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Blackstone, Craig, MD, PhD; Massachusetts General Hospital

[Spastic Paraplegia 15](#)

Blake, Kim, MD; Dalhousie University

[CHD7 Disorder](#)

Blau, Nenad, PhD; University Children's Hospital

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

Bleesing, Jack JH, MD, PhD; Cincinnati Children's Hospital

[Autoimmune Lymphoproliferative Syndrome](#)

Bley, Annette E, MD; University Children's Hospital

[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 Protoporphryia, Autosomal Recessive](#)

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Bober, Michael B, MD, PhD; Nemours Children's Health

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

[Hypochondroplasia](#)

[Microcephalic Osteodysplastic Primordial Dwarfism Type II](#)

[Rhizomelic Chondrodysplasia Punctata Type 1](#)

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Boccuto, Luigi, MD; Greenwood Genetic Center

[Phelan-McDermid Syndrome](#)

Bodell, Adria, MS; Beth Israel Deaconess Medical Center

[FLNA Deficiency](#)[Polymicrogyria Overview](#)

Boehm, Corinne D, MS; GC Johns Hopkins Hospital

[X-Linked Adrenoleukodystrophy](#)

Boerkoel, Cornelius F, MD, PhD; University of British Columbia

[NSDHL-Related Disorders](#)[Schimke Immunoosseous Dysplasia](#)[Snyder-Robinson Syndrome](#)[Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive](#)[UNC80 Deficiency](#)

Boespflug-Tanguy, Odile, MD, PhD; UMR INSERM 676 / Hôpital Robert Debré

[ALS2-Related Disorder](#)[Childhood Ataxia with Central Nervous System Hypomyelination / Vanishing White Matter](#)

Bohlega, Saeed A, MD; King Faisal Specialist Hospital and Research Center

[Woodhouse-Sakati Syndrome](#)

Bojanowski, Christine, MD; Tulane University School of Medicine

[Cystic Fibrosis](#)

Bolduc, Véronique, PhD; National Institute of Neurological Disorders and Stroke

[Collagen VI-Related Dystrophies](#)

Boman, Helge, MD, PhD; Haukeland University Hospital

[ADAMTSL4-Related Eye Disorders](#)[Cold-Induced Sweating Syndrome including Crisponi Syndrome](#)[Congenital Stromal Corneal Dystrophy](#)

Bomont, Pascale, PhD; Institute for Neurosciences of Montpellier

[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

Boon, Elles MJ, PhD; Leiden University Medical Center

CADASIL

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TEK-Related Venous Malformations

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ADAMTSL4-Related Eye Disorders

Booth, Kevin, PhD; Indiana School of Medicine

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

Bostwick, Bret, MD; Baylor College of Medicine

CDK13-Related Disorder

PORCN-Related Developmental Disorders

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SYNE1 Deficiency

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Oculopharyngeal Muscular Dystrophy

Bourdeaut, Franck, MD, PhD; Paris Sciences et Lettres Research University

Rhabdoid Tumor Predisposition Syndrome

Bourgeois, Patrice, PhD; La Timone Children's Hospital

Trichohepatoenteric Syndrome

Boussion, Simon, MD; CHU de Lille

Thrombocytopenia Absent Radius Syndrome

Bowdin, Sarah, BMed, MSc; Addenbrooke's Hospital

Cherubism

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[Pseudoxanthoma Elasticum](#)

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[FOXP1 Syndrome](#)

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[Multiple Endocrine Neoplasia Type 1](#)

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[MUTYH Polyposis](#)

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Bressman, Susan B, MD; Columbia University Medical Center

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

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[Lymphedema-Distichiasis Syndrome](#)

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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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[Hemophilia A](#)
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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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Brunetti-Pierri, Nicola, MD; University of Naples "Federico II"
[Chondrodysplasia Punctata 1, X-Linked](#)
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[SOST-Related Sclerosing Bone Dysplasias](#)

Brunner, Han, MD, PhD; Radboud University Medical Center
[Autosomal Dominant Robinow Syndrome](#)

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[Hypomyelination and Congenital Cataract](#)

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[Familial Lipoprotein Lipase Deficiency](#)

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

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[Familial Hemophagocytic Lymphohistiocytosis](#)

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

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

[TANGO2 Deficiency](#)

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

Burrell, T Lindsey, PhD; Emory University School of Medicine

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Burt, Randall W, MD; Huntsman Cancer Institute

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Cabalza, Jessica L; George Washington University School of Medicine

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[Krabbe Disease](#)

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[Ornithine Transcarbamylase Deficiency](#)

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[Riboflavin Transporter Deficiency](#)

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[Arterial Tortuosity Syndrome](#)
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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
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Campeau, Philippe M, MD; Sainte-Justine Hospital
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[Brugada Syndrome](#)

Cao, Antonio, MD; Consiglio Nazionale delle Ricerche
[Alpha-Thalassemia](#)
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[ZAP70-Related Combined Immunodeficiency](#)

Capp, Philip K; George Washington University School of Medicine
[Succinic Semialdehyde Dehydrogenase Deficiency](#)

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[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](#)

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[Sickle Cell Disease](#)

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[Salih Myopathy](#)

Caron, Nicholas S, PhD; University of British Columbia

[Huntington Disease](#)

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[DICER1 Tumor Predisposition](#)

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

Carson, Vincent J, MD; Clinic for Special Children

[Maple Syrup Urine Disease](#)

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

Carvill, Gemma, PhD; Northwestern University

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[Spastic Paraplegia 7](#)

Casey, Hannah L, BS; University of Chicago

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Cassidy, Suzanne B, MD; Children's Hospital of Philadelphia

[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

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

[BAP1 Tumor Predisposition Syndrome](#)

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

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Cederbaum, Stephen, MD; UCLA Medical Center

[Arginase Deficiency](#)

Chaix, Charlène; La Timone Children's Hospital

[Trichohepatoenteric Syndrome](#)

Chamberlain, Stormy, PhD; University of Connecticut Health Center

[Maternal 15q Duplication Syndrome](#)

