Thunell, Stig, MD, PhD; Karolinska Institute

[Acute Intermittent Porphyria](#)

Tift, Cynthia J, MD, PhD; National Human Genome Research Institute

[GLB1-Related Disorders](#)

[GM2 Activator Deficiency](#)

[HEXA Disorders](#)

[Sandhoff Disease](#)

Tijssen, Marina AJ, MD; University Medical Center Groningen

[Hereditary Hyperekplexia Overview](#)

Tiller, George E, MD, PhD; Kaiser Permanente

[Hypochondroplasia](#)

[X-Linked Spondyloepiphyseal Dysplasia Tarda](#)

Timmerman, Vincent, PhD; Institute Born Bunge

[GAN-Related Neurodegeneration](#)

Timothy, Katherine W, BS; Harvard Medical School

[CACNA1C-Related Disorders](#)

Tinkle, Brad T, MD, PhD; Advocate Children's Hospital

[Pompe Disease](#)

[Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

Tiranti, Valeria Sonia, PhD; IRCCS Foundation Carlo Besta Neurological Institute

[Ethylmalonic Encephalopathy](#)

Tkachenko, Nataliya, MD; Santo Antonio University Hospital Center

[Ellis-van Creveld Syndrome](#)

Todd, Peter K, MD, PhD; University of Michigan School of Medicine

[FMR1 Disorders](#)

Tokuda, Takahiko, MD, PhD; Kyoto Prefectural University Hospital

[Hereditary Transthyretin Amyloidosis](#)

Tolar, Jakub, MD, PhD; University of Minnesota

[Fanconi Anemia](#)

Tonduti, Davide, MD, PhD; V Buzzi Children's Hospital

[Allan-Herndon-Dudley Syndrome](#)

Toone, Jennifer, BSc; Children's and Women's Health Centre of British Columbia

[Nonketotic Hyperglycinemia](#)

Torgerson, Troy, MD, PhD; Division of Infectious Diseases, Immunology, and Rheumatology

[IPEX Syndrome](#)

Toriello, Helga V, PhD; Michigan State University

[Oral-Facial-Digital Syndrome Type I](#)

[Thrombocytopenia Absent Radius Syndrome](#)

Toro, Camilo, MD; National Human Genome Research Institute

[Chediak-Higashi Syndrome](#)

[GM2 Activator Deficiency](#)

[HEXA Disorders](#)

[Sandhoff Disease](#)

[SCARB2-Related Action Myoclonus – Renal Failure Syndrome](#)

Toro, Jorge R, MD; National Institutes of Health

[Birt-Hogg-Dube Syndrome](#)

[FH Tumor Predisposition Syndrome](#)

Torres, Vicente E, MD, PhD; Mayo Clinic

[Polycystic Kidney Disease, Autosomal Dominant](#)

Toyoshima, Yasuko, MD, PhD; Agano Hospital

[Spinocerebellar Ataxia Type 17](#)

Traboulsi, Elias I, MD, MEd; Cleveland Clinic Lerner College of Medicine

[Cohen Syndrome](#)

[Isolated and Classic Cutis Marmorata Telangiectatica Congenita](#)

Trahms, Cristine, MS, RD; University of Washington

[Tyrosinemia Type I](#)

Tran-Fadulu, Van, MS; University of Texas Medical School at Houston

[Heritable Thoracic Aortic Disease Overview](#)

Tranebjærg, Lisbeth, MD, PhD; University of Copenhagen

[Deafness-Dystonia-Optic Neuronopathy Syndrome](#)

[Jervell and Lange-Nielsen Syndrome](#)

[WFS1 Spectrum Disorder](#)

Trevisson, Eva, MD, PhD; University of Padova

[Primary Coenzyme Q10 Deficiency Overview](#)

Trifiro, Mark A, MD; McGill University

[Androgen Insensitivity Syndrome](#)

Trinh, Joanne, BSc; University of British Columbia

[LRRK2 Parkinson Disease](#)

Trollet, Capucine, PhD; Centre de Recherche en Myologie

[Oculopharyngeal Muscular Dystrophy](#)

Trouillard, Oriane, BS; Institut du Cerveau et de la Moelle épinière

[Congenital Mirror Movements](#)

Trzuppek, Karmen M, MS; InformedDNA

[Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview](#)

[Usher Syndrome Type I](#)

[Usher Syndrome Type II](#)

Tsang, Mandy HY, MMSc; University of Hong Kong

[CTNNB1 Neurodevelopmental Disorder](#)

Tsilou, Ekaterini, MD; Center for Biologics Evaluation and Research, US FDA

[Weill-Marchesani Syndrome](#)

Tsuji, Shoji, MD, PhD; University of Tokyo Graduate School of Medicine

[DRPLA](#)

[Spinocerebellar Ataxia Type 17](#)

