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Bindu, Parayil Sankaran, MD; Children's Hospital at Westmead

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Blackstone, Craig, MD, PhD; Massachusetts General Hospital

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Blau, Nenad, PhD; University Children's Hospital

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



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

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

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Bloomer, Joseph, MD; University of Alabama at Birmingham

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

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Bonafé, Luisa, MD, PhD; Lausanne University Hospital

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Bonkowski, Emily, ScM; St Jude Children's Research Hospital

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Boon, Elles MJ, PhD; Leiden University Medical Center

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Boon, Laurence M, MD, PhD; Cliniques universitaires Saint-Luc

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Brand, Randall, MD; University of Pittsburgh

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Brandt, Vicky L; Baylor College of Medicine

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Bresin, Elena, MD; Istituto di Ricerche Farmacologiche Mario Negri-IRCCS

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Breuning, Martijn H, MD, PhD; Leiden University Medical Center

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Brice, Alexis, MD; Hôpital Pitié-Salpêtrière

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Brockmann, Knut, MD; University Medical Center

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Bronicki, Lucas, PhD; Cincinnati Children's Hospital Medical Center

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Brosh, Robert M, PhD; National Institute on Aging/NIH

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Brown, Jeremy M, MD; Addenbrooke's Hospital

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Bruckner, Anna L, MD; University of Colorado School of Medicine

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Brugada, Pedro, MD, PhD; Free University of Brussels

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Brugada, Ramon, MD, PhD; University of Girona

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

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Brunkow, Mary E, PhD; Institute for Systems Biology

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Brzezinski, Jack, MD, PhD; Hospital for Sick Children

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

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Camaschella, Clara, MD; Istituto Scientifico San Raffaele



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Chaix, Charlene; La Timone Children's Hospital

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Chen, Marcus, MD; National Heart, Lung, and Blood Institute - NIH

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

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Chen, Xin, PhD; University of Texas Southwestern Medical Center

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

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Cheung, Sau Wai, MBA, PhD; Baylor College of Medicine

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Chinnery, Patrick F, BMedSci, MBBS, PhD; University of Cambridge

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Concolino, Daniela, MD; University "Magna Graecia"

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Conley, Mary Ellen, MD; University of Tennessee

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Connell, Fiona; Guy's Hospital

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Connor, Jessica A, MS; Counsyl, Inc

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Conta, Jessie, MS; Seattle Children's Hospital

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Conway, Gerard S, MD; University College London

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Conway, Robert, MD; Wayne State University

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Cook Shukla, Lola, MS; Indiana University School of Medicine

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

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Cook, Jared F, MA; Wake Forest School of Medicine

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Cooper, Edward C, MD, PhD; Baylor College of Medicine

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Cooper, Megan, MD, PhD; Washington University

[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

Copeland, William C, PhD; National Institute of Environmental Health Sciences

[POLG-Related Disorders](#)

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

[DYNC1H1-Related Disorders](#)

Coppola, Giangennaro, MD; University of Salerno

[KCNQ2-Related Disorders](#)

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Coppola, Giovanni, MD; University of California Los Angeles

[Primary Familial Brain Calcification](#)

Coppola, Stephanie, BS; Jefferson Medical College

[Krabbe Disease](#)

Corces, Victor, PhD; Emory University School of Medicine

[CTCF-Related Disorder](#)

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

[Geleophysic Dysplasia](#)

[Weill-Marchesani Syndrome](#)

Correa, Fernanda Azevedo, MD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo

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

Cortese, Andrea, MD, PhD; University College London

[RFC1 CANVAS / Spectrum Disorder](#)

Coryell, Jason, MS; Oregon Health and Science University

[Pantothenate Kinase-Associated Neurodegeneration](#)

Coucke, Paul, MD, PhD; Ghent University Hospital

[Arterial Tortuosity Syndrome](#)

Coughlin, Curtis, MS; University of Colorado School of Medicine

[Nonketotic Hyperglycinemia](#)

Couillard-Despres, Sebastien, PhD; Paracelsus Medical University Salzburg

[DCX-Related Disorders](#)

Coulter-Mackie, Marion B, PhD; British Columbia's Children's and Women's Hospital

[Primary Hyperoxaluria Type 1](#)

Cowan, Jason, MS; University of Miami Miller School of Medicine



[LMNA-Related Dilated Cardiomyopathy](#)

Cox, Diane W, PhD; University of Alberta

[Alpha-1 Antitrypsin Deficiency](#)[Wilson Disease](#)

Cragun, Deborah, MS; University of Tampa

[Cytochrome P450 Oxidoreductase Deficiency](#)

Craigen, William J, MD, PhD; Baylor College of Medicine

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Crino, Peter B, BA, MD, PhD; University of Maryland School 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

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

Crowley, William F, MD; Massachusetts General Hospital

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

Crow, Yanick J, BMedSci, MBBS, PhD; Central Manchester Foundation Trust University Hospitals

[Aicardi-Goutieres Syndrome](#)

Cruts, Marc, PhD; University of Antwerp

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

Cruz Corchado, Johnny; University of Iowa

[C3 Glomerulopathy](#)

Cruz, Vincent, MD, MS; DDC Clinic – Center for Special Needs Children

[GM3 Synthase Deficiency](#)

Cubells, Joseph F, MD, PhD; Emory University School of Medicine

[3q29 Recurrent Deletion](#)

Cudrici, Cornelia, MD; National Heart, Lung and Blood Institute

[TNF Receptor-Associated Periodic Fever Syndrome](#)

Culver, Julie O Bars, MS; Fred Hutchinson Cancer Research Center

### BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Culver, Kathy, MS; Sutter Pacific Medical Foundation/California Pacific Medical Center  
[Simpson-Golabi-Behmel Syndrome Type 1](#)

Cunniff, Christopher M, MD; Weill Cornell Medical College  
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Cunningham, Lea, MD; National Cancer Institute  
[RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies](#)

Cunningham, Michael L, MD, PhD; University of Washington Schools of Medicine and Dentistry  
[Saethre-Chotzen Syndrome](#)

Curiel, Julian, BS; Children's National Health System  
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Cutting, Garry R, MD; Johns Hopkins University  
[Cystic Fibrosis](#)  
[Thanatophoric Dysplasia](#)  
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Córdova, Lilian, MD; University of British Columbia  
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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

[ELANE-Related Neutropenia](#)

Daley, Shane M, MD; Mayo Clinic

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Dalton, Joline C, MS; Institute of Human Genetics

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

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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](#)

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Davenport, Sandra LH, MD; Sensory Genetics and Neurodevelopment

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Davies, Alexandra K, PhD; University of Cambridge

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Davis, Joie; NIH

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Day, John W, MD, PhD; Stanford University

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Dazzo, Emanuela, PhD; CNR Institute of Neuroscience

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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

CLPB Deficiency

Feingold Syndrome 1

Phosphoribosylpyrophosphate Synthetase Deficiency

Phosphoribosylpyrophosphate Synthetase Superactivity

SERAC1 Deficiency

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

VPS13A Disease

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

Fibrous Dysplasia / McCune-Albright Syndrome

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

X-Linked Hyper IgM Syndrome

de Leeuw, Nicole, PhD; Radboud University Medical Center

Kleefstra Syndrome

De Leon, Deborah, MS; Columbia University Medical Center

DYT1 Early-Onset Isolated Dystonia

De León, Diva D, MD; University of Pennsylvania

Permanent Neonatal Diabetes Mellitus

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

Arterial Tortuosity Syndrome

Classic Ehlers-Danlos Syndrome

EFEMP2-Related Cutis Laxa

de Valles-Ibáñez, Guillem, BSc, MSc, PhD; University of Otago

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

[CLCN7-Related Osteopetrosis](#)

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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DeFelice, Magee, MD; Nemours Children's Health

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Dehghani, Seyed Mohsen, MD, MPH; Shiraz University of Medical Sciences

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

[Friedreich Ataxia](#)

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

[Canavan Disease](#)

Dellefave-Castillo, Lisa, MS; University of Chicago

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Dell, Katherine MacRae, MD; Case Western Reserve University School of Medicine  
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Demain, Leigh AM, PhD; University of Manchester and Manchester University NHS Foundation Trust  
[Perrault Syndrome](#)

Demarest, Scott, MD; University of Colorado  
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Demirdas, Serwet, MD, PhD; Erasmus Medical Center  
[TNXB-Related Classical-Like Ehlers-Danlos Syndrome](#)

Dempsey, Melissa A, MS; Parkview Health  
[Allan-Herndon-Dudley Syndrome](#)  
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Demuth, Ilja, PhD; Charité – Universitätsmedizin Berlin  
[Nijmegen Breakage Syndrome](#)

Deneubourg, Celine, MSc; King's College London  
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Depienne, Christel, PhD; Hôpital Pitié-Salpêtrière  
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[Hidrotic Ectodermal Dysplasia 2](#)

Derks, Terry GJ, MD, PhD; University Medical Center Groningen  
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Derry, W Brent, PhD; The Hospital for Sick Children  
[Familial Cerebral Cavernous Malformation](#)

Desai, Jigar, PhD; Boston Children's Hospital  
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Desnick, Robert J, MD, PhD; Icahn School of Medicine at Mount Sinai  
[Congenital Erythropoietic Porphyria](#)  
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Deutch, Natalie, MS; National Human Genome Research Institute

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Deuschländer, Angela B, MD; Mayo Clinic

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

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Dgany, Orly, PhD; Felsenstein Medical Research Center, Beilinson Campus

[Alpha-Thalassemia](#)

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

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

[Episodic Ataxia Type 1](#)

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

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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

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

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Dias, Renuka, PhD; Birmingham Women's and Children's Hospital

[WFS1 Spectrum Disorder](#)

Diaz, George A, MD, PhD; Icahn School of Medicine at Mount Sinai

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Dib-Hajj, Sulayman D, PhD; Yale University School of Medicine

[SCN9A Neuropathic Pain Syndromes](#)

Dichgans, Martin, MD, PhD; Ludwig-Maximilians-Universität München

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Dietz, Harry C, MD; Johns Hopkins University School of Medicine

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DiGiovanna, John J, MD; National Cancer Institute

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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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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

[Y Chromosome Infertility](#)

DiStefano, Charlotte, PhD; University of California Los Angeles

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Dobson-Stone, Carol, DPhil; University of Sydney

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Dobyns, William B, MD; University of Minnesota

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

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



Doherty, Dan, MD, PhD; University of Washington

[MN1 C-Terminal Truncation Syndrome](#)

Doherty, Emily S, MD; Carilion Clinic

[Muenke Syndrome](#)

Doimo, Mara, PhD; University of Padova

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

[Wilms Tumor Predisposition](#)

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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[Donnai-Barrow Syndrome](#)

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

[DICER1 Tumor Predisposition](#)

Dorsey, Adrienne M; Children's National Medical Center

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Dotti, Maria Teresa, MD; University of Siena

[Cerebrotendinous Xanthomatosis](#)

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

[Sickle Cell Disease](#)

Douzgou, Sofia, MD, PhD; Haukeland University Hospital

[PPP2R1A-Related Neurodevelopmental Disorder](#)

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

[X-Linked Myotubular Myopathy](#)

Downs, Jenny, MSc, PhD; Telethon Kids Institute

[CDKL5 Deficiency Disorder](#)

Drillings, Ian; Children's National Medical Center

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

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

Angelman Syndrome

Prader-Willi Syndrome

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

ASPM Primary Microcephaly

Baraitser-Winter Cerebrofrontofacial Syndrome

WDR62 Primary Microcephaly

du Souich, Christèle, MSc; University of British Columbia

NSDHL-Related Disorders

UNC80 Deficiency

Dua Puri, Ratna, MD; Sir Ganga Ram Hospital

Primrose Syndrome

Dubois, Anna, BSc, MBChB; Royal Victoria Infirmary

CYLD Cutaneous Syndrome

Dugan, Stefanie N, MS; BloodCenter of Wisconsin

ANKRD26-Related Thrombocytopenia

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

Schinzel-Giedion Syndrome

SETBP1 Haploinsufficiency Disorder

Duker, Angela, MS; Nemours Children's Health

Microcephalic Osteodysplastic Primordial Dwarfism Type II

Rhizomelic Chondrodysplasia Punctata Type 1

RNU4atac-opathy

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

Phosphoribosylpyrophosphate Synthetase Deficiency

Phosphoribosylpyrophosphate Synthetase Superactivity

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

CSF1R-Related Disorder

DCTN1-Related Neurodegeneration

VPS35-Related Parkinson Disease

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

Allan-Herndon-Dudley Syndrome

Dunn, Clinton P, MD; University of Washington

X-Linked Hyper IgM Syndrome

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

Congenital Mirror Movements

Dunwoodie, Sally, BSc, PhD; Victor Chang Cardiac Research Institute  
Congenital NAD Deficiency Disorder  
Spondylocostal Dysostosis, Autosomal Recessive