Chamova, Teodora, MD, PhD; Medical University of Sofia

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

Chander, Varuna, MS; Baylor College of Medicine

[Xia-Gibbs Syndrome](#)

Chandra, Sharat, MD; Cincinnati Children's Hospital Medical Center

[WAS-Related Disorders](#)

Chang, Bernard, MD; Harvard Medical School

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

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

Chanprasert, Sirisak, MD; Baylor College of Medicine

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

Chansky, Howard A, MD; University of Washington

[Hereditary Multiple Osteochondromas](#)

Chan, Sophelia HS, MD; University of Hong Kong

[CHKB-Related Muscular Dystrophy](#)

Chao, Daniel L, MD, PhD; Shiley Eye Institute / UCSD

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

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

[Urea Cycle Disorders Overview](#)

Charles, Jane, MD; Medical University of South Carolina

[Smith-Magenis Syndrome](#)

Chaussonot, Annabelle, MD; Nice Hospital

CHCHD10-Related Disorders

Chelban, Viorica, MD; University College London Institute of Neurology

NKX6-2-Related Disorder

Chen, Dong-Hui, MD, PhD; University of Washington Medical Center

SAMD9L Ataxia-Pancytopenia Syndrome

Spinocerebellar Ataxia Type 14

Cheng, Edith, MD, MS; University of Washington School of Medicine

Cystic Fibrosis

Cheng, Shirley SW, MD; Hong Kong Children's Hospital

CTNNB1 Neurodevelopmental Disorder

FAM111A-Related Skeletal Dysplasias

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Craniometaphyseal Dysplasia, Autosomal Dominant

Chen, Karin, MD; University of Washington

X-Linked Severe Combined Immunodeficiency

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Alkaptonuria

Chen, Ming Hui, MD, MMSc; Boston Children's Hospital

FLNA Deficiency

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Aspartylglucosaminuria

Chen, Yuan-Tsong, MD, PhD; Duke University Medical Center

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

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[Huppke-Brendel Syndrome](#)

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[Autosomal Dominant Robinow Syndrome](#)

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

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[Spinocerebellar Ataxia Type 8](#)

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[ALS2-Related Disorder](#)

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[Diamond-Blackfan Anemia](#)

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[DYRK1A Syndrome](#)

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[LAMA2 Muscular Dystrophy](#)

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[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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[GNB1 Encephalopathy](#)

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[Pelizaeus-Merzbacher-Like Disease 1](#)
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[Nijmegen Breakage Syndrome](#)

Concolino, Daniela, MD; University "Magna Graecia"
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[X-Linked Agammaglobulinemia](#)

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[Milroy Disease](#)

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

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[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

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[Primary Familial Brain Calcification](#)

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- Cortese, Andrea, MD, PhD; University College London
[RFC1 CANVAS / Spectrum Disorder](#)
- Coryell, Jason, MS; Oregon Health and Science University
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- 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
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- 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](#)
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- Cragun, Deborah, MS; University of Tampa
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- Craigen, William J, MD, PhD; Baylor College of Medicine
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- Crombez, Eric A, MD; Ultragenyx Pharmaceutical Inc
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- Crosby, Andrew H, BSc, PhD; University of Exeter Medical School
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- Crowley, Leon, MSc; King's College London
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- 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

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Cunningham, Michael L, MD, PhD; University of Washington Schools of Medicine and Dentistry

[Saethre-Chotzen Syndrome](#)

Curiel, Julian, BS; Children's National Health System

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Cutting, Garry R, MD; Johns Hopkins University

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D'Adamo, Maria Cristina, PhD; University of Malta

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D'Andrea, Alan, MD; Dana Farber Cancer Institute

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

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Dafsari, Hormos Salimi, MD; University of Cologne

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

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Dai, Hongzheng, PhD; Baylor College of Medicine

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

[Progressive Pseudorheumatoid Dysplasia](#)

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

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Daley, Shane M, MD; Mayo Clinic

[Jervell and Lange-Nielsen Syndrome](#)

Dalton, Joline C, MS; Institute of Human Genetics

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

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

Danek, Adrian, MD; Ludwig-Maximilians-Universität

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

[GAA-FGF14-Related Ataxia](#)

Darras, Basil T, MD; Boston Children's Hospital / Harvard Medical School

[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](#)

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Day, John W, MD, PhD; Stanford University

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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](#)

de Brouwer, Arjan PM, PhD; Radboud University Nijmegen Medical Center

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

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

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

De Castro, Mauricio, MD; Keesler Air Force Base

[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](#)

de Koning, Tom J, MD, PhD; Lund University

[Serine Deficiency Disorders](#)

de la Morena, M Teresa, MD; Seattle Children's Hospital

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de Leeuw, Nicole, PhD; Radboud University Medical Center

[Kleefstra Syndrome](#)

De Leon, Deborah, MS; Columbia University Medical Center

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De León, Diva D, MD; University of Pennsylvania

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De Paepe, Anne, MD, PhD; Ghent University Hospital

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

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De Vivo, Darryl, MD; Columbia University

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De Voer, Richarda M, PhD; Radboud University Medical Center

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de Vries, Bert BA, MD, PhD; Radboud University Medical Center

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Deardorff, Matthew A, MD, PhD; Children's Hospital Los Angeles

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DeFelice, Magee, MD; Nemours Children's Health

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Dehner, Louis P; Washington University Medical Center

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del Gaudio, Daniela, PhD; University of Chicago

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Delgado, Lisvania, BS; University of Texas - Medical Branch

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Demain, Leigh AM, PhD; University of Manchester and Manchester University NHS Foundation Trust

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Demarest, Scott, MD; University of Colorado

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Demirdas, Serwet, MD, PhD; Erasmus Medical Center

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Dempsey, Melissa A, MS; Parkview Health

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Demuth, Ilja, PhD; Charité – Universitätsmedizin Berlin

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Depienne, Christel, PhD; Hôpital Pitié-Salpêtrière