Tsunogai, Toshiki, MD; Jikei University School of Medicine

[Thiamine-Responsive Megaloblastic Anemia Syndrome](#)

Tuchman, Mendel, MD; Children's National Medical Center

[Urea Cycle Disorders Overview](#)

Turner, Joyce T, MS; Children's National Hospital

[Wilms Tumor Predisposition](#)

Turner, Stefanie, MS; 17q12 Foundation

[17q12 Recurrent Deletion Syndrome](#)

Turnpenny, Peter D, BSc, ChB, MB; Royal Devon University Healthcare NHS Foundation Trust

[Spondylocostal Dysostosis, Autosomal Recessive](#)

Tuschl, Karin, MD, PhD; UCL Great Ormond Street Institute of Child Health

[Hypermanganesemia with Dystonia 1](#)

[SLC39A14 Deficiency](#)

Tuupanen, Sari, PhD; Blueprint Genetics

[X-Linked Congenital Stationary Night Blindness](#)

Tümer, Zeynep, DMSc, MD, PhD; Rigshospitalet

[DLG4-Related Synaptopathy](#)

Tüysüz, Beyhan, MD, PhD; Istanbul University

[Trichorhinophalangeal Syndrome](#)

U

Udd, Bjarne, MD, PhD; Tampere University Hospital

[ANO5 Muscle Disease](#)

[Salih Myopathy](#)

[Udd Distal Myopathy - Tibial Muscular Dystrophy](#)

Uhlig, Holm; NIHR Oxford Biomedical Research Centre

[PI4KA-Related Disorder](#)

Uitto, Jouni, MD, PhD; Thomas Jefferson University

[Kindler Syndrome](#)

[Lipoid Proteinosis](#)

[Pseudoxanthoma Elasticum](#)

Umair, Muhammad, PhD; King Saud Bin Abdulaziz University for Health Sciences

[EMC10-Related Neurodevelopmental Disorder](#)

Unger, Sheila, MD; University of Lausanne

[Achondrogenesis Type 1B](#)

[Atelosteogenesis Type 2](#)

[Campomelic Dysplasia](#)

[CHST3-Related Skeletal Dysplasia](#)

[Diastrophic Dysplasia](#)

[SLC26A2-Related Multiple Epiphyseal Dysplasia](#)

Upadia, Jariya, MD; Tulane University School of Medicine

[Cystic Fibrosis](#)

Urban, Zsolt, PhD; University of Pittsburgh

[EFEMP2-Related Cutis Laxa](#)

[ELN-Related Cutis Laxa](#)

[LTBP4-Related Cutis Laxa](#)

Urion, David K, MD; Boston Children's Hospital / Harvard Medical School

[Dystrophinopathies](#)

Ursini, Matilde Valeria, PhD; Institute of Genetics and Biophysics - CNR

[Incontinentia Pigmenti](#)

Usami, Shin-ichi, MD, PhD; Shinshu University School of Medicine

[Nonsyndromic Hearing Loss and Deafness, Mitochondrial](#)

Uyanik, Gökhan, MD; Hanusch Hospital

[CASK Disorders](#)

[DCX-Related Disorders](#)

V

Vahidnezhad, Hassan, MSc, PhD; Pasteur Institute of Iran

[Kindler Syndrome](#)

[Lipoid Proteinosis](#)

Vakkilainen, Svetlana, MD, PhD; University of Helsinki and Helsinki University Hospital

[Cartilage-Hair Hypoplasia - Anauxetic Dysplasia Spectrum Disorders](#)

Valdmanis, Paul N, PhD; University of Washington

[Spastic Paraplegia 8](#)

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[Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

Valentin, Ilia, MD; Heidelberg University

[NR2F1-Related Neurodevelopmental Disorder](#)

Valeriani, Emanuele, MD; Sapienza University of Rome

[Factor V Leiden Thrombophilia](#)

Valle, Laura, PhD; Catalan Institute of Oncology

[Lynch Syndrome](#)

Valverde de Morales, Hannah Gabriela, MD; Integra Genetics

[CTCF-Related Disorder](#)

van Belzen, Martine, PhD; Leiden University Medical Center

[Peters Plus Syndrome](#)

van Bokhoven, Hans, PhD; Radboud University Medical Center

[TP63-Related Disorders](#)

van Bon, Bregje WM, MD, PhD; Radboud University Medical Center

[15q13.3 Recurrent Deletion](#)

[DYRK1A Syndrome](#)

[Schinzel-Giedion Syndrome](#)

[SETBP1 Haploinsufficiency Disorder](#)

Van Broeckhoven, Christine, DSc, PhD; University of Antwerp

[C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia](#)

Van Camp, Guy, PhD; University of Antwerp

[Genetic Hearing Loss Overview](#)

[GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss](#)