Dunø, Morten, PhD; Rigshospitalet  
Myotonia Congenita

Dupré, Nicolas, MD, MSc; Université Laval  
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum  
SYNE1 Deficiency

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

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

Duquette, Antoine, MD, MSc; Centre hospitalier de l'Université de Montréal  
Friedreich Ataxia

Durda, Kate M, MS; University of Iowa  
IRF6-Related Disorders

Durie, Peter R, MD; The Hospital for Sick Children, University of Toronto  
Shwachman-Diamond Syndrome

Durr, Alexandra, MD, PhD; Hôpital de la Pitié-Salpêtrière  
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Dykens, Elisabeth, PhD; University of California, Los Angeles  
Smith-Magenis Syndrome

Dyment, David A, DPhil, MD; Children's Hospital of Eastern Ontario  
ASAH1-Related Disorders  
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TRPM3-Related Neurodevelopmental Disorder

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

[AP-4-Associated Hereditary Spastic Paraplegia](#)[EPG5-Related Disorder](#)[PRRT2-Related Disorder](#)[Spastic Paraplegia 15](#)[TECPR2-Related Hereditary Sensory and Autonomic Neuropathy with Intellectual Disability](#)

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

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

[HFE-Related Hemochromatosis](#)

Efthymiou, Stephanie, MSc, PhD; University College London

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Eggen, Veerle RC, MSc; University of Amsterdam

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

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Eichler, Florian S, MD; Massachusetts General Hospital

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Ejaz, Resham, MD; McMaster Children's Hospital

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El Feghaly, Jinia, MD; University of Rochester Medical Center

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

## Glycogen Storage Disease Type I

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

Asparagine Synthetase Deficiency

Deoxyguanosine Kinase Deficiency

El-Hattab-Alkuraya Syndrome

FARS2 Deficiency

FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion Syndrome

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MELAS

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Primary Carnitine Deficiency

SUCLA2-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria

SUCLG1-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria

TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated

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

LRRK2 Parkinson Disease

Elizondo, Leah I; Baylor College of Medicine

Schimke Immunoosseous Dysplasia

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

Ritscher-Schinzel Syndrome

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

Pseudohypoaldosteronism Type II

Elsas, Louis J, MD; University of Miami

Classic Galactosemia and Clinical Variant Galactosemia

Elsa, Sarah H, PhD; Baylor College of Medicine

Aromatic L-Amino Acid Decarboxylase Deficiency

MBD5 Haploinsufficiency

Smith-Magenis Syndrome

Else, Tobias, MD; University of Michigan

Hereditary Paraganglioma-Pheochromocytoma Syndromes

POT1 Tumor Predisposition

Elsharkawi, Ibrahim, MD; Washington University

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

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Eng, Charis, MD, PhD; Cleveland Clinic

[Multiple Endocrine Neoplasia Type 2](#)

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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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[Duane Syndrome](#)

Enns, Gregory M, MB ChB; Stanford University

[Carnitine-Acylcarnitine Translocase Deficiency](#)

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

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

[Argininosuccinate Lyase Deficiency](#)

Ernst Florido, Michelle, MS; Columbia University Vagelos College of Physicians and Surgeons

[CSNK2B-Related Neurodevelopmental Disorder](#)

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](#)

Evans, D Gareth, MD; Manchester University Hospitals

[Nevoid Basal Cell Carcinoma Syndrome](#)  
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Evans, Kelly N, MD; Seattle Children's Hospital  
[Apert Syndrome](#)  
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Everett, Lorraine A; National Human Genome Research Institute  
[Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct](#)

Evidente, Virgilio Gerald H, MD; Movement Disorders Center of Arizona  
[X-Linked Dystonia-Parkinsonism Syndrome](#)

Ewbank, Clifton, MD; University of California San Francisco  
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Eymard-Pierre, Eleonore, PhD; Unité mixte de recherche 384 et Fédération de génétique humaine Auvergne  
[ALS2-Related Disorder](#)

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Fabre, Alexandre, MD, PhD; La Timone Children's Hospital  
[Trichohepatoenteric Syndrome](#)

Fahim, Abigail T, MD, PhD; University of Michigan Medical School  
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Fahrner, Jill A, MD, PhD; Johns Hopkins School of Medicine  
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Falchetti, Alberto, MD; University Hospital of Careggi  
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Falk, Marni J, MD; Children's Hospital of Philadelphia  
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Fallatah, Wedad, MD; McGill University  
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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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Fan, Yuting, MD; St Luke's Hospital

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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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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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Fathi, Esmat, PhD; St Jude Children's Research Hospital

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

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Fish, Jennifer, PhD; University of Massachusetts Lowell

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Fiskerstrand, Torunn, MD, PhD; University of Bergen

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FitzPatrick, David R, MD; University of Edinburgh

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Foss, Kimberly, MS; Seattle Children's Hospital

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Gadea, Randi, MS; Children's Mercy Hospitals and Clinics

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Gafni, Rachel I, MD; National Institute of Dental and Craniofacial Research

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Galanello, Renzo, MD; Ospedale Regionale Microcitemie

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Gallie, Brenda L, MD; Hospital for Sick Children

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

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Garber, Kathryn, PhD; Emory University School of Medicine

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

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Garcia, Christine Kim, MD, PhD; Columbia University Medical Center

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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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Garland, Emily M, PhD; Vanderbilt University Medical Center

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

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Gassner, Christoph, PhD; Swiss Red Cross

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Gear, Russell, MB ChB; Victorian Clinical Genetics Services

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Geberhiwot, Tarekegn, MD; University Hospitals

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

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Geramizadeh, Bitra, MD; Shiraz University of Medical Sciences

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German, James, MD; Weill Cornell Medical College

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

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

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

#### TNXB-Related Classical-Like Ehlers-Danlos Syndrome

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

#### Dystrophinopathies

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

#### GARS-Associated Axonal Neuropathy

Gibbs, Richard A, PhD; Baylor College of Medicine

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Gibson, K Michael, PhD; Washington State University Spokane

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

#### Cystic Fibrosis

Gibson, William Thomas, MD, PhD; BC Children's Hospital

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

#### Oculopharyngeal Muscular Dystrophy

Gil-Peña, Helena, PhD; Hospital Universitario Central de Asturias

#### Hereditary Distal Renal Tubular Acidosis

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

#### Alagille Syndrome

Giles, Rachel H, PhD; University Medical Center Utrecht

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Gillis, David, MD; Hadassah Hebrew University Hospital

#### Nonsyndromic Genetic Hyperinsulinism Overview

Gimpel, Charlotte, MD; Heidelberg University

#### Autosomal Recessive Polycystic Kidney Disease – PKHD1

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

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Giovanni, Monica A, MS; Geisinger Health System

#### Lysosomal Acid Lipase Deficiency

Girirajan, Santhosh, MBBS, PhD; Pennsylvania State University

#### 16p12.2 Recurrent Deletion

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- Girisha, Katta Mohan, MD, PhD; Manipal Academy of Higher Education  
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- Glaser, Benjamin, MD; Hadassah-Hebrew University Medical Center  
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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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- Godfrey, Maurice, PhD; University of Nebraska Medical Center  
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- Gogineni, Kishore Sai, MSc; Kasturba Medical College, Manipal  
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- 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  
[SCN3A-Related Neurodevelopmental Disorder](#)
- Goldberg, Y Paul, MBChB, PhD; Xenon Pharmaceuticals Inc



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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

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

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

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

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Goodman, Stephen I, MD; University of Colorado

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

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

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Gordon, Christopher T, PhD; Institut Imagine

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Gordon, Kristiana, MD; St George's University Hospitals  
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Gordon, Leslie B, MD, PhD; Alpert Medical School of Brown University  
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Grand, Katheryn L, MS; Children's Hospital of Philadelphia

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

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Greally, Marie T, MD, MSc; Our Lady's Children's Hospital, Crumlin

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Gupta, Rajat, MD; Birmingham Women's and Children's Hospital

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Haack, Tobias, MD; Helmholtz Zentrum & Technische Universität München

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Haas-Givler, Barbara, MEd; Elwyn Training and Research Institute

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Hackman, Peter, PhD; University of Helsinki

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Hack, Remco, MD; Leiden University Medical Center

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Hainque, Bernard, PhD; Assistance Publique - Hôpitaux de Paris

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Hakola, Panu, MD, PhD; Niuvanniemi Hospital

[Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy](#)

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Hamie, Lamiaa, MD, MSc; Mediclinic Middle East

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Hamilton, Eline, MD; VU University Medical Center

[TUBB4A-Related Leukodystrophy](#)

Hammer, Michael F, PhD; University of Arizona

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

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

[Nonketotic Hyperglycinemia](#)

Hampel, Heather, MS; The Ohio State University

[PTEN Hamartoma Tumor Syndrome](#)

Handley, Mark, PhD; University of Leeds

[RAB18 Deficiency](#)

Hanna, Michael G, BSc (Hons), MD; UCL Institute of Neurology Queen Square

[Episodic Ataxia Type 1](#)

Hannibal, Mark C, MD, PhD; University of Michigan Medical School

[IPEX Syndrome](#)

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Hannig, Vickie L, MS; Vanderbilt Medical Center

[X-Linked Spondyloepiphyseal Dysplasia Tarda](#)

Hansen-Kiss, Emily, MA, MS; UTHealth School of Dentistry at Houston

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Hansen, C David, MD; University of Utah

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Hanson, Ellen, PhD; Boston Children's Hospital

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Hanson, Nancy, MS; University of Washington

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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

[Troyer Syndrome](#)

Harms, Matthew M, MD; Washington University School of Medicine

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

Harris, Anne K, MPH; Children's Minnesota

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

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Harris, Peter C, PhD; Mayo Clinic

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Hartig, Monika, MD; Technische Universität München

[Mitochondrial Membrane Protein-Associated Neurodegeneration](#)

Hartka, Thomas R, MS; George Washington University School of Medicine

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Hartley, Taila, MSc; Children's Hospital of Eastern Ontario Research Institute

[Mandibulofacial Dysostosis with Microcephaly](#)

Hart, P Suzanne, PhD; National Institutes of Health

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

Harvengt, Pol, PhD; XLH Belgium

[X-Linked Hypophosphatemia](#)

Hasan, Sonia M, PhD; Kuwait University

[Episodic Ataxia Type 1](#)

Hasselblatt, Martin, MD; University Hospital Münster

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

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

Haviland, Isabel, MD; Boston Children's Hospital

[CDKL5 Deficiency Disorder](#)

Hayashi, Masahiro, MD, PhD; Yamagata University

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

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

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Hedera, Peter, MD, PhD; University of Louisville

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

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Hegele, Robert A, MD; Western University

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Hehr, Ute, MD; Center for Human Genetics



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Heidlebaugh, Alexis R, ScM; Geisinger Health System

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

Heiman, Meadow, MS; Indiana Hemophilia & Thrombosis Center

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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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He, Miao, PhD; University of Pennsylvania

[Epimerase Deficiency Galactosemia](#)

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

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

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Henry, Marie-Louise, MS; University of Michigan

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Heppner, Jonathan, PhD; University of British Columbia

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Herbert, Mrudu, MD, MPH; University of Kentucky

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

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

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

[ENTPD1-Related Neurodevelopmental Disorder](#)

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

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Hershberger, Ray E, MD; The Ohio State University

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Hershfield, Michael, MD; Duke University Medical Center