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Derks, Terry GJ, MD, PhD; University Medical Center Groningen

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Desai, Jigar, PhD; Boston Children's Hospital

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

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Deutch, Natalie, MS; National Human Genome Research Institute
RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies
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Deuschländer, Angela B, MD; Mayo Clinic
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Maternal 15q Duplication Syndrome

Dgany, Orly, PhD; Felsenstein Medical Research Center, Beilinson Campus
Alpha-Thalassemia
Congenital Dyserythropoietic Anemia Type I

Dhamija, Radhika, MD; Mayo Clinic
FGFR1-Related Hartsfield Syndrome
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Di Donato, Nataliya, MD, PhD; Universitätsklinikum Carl Gustav Carus
Baraitser-Winter Cerebrofrontofacial Syndrome

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Di Gregorio, Eleonora, PhD; Città della Salute e della Scienza University Hospital
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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
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Diaz, George A, MD, PhD; Icahn School of Medicine at Mount Sinai
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Dichgans, Martin, MD, PhD; Ludwig-Maximilians-Universität München

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Dickerson, Kathryn E, MD; University of Texas Southwestern Medical Center

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Digweed, Martin, PhD; Charité – Universitätsmedizin Berlin

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DiMauro, Salvatore, MD; Columbia University Medical Center

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Dindot, Scott, PhD; Texas A&M University

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Diop-Bove, Ndeye, PhD; Albert Einstein College of Medicine

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Dirksen, Robert T, PhD; University of Rochester School of Medicine and Dentistry

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DiStasio, Andrew T, PhD; Nationwide Children's Hospital

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Disteche, Christine M, PhD; University of Washington

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DiStefano, Charlotte, PhD; University of California Los Angeles

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Dobson-Stone, Carol, DPhil; University of Sydney

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Doherty, Dan, MD, PhD; University of Washington

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Doherty, Emily S, MD; Carilion Clinic

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Doimo, Mara, PhD; University of Padova

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

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

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

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

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

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Dorsey, Adrienne M; Children's National Medical Center

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Dotti, Maria Teresa, MD; University of Siena

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

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Douzgou, Sofia, MD, PhD; Haukeland University Hospital

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- Dowling, James J, MD, PhD; Hospital for Sick Children
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- Downs, Jenny, MSc, PhD; Telethon Kids Institute
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- Drillings, Ian; Children's National Medical Center
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- Driscoll, Daniel J, MD, PhD; University of Florida College of Medicine
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- Drunat, Séverine, PharmD, PhD; APHP-Robert Debré University Hospital
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- du Souich, Christèle, MSc; University of British Columbia
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- Dua Puri, Ratna, MD; Sir Ganga Ram Hospital
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- Dubois, Anna, BSc, MBChB; Royal Victoria Infirmary
[CYLD Cutaneous Syndrome](#)
- Dugan, Stefanie N, MS; BloodCenter of Wisconsin
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- Duis, Jessica, MD, MS; Children's Hospital Colorado
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- Duker, Angela, MS; Nemours Children's Health
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- Duley, John A, PhD; University of Queensland and Mater Research
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- Dulski, Jaroslaw, MD, PhD; Medical University of Gdansk
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Dumitrescu, Alexandra M, MD, PhD; University of Chicago Medical Center

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Dunn, Clinton P, MD; University of Washington

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

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

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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](#)

Durda, Kate M, MS; University of Iowa

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

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

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

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

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

Nonsyndromic 46,XX Testicular Disorders/Differences of Sex Development

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Earl, Rachel, PhD; University of Washington

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Ebens, Christen, MD, MPH; University of Minnesota

Fanconi Anemia

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

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Eccles, Michael R, PhD; University of Otago

PAX2-Related Disorder

Edvardsson, Vidar Orn, MD; The National University Hospital of Iceland

Adenine Phosphoribosyltransferase Deficiency

Edward, Deepak P, MD; University of Illinois at Chicago

Primary Congenital Glaucoma

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

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Efthymiou, Stephanie, MSc, PhD; University College London

GRIA2-Related Neurodevelopmental Disorder

WARS2 Deficiency

Eggen, Veerle RC, MSc; University of Amsterdam

EXOSC3 Pontocerebellar Hypoplasia

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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

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Ejaz, Resham, MD; McMaster Children's Hospital

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

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Elizondo, Leah I; Baylor College of Medicine

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

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

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Elsharkawi, Ibrahim, MD; Washington University

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

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Emanuel Syndrome

Eng, Charis, MD, PhD; Cleveland Clinic

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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

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Enns, Gregory M, MB ChB; Stanford University

Carnitine-Acylcarnitine Translocase Deficiency

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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

Esposito, Dario, MD; Sapienza Università di Roma

PRICKLE1-Related Disorders

Evangelista, Teresinha, MD; Institut de Myologie

Oculopharyngeal Muscular Dystrophy

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Evans, Kelly N, MD; Seattle Children's Hospital

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Everett, Lorraine A; National Human Genome Research Institute

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

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Ewbank, Clifton, MD; University of California San Francisco

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Fabre, Alexandre, MD, PhD; La Timone Children's Hospital

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Falchetti, Alberto, MD; University Hospital of Careggi

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Falik Zaccai, Tzipora C, MD; Galilee Medical Center

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Fam, Hok Khim, BSc; University of British Columbia

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Fanin, Marina, PhD; University of Padova

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Fanto, Manolis, PhD; King's College London

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Faqeih, Eissa, MD; King Fahad Medical City

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

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

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Farrer, Matthew, PhD; University of British Columbia

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

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Fatemi, Ali, MD; Kennedy Krieger Institute

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Fauth, Christine, MD; Medical University of Innsbruck

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Fecarotta, Simona, MD; Federico II University

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Federico, Antonio, MD; University of Siena

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

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Feldman, Howard H, MD; University of British Columbia

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Felton, Thomas, MS; McLendon Clinical Laboratories

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

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

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Field, Amanda, MPH; ResourcePath