[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

van de Warrenburg, Bart P, MD, PhD; Radboud University Nijmegen Medical Center

[ARSACS](#)

Van den Veyver, Ignatia Barbara, MD; Baylor College of Medicine

[Aicardi Syndrome](#)

[PORCN-Related Developmental Disorders](#)

van der Crabben, Saskia N, MD, PhD; Amsterdam UMC

[Serine Deficiency Disorders](#)

van der Knaap, Marjo S, MD, PhD; Amsterdam University Medical Centers

Childhood Ataxia with Central Nervous System Hypomyelination / Vanishing White Matter
CLCN2-Related Leukoencephalopathy
Hypomyelination and Congenital Cataract
Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation
Megalencephalic Leukoencephalopathy with Subcortical Cysts
TUBB4A-Related Leukodystrophy

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Facioscapulohumeral Muscular Dystrophy

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ARID1B-Related Disorder

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C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia

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ADNP-Related Disorder

van Dijk, Fleur S, MD, PhD; London North West University Healthcare NHS Trust
Periodontal Ehlers-Danlos Syndrome
TNXB-Related Classical-Like Ehlers-Danlos Syndrome

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EXOSC3 Pontocerebellar Hypoplasia
TSEN54 Pontocerebellar Hypoplasia

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MECP2 Duplication Syndrome

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Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

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Nonketotic Hyperglycinemia

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Carbonic Anhydrase VA Deficiency
PLPBP Deficiency

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Von Hippel-Lindau Syndrome

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SOST-Related Sclerosing Bone Dysplasias

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ATP6V0A2-Related Cutis Laxa

[Baller-Gerold Syndrome](#)

[Berardinelli-Seip Congenital Lipodystrophy](#)

[FBLN5-Related Cutis Laxa](#)

van Nesselrooij, Bernadette, MD, PhD; University Medical Center Utrecht

[Von Hippel-Lindau Syndrome](#)

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[Ataxia-Telangiectasia](#)

van Ravenswaaij-Arts, Conny M, MD, PhD; University Medical Center Groningen

[CHD7 Disorder](#)

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[PLP1 Disorders](#)

van Steensel, Maurice, MD, PhD; University Hospital Maastricht

[Pachyonychia Congenita](#)

van Swieten, John C, MD, PhD; Erasmus Medical Center

[MAPT-Related Frontotemporal Dementia](#)

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[Milroy Disease](#)

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[GDAP1-Related Hereditary Motor and Sensory Neuropathy](#)

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[Huppke-Brendel Syndrome](#)

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[ZAP70-Related Combined Immunodeficiency](#)

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[LMNB1-Related Autosomal Dominant Leukodystrophy](#)

[Pelizaeus-Merzbacher-Like Disease 1](#)

[POLR3-Related Leukodystrophy](#)

[TUBB4A-Related Leukodystrophy](#)

Vandeweyer, Geert, PhD; Antwerp University Hospital

[ADNP-Related Disorder](#)

VanSickle, Elizabeth, MS; Spectrum Health

[Bachmann-Bupp Syndrome](#)

Vargas, Mauricio, MD, PhD; Oregon Health Sciences University

[Bietti Crystalline Dystrophy](#)

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[Nijmegen Breakage Syndrome](#)

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[TRIO-Related Neurodevelopmental Disorder](#)

[WAC-Related Intellectual Disability](#)

Vaughan, Douglas E, MD; Northwestern University Feinberg School of Medicine

[Complete Plasminogen Activator Inhibitor 1 Deficiency](#)

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[Ataxia-Telangiectasia](#)

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[Andersen-Tawil Syndrome](#)

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[ESCO2 Spectrum Disorder](#)

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[RNU4atac-opathy](#)

[SLC25A24 Fontaine Progeroid Syndrome](#)

Velayos Baeza, Antonio, PhD; Wellcome Trust Centre for Human Genetics

[VPS13A Disease](#)

Velez-Bartolomei, Frances, MD; Stanford University

[MERRF](#)

Velleman, Shelley L, PhD; University of Vermont

[7q11.23 Duplication Syndrome](#)

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[Isolated Methylmalonic Acidemia](#)

[Propionic Acidemia](#)

Veneziano, Liana, PhD; Italian National Research Council

[DRPLA](#)

Venkateswaran, Sunita, MD; Children's Hospital of Eastern Ontario

[Fatty Acid Hydroxylase-Associated Neurodegeneration](#)

Verdin, Hannah, MSc, PhD; Ghent University Hospital

[Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome](#)

Verloes, Alain, MD, PhD; APHP-Robert Debré University Hospital

[ASPM Primary Microcephaly](#)

[Baraitser-Winter Cerebrofrontofacial Syndrome](#)

[WDR62 Primary Microcephaly](#)