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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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High, Frances A, MD, PhD; MassGeneral Hospital for Children

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Hildebrand, Michael S, PhD; University of Melbourne

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Hildebrandt, Friedhelm, MD; Boston Children's Hospital

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[Apert Syndrome](#)

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[PAX6-Related Aniridia](#)

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[MELAS](#)

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Hirano, Ryuki, MD, PhD; Kagoshima University Graduate School of Medical and Dental Sciences

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

Hirose, Shinichi, MD, PhD; Fukuoka University

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

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[FRMD7-Related Infantile Nystagmus](#)

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

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

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Hodge, Jennelle, PhD; Cedars-Sinai Medical Center

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Ho, Gladys; The Children's Hospital at Westmead

[MECP2 Disorders](#)

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

[Three M Syndrome](#)

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

[SYNGAP1-Related Intellectual Disability](#)

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[Chronic Granulomatous Disease](#)

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Holm, Ida E, DMSc, MD; Aarhus University

[CHMP2B Frontotemporal Dementia](#)

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

[Brugada Syndrome](#)

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

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Hooper, Amanda J, PhD; University of Western Australia

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

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Hopfer, Suellen, MS, PhD; Pennsylvania State University

[Primary Familial Brain Calcification](#)

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

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

[Alstrom Syndrome](#)

Hopp, Katharina, PhD; Mayo Clinic - Minnesota

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Horsthemke, Bernhard, PhD; University of Essen

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Horvath, Anelia, PhD; National Institutes of Health

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Hosoi, Yasushi, MD; Hamamatsu University School of Medicine

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Houck, Kimberly, MD; Baylor College of Medicine

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Houge, Gunnar, MD, PhD; Haukeland University Hospital

[PPP2R1A-Related Neurodevelopmental Disorder](#)

Houlden, Henry, MD, PhD; University College London

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

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

[X-Linked Agammaglobulinemia](#)

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

[Juvenile Polyposis Syndrome](#)

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

[Hyaline Fibromatosis Syndrome](#)

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

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

[GRN Frontotemporal Dementia](#)

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

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Hudgins, Louanne, MD; Stanford University

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

[Wilms Tumor Predisposition](#)

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

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

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Huizing, Marjan, PhD; National Human Genome Research Institute

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

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

Hull, Peter R, MD, PhD; Dalhousie University

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Hulton, Sally-Anne, MD; Birmingham Children's Hospital

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Hume, Stacey, PhD; University of Alberta

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Hu, Mimi I, MD; The University of Texas MD Anderson Cancer Center

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Hundley, Rachel, PhD; Boston Children's Hospital

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Hunt, David, MBBS, PhD; Princess Anne Hospital

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Hunter, Alasdair GW, MD; Children's Hospital of Eastern Ontario

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Hunter, David G, MD, PhD; Children's Hospital Boston

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Hunter, Elizabeth, BS; UTHealth School of Dentistry at Houston

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Hunter, Jessica Ezzell, PhD; Kaiser Permanente Northwest

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Hunter, JM, PhD; Nationwide Children's Hospital

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

Huntsman, David G, MD; University of British Columbia

[Diffuse Gastric and Lobular Breast Cancer Syndrome](#)

Hupertz, Vera, MD; Cleveland Clinic

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Huq, Mahbul, MD, PhD; University of British Columbia

[Huntington Disease](#)

Hurley, R Morrison, MD; British Columbia's Children's and Women's Hospital

[Primary Hyperoxaluria Type 1](#)

Hurst, Anna CE, MD, MS; University of Alabama at Birmingham

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Huryn, Laryssa A, MD; National Eye Institute

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Huston, Haley, BS; Bloodworks Northwest

[Hemophilia A](#)

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Huston, Sara, MS; Northwestern University

[Treacher Collins Syndrome](#)

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

[Alagille Syndrome](#)

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

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

[Carbonic Anhydrase VA Deficiency](#)

Iacovazzo, Donato, MD; Queen Mary University of London

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Idkowiak, Jan, MD, PhD; University of Birmingham

[Cytochrome P450 Oxidoreductase Deficiency](#)

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

[Lynch Syndrome](#)

Iffland, Philip H, BA, MA, PhD; University of Maryland School of Medicine

[KPTN-Related Disorder](#)

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

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

### Hereditary Transthyretin Amyloidosis

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

Spinocerebellar Ataxia Type 8

Indo, Yasuhiro, MD, PhD; Kumamoto University Hospital

NTRK1 Congenital Insensitivity to Pain with Anhidrosis

Infante, Elena, MS; University of Pittsburgh

MUTYH Polyposis

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

Au-Kline Syndrome

SHORT Syndrome

THOC6 Intellectual Disability Syndrome

TRPM3-Related Neurodevelopmental Disorder

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

Hand-Foot-Genital Syndrome

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

Alkaptonuria

Chediak-Higashi Syndrome

Hermansky-Pudlak Syndrome

Irons, Mira, MD; Children's Hospital Boston

Smith-Lemli-Opitz Syndrome

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

Three M Syndrome

Isaacs, Adrian M, DPhil; University College London

CHMP2B Frontotemporal Dementia

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

Familial Hypercholesterolemia

Ito, Daisuke, MD, PhD; Keio University

BSCL2-Related Neurologic Disorders / Seipinopathy

Iwasa, Yoichiro, MD, PhD; University of Iowa

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## J

Jabs, Ethylin Wang, MD; Mayo Clinic

ESCO2 Spectrum Disorder

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- Jackson, Andrew, PhD; University of Edinburgh  
Microcephalic Osteodysplastic Primordial Dwarfism Type II  
RNU4atac-opathy
- Jackson, Michelle A, MS; University of Texas MD Anderson Cancer Center  
CDC73-Related Disorders
- Jacob, Prince, MSc, PhD; JSS Academy of Higher Education and Research  
HOXA1-Related Disorders
- James, Aaron; University of California San Francisco  
Simpson-Golabi-Behmel Syndrome Type 1
- James, Paula, MD; Queen's University  
Von Willebrand Disease
- Jankovic, Joseph, MD; Baylor College of Medicine  
NKX2-1-Related Disorders
- Jansen, Anna C, MD, PhD; Universitair Ziekenhuis Brussel  
PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia  
Progressive Myoclonus Epilepsy, Lafora Type
- Janssens, Veerle, PhD; KU Leuven  
PPP2R1A-Related Neurodevelopmental Disorder
- Jaspersen, Kory W; University of Utah  
APC-Associated Polyposis Conditions
- Jeffery, Steve, PhD; St George's University of London  
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Milroy Disease
- Jen, Joanna C, MD, PhD; Icahn School of Medicine at Mount Sinai  
Familial Hemiplegic Migraine
- Jerves Serrano, Teodoro, MD; Yale School of Medicine  
Propionic Acidemia
- Jeste, Shafali Spurling, MD; Children's Hospital Los Angeles  
Maternal 15q Duplication Syndrome
- Jethva, Reena, MD; St Christopher's Hospital for Children  
Short-Chain Acyl-CoA Dehydrogenase Deficiency
- Jewett, Tamison, MD; Wake Forest University School of Medicine  
1q21.1 Recurrent Microdeletion
- Jinnah, Hyder A, MD, PhD; Emory University  
HPRT1 Disorders

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

[Rotor Syndrome](#)

Johannesen, Katrine, MD, PhD; Rigshospitalet

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

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Johannsen, Peter, MD, PhD; Rigshospitalet

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Johnsen, Jill, MD; University of Washington

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

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Johnson, Amanda; ARRE Foundation

[Shashi-Pena Syndrome](#)

Johnson, Eric W, PhD; Barrow Neurological Institute

[Familial Cerebral Cavernous Malformation](#)

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

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

[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

[Greig Cephalopolysyndactyly Syndrome](#)

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

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Jordan, Elizabeth, MMSc; The Ohio State University

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

[Hypohidrotic Ectodermal Dysplasia](#)

Josephson, Neil C, MD; Seattle Genetics

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

Joshi, Nishtha; Baylor College of Medicine

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

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

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Jäggle, Herbert, MD; University of Regensburg

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## K

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

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

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Kagami, Masayo, MD, PhD; National Research Institute for Child Health and Development

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

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

[GBE1 Adult Polyglucosan Body Disease](#)

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

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

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

[ATP7A-Related Copper Transport Disorders](#)

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

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

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

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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

[ARSACS](#)

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Kang, Jingqiong, PhD; Vanderbilt University Medical Center

[SLC6A1-Related Neurodevelopmental Disorder](#)

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[Cherubism](#)

Kantarci, Sibel, PhD; Quest Diagnostics Nichols Institute

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

[Periodontal Ehlers-Danlos Syndrome](#)

Kaplan, Julie D, MD; Cleveland Clinic Foundation

[MYRF-Related Cardiac Urogenital Syndrome](#)

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

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Karpati, George, MD; Montreal Neurological Institute

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Kashtan, Clifford E, MD; University of Minnesota

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Kaspar, Roger L, PhD; Transderm, Inc

[Pachyonychia Congenita](#)

Kastner, Dan, MD, PhD; National Human Genome Research Institute

[ALPK1-Related Autoinflammatory Disease](#)

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

[Diffuse Gastric and Lobular Breast Cancer Syndrome](#)

Kaur, Parneet, MSc; Manipal Academy of Higher Education

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

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

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Kaylor, Julie, MS; Arkansas Children's Hospital

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[Usher Syndrome Type I](#)

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

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

[OTOF-Related Deafness](#)

Kelly, Edward J, PhD; University of Washington

[Bietti Crystalline Dystrophy](#)

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

[Hypochondroplasia](#)

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

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

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[STXBP1 Encephalopathy with Epilepsy](#)

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

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

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

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

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

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

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

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

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

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Kirk, Jessica, BS; Yale University School of Medicine

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Kirmani, Salman, MBBS; Aga Khan University

[Hereditary Paraganglioma-Pheochromocytoma Syndromes](#)

Kirschner, Lawrence, MD, PhD; The Ohio State University Comprehensive Cancer Center

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Kishnani, Priya S, MD; Duke University Medical Center

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Leach, Meganne E, MSN, PNP; Oregon Health and Science University

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Lee, Kristen N, MD; University of Michigan

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Lee, Mianne, MSc; Univeristy of Hong Kong

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Lehman, April N, MD; The Ohio State University College of Medicine

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#### 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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Lemmink, Henny, PhD; University Medical Center

#### Hereditary Nephrogenic Diabetes Insipidus

Lentz, Jennifer J, PhD; Louisiana State University Health Sciences Center

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Leonard, Laura D, BA; Children's Hospital of Philadelphia

#### Alagille Syndrome

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

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Leslie, Elizabeth J, PhD; Emory 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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Letra, Ariadne, DDS, MS, PhD; UTHealth School of Dentistry at Houston

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MUTYH Polyposis

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Mahtani, Karishma, BSc; Atwal Clinic: Genomic & Personalized Medicine

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Mak, Christopher CY, MB ChB, PhD; University of Hong Kong

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Malfait, Fransiska, MD, PhD; Ghent University Hospital

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Malicdan, May Christine V, MD, PhD; National Human Genome Research Institute

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Malm, Dag, MD, PhD; Tromsø Center of Internal Medicine

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Mancini, Cecilia, PhD; University of Turin

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Mandell, Roseann; Massachusetts General Hospital

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Mao, Rong, MD; University of Utah

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Marazita, Mary L, PhD; University of Pittsburgh

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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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Marcogliese, Paul C, PhD; University of Manitoba

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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

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Markovitz, Rebecca, MD, PhD; Baylor College of Medicine

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Mark, Paul, MD; Michigan State University

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Marquard, Jessica, MS; Cleveland Clinic

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Marquardt, Thorsten, Dr. med.; Universitätsklinikum Münster

#### SLC39A8-CDG

Marras, Connie, MD, PhD; University of Toronto

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Marshall, Susan G, MD; University of Washington School of Medicine

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Marsh, Rebecca A, MD; Cincinnati Children's Hospital

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Martin, Christa L, PhD; Geisinger Health System



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Martin, George M, MD; University of Washington

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Martin, Jack W, MD; The University of Texas MD Anderson Cancer Center

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Martin, Rick A, MD; Saint Louis University