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Fish, Jennifer, PhD; University of Massachusetts Lowell

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FitzPatrick, David R, MD; University of Edinburgh

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

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Flemming, Kelly D, MD; Mayo Clinic

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Frontali, Marina, MD; Italian National Research Council

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Gahl, William A, MD, PhD; National Human Genome Research Institute

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Gallus, Gian Nicola, Dsci; University of Siena

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

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

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Gardner, Kathy Lou, MD; University of Pittsburgh

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Gardner, RJM, MB ChB; University of Otago

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Gassner, Christoph, PhD; Swiss Red Cross

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

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

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

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Gibbs, Richard A, PhD; Baylor College of Medicine

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

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

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Giles, Rachel H, PhD; University Medical Center Utrecht

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Gillis, David, MD; Hadassah Hebrew University Hospital

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Gimpel, Charlotte, MD; Heidelberg University

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

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Glaser, Benjamin, MD; Hadassah-Hebrew University Medical Center

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Glassford, Megan, MMSc; Emory University School of Medicine

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Glass, Ian, MBChB, MD; University of Washington

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Glinton, Kevin, MD, PhD; Baylor College of Medicine

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Gochuico, Bernadette R, MD; National Human Genome Research Institute

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Godfrey, Maurice, PhD; University of Nebraska Medical Center

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Gogineni, Kishore Sai, MSc; Kasturba Medical College, Manipal

[LPIN2-Related Majeed Syndrome](#)

Golabi, Mahin, MD, MPH; San Francisco General Hospital

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Golas, Gretchen A, MS, RN; National Institutes of Health

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Gold-Von Simson, Gabrielle, BA, MD; NYU Medical Center

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Goldberg, Ethan M, MD, PhD; Children's Hospital of Philadelphia

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Goldberg, Y Paul, MBChB, PhD; Xenon Pharmaceuticals Inc

[Juvenile Hemochromatosis](#)

Goldfarb, Lev G, MD; National Institute of Neurological Disorders and Stroke / NIH

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Goldin, Ehud, PhD; National Institutes of Health

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Goldman, I David, MD; Albert Einstein College of Medicine

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

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

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

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Gomes, Alicia, MS; University of Alabama

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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

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

von Willebrand Disease

Goodman, Stephen I, MD; University of Colorado

Glutaric Acidemia Type I

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Factor V Leiden Thrombophilia

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

Aspartylglucosaminuria

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Gordon, Christopher T, PhD; Institut Imagine

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Milroy Disease

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

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

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Gorman, Gráinne S, MBBCh, PhD; Newcastle University

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

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Gosky, Michael, BS, MS; University of Pittsburgh

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

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

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Greenbaum, Larry A, MD, PhD; Emory University

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

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

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

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

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

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Gupta, Karn, MD; Wake Forest University School of Medicine

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[SYNGAP1-Related Intellectual Disability](#)

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Hampel, Heather, MS; The Ohio State University

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Hansen, C David, MD; University of Utah

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

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

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DICER1 Tumor Predisposition

Harris, James C, MD; Johns Hopkins University School of Medicine

HPRT1 Disorders

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Mitochondrial Membrane Protein-Associated Neurodegeneration

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Mandibulofacial Dysostosis with Microcephaly

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X-Linked Hypophosphatemia

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Hayashi, Masahiro, MD, PhD; Yamagata University

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Hehr, Ute, MD; Center for Human Genetics

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Heidlebaugh, Alexis R, ScM; Geisinger Health System

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Heiman, Meadow, MS; Indiana Hemophilia & Thrombosis Center

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

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

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

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Helsmoortel, Céline, MSc; University of Antwerp

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Herbert, Mrudu, MD, MPH; University of Kentucky

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

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

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

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Hershberger, Ray E, MD; The Ohio State University

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

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Hicks, Debbie, PhD; University of Newcastle upon Tyne

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Ho, Gladys; The Children's Hospital at Westmead

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

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

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

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

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

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

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

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

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Isaacs, Adrian M, DPhil; University College London

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Familial Hypercholesterolemia

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Iwasa, Yoichiro, MD, PhD; University of Iowa

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Jackson, Andrew, PhD; University of Edinburgh

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

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James, Aaron; University of California San Francisco

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James, Paula, MD; Queen's University

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Jankovic, Joseph, MD; Baylor College of Medicine

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

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

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

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

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

Short-Chain Acyl-CoA Dehydrogenase Deficiency

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Rotor Syndrome

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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

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X-Linked Lymphoproliferative Disease

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

Smith-Magenis Syndrome

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Hemophilia A

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

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

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

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

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Josephson, Neil C, MD; Seattle Genetics

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Joshi, Nishtha; Baylor College of Medicine

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

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Jung, Hans H, MD; University Hospital Zurich

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Jägle, Herbert, MD; University of Regensburg

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

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

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

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

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

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

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

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

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

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Kang, Jingqiong, PhD; Vanderbilt University Medical Center

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

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Kantarci, Sibel, PhD; Quest Diagnostics Nichols Institute

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

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Kaplan, Julie D, MD; Cleveland Clinic Foundation

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

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

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Khaytin, Ilya, MD, PhD; Northwestern University

[Congenital Central Hypoventilation Syndrome](#)

Kibrom, Sara, MD; Stanford University School of Medicine

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

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

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

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Klar, Aharon, MD; Hebrew University

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Kleta, Robert, MD, PhD; National Institutes of Health

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

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

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

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

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

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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

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Kraemer, Kenneth H, MD; National Cancer Institute

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Krahn, Katherine Nash, MS; University of Iowa

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

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

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

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

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

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Krey, Ilona, MD; Univeristy of Leipzig Medical Center

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

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Kronn, David F, MD; New York Medical College

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

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Kucine, Nicole, MD, MS; Weill Cornell Medicine

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Kuhlenbäumer, Gregor, MD, PhD; University of Kiel

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

[NTHL1 Tumor Syndrome](#)

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

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Kumar, Ajith V, MD; Great Ormond Street Hospital