Verma, Ishwar Chandar, MD; Sir Ganga Ram Hospital

[Primrose Syndrome](#)

Vermeer, Sascha, MD, PhD; University Hospital Leuven

[ARSACS](#)

Vernon, Hilary, MD, PhD; Johns Hopkins University

[Barth Syndrome](#)

Vicart, Savine, MD; Assistance Publique – Hôpitaux de Paris

[Hypokalemic Periodic Paralysis](#)

Vichinsky, Elliott, MD; Children's Hospital & Research Center Oakland

[Sickle Cell Disease](#)

Vig, Parminder JS, PhD; University of Mississippi Medical Center

[Spinocerebellar Ataxia Type 3](#)

Vihola, Anna, PhD; Fimlab Laboratories

[ANO5 Muscle Disease](#)

Vikkula, Miikka, MD, PhD; Université catholique de Louvain

[TEK-Related Venous Malformations](#)

Vilain, Eric J, MD, PhD; Children's National Hospital

[Nonsyndromic 46,XX Testicular Disorders/Differences of Sex Development](#)

[NR0B1-Related Adrenal Hypoplasia Congenita](#)

Villa, Anna, MD, PhD; Humanitas Clinical and Research Center

[CLCN7-Related Osteopetrosis](#)

Villanueva, Joyce, MBA, MT; Cincinnati Children's Hospital Medical Center

[Familial Hemophagocytic Lymphohistiocytosis](#)

Vincent, G Michael, MD; University of Utah School of Medicine

[Long QT Syndrome Overview](#)

Viskochil, David H, MD, PhD; University of Utah

[BCL11A-Related Intellectual Disability](#)

[Legius Syndrome](#)

Vissers, Lisenka ELM, PhD; Radboud University Medical Center

[Squalene Synthase Deficiency](#)

[TRIO-Related Neurodevelopmental Disorder](#)

[WAC-Related Intellectual Disability](#)

Vissing, John, DMSc, MD; Rigshospitalet

Myotonia Congenita

Vnencak-Jones, Cindy, PhD; GC Vanderbilt University Medical Center

[PROP1-Related Combined Pituitary Hormone Deficiency](#)

Vockley, Jerry, MD, PhD; University of Pittsburgh

[Short-Chain Acyl-CoA Dehydrogenase Deficiency](#)

Vogel-Farley, Vanessa, BA; Dup15q Alliance

[Maternal 15q Duplication Syndrome](#)

Volpini, Victor, MD, PhD; Bellvitge Biomedical Research Institute

[Spinocerebellar Ataxia Type 37](#)

Vos, Yvonne J, PhD; University Medical Center Groningen

[L1 Syndrome](#)

W

Wagner, Klaus, MD, PhD; Medical University Graz

[BSCL2-Related Neurologic Disorders / Seipinopathy](#)

Wagner, Victoria F, MS; McGovern Medical School

[Mucopolysaccharidosis Type III](#)

Wagnon, Jacy L, PhD; University of Michigan

[SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders](#)

Waguespack, Steven G, MD; University of Texas MD Anderson Cancer Center

[CDC73-Related Disorders](#)

Wakefield, Emily, MS; Cincinnati Children's Hospital Medical Center

[X-Linked Lymphoproliferative Disease](#)

Waldman, Amy; Children's Hospital of Philadelphia

[Alexander Disease](#)

Walker, Elaine F, PhD; Emory University

[3q29 Recurrent Deletion](#)

Walker, Ruth H, MBBS, MD, PhD; Mount Sinai School of Medicine

[McLeod Neuroacanthocytosis Syndrome](#)

[VPS13A Disease](#)

Walkovich, Kelly, MD; University of Michigan Medical School

[ZAP70-Related Combined Immunodeficiency](#)

Wallace, Stephanie E, MD; Seattle Children's Hospital

[Camurati-Engelmann Disease](#)

Wallefeld, William, BSc (Hons); Royal Perth Hospital

[Laing Distal Myopathy](#)

Walsh, Christopher A, MD, PhD; Harvard Medical School

[FLNA Deficiency](#)

[Polymicrogyria Overview](#)

Wanders, Ronald JA, PhD; Academic Medical Center, University of Amsterdam

[Adult Refsum Disease](#)

Wang, Bruce, MD; University of California San Francisco

[Hereditary Coproporphryia](#)

Wang, Dong, MD; Emory University Affiliated Hospital

[Glucose Transporter Type 1 Deficiency Syndrome](#)

[Pyruvate Carboxylase Deficiency](#)

Wang, Heng, MD, PhD; DDC Clinic – Center for Special Needs Children

[Cohen Syndrome](#)

[GM3 Synthase Deficiency](#)

[Prolidase Deficiency](#)

Wang, Hsiao-Lin, PhD; Emory University School of Medicine

[CTCF-Related Disorder](#)