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C3 Glomerulopathy

Martín, Miguel A, PhD; Hospital 12 de Octubre Research Institute

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Matalon, Reuben, MD, PhD; University of Texas Medical Branch

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Mathieu, Jean, MD; Complexe Hospitalier de la Sagamie

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Moeschler, John, MD, MS; Geisel School of Medicine/Dartmouth College

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Moreau, Lisa, MS; Dana Farber Cancer Institute

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Moreira Marques, Juliana, PhD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo

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Moreira, Maria-Ceu, MSc, PhD; Professor of Molecular Biology and Genetics

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Morizono, Hiroki, PhD; Children's National Hospital

Ornithine Transcarbamylase Deficiency

Morleo, Manuela, PhD; Federico II University

Microphthalmia with Linear Skin Defects Syndrome

Mornet, Etienne, PhD; Centre Hospitalier de Versailles

Hypophosphatasia

Morotti, Raffaella, MD; Yale School of Medicine

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Morris, Colleen A, MD; University of Nevada, Reno School of Medicine

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Williams Syndrome

Morrison, Leslie, MD; University of New Mexico

Familial Cerebral Cavernous Malformation

Morrow, Eric M, MD, PhD; Brown University

Christianson Syndrome

Mortier, Geert R, MD, PhD; University Hospitals Leuven

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Stickler Syndrome

X-Linked Hypophosphatemia

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Morton, D Holmes, MD; Clinic for Special Children

Maple Syrup Urine Disease

Moser, Ann B, BA; Kennedy Krieger Institute

Rhizomelic Chondrodysplasia Punctata Type 1

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Mucha, Bettina E, MD; Sainte-Justine Hospital  
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Nielsen, Maartje, MD, PhD; Leiden University Medical Center

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Nielsen, Troels T, MSc, PhD; Rigshospitalet

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Niewisch, Marena R, MD; National Cancer Institute, NIH

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Niinikoski, Harri, MD, PhD; University of Turku

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Nillesen, Willy M, BSc; Radboud University Medical Center

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Nimkarn, Saroj, MD; Bumrungrad Hospital

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Nisbet, Alex F, BS; Children's Hospital of Philadelphia

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Nishi, Mirian Yumie, PhD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo

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Nobile, Carlo; CNR Institute of Neuroscience

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Noon, Sarah Elizabeth, MS; The Children's Hospital of Philadelphia

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Ocansey, Sharon, BSc, MBBS, MSc; St George's University Hospital NHS Foundation Trust

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Oddoux, Carole, PhD; New York University School of Medicine

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Odell, Amanda M, MS; University of Iowa Hospitals and Clinics

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Oetgen, Matthew, MD; Children's National Medical Center

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Ogata, Tsutomu, MD, PhD; Hamamatsu University School of Medicine

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Oglesbee, Devin, PhD; Mayo Clinic

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Oishi, Kimihiko, MD; Jikei University School of Medicine

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Okialda, Krystle A, BS; University of Washington

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Okur, Volkan, MD; New York Genome Center

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Oliveira, Carla, PhD; University of Porto

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Oliveira, Joao, MD, PhD; Federal University of Pernambuco

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Oliveira, Jorge, MSc, PhD; Universidade do Porto

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Oliver Petit, Isabelle, MD; Children's Hospital

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Olivucci, Giulia, MD; IRCCS Azienda Ospedaliero-Universitaria di Bologna

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Olney, Ann Haskins, MD; University of Nebraska Medical Center

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Olson, Heather, MD, MS; Boston Children's Hospital

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Olson, Timothy S, MD, PhD; Children's Hospital of Philadelphia

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Ong, Thida, MD; University of Washington School of Medicine

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Onodera, Osamu, MD, PhD; Niigata University

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

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

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Origa, Raffaella, MD; Ospedale Pediatrico Microcitemico Antonio Cao

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

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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

[SPTBN4 Disorder](#)

Osborne, Jenae, MS; University of Michigan

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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, Pia; St George's, University of London

[Milroy Disease](#)

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

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

[Tatton-Brown-Rahman Syndrome](#)

Oswald, Gretchen L, MS; Johns Hopkins Medical Center

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

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Ottman, Ruth, PhD; Columbia University

[Autosomal Dominant Epilepsy with Auditory Features](#)

Ozelius, Laurie, PhD; Massachusetts General Hospital

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Padiath, Quasar, MBBS, PhD; University of Pittsburgh  
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Pagon, Roberta A, MD; University of Washington  
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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  
[Isolated Gonadotropin-Releasing Hormone \(GnRH\) Deficiency](#)

Palmer, Elizabeth, MBBS, PhD; University of New South Wales  
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Palmio, Johanna, MD, PhD; University of Tampere  
[ANO5 Muscle Disease](#)

Paloneva, Juha, MD, PhD; Central Finland Hospital Nova  
[Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy](#)

Palsson, Runolfur, MD; The National University Hospital of Iceland  
[Adenine Phosphoribosyltransferase Deficiency](#)

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

Pandya, Arti, MBA, MD; Virginia Commonwealth University Health System  
[Nonsyndromic Hearing Loss and Deafness, Mitochondrial](#)

Pankratz, Nathan D, PhD; Indiana University School of Medicine  
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Papanikalaou, George, MD, PhD; National and Kapodistrian University of Athens  
[Juvenile Hemochromatosis](#)

Papapoulos, Socrates, MD, PhD; Leiden University Medical Center

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Pappas, John G, MD, MS; NYU Grossman School of Medicine

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

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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

[Familial Hypercholesterolemia](#)

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

[HFE-Related Hemochromatosis](#)

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

[ALK-Related Neuroblastic Tumor Susceptibility](#)

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

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Parodi, Livia, PhD; Sorbonne Université – Pitié-Salpêtrière University Hospital

[Spastic Paraplegia 4](#)

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

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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](#)

Pastori, Daniele, MD, PhD; Sapienza University of Rome

[Factor V Leiden Thrombophilia](#)

Patel, Heema, PhD; St George's, University of London

[Troyer Syndrome](#)

Patel, Millan S, MD, MSc; University of British Columbia

[Isolated and Classic Cutis Marmorata Telangiectatica Congenita](#)

Patel, Neepa Jayant, MD; Henry Ford Health System

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

[Friedreich Ataxia](#)

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

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Patil, Siddaramappa J, MD; Mazumdar Shaw Medical Centre / Narayana Hrudayalaya Hospitals

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Patterson, Marc, MD; Mayo Clinic

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Patwari, Pallavi P, MD; Children's Memorial Hospital

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

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

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Pearl, Phillip L, MD; George Washington University School of Medicine

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Pearson, Deborah A, PhD; McGovern Medical School

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Pearson, Toni S, MBBS, MD; Nationwide Children's Hospital

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

[MYH9-Related Disease](#)

Peikert, Kevin, MD; University Medical Center Rostock

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Pellerin, David, MD, MSc; University College London

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

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Pena, Izabella, PhD; Massachusetts Institute of Technology

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Pena, Loren DM, MD, PhD; Cincinnati Children's Hospital and Medical Center

[IRF2BPL-Related Disorder](#)

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Pencheva, Bojana, MMSc; Emory University School of Medicine

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Pendyal, Surekha, MEd, MSc, RD; Duke University Medical Center

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

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Penttilä, Sini, PhD; Fimlab Laboratories

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Pepin, Melanie G, MS; University of Washington Medical Center

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Percy, Melanie, PhD; Belfast City Hospital

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Perez Botero, Juliana, MD; BloodCenter of Wisconsin

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Perlman, Susan, MD; David Geffen School of Medicine at UCLA

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

[BCL11A-Related Intellectual Disability](#)

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Perrier, Nancy D, MD; University of Texas MD Anderson Cancer Center  
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Perry, Monique, MD; Clinical Center - National Institutes of Health  
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Pescosolido, Matthew F, BA; Brown University  
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Petrucelli, Nancie, MS; Karmanos Cancer Institute, Cancer Genetic Counseling Service  
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Pierre, Germaine, MBBS, MSc; Bristol Royal Hospital for Children

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

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Plitt, Gilman, MD; Cleveland Clinic

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Prabhu, Sandeep, MD; University of Oklahoma

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Prada, Carlos, MD; Ann & Robert H Lurie Children's Hospital of Chicago

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Prades, Silvia, PhD; Ataxia UK

[DRPLA](#)

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

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

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Prasov, Lev, MD, PhD; University of Michigan Medical School

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Prasun, Pankaj, MD; Icahn School of Medicine at Mount Sinai

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Prokisch, Holger, PhD; Technische Universität München

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Pronk, JC, PhD; Vrije Universiteit Medical Center

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Proudlock, Frank A, BSc, MSc, PhD; University of Leicester

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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

Maple Syrup Urine Disease

Pulst, Stefan M, MD; University of Utah

Spinocerebellar Ataxia Type 13

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21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

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Spinocerebellar Ataxia Type 11

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Hereditary Hemorrhagic Telangiectasia

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

MYRF-Related Cardiac Urogenital Syndrome

Périé, Sophie, MD, PhD; Université Pierre et Marie Curie

Oculopharyngeal Muscular Dystrophy

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

LAMA2 Muscular Dystrophy

Quinonez, Shane C, MD; University of Michigan

Citrullinemia Type I

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

NFIA-Related Disorder

Wiedemann-Steiner Syndrome

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

Spinocerebellar Ataxia Type 36

Quiroz, Vincente, MD; Harvard Medical School

PRRT2-Related Disorder

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

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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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Ramos, Eliana Marisa, PhD; University of California Los Angeles

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Rampoldi, Luca, PhD; Ospedale San Raffaele

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Rana, Huma Q, MD; Dana-Farber Cancer Institute

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Rand, Casey M, BS; Ann & Robert H Lurie Children's Hospital of Chicago

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Saethre-Chotzen Syndrome

Rauen, Katherine A, MD, PhD; University of California, Davis  
Cardiofaciocutaneous Syndrome  
HRAS-Related Costello Syndrome

Rawlings, David J, MD; University of Washington  
X-Linked Severe Combined Immunodeficiency

Rawlins, Lettie E, BSc, MBChB, PhD; University of Exeter Medical School  
KPTN-Related Disorder

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TANGO2 Deficiency

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

Raymond, Deborah, MS; Icahn School of Medicine at Mount Sinai / Mount Sinai Beth Israel  
DYT1 Early-Onset Isolated Dystonia  
LRRK2 Parkinson Disease  
SGCE Myoclonus-Dystonia

Raymond, F Lucy, MD, PhD; University of Cambridge  
NSDHL-Related Disorders

Raymond, Gerald V, MD; Johns Hopkins Hospital  
X-Linked Adrenoleukodystrophy  
Zellweger Spectrum Disorder

Ray, Peter N, PhD; The Hospital for Sick Children  
Cherubism

Rea, Hannah, PhD; University of Washington  
CHD8-Related Neurodevelopmental Disorder with Overgrowth

Redfield, Shelby, MS; Boston Children's Hospital  
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Reding, Madeline Q, MPH, MS; Mayo Clinic  
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Redman, Colvin M, PhD; The New York Blood Center

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Reehal, Tom; Sheffield University School of Medicine & Biomedical Sciences

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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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Reichenberger, Ernst, PhD; UConnHealth

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

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Remuzzi, Giuseppe, MD; Istituto di Ricerche Farmacologiche Mario Negri-IRCCS

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Renaud, Mathilde, MD, PhD; CHRU de Nancy

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Rendtorff, Nanna Dahl, PhD; University Hospital / Kennedy Center

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Richard, Gabriele, MD; GeneDx, Inc

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Richmond, Christopher Mark, MBBS; Royal Brisbane & Women's Hospital

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Rich, Thereasa A, MS; University of Texas MD Anderson Cancer Center

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

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Sahota, Amrik, PhD; Rutgers University

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Sajorda, Brian J, BA; Children's Hospital of Philadelphia

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Saunders, Carol J, PhD; University of Missouri

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Sauvé, Yves, PhD; University of Alberta

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Savant, Adrienne, MD, MS; Tulane University School of Medicine

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Savarese, Marco, PhD; University of Helsinki

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Savarirayan, Ravi, MBBS, MD; University of Melbourne