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Kumar, Anil; University of Leicester

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Kumaran, Neruban, BSc, MBBS; University College London

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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

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[GLYT1 Encephalopathy](#)

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

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

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

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

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Kälviäinen, Reetta, MD, PhD; Kuopio University Hospital

Progressive Myoclonic Epilepsy Type 1

Kölker, Stefan, MD; University Hospital Heidelberg

Classic Isovaleric Acidemia

König, Arne, MD; Philipps-Universität

NSDHL-Related Disorders

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

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

Spinal and Bulbar Muscular Atrophy

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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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Hereditary Coproporphyrinuria

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Laing Distal Myopathy

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Lam, Christina, MD; University of Washington

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Lamont, Phillipa, PhD; Royal Perth Hospital

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Lampe, Anne Katrin, MD; Western General Hospital

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Lamperti, Costanza, MD; IRCCS Foundation Carlo Besta Neurological Institute

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Lange, Lara M, MD; University of Lübeck

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

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Lanpher, Brendan C, MD; Mayo Clinic

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Larson, Austin, MD; University of Colorado

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LaRusch, Jessica, PhD; Ariel Precision Medicine

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Laugel, Vincent, MD, PhD; Laboratory of Medical Genetics

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Laurent, Michaël R, MD, PhD; University Hospitals Leuven

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

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Lawrence, Jessica, MSc; Alberta Health Services

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Layman, Lawrence C, MD; Medical College of Georgia

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Le Goff, Carine, PhD; INSERM U781

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LeBlanc, Shannon, MBBS; University of Melbourne

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Lebwohl, Benjamin, MD, MS; Columbia University

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

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Lee, Chung U, MD; Stanford University

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Lee, Kristen N, MD; University of Michigan

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- Lee, Soohee, PhD; The New York Blood Center
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
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- LeGuern, Eric, MD, PhD; Hôpital de la Pitié-Salpêtrière
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- Lehesjoki, Anna-Elina, MD, PhD; University of Helsinki
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- Lehman, Christopher, MS; Geisinger Health System
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- Lehmann-Horn, Frank, MD, MS, PhD; Ulm University
Hyperkalemic Periodic Paralysis
Hypokalemic Periodic Paralysis
- Lehmberg, Kai, MD; University Medical Center Eppendorf
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- 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
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- Lemke, Johannes R, MD; University of Leipzig Medical Center
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Lemmers, Richard JLF, PhD; Leiden University Medical Center

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Lemmink, Henny, PhD; University Medical Center

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[CDKL5 Deficiency Disorder](#)

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Leroy, Bart P, MD, PhD; Ghent University Hospital & Ghent University

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

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

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Leturcq, France, MD; Hôpital Cochin

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Leung, Alva, BS; California Pacific Medical Center

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

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Leuzzi, Vincenzo, MD; Sapienza Università di Roma

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Leventer, Richard J, BMedSci, MBBS, PhD; Royal Children's Hospital

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

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

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Levy, Amanda M, MSc; Rigshospitalet

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

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

[Hypermobility Ehlers-Danlos Syndrome](#)

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[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 Immunosseous Dysplasia](#)

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

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

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Li, Hong, MD, PhD; Emory University School of Medicine
[CTCF-Related Disorder](#)

Lilien, Marc, MD, PhD; University Medical Center Utrecht
[Nephronophthisis-Related Ciliopathies](#)

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[RRM2B Mitochondrial DNA Maintenance Defects](#)

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[Branchiooculofacial Syndrome](#)
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Lindhurst, Marjorie J, PhD; National Institutes of Health
[SLC25A19-Related Thiamine Metabolism Dysfunction](#)

Lindor, Noralane M, MD; Mayo Clinic
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Lindsay, Mark E, MD, PhD; MassGeneral Hospital for Children
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Lines, Matthew, MD, MSc; University of Calgary
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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
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Liu, Lawrence U, MD; Icahn School of Medicine at Mount Sinai
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Liu, Paul, MD, PhD; National Human Genome Research Institute

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Lloyd, Ricardo V, MD, PhD; Mayo Clinic

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

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

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Lohmann, Katja, PhD; University of Lübeck

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Loomes, Kathleen M, MD; University of Pennsylvania

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Lopez, Christina, BS; California Pacific Medical Center

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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

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Loyd, James E, MD; Vanderbilt University Medical Center

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Lubarr, Naomi, MD; Beth Israel Medical Center

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Luchsinger, Kadi, BS; Dup15q Alliance

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

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Lucking, Christoph, MD; Ludwig-Maximilians-University

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

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

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

[Smith-Magenis Syndrome](#)

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

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Lyman, Benjamin, DO; LSU Health Science Center

[Cystic Fibrosis](#)

Lynch, Henry, MD; Creighton University Hereditary Cancer Center

[MUTYH Polyposis](#)

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MED12-Related Disorders

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Infantile-Onset Spinocerebellar Ataxia

Lücke, Thomas, MD, PhD; Hannover Medical School

Schimke Immunoosseous Dysplasia

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TXNL4A-Related Craniofacial Disorders

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Maas, Saskia, MD; University of Amsterdam

Trichorhinophalangeal Syndrome

Maccollin, Mia M, MD; Harvard Medical School/Massachusetts General Hospital

NF2-Related Schwannomatosis

Macdermott, Seamus, MD; Torbay Hospital

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

MacFarlane, Julie, MS; Xenon Pharmaceuticals Inc

Juvenile Hemochromatosis

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Cleidocranial Dysplasia Spectrum Disorder

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Familial Hemophagocytic Lymphohistiocytosis

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Diabetes Mellitus, 6q24-Related Transient Neonatal

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Maconachie, Gail, BSc, PhD; University of Leicester

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Madhok, Sehajvir, MD; Columbia University Irving Medical Center

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Magenis, Ellen, MD; Oregon Health and Science University

[Smith-Magenis Syndrome](#)

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Mahadeo, Kris M, MD, MPH; Albert Einstein College of Medicine