Wang, Hua, MD, PhD; Loma Linda University

[MBTPS1-Related Spondyloepimetaphyseal Dysplasia with Elevated Lysosomal Enzymes](#)

Wang, Jing, MD; Baylor College of Medicine

[TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form](#)

Wang, Julia, BS; Baylor College of Medicine

[MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect](#)

[TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form](#)

Wang, Leo H, MD, PhD; University of Washington

[Facioscapulohumeral Muscular Dystrophy](#)

Wangler, Michael F, MD; Baylor College of Medicine

[ACTG2 Visceral Myopathy](#)

[Xia-Gibbs Syndrome](#)

Wang, Lisa L, MD; Baylor College of Medicine

[Baller-Gerold Syndrome](#)

[Poikiloderma with Neutropenia](#)

[Rothmund-Thomson Syndrome](#)

Wang, Raymond, MD; University of California, Irvine

[Mucopolysaccharidosis Type VII](#)

Wang, Zhao, MD; Beijing Friendship Hospital

[Familial Hemophagocytic Lymphohistiocytosis](#)

Wappner, Rebecca S, MD; Riley Hospital for Children, Indiana University School of Medicine

[Lowe Syndrome](#)

Warby, Simon C, PhD; University of Montreal

[Huntington Disease](#)

Warman, Matthew L, MD; Boston Children's Hospital

[Stickler Syndrome](#)

Wasserstein, Melissa P, MD; Albert Einstein College of Medicine

[Acid Sphingomyelinase Deficiency](#)

[Free Sialic Acid Storage Disorders](#)

[Krabbe Disease](#)

Wassif, Christopher A, PhD; National Institute of Child Health and Human Development

[Smith-Lemli-Opitz Syndrome](#)

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[Adult Refsum Disease](#)

[Squalene Synthase Deficiency](#)

Waters, Aoife M, BCh, MB BAO, MSc; University College London

[Bardet-Biedl Syndrome Overview](#)

Waters, Michael F, MD, MS, PhD; Barrow Neurological Institute

[Spinocerebellar Ataxia Type 13](#)

Wattendorf, Daniel J, MD; National Institutes of Health

[Xeroderma Pigmentosum](#)

Watts, Giles, PhD; University of East Anglia

[Inclusion Body Myopathy with Paget Disease of Bone and/or Frontotemporal Dementia](#)

Waxman, Stephen G, MD, PhD; Yale University School of Medicine

[SCN9A Neuropathic Pain Syndromes](#)

Weaver, K Nicole, MD; Cincinnati Children's Hospital Medical Center

[HRAS-Related Costello Syndrome](#)

[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

Weaver, W Curtis, BS; Geisinger Health System

[16p11.2 Recurrent Deletion](#)

Webb, Bryn D, MD, PhD; Icahn School of Medicine at Mount Sinai

[STAC3 Disorder](#)

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[Hyperkalemic Periodic Paralysis](#)

[Hypokalemic Periodic Paralysis](#)

Weckhuysen, Sarah, MD, PhD; University of Antwerp

[DEPDC5-Related Epilepsy](#)

[KCNQ2-Related Disorders](#)

[KCNQ3-Related Disorders](#)

Weemaes, Corry, MD, PhD; Radboud University Medical Center

[Ataxia-Telangiectasia](#)

Weese-Mayer, Debra E, MD; Children's Memorial Hospital

[Congenital Central Hypoventilation Syndrome](#)

Wei, Chongjuan, PhD; University of Texas MD Anderson Cancer Center

[Peutz-Jeghers Syndrome](#)

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[Glycogen Storage Disease Type III](#)

[Glycogen Storage Disease Type VI](#)

Weisfeld-Adams, James, MB ChB; Children's Hospital Colorado

[Hereditary Fructose Intolerance](#)

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[CHD4 Neurodevelopmental Disorder](#)

Weiss, Karl Heinz, MD; Salem Medical Center

[Wilson Disease](#)

Weiss, Marjan M, MD, PhD; Leiden University Medical Center

[Peters Plus Syndrome](#)

Weiss, Mitchell J, MD, PhD; St Jude Children's Research Hospital

[GATA1-Related Cytopenia](#)

Weisz Hubshman, Monika, MD, PhD; Rabin Medical Center

[Familial Dysautonomia](#)

Weksberg, Rosanna, MD, PhD; The Hospital for Sick Children

[Beckwith-Wiedemann Syndrome](#)

Weleber, Richard G, MD; Casey Eye Institute of Oregon Health Sciences University

[Bietti Crystalline Dystrophy](#)

[Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview](#)

[Nonsyndromic Retinitis Pigmentosa Overview](#)

Welsh, Holly I, MS; University of Missouri

[Pitt-Hopkins Syndrome](#)