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Scaglia, Fernando, MD; Baylor College of Medicine

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Scarpa, Maurizio, MD, PhD; Horst Schmidt Klinik  
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Schaaf, Christian P, MD, PhD; Heidelberg University  
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Schaefer, Franz, MD; Heidelberg University  
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Schanen, N Carolyn, MD, PhD; Dup15q Alliance  
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Scharenberg, Andrew M, MD; University of Washington  
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Scharer, Gunter, MD, PhD; Denver Genetic Laboratories, University of Colorado  
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Scheffer, Ingrid E, MBBS, PhD; University of Melbourne  
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Schiffmann, Raphael, MD, MHSc; Baylor Scott & White Research Institute

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Schlechter, Catherine, MS; Casey Eye Institute - Oregon Health Sciences University

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Schrier Vergano, Samantha A, MD; Children's Hospital of the King's Daughters

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Schroer, Richard, MD; Greenwood Genetic Center

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Schultz, Kris Ann P, MD; Children's Minnesota

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Schulz, Ansgar, MD; Universitätsklinik für Kinder- und Jugendmedizin

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Schwarz, Guenter; Cologne University

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Sims, Katherine B, MD; Massachusetts General Hospital / Harvard Medical School

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Singal, Ashwani K, MD, MSc; University of South Dakota Sanford School of Medicine

[Variegate Porphyria](#)

Singer, Kaitlyn, MS; Geisinger Health System

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Singh, Gautam K, MD; Central Michigan University School of Medicine

[Cantú syndrome](#)

Singh, Rani H, PhD, RD; Emory University School of Medicine

[Duarte Variant Galactosemia](#)

Singh, Swati, MSc; Manipal Academy of Higher Education

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

Sinnreich, Michael, MD, PhD; Montreal Neurological Institute

[GNE Myopathy](#)

Siper, Paige, PhD; Icahn School of Medicine at Mount Sinai

[FOXP1 Syndrome](#)

Sisodiya, Sanjay, MD, PhD; UCL Queen Square Institute of Neurology

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Sivakumar, Kumaraswamy, MD; St Joseph's Hospital and Medical Center

[GARS-Associated Axonal Neuropathy](#)

Skefos, Catherine M, MA, MS; The University of Texas MD Anderson Cancer Center

[CDC73-Related Disorders](#)

Skinner, Jonathan Robert, MB ChB, MD; University of Auckland

[GNB5-Related Neurodevelopmental Disorder](#)

Skraban, Cara M, MD; Children's Hospital of Philadelphia

[WDR26-Related Intellectual Disability](#)

Slattery, Susan M, MD, MS; Northwestern University

[Congenital Central Hypoventilation Syndrome](#)

Slaugenhaupt, Susan A, PhD; MGH Center for Human Genetic Research / Harvard Medical School

[Mucopolidosis IV](#)

Slavotinek, Anne M, MBBS, PhD; University of California, San Francisco

[FREM1 Autosomal Recessive Disorders](#)

[Fryns Syndrome](#)

[McKusick-Kaufman Syndrome](#)

Sleasman, John W, MD; Duke University Medical Center

[IPEX Syndrome](#)

Sloan, Jennifer L, MS, PhD; National Human Genome Research Institute

[Disorders of Intracellular Cobalamin Metabolism](#)

[Isolated Methylmalonic Acidemia](#)

Sloman, Melissa, BSc; Royal Devon University Healthcare NHS Foundation Trust

[Spondylocostal Dysostosis, Autosomal Recessive](#)

Smaoui, Nizar, MD; GeneDx, Inc

[Choroideremia](#)

[X-Linked Congenital Retinoschisis](#)

Smith-Simmer, Kelcy, MMSc; University of Wisconsin Carbone Cancer Center

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

Smith, Adam C, MS; University of Toronto

[Beckwith-Wiedemann Syndrome](#)

Smith, Ann CM, DSc (Hon), MA; National Institutes of Health

[Smith-Magenis Syndrome](#)

Smith, CI Edvard, MD, PhD; Karolinska Institutet

[X-Linked Agammaglobulinemia](#)

Smith, Edward R, MD; Boston Children's Hospital

[Familial Cerebral Cavernous Malformation](#)

Smith, Frances JD, PhD; University of Dundee

[Pachyonychia Congenita](#)

Smith, Janine, MD; National Institutes of Health

[Mucopolipidosis IV](#)

Smith, Rebecca, MS; Geisinger Health System

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Smith, Richard JH, MD; University of Iowa Hospitals and Clinics

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Smith, Simone, MSc; Geisinger Health System

[PACS1 Neurodevelopmental Disorder](#)

Smith, Stephanie C, MS; University of Mississippi Medical Center

[Spinocerebellar Ataxia Type 3](#)

Sniderman King, Lisa, MSc; Genzyme Corporation

[Tyrosinemia Type I](#)

Snijders Blok, Lot, MD; Radboud University Medical Center

[DDX3X-Related Neurodevelopmental Disorder](#)

Snow-Bailey, Karen, PhD; Auckland City Hospital

[Multiple Endocrine Neoplasia Type 2](#)

Snyder, Cara L, MS; CLS Genetic Consulting, LLC

[Celiac Disease](#)

Soares, Ana Rita, MD; Santo Antonio University Hospital Center

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Sobacchi, Cristina, MS; Humanitas Clinical and Research Center

[CLCN7-Related Osteopetrosis](#)

Sobeih, Magdi M, MD, PhD; Boston Children's Hospital

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Sobrido, Maria J, MD, PhD; Fundación Pública Galega de Medicina Xenómica-SERGAS

[Primary Familial Brain Calcification](#)

[Spinocerebellar Ataxia Type 36](#)

So, Jodi Y, BA; Stanford School of Medicine

[Epidermolysis Bullosa Simplex](#)

Soldovieri, Maria Virginia, PhD; University of Molise

[KCNQ2-Related Disorders](#)

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Soler-Alfonso, Claudia, MD; Baylor College of Medicine

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Solomon, Benjamin D, MD; National Institutes of Health

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Solomon, Beth, MS; National Institutes of Health

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Solomon, Cindy, MS; Myriad Genetic Laboratories, Inc

[APC-Associated Polyposis Conditions](#)

Solomon, Sheila, MS; GeneDx, Inc

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Song, Yuan-Zong, MD, PhD; Jinan University

[Citrin Deficiency](#)

Sotero de Menezes, Marcio A, MD; Swedish Neuroscience Institute

[SCN1A-Related Seizure Disorders](#)

Spacey, Sian, MD; University of British Columbia

[Familial Paroxysmal Nonkinesigenic Dyskinesia](#)

Sparks, Susan E, MD, PhD; Sanofi Genzyme

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Spence, Sarah J, MD, PhD; Children's Hospital Boston

[Maternal 15q Duplication Syndrome](#)

Sperandeo, Maria Pia, PhD; Federico II University

[Lysinuric Protein Intolerance](#)

Spiegel, Ronen, MD; Emek Medical Center

[Mucopolidosis III Gamma](#)

Spillmann, Rebecca C, MS; Duke University

[Shashi-Pena Syndrome](#)

Spinner, Nancy B, PhD; Children's Hospital of Philadelphia

[Alagille Syndrome](#)

Splawski, Igor, PhD; Assistant, Children's Hospital Boston

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Sprecher, Eli, MD, PhD; Tel Aviv Medical Center

[Pachyonychia Congenita](#)

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

Srivastava, Siddharth, MD; Boston Children's Hospital

[Alexander Disease](#)

[SETBP1 Haploinsufficiency Disorder](#)



Stanich, Peter P, MD; The Ohio State University Wexner Medical Center

[APC-Associated Polyposis Conditions](#)

Stanley, Charles A, MD; University of Pennsylvania

[Permanent Neonatal Diabetes Mellitus](#)

Starr, Lois J, MD, PhD; University of Nebraska Medical Center

[Myhre Syndrome](#)

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Statland, Jeffrey M, MD; University of Kansas Medical Center

[Andersen-Tawil Syndrome](#)

Staudt, Martin, MD; LMU Munich

[FOXG1 Syndrome](#)

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[Alstrom Syndrome](#)

Steeves, Marcie, MS; MassGeneral Hospital for Children

[Pitt-Hopkins Syndrome](#)

Steinberg, Steven J, PhD; ARUP Laboratories

[Rhizomelic Chondrodysplasia Punctata Type 1](#)

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Steiner, Robert D, MD; University of Wisconsin School of Medicine and Public Health

[COL1A1/2 Osteogenesis Imperfecta](#)

[Sitosterolemia](#)

Stein, Jennifer L, MS; Cleveland Clinic Genomic Medicine Institute

[PTEN Hamartoma Tumor Syndrome](#)

Steinlein, Ortrud K, MD, PhD; Ludwig Maximilian University of Munich

[Birt-Hogg-Dube Syndrome](#)

Steinman, Kyle J, MD; Seattle Children's Hospital

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Steinmann, Beat, MD; University Children's Hospital

[PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome](#)

Stephens, Karen, PhD; University of Washington Medical Center

[Epidermolysis Bullosa Simplex](#)

Stephenson, John BP; Royal Hospital for Sick Children

[Aicardi-Goutieres Syndrome](#)

Stepien, Karolina M, MD, PhD; Salford Royal NHS Foundation Trust

[Alpha-Mannosidosis](#)

Sternberg, Damien, MD, PhD; Assistance Publique - Hôpitaux de Paris

[Hypokalemic Periodic Paralysis](#)

Sternen, Darci L, MS; Children's Hospital and Regional Medical Center

[Cystic Fibrosis](#)

Stevanin, Giovanni, PhD; Hôpital de la Pitié-Salpêtrière

[Spastic Paraplegia 11](#)

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

[Rubinstein-Taybi Syndrome](#)

Stevenson, David A, MD; Stanford University

[Capillary Malformation-Arteriovenous Malformation Syndrome](#)[Hereditary Hemorrhagic Telangiectasia](#)[Legius Syndrome](#)

Stevenson, Roger E, MD; Greenwood Genetic Center

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

Stewart, Blythe; University of Edinburgh

[MYRF-Related Cardiac Urogenital Syndrome](#)

Stewart, Douglas R, MD; National Cancer Institute

[DICER1 Tumor Predisposition](#)

Stojinski, Carol, MD, MS; Drexel University College of Medicine

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

Stojkovic, Tanya, MD; Institut de Myologie

[Oculopharyngeal Muscular Dystrophy](#)

Stokman, Marijn, MD, PhD; Radboud University Medical Centre

[Nephronophthisis-Related Ciliopathies](#)

Stolle, Catherine A, PhD; Children's Hospital of Philadelphia

[Von Hippel-Lindau Syndrome](#)

Stoller, James K, MD, MS; Cleveland Clinic

[Alpha-1 Antitrypsin Deficiency](#)

Stone, Jeremy T, BSc; Mayo Clinic

[LRRK2 Parkinson Disease](#)

Storey, Elsdon, DPhil; Monash University

[Spinocerebellar Ataxia Type 20](#)

Stover, Niamh B, MS; Banner Health

[Bietti Crystalline Dystrophy](#)

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

[Carney Complex](#)

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

[DICER1 Tumor Predisposition](#)

Strauss, Arnold W, MD; Cincinnati Children's Hospital Medical Center

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

Strauss, Kevin A, MD; Clinic for Special Children

[Maple Syrup Urine Disease](#)

Strehlow, Vincent, MD; University of Leipzig Medical Center

[GRIN2A-Related Disorders](#)

Strober, Jonathan, MD; UCSF / Benioff Children's Hospital

[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

Stuart, Helen M, MD; University of Manchester and Central Manchester University Hospitals NHS Foundation Trust

[Urofacial Syndrome](#)

Stumpel, Connie, MD, PhD; Academic Hospital Maastricht

[L1 Syndrome](#)

Stöckler-Ipsiroglu, Sylvia, MBA, MD, PhD; British Columbia Children's Hospital

[Creatine Deficiency Disorders](#)

Subramony, SH, MD; University of Florida

[Spinocerebellar Ataxia Type 1](#)[Spinocerebellar Ataxia Type 3](#)[Spinocerebellar Ataxia Type 8](#)