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[Classic Ehlers-Danlos Syndrome](#)

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Mao, Rong, MD; University of Utah

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Marafi, Dana, MD, MSc; Kuwait University

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Marazita, Mary L, PhD; University of Pittsburgh

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Marbach, Felix, MD; Heidelberg University

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Marcelis, Carlo LM, MD; Radboud University Nijmegen Medical Center

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Marchuk, Douglas, PhD; Duke University

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Marconi, Roberto, MD; Ospedale Misericordia

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Marcucci, Guido, MD; Ohio State University

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Margolis, Russell L, MD; Johns Hopkins University School of Medicine

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- Marini, Francesca, PhD; Italian Foundation for Research on Bone Diseases
[Multiple Endocrine Neoplasia Type 1](#)
- Markovitz, Rebecca, MD, PhD; Baylor College of Medicine
[GARS-Associated Axonal Neuropathy](#)
- Mark, Paul, MD; Michigan State University
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- Marquard, Jessica, MS; Cleveland Clinic
[Multiple Endocrine Neoplasia Type 2](#)
- Marquardt, Thorsten, Dr. med.; Universitätsklinikum Münster
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- Marras, Connie, MD, PhD; University of Toronto
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- Marshall, Jan D, MS; The Jackson Laboratory
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[Familial Hemophagocytic Lymphohistiocytosis](#)
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- Martin, Christa L, PhD; Geisinger Health System
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- Martin, Donna M, MD, PhD; University of Michigan
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- Martin, George M, MD; University of Washington
[Werner Syndrome](#)
- Martin, Jack W, MD; The University of Texas MD Anderson Cancer Center
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- Martin, Rick A, MD; Saint Louis University
[Mucopolysaccharidosis Type II](#)
- Martín, Bertha, PhD; University of Iowa
[C3 Glomerulopathy](#)

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[Geleophysic Dysplasia](#)
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Mastrangelo, Mario, MD, PhD; Sapienza Università di Roma
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Mastrianni, James A, MD, PhD; University of Chicago
[Genetic Prion Disease](#)

Matalon, Reuben, MD, PhD; University of Texas Medical Branch
[Canavan Disease](#)

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

Mathews, Jennifer, MS; University of North Carolina at Chapel Hill
[Angelman Syndrome](#)

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[Spinocerebellar Ataxia Type 37](#)

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Matsuura, Tohru, MD; Jichi Medical University
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Matton, Charlotte, MSc; Ghent University
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Mavrogiannis, Lampros A, DPhil, MSc; St James's University Hospital
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Mazzanti, Andrea, MD, PhD; ICS Maugeri
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Mazzeu, Juliana, PhD; University of Brasília
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McCarthy, Liam, MD; Birmingham Women's and Children's Hospital
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McCormick, Elizabeth M, MS; Children's Hospital of Philadelphia
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McCormick, Sally PA, PhD; University of Otago
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McCray, Brett A, MD, PhD; Johns Hopkins University School of Medicine
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McDaniel, D Olga, PhD; University of Mississippi Medical Center
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McDermott, Deborah A, MS; Consultant, Human Genetics and Genetic Counseling
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McDonald-McGinn, Donna M, MS; Children's Hospital of Philadelphia
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McDonald, Jamie, MS; University of Utah Medical Center
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McDonell, Laura M, MD, PhD; Dalhousie University
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McFarland, Robert, MA, MBBS, PhD; Newcastle University
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McFarquhar, Ashley, BSc; McGill University
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McGarrity, Thomas J, MD; Milton S Hershey Medical Center
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McGovern, Margaret M, MD, PhD; Stony Brook University School of Medicine
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McIntosh, Iain, PhD; American University of the Caribbean
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McLean, WH Irwin, DSc; University of Dundee
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McMullin, Mary Frances, MD; Queens University
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McNiven, Vanda, MD, MSc; McMaster Children's Hospital
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Nagappa, Madhu, MD; National Institute of Mental Health & Neurosciences

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Namavar, Yasmin, MSc; University of Amsterdam

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Nance, Martha A, MD; Hennepin County Medical Center

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

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

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

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

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Osborne, Lucy R, PhD; University of Toronto

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Oshima, Junko, MD, PhD; University of Washington

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Ostergaard, Elsebet, MD, PhD; National University Hospital Rigshospitalet

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

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Oswald, Gretchen L, MS; Johns Hopkins Medical Center

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Ottman, Ruth, PhD; Columbia University

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Paisey, Richard B, MD; Torbay Hospital

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

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Pallais, J Carl, MD, MPH; Massachusetts General Hospital

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Palmer, Elizabeth, MBBS, PhD; University of New South Wales

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Palsson, Runolfur, MD; The National University Hospital of Iceland

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Pal, Tuya, MD; Vanderbilt-Ingram Cancer Center

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Pandya, Arti, MBA, MD; Virginia Commonwealth University Health System

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Parboosingh, Jillian S, PhD; University of Calgary

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Parente Freixo, João, MD; Universidade do Porto

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Pariani, Mitchel, MS; Stanford Center for Inherited Cardiovascular Disease

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Parisi, Melissa A, MD, PhD; National Institute of Child Health & Human Development

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Parker, Alasdair Patrick John, MA, MBBS, MD; Addenbrooke's Hospital

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Parkin, Kimberly; Massachusetts General Hospital

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Park, Julien H, Dr. med.; Universitätsklinikum Münster

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Park, Julie R, MD; Seattle Children's Hospital

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Pascotto, Antonio, MD; Second University of Naples

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Pascual, Juan M, MD, PhD; University of Texas Southwestern Medical Center

Glucose Transporter Type 1 Deficiency Syndrome

Pasini, Elena, MD; Bellaria Hospital

Autosomal Dominant Epilepsy with Auditory Features

Passarge, Eberhard, MD; University of Essen

Retinoblastoma

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

ASPM Primary Microcephaly

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Pastores, Gregory M, MD; University College

Gaucher Disease

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Factor V Leiden Thrombophilia