Wenger, David A, PhD; Thomas Jefferson University Hospital
[Krabbe Disease](#)

Wenger, Olivia K, MD; New Leaf Center
[HYAL2 Deficiency](#)

Wenger, Tara L, MD, PhD; Seattle Children's Hospital
[Apert Syndrome](#)
[FGFR Craniosynostosis Syndromes Overview](#)

Wensel, Christine, MS; DDC Clinic – Center for Special Needs Children
[Cohen Syndrome](#)

Wenstrup, Richard, MD; Cincinnati Children's Hospital Medical Center
[Classic Ehlers-Danlos Syndrome](#)
[PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome](#)

Westbroek, Wendy, PhD; National Institutes of Health
[Chediak-Higashi Syndrome](#)

Weston, Michael D, MA; Boys Town National Research Hospital
[Usher Syndrome Type I](#)
[Usher Syndrome Type II](#)

Wevers, Ron A, PhD; Radboud University Nijmegen Medical Center
[CLPB Deficiency](#)
[SERAC1 Deficiency](#)
[Squalene Synthase Deficiency](#)

Whatley, Sharon D, MSc, PhD; University Hospital of Wales
[Acute Intermittent Porphyrin](#)

Whitcomb, David C, MD, PhD; University of Pittsburgh & University of Pittsburgh Medical Center
[Pancreatitis Overview](#)
[PRSS1-Related Hereditary Pancreatitis](#)

White-Brown, Alexandre M, MSc; Children's Hospital of Eastern Ontario
[DLG4-Related Synaptopathy](#)

White, Colin T, MD; British Columbia's Children's and Women's Hospital
[Primary Hyperoxaluria Type 1](#)

White, Janson, PhD; Invitae Corporation
[White-Sutton Syndrome](#)

White, Stormi; Emory University School of Medicine
[3q29 Recurrent Deletion](#)

White, Susan M, MD; Victorian Clinical Genetics Service

[Floating-Harbor Syndrome](#)

Whitman, Mary C, MD, PhD; Harvard Medical School

[Congenital Fibrosis of the Extraocular Muscles Overview](#)

[Duane Syndrome](#)

Whitton, Chloe, BMed, MD; University of New South Wales

[ATN1-Related Neurodevelopmental Disorder](#)

Wider, Christian, MD; Centre Hospitalier Universitaire Vaudois

[DCTN1-Related Neurodegeneration](#)

Wieczorek, Dagmar, MD; Universitätsklinikum Düsseldorf

[Coffin-Siris Syndrome](#)

[TXNL4A-Related Craniofacial Disorders](#)

Wierenga, Andrea, PhD; SIVOTEC Bioinformatics

[MBTPS1-Related Spondyloepimetaphyseal Dysplasia with Elevated Lysosomal Enzymes](#)

Wierenga, Klaas, MD; Mayo Clinic Florida

[MBTPS1-Related Spondyloepimetaphyseal Dysplasia with Elevated Lysosomal Enzymes](#)

[SPTBN4 Disorder](#)

Wieser, Thomas, MD; Fachkrankenhaus Jerichow

[Carnitine Palmitoyltransferase II Deficiency](#)

Wiesner, Georgia L, MD, MS; Children's Hospital of Philadelphia

[Multiple Endocrine Neoplasia Type 2](#)

Wilcox, William R, MD, PhD; Emory University School of Medicine

[Camurati-Engelmann Disease](#)

Wildin, Robert S, MD; Oregon Health Sciences University

[Hereditary Nephrogenic Diabetes Insipidus](#)

Wilkie, Andrew OM, MA; University of Oxford

[Enlarged Parietal Foramina](#)

Wilkin, Douglas J; Federal Bureau of Investigation

[Achondroplasia](#)

Willemsen, Michèl, MD, PhD; Radboud University Medical Center

[Ataxia-Telangiectasia](#)

Williams, Charles A, MD; University of Florida College of Medicine

[Angelman Syndrome](#)

Williams, Denise, MD; Birmingham Women's and Children's Hospital

[Alstrom Syndrome](#)

[WFS1 Spectrum Disorder](#)

Williams, Gretchen, BS; Children's Hospitals and Clinics of Minnesota

[DICER1 Tumor Predisposition](#)

Williamson, Kathleen A, PhD; University of Edinburgh

[SOX2 Disorder](#)

Wilson, Frederick H, MD, PhD; Harvard Medical School

[Pseudohypoaldosteronism Type II](#)

Winkler, Jürgen, MD; University Hospital Erlangen

[DCX-Related Disorders](#)

Winter, Robin M; Institute of Child Health

[Three M Syndrome](#)

Wise, Anastasia L, PhD; National Jewish Health and University of Colorado Denver

[Pulmonary Fibrosis Predisposition Overview](#)