Suchy, Sharon F, PhD; GeneDx, Inc

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

Suit, Lindsey; University of California, San Francisco

[DDX3X-Related Neurodevelopmental Disorder](#)

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

[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

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

[X-Linked Lymphoproliferative Disease](#)

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

[Urea Cycle Disorders Overview](#)

Sumner, Charlotte J, MD; Johns Hopkins University School of Medicine

### Autosomal Dominant TRPV4 Disorders

Sun, Angela, MD; Seattle Children's Hospital

Arginase Deficiency

Mucopolysaccharidosis Type VII

Sundal, Christina, MD, PhD; Gothenburg University

CSF1R-Related Disorder

Suominen, Tiina, MSc; University of Tampere

Salih Myopathy

Udd Distal Myopathy - Tibial Muscular Dystrophy

Superti-Furga, Andrea, MD; University of Lausanne

Achondrogenesis Type 1B

Atelosteogenesis Type 2

Campomelic Dysplasia

Chondrodysplasia with Congenital Joint Dislocations, CHST3-Related

Diastrophic Dysplasia

SLC26A2-Related Multiple Epiphyseal Dysplasia

Sutton, V Reid, MD; Baylor College of Medicine

Aicardi Syndrome

PORCN-Related Developmental Disorders

TP63-Related Disorders

White-Sutton Syndrome

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

Alkaptonuria

Suzuki, Tamio, MD, PhD; Yamagata University

Oculocutaneous Albinism Type 4

Sveden, Abigail, MS; Boston Children's Hospital

ANKRD17-Related Neurodevelopmental Syndrome

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

Nonketotic Hyperglycinemia

Swadner, Kathleen J, PhD; Massachusetts General Hospital

ATP1A3-Related Neurologic Disorders

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

Nail-Patella Syndrome

Sweeney, William E, MS; Medical College and Children's Health System of Wisconsin

Autosomal Recessive Polycystic Kidney Disease – PKHD1

Sweetser, David A, MD, PhD; Massachusetts General Hospital

[GNAO1-Related Disorder](#)

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Swoboda, Kathryn J, MD; Massachusetts General Hospital

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[Tyrosine Hydroxylase Deficiency](#)

Sybert, Virginia P, MD; University of Washington

[Epidermolysis Bullosa Simplex](#)

Symoens, Sofie, PhD; Ghent University Hospital

[Classic Ehlers-Danlos Syndrome](#)

Synofzik, Matthis, MD; Center for Neurology & Hertie Institute for Clinical Brain Research

[ARSACS](#)

[GAA-FGF14-Related Ataxia](#)

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Syx, Delfien, PhD; Ghent University Hospital

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## T

Tabarki, Brahim, MD; Prince Sultan Military Medical City

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

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Tabti, Nacira, MD, PhD; Assistance Publique - Hôpitaux de Paris

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

[KCNQ2-Related Disorders](#)

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Tait, Jonathan F, MD, PhD; University of Washington School of Medicine

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Takahashi, Hitoshi, MD, PhD; Niigata University

[Spinocerebellar Ataxia Type 17](#)

Takahashi, Toshiaki, MD, PhD; National Hospital Organization – Sendai-Nishitaga Hospital

[Dysferlinopathy](#)

Takasaki, Kaoru, MD; Children's Hospital of Philadelphia

[GATA1-Related Cytopenia](#)

Takashima, Hiroshi, MD, PhD; Kagoshima University Graduate School of Medical and Dental Sciences

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

Talbert, Janet L, MS; Vanderbilt University Medical Center

[Pulmonary Fibrosis Predisposition Overview](#)

Tallaksen, Chantal; Hôpital de la Pitié-Salpêtrière

[Spastic Paraplegia 4](#)

Taly, Arun B, MD; National Institute of Mental Health & Neurosciences

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Tamary, Hannah, MD; Schneider Children's Medical Center of Israel

[Alpha-Thalassemia](#)

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Tamburro, Joan, DO; Cleveland Clinic

[Isolated and Classic Cutis Marmorata Telangiectatica Congenita](#)

Tamura, Deborah, RN; National Cancer Institute

[Xeroderma Pigmentosum](#)

Tanase-Nakao, Kanako, MD, PhD; National Research Institute for Child Health and Development

[MIRAGE Syndrome](#)

Tan, Christopher, MS; University of Chicago

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

Taniguchi, Toshiyasu, MD, PhD; Fred Hutchinson Cancer Research Center

[Fanconi Anemia](#)

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

[Mucopolysaccharidosis Type IVA](#)

Tan, Queenie K-G, MD, PhD; Mayo Clinic

[IPEX Syndrome](#)

Tan, Weizhen, MD; MassGeneral Hospital for Children

[Cranioectodermal Dysplasia](#)

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

[Bohring-Opitz Syndrome](#)

Tarleton, Jack C, PhD; Fullerton Genetics Center

[FMR1 Disorders](#)

Tarrant, Teresa, MD; Duke University Medical Center

[Adenosine Deaminase Deficiency](#)

Tartaglia, Marco, PhD; Bambino Gesù Children's Hospital IRCSS

[Noonan Syndrome with Multiple Lentigines](#)

Tatton-Brown, Katrina, BM BCh, MD; St George's University of London

[EZH2-Related Overgrowth](#)

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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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[Faciocapulohumeral Muscular Dystrophy](#)

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

[Celiac Disease](#)

Taylor, Cora M, PhD; Geisinger Health System

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

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Taylor, Kelly A, MS; Vanderbilt Medical Center

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

[Danon Disease](#)

Taylor, Robert W, PhD; Newcastle University

[RRM2B Mitochondrial DNA Maintenance Defects](#)

Taylor, Rohan, BSc (Hons); St George's Healthcare NHS Trust

[ROR2-Related Robinow Syndrome](#)

Tchan, Michel, BMedSci, MBBS, PhD; Westmead Hospital

[Aspartylglucosaminuria](#)

Teebi, Ahmad, MD; Education City

[Cherubism](#)

Tekendo-Ngongang, Cedrik, MD; National Human Genome Research Institute

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Tekin, Mustafa, MD; John P Hussman Institute for Human Genomics

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

[Deafness and Myopia Syndrome](#)

[KBG Syndrome](#)

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

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

Teng, Joyce, MD, PhD; Stanford School of Medicine

[Epidermolysis Bullosa Simplex](#)

Terry, Sharon F, MA; PXE International

[Pseudoxanthoma Elasticum](#)

Terwindt, Gisela M, MD, PhD; Leiden University Medical Center

[CADASIL](#)

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

Testa, Giuseppe, MA, MD, PhD; European Institute of Oncology

[Gabriele-de Vries Syndrome](#)

Thabet, Farah, MD; Fattouma Bourguiba University Hospital

[SLC25A19-Related Thiamine Metabolism Dysfunction](#)

Thauvin-Robinet, Christel, MD, PhD; University of Burgundy

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

Thibert, Ronald L, DO, MSPH; Massachusetts General Hospital

[Maternal 15q Duplication Syndrome](#)

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Thiel, Christian T; Friedrich-Alexander Universität Erlangen-Nürnberg

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

Thiel, Moritz, MD; University of Cologne

[GNAO1-Related Disorder](#)

Thoene, Jess G, MD; University of Michigan

[Citrullinemia Type I](#)

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

[DDX3X-Related Neurodevelopmental Disorder](#)

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

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

[Hemophilia A](#)



[Hemophilia B](#)

Thompson, Reid, MD; Johns Hopkins University

[Barth Syndrome](#)

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

[Mitochondrial DNA-Associated Leigh Syndrome Spectrum](#)[Nuclear Gene-Encoded Leigh Syndrome Spectrum Overview](#)

Thorpe, Ryan K, MD; University of Iowa

[OTOF-Related Deafness](#)

Thunell, Stig, MD, PhD; Karolinska Institute

[Acute Intermittent Porphyria](#)

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

[GLB1-Related Disorders](#)[GM2 Activator Deficiency](#)[HEXA Disorders](#)[Sandhoff Disease](#)

Tijssen, Marina AJ, MD; University Medical Center Groningen

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Tiller, George E, MD, PhD; Kaiser Permanente

[Hypochondroplasia](#)[X-Linked Spondyloepiphyseal Dysplasia Tarda](#)

Timmerman, Vincent, PhD; Institute Born Bunge

[GAN-Related Neurodegeneration](#)

Timothy, Katherine W, BS; Harvard Medical School

[CACNA1C-Related Disorders](#)

Tinkle, Brad T, MD, PhD; Advocate Children's Hospital

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

Tiranti, Valeria Sonia, PhD; IRCCS Foundation Carlo Besta Neurological Institute

[Ethylmalonic Encephalopathy](#)

Tkachenko, Nataliya, MD; Santo Antonio University Hospital Center

[Ellis-van Creveld Syndrome](#)

Todd, Peter K, MD, PhD; University of Michigan School of Medicine

[FMR1 Disorders](#)

Tokuda, Takahiko, MD, PhD; Kyoto Prefectural University Hospital

[Hereditary Transthyretin Amyloidosis](#)

Tolar, Jakob, 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](#)

Toriello, Helga V, PhD; Michigan State University

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

[Thrombocytopenia Absent Radius Syndrome](#)

Toro, Camilo, MD; National Human Genome Research Institute

[Chediak-Higashi Syndrome](#)

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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

[Spinocerebellar Ataxia Type 17](#)

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[Cohen Syndrome](#)

[Isolated and Classic Cutis Marmorata Telangiectatica Congenita](#)

Trahms, Cristine, MS, RD; University of Washington

[Tyrosinemia Type I](#)

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

[Heritable Thoracic Aortic Disease Overview](#)

Tranebjærg, Lisbeth, MD, PhD; University of Copenhagen

[Deafness-Dystonia-Optic Neuronopathy Syndrome](#)

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[WFS1 Spectrum Disorder](#)

Trevisson, Eva, MD, PhD; University of Padova

[Primary Coenzyme Q10 Deficiency Overview](#)

Trifiro, Mark A, MD; McGill University

[Androgen Insensitivity Syndrome](#)

Trinh, Joanne, BSc; University of British Columbia

[LRRK2 Parkinson Disease](#)

Trollet, Capucine, PhD; Centre de Recherche en Myologie

[Oculopharyngeal Muscular Dystrophy](#)

Trouillard, Oriane, BS; Institut du Cerveau et de la Moelle épinière

[Congenital Mirror Movements](#)

Trzupek, Karmen M, MS; InformedDNA

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

[Usher Syndrome Type I](#)

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Tsang, Mandy HY, MMSc; University of Hong Kong

[CTNNB1 Neurodevelopmental Disorder](#)

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

[Weill-Marchesani Syndrome](#)

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

[DRPLA](#)

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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

[Urea Cycle Disorders Overview](#)

Turner, Joyce T, MS; Children's National Hospital

[Wilms Tumor Predisposition](#)

Turner, Stefanie, MS; 17q12 Foundation

[17q12 Recurrent Deletion Syndrome](#)

Turnpenny, Peter D, BSc, ChB, MB; Royal Devon University Healthcare NHS Foundation Trust

[Spondylocostal Dysostosis, Autosomal Recessive](#)

Tuschl, Karin, MD, PhD; UCL Great Ormond Street Institute of Child Health

[Hypermanganesemia with Dystonia 1](#)

[SLC39A14 Deficiency](#)

Tuupanen, Sari, PhD; Blueprint Genetics

### X-Linked Congenital Stationary Night Blindness

Tümer, Zeynep, DMSc, MD, PhD; Rigshospitalet

[DLG4-Related Synaptopathy](#)

Tüysüz, Beyhan, MD, PhD; Istanbul University

[Trichorhinophalangeal Syndrome](#)

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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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[Pseudoxanthoma Elasticum](#)

Ulm Seiwert, Elizabeth, MS; Cincinnati Children's Hospital and Medical Center

[IRF2BPL-Related Disorder](#)

Umair, Muhammad, PhD; King Saud Bin Abdulaziz University for Health Sciences

[EMC10-Related Neurodevelopmental Disorder](#)

Unger, Sheila, MD; Genetica AG

[Achondrogenesis Type 1B](#)

[Atelosteogenesis Type 2](#)