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Troyer Syndrome

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Isolated and Classic Cutis Marmorata Telangiectatica Congenita

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NKX2-1-Related Disorders

Patel, Pragna I, PhD; Nagoya University Graduate School of Medicine

Friedreich Ataxia

Patel, Swati G, MD, MS; University of Colorado Anschutz Medical Center

APC-Associated Polyposis Conditions

Patterson, Marc, MD; Mayo Clinic

Niemann-Pick Disease Type C

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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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Tuberous Sclerosis Complex

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

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Pizzo, Lucilla, MS; Pennsylvania State University

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Plotkin, Scott, MD, PhD; Harvard Medical School and Massachusetts General Hospital

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Prades, Silvia, PhD; Ataxia UK

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Prasad, Rathi, MBBS, PhD; Queen Mary University of London

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Prasad, Sai D, BS; University of Iowa Hospitals and Clinics

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Priori, Silvia G, MD, PhD; ICS Maugeri

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Prokisch, Holger, PhD; Technische Universität München

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Ptacek, Louis J, MD; University of California at San Francisco

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Puck, Jennifer M, MD; University of California San Francisco

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Puffenberger, Erik G, PhD; Clinic for Special Children

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Pulst, Stefan M, MD; University of Utah

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

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Pyeritz, Reed E, MD, PhD; Hospital of the University of Pennsylvania

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Pyle, Louise C, MD, PhD; Children's National Medical Center

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Quintáns, Beatriz, PhD; Centro de Investigación Biomédica en red de Enfermedades Raras

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Rabin, Rachel, MS; NYU Grossman School of Medicine

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Raininko, Raili, MD, PhD; Uppsala University

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Raygada, Margarita, MSc, PhD; National Institutes of Health

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Rea, Hannah, PhD; University of Washington

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Redfield, Shelby, MS; Boston Children's Hospital

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Redman, Colvin M, PhD; The New York Blood Center

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Rees, Mark I, PhD; Swansea University

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Refetoff, Samuel, MD; University of Chicago Medical Center

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Regalado, Ellen, MS; University of Texas Health Science Center at Houston McGovern Medical School

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Rehder, Catherine, PhD; Duke University Medical Center

Phosphorylase Kinase Deficiency

Reichenberger, Ernst, PhD; UConnHealth

Cranio metaphyseal Dysplasia, Autosomal Dominant

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PURA-Related Neurodevelopmental Disorders

Reilly, Mary M, MBBCh, MD; University College London

RFC1 CANVAS / Spectrum Disorder

Reinhart, Michaela, MD; Children's Hospital of Philadelphia

TRMU Deficiency

Reischl-Hajiabadi, Anna, MD; University Hospital Heidelberg

Classic Isovaleric Acidemia

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Maternal 15q Duplication Syndrome

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Genetic Atypical Hemolytic-Uremic Syndrome

Renaud, Mathilde, MD, PhD; CHRU de Nancy

GAA-FGF14-Related Ataxia

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WFS1 Spectrum Disorder

Revah-Politi, Anya, MS; Columbia University Irving Medical Center

GNB1 Encephalopathy

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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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Roberts, Amy E, MD; Children's Hospital Boston

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Robitaille, Yves, MD; Enfant-Jesus Hospital, Quebec City

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Rogers, R Curtis, MD; Greenwood Genetic Center

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Scherer, Gerd, PhD; University of Freiburg

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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

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Stevens, Cathy A, MD; University of Tennessee College of Medicine

[Rubinstein-Taybi Syndrome](#)

Stevenson, David A, MD; Stanford University

[Capillary Malformation-Arteriovenous Malformation Syndrome](#)

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Stevenson, Roger E, MD; Greenwood Genetic Center

[Alpha-Thalassemia X-Linked Intellectual Disability Syndrome](#)

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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

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Stoller, James K, MD, MS; Cleveland Clinic

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Stone, Jeremy T, BSc; Mayo Clinic

[LRRK2 Parkinson Disease](#)

Storey, Elsdon, DPhil; Monash University

[Spinocerebellar Ataxia Type 20](#)

Stover, Niamh B, MS; Banner Health

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Stratakis, Constantine A, DSc, MD; Foundation for Research & Technology Hellas

[Carney Complex](#)

Stratton, Pamela, MD; National Institute of Neurological Disorders and Stroke

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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

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Stumpel, Connie, MD, PhD; Academic Hospital Maastricht

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Subramony, SH, MD; University of Florida

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Suchy, Sharon F, PhD; GeneDx, Inc

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

Suit, Lindsey; University of California, San Francisco

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Sullivan, Bonnie, MD; Children's Mercy Kansas City

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Sumegi, Janos, MD, PhD; Cincinnati Children's Hospital Medical Center

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Summar, Marshall L, MD; Children's National Health System

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Sutton, V Reid, MD; Baylor College of Medicine

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Suwannarat, Pim, MD; Faculty of Medicine, Ramathibodi Hospital

Alkaptonuria

Suzuki, Tamio, MD, PhD; Yamagata University

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Sveden, Abigail, MS; Boston Children's Hospital

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Swanson, Michael, PhD; University of Colorado School of Medicine

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Sweadner, Kathleen J, PhD; Massachusetts General Hospital

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Sweeney, Elizabeth, MB ChB, MD; Liverpool Women's Hospital

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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

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Tabarki, Brahim, MD; Prince Sultan Military Medical City

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Taglialatela, Maurizio, MD, PhD; University of Naples Federico II

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Tamburro, Joan, DO; Cleveland Clinic

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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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[Fanconi Anemia](#)

Tanpaiboon, Pranoot, MD; Children's National Medical Center

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[IPEX Syndrome](#)

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Tan, Wen-Hann, MBBS; Boston Children's Hospital

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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

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Taylor, Annette K, MS, PhD; Laboratory Corporation of America Holdings

[Celiac Disease](#)

Taylor, Cora M, PhD; Geisinger Health System

[16p11.2 Recurrent Deletion](#)