Wissinger, Bernd, MSc, PhD; University of Tübingen

[Achromatopsia](#)

Wiwattanadittakul, Natrujee, MD; Chiang Mai University

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Wlodarski, Marcin, MD, PhD; St Jude Children's Research Hospital

[Monosomy 7 Predisposition Syndromes Overview](#)

Wojcieszek, Joanne, MD; Indiana University School of Medicine

[Parkinson Disease Overview](#)

Wolf, Barry, MD, PhD; Ann and Robert H Lurie Children's Hospital of Chicago

[Biotinidase Deficiency](#)

Wolfe, Lynne, ARNP, MS; National Human Genome Research Institute

[NGLY1-Related Congenital Disorder of Deglycosylation](#)

[Short-Chain Acyl-CoA Dehydrogenase Deficiency](#)

Wolf, Nicole I, MD, PhD; Amsterdam University Medical Centers

[Hypomyelination and Congenital Cataract](#)

[PI4KA-Related Disorder](#)

[PLP1 Disorders](#)

Wollnik, Bernd, MD; University of Cologne

[Coffin-Siris Syndrome](#)

Wong, Derek, MD; University of California Los Angeles Medical Center

[Arginase Deficiency](#)

Wong, Lee-Jun C, PhD; Baylor College of Medicine

[Deoxyguanosine Kinase Deficiency](#)

[FARS2 Deficiency](#)

[FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion Syndrome](#)

[Mitochondrial DNA Maintenance Defects Overview](#)

[MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect](#)

[TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form](#)

Wong, Melanie, MBBS, PhD; Children's Hospital Westmead

[Hepatic Veno-Occlusive Disease with Immunodeficiency](#)

Woods, Christopher Geoffrey, MB ChB; University of Cambridge

[Congenital Insensitivity to Pain Overview](#)

Woolf, Adrian S, MA, MD; University of Manchester and Central Manchester University Hospitals NHS Foundation Trust

[Urofacial Syndrome](#)

Wortmann, Saskia B, MD, PhD; Salzburger Landeskliniken and Paracelsus Medical University

[CLPB Deficiency](#)

[SERAC1 Deficiency](#)

Wright, Benjamin, MD; University Hospitals Birmingham

[WFS1 Spectrum Disorder](#)

Wright, Galen EB, PhD; University of British Columbia

[Huntington Disease](#)

Wright, J Timothy, DDS, MS; University of North Carolina

[Hypohidrotic Ectodermal Dysplasia](#)

Wright, Michael J, ChB, MB, MSc; Institute of Human Genetics

[COMP-Related Pseudoachondroplasia](#)

[Multiple Epiphyseal Dysplasia, Dominant](#)

Wszolek, Zbigniew K, MD; Mayo Clinic

[CSF1R-Related Disorder](#)

[DCTN1-Related Neurodegeneration](#)

[DYT-GNAL](#)

[VPS35-Related Parkinson Disease](#)

Wu, Bai-Lin, M Med, PhD; Boston Children's Hospital

[16p11.2 Recurrent Deletion](#)

Wuyts, Wim, PhD; University and University Hospital of Antwerp

[Hereditary Multiple Osteochondromas](#)

X

Xia, Maya, BA; COMBINEDBrain

[SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders](#)

Xiao, Changrui, MD; University of California Irvine

[GM2 Activator Deficiency](#)

[Sandhoff Disease](#)

[SCARB2-Related Action Myoclonus – Renal Failure Syndrome](#)

Xin, Baozhong, PhD; DDC Clinic – Center for Special Needs Children

[GM3 Synthase Deficiency](#)

Xu, Manlong, MD, PhD; University of Alberta

[Choroideremia](#)

Y

Yamada, Koki, MD, PhD; Children's Hospital Boston

[Congenital Fibrosis of the Extraocular Muscles Overview](#)

Yamada, Mitsunori, MD, PhD; Shinshu University School of Medicine

[Spinocerebellar Ataxia Type 17](#)

Yamanaka, Koji, MD, PhD; RIKEN Brain Science Institute

[ALS2-Related Disorder](#)

Yang, Jiandong, PhD; George Washington University School of Medicine

[DICER1 Tumor Predisposition](#)

Yang, Paul, MD, PhD; Casey Eye Institute - Oregon Health Sciences University

[Bietti Crystalline Dystrophy](#)

[Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview](#)

Yang, Yaping, PhD; AiLife Diagnostics

[TANGO2 Deficiency](#)

Yap, Kai Lee, PhD; Northwestern University

[Congenital Central Hypoventilation Syndrome](#)

Yates, T Michael, MBChB; University of Edinburgh

[SOX2 Disorder](#)

Yau, Mabel, MD; Mount Sinai School of Medicine

[21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia](#)

Yehia, Lamis, PhD; Cleveland Clinic

[PTEN Hamartoma Tumor Syndrome](#)