[Campomelic Dysplasia](#)

[Chondrodysplasia with Congenital Joint Dislocations, CHST3-Related](#)

[Diastrophic Dysplasia](#)

[SLC26A2-Related Multiple Epiphyseal Dysplasia](#)

Upadia, Jariya, MD; Tulane University School of Medicine

[Cystic Fibrosis](#)

Urban, Zsolt, PhD; University of Pittsburgh

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Urion, David K, MD; Boston Children's Hospital / Harvard Medical School

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Ursini, Matilde Valeria, PhD; Institute of Genetics and Biophysics - CNR

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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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## V

Vahidnezhad, Hassan, MSc, PhD; Pasteur Institute of Iran

[Kindler Syndrome](#)

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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

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

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Valentin, Ilia, MD; Heidelberg University

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Valeriani, Emanuele, MD; Sapienza University of Rome

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Valle, Laura, PhD; Catalan Institute of Oncology

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Valverde de Morales, Hannah Gabriela, MD; Integra Genetics

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van Belzen, Martine, PhD; Leiden University Medical Center

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van Bokhoven, Hans, PhD; Radboud University Medical Center

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van Bon, Bregje WM, MD, PhD; Radboud University Medical Center

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Van Broeckhoven, Christine, DSc, PhD; University of Antwerp

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Van Camp, Guy, PhD; University of Antwerp

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van de Warrenburg, Bart P, MD, PhD; Radboud University Nijmegen Medical Center

[ARSACS](#)

Van den Veyver, Ignatia Barbara, MD; Baylor College of Medicine

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

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van der Knaap, Marjo S, MD, PhD; Amsterdam University Medical Centers

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van der Maarel, Silvere M, MD; Leiden University Medical Center

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

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van der Zee, Julie, PhD; University of Antwerp

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Van Dijck, Anke, MD, PhD; University of Antwerp

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

[Periodontal Ehlers-Danlos Syndrome](#)

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van Dijk, Tessa, MD, PhD; Leiden University Medical Center

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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

[Nonketotic Hyperglycinemia](#)

van Karnebeek, Clara, MD, PhD; Amsterdam University Medical Centers

[Carbonic Anhydrase VA Deficiency](#)

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van Leeuwen, Rachel S, MD, PhD; University Medical Center Utrecht

[Von Hippel-Lindau Syndrome](#)

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

[SOST-Related Sclerosing Bone Dysplasias](#)

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

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

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van Os, Nienke, MD, PhD; Radboud University Medical Center

[Ataxia-Telangiectasia](#)

van Ravenswaaij-Arts, Conny M, MD, PhD; University Medical Center Groningen

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van Spaendonk, Rosalina ML, PhD; Amsterdam University Medical Center

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van Steensel, Maurice, MD, PhD; University Hospital Maastricht

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van Swieten, John C, MD, PhD; Erasmus Medical Center

[MAPT-Related Frontotemporal Dementia](#)

Van Zanten, Malou, PhD; St George's, University of London

[Milroy Disease](#)

Vanagunas, Tomas, MD, PhD; Cincinnati Children's Hospital and Medical Center

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Vance, Jeffery M, MD, PhD; Duke University Medical Center

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

Vandana, VP, PhD; National Institute of Mental Health & Neurosciences

### Huppke-Brendel Syndrome

Vander Lugt, Mark, MD; University of Michigan Medical School

[ZAP70-Related Combined Immunodeficiency](#)

Vanderver, Adeline, MD; Children's Hospital of Philadelphia

[LMNB1-Related Autosomal Dominant Leukodystrophy](#)

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Vandeweyer, Geert, PhD; Antwerp University Hospital

[ADNP-Related Disorder](#)

VanSickle, Elizabeth, MS; Spectrum Health

[Bachmann-Bupp Syndrome](#)

Vargas, Mauricio, MD, PhD; Oregon Health Sciences University

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Varon, Raymonda, PhD; Charité – Universitätsmedizin Berlin

[Nijmegen Breakage Syndrome](#)

Varvagiannis, Konstantinos, MD; Cyprus Institute of Neurology & Genetics

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Vaughan, Douglas E, MD; Northwestern University Feinberg School of Medicine

[Complete Plasminogen Activator Inhibitor 1 Deficiency](#)

Veenhuis, Stefanie, MD; Radboud University Medical Center

[Ataxia-Telangiectasia](#)

Veerapandiyan, Aravindhan, MBBS; University of Rochester Medical Center

[Andersen-Tawil Syndrome](#)

Vega, Hugo H, MD, PhD; City University of New York

[ESCO2 Spectrum Disorder](#)

Velasco, Danita, MD; University of Nebraska Medical Center

[RNU4atac-opathy](#)

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

[VPS13A Disease](#)

Velez-Bartolomei, Frances, MD; Stanford University

[MERRF](#)

Velleman, Shelley L, PhD; University of Vermont



[7q11.23 Duplication Syndrome](#)

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

[Andersen-Tawil Syndrome](#)

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

[Disorders of Intracellular Cobalamin Metabolism](#)[Isolated Methylmalonic Acidemia](#)[Propionic Acidemia](#)

Veneziano, Liana, PhD; Italian National Research Council

[DRPLA](#)

Venkateswaran, Sunita, MD; Children's Hospital of Eastern Ontario

[Fatty Acid Hydroxylase-Associated Neurodegeneration](#)

Verdin, Hannah, MSc, PhD; Ghent University Hospital

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

Verloes, Alain, MD, PhD; APHP-Robert Debré University Hospital

[ASPM Primary Microcephaly](#)[Baraitser-Winter Cerebrofrontofacial Syndrome](#)[WDR62 Primary Microcephaly](#)

Verma, Ishwar Chandar, MD; Sir Ganga Ram Hospital

[Primrose Syndrome](#)

Vermeer, Sascha, MD, PhD; University Hospital Leuven

[ARSACS](#)

Vernon, Hilary, MD, PhD; Johns Hopkins University

[Barth Syndrome](#)

Vicart, Savine, MD; Assistance Publique – Hôpitaux de Paris

[Hypokalemic Periodic Paralysis](#)

Vichinsky, Elliott, MD; Children's Hospital & Research Center Oakland

[Sickle Cell Disease](#)

Vig, Parminder JS, PhD; University of Mississippi Medical Center

[Spinocerebellar Ataxia Type 3](#)

Vihola, Anna, PhD; Fimlab Laboratories

[ANO5 Muscle Disease](#)

Vikkula, Miikka, MD, PhD; Université catholique de Louvain

[TEK-Related Venous Malformations](#)

Vilain, Eric J, MD, PhD; Children's National Hospital

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

[NR0B1-Related Adrenal Hypoplasia Congenita](#)

Villa, Anna, MD, PhD; Humanitas Clinical and Research Center

[CLCN7-Related Osteopetrosis](#)

Villanueva, Joyce, MBA, MT; Cincinnati Children's Hospital Medical Center

[Familial Hemophagocytic Lymphohistiocytosis](#)

Vincent, G Michael, MD; University of Utah School of Medicine

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Viskochil, David H, MD, PhD; University of Utah

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Visser, Lisenka ELM, PhD; Radboud University Medical Center

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Vissing, John, DMSc, MD; Rigshospitalet

[Myotonia Congenita](#)

Vnencak-Jones, Cindy, PhD; GC Vanderbilt University Medical Center

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

Vockley, Jerry, MD, PhD; University of Pittsburgh

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

Vogel-Farley, Vanessa, BA; Dup15q Alliance

[Maternal 15q Duplication Syndrome](#)

Volpini, Victor, MD, PhD; Bellvitge Biomedical Research Institute

[Spinocerebellar Ataxia Type 37](#)

Vos, Yvonne J, PhD; University Medical Center Groningen

[L1 Syndrome](#)

## W

Wagner, Klaus, MD, PhD; Medical University Graz

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

Wagner, Victoria F, MS; McGovern Medical School

[Mucopolysaccharidosis Type III](#)

Wagon, Jacy L, PhD; University of Michigan

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

Waguespack, Steven G, MD; University of Texas MD Anderson Cancer Center  
[CDC73-Related Disorders](#)

Wakefield, Emily, MS; Cincinnati Children's Hospital Medical Center  
[X-Linked Lymphoproliferative Disease](#)

Waldman, Amy; Children's Hospital of Philadelphia  
[Alexander Disease](#)

Walker, Elaine F, PhD; Emory University  
[3q29 Recurrent Deletion](#)

Walker, Ruth H, MBBS, MD, PhD; Mount Sinai School of Medicine  
[McLeod Neuroacanthocytosis Syndrome](#)  
[VPS13A Disease](#)

Walkovich, Kelly, MD; University of Michigan Medical School  
[ZAP70-Related Combined Immunodeficiency](#)

Wallace, Stephanie E, MD; Seattle Children's Hospital  
[Camurati-Engelmann Disease](#)

Wallefeld, William, BSc (Hons); Royal Perth Hospital  
[Laing Distal Myopathy](#)

Walsh, Christopher A, MD, PhD; Harvard Medical School  
[FLNA Deficiency](#)

Wanders, Ronald JA, PhD; Academic Medical Center, University of Amsterdam  
[Adult Refsum Disease](#)

Wang, Bruce, MD; University of California San Francisco  
[Hereditary Coproporphria](#)

Wang, Dong, MD; Emory University Affiliated Hospital  
[Glucose Transporter Type 1 Deficiency Syndrome](#)  
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Wang, Heng, MD, PhD; DDC Clinic – Center for Special Needs Children  
[Cohen Syndrome](#)  
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Wang, Hsiao-Lin, PhD; Emory University School of Medicine  
[CTCF-Related Disorder](#)

Wang, Hua, MD, PhD; Loma Linda University  
[MBTPS1-Related Spondyloepimetaphyseal Dysplasia with Elevated Lysosomal Enzymes](#)

Wang, Jing, MD; Baylor College of Medicine

TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Wang, Julia, BS; Baylor College of Medicine

MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect

TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Wang, Leo H, MD, PhD; University of Washington

Facioscapulohumeral Muscular Dystrophy

Wangler, Michael F, MD; Baylor College of Medicine

ACTG2 Visceral Myopathy

Xia-Gibbs Syndrome

Wang, Lisa L, MD; Baylor College of Medicine

Baller-Gerold Syndrome

Poikiloderma with Neutropenia

Rothmund-Thomson Syndrome

Wang, Raymond, MD; University of California, Irvine

Mucopolysaccharidosis Type VII

Wang, Zhao, MD; Beijing Friendship Hospital

Familial Hemophagocytic Lymphohistiocytosis

Wappner, Rebecca S, MD; Riley Hospital for Children, Indiana University School of Medicine

Lowe Syndrome

Warby, Simon C, PhD; University of Montreal

Huntington Disease

Warman, Matthew L, MD; Boston Children's Hospital

Stickler Syndrome

Warner, Blake M, DDS, MPH, PhD; National Institute of Dental and Craniofacial Research

ALPK1-Related Autoinflammatory Disease

Wasserstein, Melissa P, MD; Albert Einstein College of Medicine

Acid Sphingomyelinase Deficiency

Free Sialic Acid Storage Disorders

Krabbe Disease

Wassif, Christopher A, PhD; National Institute of Child Health and Human Development

Smith-Lemli-Opitz Syndrome

Waterham, Hans R, PhD; Academic Medical Center, University of Amsterdam

Adult Refsum Disease

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Waters, Aoife M, BCh, MB BAO, MSc; University College London

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Waters, Michael F, MD, MS, PhD; Barrow Neurological Institute

[Spinocerebellar Ataxia Type 13](#)

Wattendorf, Daniel J, MD; National Institutes of Health

[Xeroderma Pigmentosum](#)

Watts, Giles, PhD; University of East Anglia

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

Waxman, Stephen G, MD, PhD; Yale University School of Medicine

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Weaver, K Nicole, MD; Cincinnati Children's Hospital Medical Center

[HRAS-Related Costello Syndrome](#)[Sphingosine Phosphate Lyase Insufficiency Syndrome](#)

Weaver, W Curtis, BS; Geisinger Health System

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Webb, Bryn D, MD, PhD; Icahn School of Medicine at Mount Sinai