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Taylor, Matthew RG, MD, PhD; University of Colorado Anschutz Medical Campus

[Danon Disease](#)

Taylor, Robert W, PhD; Newcastle University

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[Aspartylglucosaminuria](#)

Teebi, Ahmad, MD; Education City

[Cherubism](#)

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Temple, Isabel Karen, MD; Southampton University Hospitals NHS Trust

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

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Terry, Sharon F, MA; PXE International

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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

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Thoene, Jess G, MD; University of Michigan

[Citrullinemia Type I](#)

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Thomas, Julian, MD; Children's Hospital Orange County

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Thomas, Mervyn G, MBChB, PhD; University of Leicester

[FRMD7-Related Infantile Nystagmus](#)

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Thomas, Rhys, PhD; Newcastle University

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Thomas, Shery; University of Leicester

[FRMD7-Related Infantile Nystagmus](#)

Thompson, Arthur R, MD, PhD; University of Washington

[Hemophilia A](#)

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Thompson, Reid, MD; Johns Hopkins University

[Barth Syndrome](#)

Thorburn, David R, PhD; Royal Children's Hospital and University of Melbourne

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[OTOF-Related Deafness](#)

Thunell, Stig, MD, PhD; Karolinska Institute

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Tift, Cynthia J, MD, PhD; National Human Genome Research Institute

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Tiller, George E, MD, PhD; Kaiser Permanente

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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

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Nonketotic Hyperglycinemia

Torgerson, Troy, MD, PhD; Division of Infectious Diseases, Immunology, and Rheumatology

IPEX Syndrome

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Oral-Facial-Digital Syndrome Type I

Thrombocytopenia Absent Radius Syndrome

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Chediak-Higashi Syndrome

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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

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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

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Tran-Fadulu, Van, MS; University of Texas Medical School at Houston

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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

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Trzupek, Karmen M, MS; InformedDNA

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Tsang, Mandy HY, MMSc; University of Hong Kong

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Tsilou, Ekaterini, MD; Center for Biologics Evaluation and Research, US FDA

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Tsuji, Shoji, MD, PhD; University of Tokyo Graduate School of Medicine

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Tsunogai, Toshiki, MD; Jikei University School of Medicine
[Thiamine-Responsive Megaloblastic Anemia Syndrome](#)

Tuchman, Mendel, MD; Children's National Medical Center
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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](#)
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Tuupanen, Sari, PhD; Blueprint Genetics
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Tümer, Zeynep, DMSc, MD, PhD; Rigshospitalet
[DLG4-Related Synaptopathy](#)

Tüysüz, Beyhan, MD, PhD; Istanbul University
[Trichorhinophalangeal Syndrome](#)

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Udd, Bjarne, MD, PhD; Tampere University Hospital
[ANO5 Muscle Disease](#)
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Uhlig, Holm; NIHR Oxford Biomedical Research Centre
[PI4KA-Related Disorder](#)

Uitto, Jouni, MD, PhD; Thomas Jefferson University
[Kindler Syndrome](#)
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Umair, Muhammad, PhD; King Saud Bin Abdulaziz University for Health Sciences
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Unger, Sheila, MD; University of Lausanne
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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

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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

Valencia, C Alexander, PhD; Cincinnati Children's Hospital Medical Center

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

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Lynch Syndrome

Valverde de Morales, Hannah Gabriela, MD; Integra Genetics

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Van Camp, Guy, PhD; University of Antwerp

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van der Crabben, Saskia N, MD, PhD; Amsterdam UMC

[Serine Deficiency Disorders](#)

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

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van der Sluijs, Pleuntje J, MSc; Leiden University Medical Center

[ARID1B-Related Disorder](#)

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[ADNP-Related Disorder](#)

van Dijk, Fleur S, MD, PhD; London North West University Healthcare NHS Trust

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Van Esch, Hilde, MD, PhD; University Hospitals Leuven

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Van Hauwe, Peter; University of Antwerp

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Van Hove, Johan LK, MD, PhD; University of Colorado School of Medicine

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Van Lierop, Antoon, MD, PhD; Leiden University Medical Center

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Van Maldergem, Lionel, MD, PhD; Université de Franche-Comté

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van Nesselrooij, Bernadette, MD, PhD; University Medical Center Utrecht

[Von Hippel-Lindau Syndrome](#)

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[Ataxia-Telangiectasia](#)

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- van Steensel, Maurice, MD, PhD; University Hospital Maastricht
[Pachyonychia Congenita](#)
- van Swieten, John C, MD, PhD; Erasmus Medical Center
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- Van Zanten, Malou, PhD; St George's, University of London
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- Vance, Jeffery M, MD, PhD; Duke University Medical Center
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- Vandana, VP, PhD; National Institute of Mental Health & Neurosciences
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- Vander Lugt, Mark, MD; University of Michigan Medical School
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- Vanderver, Adeline, MD; Children's Hospital of Philadelphia
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- Varon, Raymonda, PhD; Charité – Universitätsmedizin Berlin
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- Varvagiannis, Konstantinos, MD; Cyprus Institute of Neurology & Genetics
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- Vaughan, Douglas E, MD; Northwestern University Feinberg School of Medicine
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- Veenhuis, Stefanie, MD; Radboud University Medical Center
[Ataxia-Telangiectasia](#)
- Veerapandiyan, Aravindhyan, MBBS; University of Rochester Medical Center
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- Vega, Hugo H, MD, PhD; City University of New York

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[RNU4atac-opathy](#)

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Velayos Baeza, Antonio, PhD; Wellcome Trust Centre for Human Genetics

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Velez-Bartolomei, Frances, MD; Stanford University

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Velleman, Shelley L, PhD; University of Vermont

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Venance, Shannon L, MD, PhD; University of Western Ontario

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Venditti, Charles P, MD, PhD; National Human Genome Research Institute

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Veneziano, Liana, PhD; Italian National Research Council

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[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

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Verma, Ishwar Chandar, MD; Sir Ganga Ram Hospital

[Primrose Syndrome](#)

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