Yen, Timothy, MD; University of Colorado Anschutz Medical Center

[APC-Associated Polyposis Conditions](#)

Yeowell, Heather N, PhD; Duke University Medical Center

[PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome](#)

Yntema, Helger G, PhD; Radboud University Medical Center

[Kleefstra Syndrome](#)

Yonker, Lael, MD; MassGeneral Hospital for Children

[Pitt-Hopkins Syndrome](#)

Yoshida, Kunihiro, MD, PhD; Shinshu University Hospital

[Hereditary Transthyretin Amyloidosis](#)

Youngblom, Emily, BA; University of Washington School of Public Health

[Familial Hypercholesterolemia](#)

Young, Danielle O, MS; Kimball Genetics, Inc

[Celiac Disease](#)

Young, Elizabeth, BSc, PhD; Royal Devon and Exeter Healthcare NHS Trust

[Spondylocostal Dysostosis, Autosomal Recessive](#)

Young, William F, MD, MSc; Mayo Clinic

[Hereditary Paraganglioma-Pheochromocytoma Syndromes](#)

Youssefian, Leila, MSc, PhD; Thomas Jefferson University

[Kindler Syndrome](#)

[Lipoid Proteinosis](#)

Yu-Wai-Man, Patrick, BMedSci, MBBS, PhD; University of Cambridge

[Leber Hereditary Optic Neuropathy](#)

[NR2F1-Related Neurodevelopmental Disorder](#)

Yuan, Bo, PhD; Baylor College of Medicine

[Potocki-Lupski Syndrome](#)

Z

Zackai, Elaine H, MD; Children's Hospital of Philadelphia

[22q11.2 Deletion Syndrome](#)

[Emanuel Syndrome](#)

Zadeh, Neda, MD; CHOC Children's Hospital

[KCNK9 Imprinting Syndrome](#)

Zaki, Maha S, MD, PhD; National Research Centre

[UNC80 Deficiency](#)

Zandee, Wouter, MD, PhD; University Medical Center Groningen

[Von Hippel-Lindau Syndrome](#)

Zara, Federico, PhD; G Gaslini Pediatric Institute and University of Genova

[Hypomyelination and Congenital Cataract](#)

Zarate, Yuri A, MD; University of Arkansas for Medical Sciences

[SATB2-Associated Syndrome](#)

Zariwala, Maimoona A, MSc, PhD; University of North Carolina at Chapel Hill

[Primary Ciliary Dyskinesia](#)

Zbuk, Kevin M, MD; Cleveland Clinic Genomic Medicine Institute

[PTEN Hamartoma Tumor Syndrome](#)

Zelley, Kristin, MS; Children's Hospital of Philadelphia

[Li-Fraumeni Syndrome](#)

Zerr, Inga, MD; University Medical School

[Genetic Prion Disease](#)

Zhai, Yi, MD, PhD; University of Alberta

[Choroideremia](#)

Zhang, Kejian, MBA, MD; GoBroad Healthcare Group

[Autoimmune Lymphoproliferative Syndrome](#)

[Familial Hemophagocytic Lymphohistiocytosis](#)

[Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency](#)

[WAS-Related Disorders](#)

[X-Linked Hyper IgM Syndrome](#)

[X-Linked Lymphoproliferative Disease](#)

Ziegler, John B, MBBS, MD; Sydney Children's Hospital

[Hepatic Veno-Occlusive Disease with Immunodeficiency](#)

Ziegler, Shira G, MD, PhD; Johns Hopkins University School of Medicine

[Generalized Arterial Calcification of Infancy](#)

Zipfel, Peter F, PhD; Hans Knoll Institute

[C3 Glomerulopathy](#)

Zippin, Jonathan, MD, PhD; Joan and Sanford I Weill Medical College of Cornell University

[Oculocutaneous Albinism and Ocular Albinism Overview](#)

Zobor, Ditta, MD, PhD; University of Tübingen

[Achromatopsia](#)

Zoghbi, Huda Y, MD; Baylor College of Medicine

[MECP2 Disorders](#)

[Spinocerebellar Ataxia Type 1](#)

Zschocke, Johannes, MD, PhD; Medical University of Innsbruck

[Periodontal Ehlers-Danlos Syndrome](#)

Züchner, Stephan, MD, PhD; University of Miami Miller School of Medicine

GAA-FGF14-Related Ataxia

GDAP1-Related Hereditary Motor and Sensory Neuropathy

MFN2 Hereditary Motor and Sensory Neuropathy

PNPLA6 Disorders

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Živná, Martina, PhD; Charles University

Autosomal Dominant Tubulointerstitial Kidney Disease -- MUC1

Autosomal Dominant Tubulointerstitial Kidney Disease -- REN

Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD

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