[STAC3 Disorder](#)

Weber, Frank, MD, PhD; German Air Force Center of Aviation Medicine

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

Weckhuysen, Sarah, MD, PhD; University of Antwerp

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Weemaes, Corry, MD, PhD; Radboud University Medical Center

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Weese-Mayer, Debra E, MD; Children's Memorial Hospital

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Wei, Chongjuan, PhD; University of Texas MD Anderson Cancer Center

[Peutz-Jeghers Syndrome](#)

Weinstein, David A, MD, MMSc; University of Connecticut

[Glycogen Storage Disease Type III](#)[Glycogen Storage Disease Type VI](#)

Weisfeld-Adams, James, MB ChB; Children's Hospital Colorado

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Weiss, Karin, MD; Technion-Israel Institute of Technology

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Weiss, Karl Heinz, MD; Salem Medical Center

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Weiss, Marjan M, MD, PhD; Leiden University Medical Center

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Weiss, Mitchell J, MD, PhD; St Jude Children's Research Hospital

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Weisz Hubshman, Monika, MD, PhD; Rabin Medical Center

[Familial Dysautonomia](#)

Weksberg, Rosanna, MD, PhD; The Hospital for Sick Children

[Beckwith-Wiedemann Syndrome](#)

Weleber, Richard G, MD; Casey Eye Institute of Oregon Health Sciences University

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Welsh, Holly I, MS; University of Missouri

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Wenger, David A, PhD; Thomas Jefferson University Hospital

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Wenger, Olivia K, MD; New Leaf Center

[HYAL2 Deficiency](#)

Wenger, Tara L, MD, PhD; Seattle Children's Hospital

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Wensel, Christine, MS; DDC Clinic – Center for Special Needs Children

[Cohen Syndrome](#)

Wenstrup, Richard, MD; Cincinnati Children's Hospital Medical Center

[Classic Ehlers-Danlos Syndrome](#)[PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome](#)

Westbroek, Wendy, PhD; National Institutes of Health

[Chediak-Higashi Syndrome](#)

Weston, Michael D, MA; Boys Town National Research Hospital

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

Wevers, Ron A, PhD; Radboud University Nijmegen Medical Center

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Whatley, Sharon D, MSc, PhD; University Hospital of Wales

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Whitcomb, David C, MD, PhD; University of Pittsburgh & University of Pittsburgh Medical Center

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White-Brown, Alexandre M, MSc; Children's Hospital of Eastern Ontario

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White, Colin T, MD; British Columbia's Children's and Women's Hospital

[Primary Hyperoxaluria Type 1](#)

White, Janson, PhD; Invitae Corporation

[White-Sutton Syndrome](#)

White, Stormi; Emory University School of Medicine

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White, Susan M, MD; Victorian Clinical Genetics Service

[Floating-Harbor Syndrome](#)

Whitman, Mary C, MD, PhD; Harvard Medical School

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Whitton, Chloe, BMed, MD; University of New South Wales

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Wider, Christian, MD; Centre Hospitalier Universitaire Vaudois

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Wieczorek, Dagmar, MD; Universitätsklinikum Düsseldorf

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Wierenga, Andrea, PhD; SIVOTEC Bioinformatics

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Wierenga, Klaas, MD; Mayo Clinic Florida

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Wieser, Thomas, MD; Fachkrankenhaus Jerichow

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Wiesner, Georgia L, MD, MS; Children's Hospital of Philadelphia

[Multiple Endocrine Neoplasia Type 2](#)

Wilcox, William R, MD, PhD; Emory University School of Medicine

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Wildin, Robert S, MD; Oregon Health Sciences University

[Hereditary Nephrogenic Diabetes Insipidus](#)

Wilkie, Andrew OM, MA; University of Oxford

[Enlarged Parietal Foramina](#)

Wilkin, Douglas J; Federal Bureau of Investigation

[Achondroplasia](#)

Willemsen, Michèl, MD, PhD; Radboud University Medical Center

[Ataxia-Telangiectasia](#)

Williams, Charles A, MD; University of Florida College of Medicine

[Angelman Syndrome](#)

Williams, Denise, MD; Birmingham Women's and Children's Hospital

[Alstrom Syndrome](#)

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Williams, Gretchen, BS; Children's Hospitals and Clinics of Minnesota

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Williamson, Kathleen A, PhD; University of Edinburgh

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Wilson, Frederick H, MD, PhD; Harvard Medical School

[Pseudohypoaldosteronism Type II](#)

Winkler, Jürgen, MD; University Hospital Erlangen

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Winter, Robin M; Institute of Child Health

[Three M Syndrome](#)

Wise, Anastasia L, PhD; National Jewish Health and University of Colorado Denver

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Wissinger, Bernd, MSc, PhD; University of Tübingen

[Achromatopsia](#)

Wiwattanadittakul, Natrujee, MD; Chiang Mai University

[Succinic Semialdehyde Dehydrogenase Deficiency](#)

Wlodarski, Marcin, MD, PhD; St Jude Children's Research Hospital

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Wojcieszek, Joanne, MD; Indiana University School of Medicine

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Wolf, Barry, MD, PhD; Ann and Robert H Lurie Children's Hospital of Chicago

[Biotinidase Deficiency](#)

Wolfe, Lynne, ARNP, MS; National Human Genome Research Institute

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Wolf, Nicole I, MD, PhD; Amsterdam University Medical Centers

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Wollnik, Bernd, MD; University of Cologne

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Wong, Derek, MD; University of California Los Angeles Medical Center

[Arginase Deficiency](#)

Wong, Lee-Jun C, PhD; Baylor College of Medicine

[Deoxyguanosine Kinase Deficiency](#)

[FARS2 Deficiency](#)

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Wong, Melanie, MBBS, PhD; Children's Hospital Westmead

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

Woods, Christopher Geoffrey, MB ChB; University of Cambridge

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Woolf, Adrian S, MA, MD; University of Manchester and Central Manchester University Hospitals NHS Foundation Trust

[Urofacial Syndrome](#)

Wortmann, Saskia B, MD, PhD; Salzburger Landeskliniken and Paracelsus Medical University

[CLPB Deficiency](#)

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Wright, Benjamin, MD; University Hospitals Birmingham

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Wright, Galen EB, PhD; University of British Columbia

[Huntington Disease](#)

Wright, J Timothy, DDS, MS; University of North Carolina

[Hypohidrotic Ectodermal Dysplasia](#)

Wright, Michael J, ChB, MB, MSc; Institute of Human Genetics

[COMP-Related Pseudoachondroplasia](#)

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Wszolek, Zbigniew K, MD; Mayo Clinic

[AARS2-Related Disorder](#)

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[DYT-GNAL](#)

[VPS35-Related Parkinson Disease](#)

Wu, Bai-Lin, M Med, PhD; Boston Children's Hospital

[16p11.2 Recurrent Deletion](#)

Wuyts, Wim, PhD; University and University Hospital of Antwerp

[Hereditary Multiple Osteochondromas](#)

## X

Xia, Maya, BA; COMBINEDBrain

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

Xiao, Changrui, MD; University of California Irvine

[GM2 Activator Deficiency](#)

[Sandhoff Disease](#)

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Xin, Baozhong, PhD; DDC Clinic – Center for Special Needs Children

[GM3 Synthase Deficiency](#)

Xu, Manlong, MD, PhD; University of Alberta

[Choroideremia](#)

## Y

Yamada, Koki, MD, PhD; Children's Hospital Boston

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Yamada, Mitsunori, MD, PhD; Shinshu University School of Medicine

[Spinocerebellar Ataxia Type 17](#)

Yamanaka, Koji, MD, PhD; RIKEN Brain Science Institute

[ALS2-Related Disorder](#)

Yang, Jiandong, PhD; George Washington University School of Medicine

[DICER1 Tumor Predisposition](#)

Yang, Kathryn, MD; Harvard Medical School

[PRRT2-Related Disorder](#)

Yang, Paul, MD, PhD; Casey Eye Institute - Oregon Health Sciences University

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

Yang, Yaping, PhD; AiLife Diagnostics

[TANGO2 Deficiency](#)

Yap, Kai Lee, PhD; Northwestern University

[Congenital Central Hypoventilation Syndrome](#)

Yates, T Michael, MBChB; University of Edinburgh

[SOX2 Disorder](#)

Yau, Mabel, MD; Mount Sinai School of Medicine

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

Yehia, Lamis, PhD; Cleveland Clinic

[PTEN Hamartoma Tumor Syndrome](#)

Yen, Timothy, MD; University of Colorado Anschutz Medical Center

[APC-Associated Polyposis Conditions](#)

Yeowell, Heather N, PhD; Duke University Medical Center

[PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome](#)

Yntema, Helger G, PhD; Radboud University Medical Center

[Kleefstra Syndrome](#)

Yonker, Lael, MD; MassGeneral Hospital for Children

[Pitt-Hopkins Syndrome](#)

Yoshida, Kunihiko, MD, PhD; Shinshu University Hospital

[Hereditary Transthyretin Amyloidosis](#)

Youngblom, Emily, BA; University of Washington School of Public Health

[Familial Hypercholesterolemia](#)

Young, Danielle O, MS; Kimball Genetics, Inc

[Celiac Disease](#)

Young, Elizabeth, BSc, PhD; Royal Devon and Exeter Healthcare NHS Trust

[Spondylocostal Dysostosis, Autosomal Recessive](#)

Young, William F, MD, MSc; Mayo Clinic

[Hereditary Paraganglioma-Pheochromocytoma Syndromes](#)

Youssefian, Leila, MSc, PhD; Thomas Jefferson University

Kindler Syndrome

Lipoid Proteinosis

Yu-Wai-Man, Patrick, BMedSci, MBBS, PhD; University of Cambridge

Leber Hereditary Optic Neuropathy

NR2F1-Related Neurodevelopmental Disorder

Yuan, Bo, PhD; Baylor College of Medicine

Potocki-Lupski Syndrome

## Z

Zackai, Elaine H, MD; Children's Hospital of Philadelphia

22q11.2 Deletion Syndrome

Emanuel Syndrome

Zadeh, Neda, MD; CHOC Children's Hospital

KCNK9 Imprinting Syndrome

Zaki, Maha S, MD, PhD; National Research Centre

UNC80 Deficiency

Zandee, Wouter, MD, PhD; University Medical Center Groningen

Von Hippel-Lindau Syndrome

Zara, Federico, PhD; G Gaslini Pediatric Institute and University of Genova

Hypomyelination and Congenital Cataract

Zarate, Yuri A, MD; University of Arkansas for Medical Sciences

SATB2-Associated Syndrome

Zariwala, Maimoona A, MSc, PhD; University of North Carolina at Chapel Hill

Primary Ciliary Dyskinesia

Zbuk, Kevin M, MD; Cleveland Clinic Genomic Medicine Institute

PTEN Hamartoma Tumor Syndrome

Zelley, Kristin, MS; Children's Hospital of Philadelphia

Li-Fraumeni Syndrome

Zerr, Inga, MD; University Medical School

Genetic Prion Disease

Zhai, Yi, MD, PhD; University of Alberta

Choroideremia

Zhang, Kejian, MBA, MD; GoBroad Healthcare Group

Autoimmune Lymphoproliferative Syndrome

Familial Hemophagocytic Lymphohistiocytosis

Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

WAS-Related Disorders

X-Linked Hyper IgM Syndrome

X-Linked Lymphoproliferative Disease

Ziegler, John B, MBBS, MD; Sydney Children's Hospital

Hepatic Veno-Occlusive Disease with Immunodeficiency

Ziegler, Shira G, MD, PhD; Johns Hopkins University School of Medicine

Generalized Arterial Calcification of Infancy

Zipfel, Peter F, PhD; Hans Knoll Institute

C3 Glomerulopathy

Zippin, Jonathan, MD, PhD; Joan and Sanford I Weill Medical College of Cornell University

Oculocutaneous Albinism and Ocular Albinism Overview

Zobor, Ditta, MD, PhD; University of Tübingen

Achromatopsia

Zoghbi, Huda Y, MD; Baylor College of Medicine

MECP2 Disorders

Spinocerebellar Ataxia Type 1

Zschocke, Johannes, MD, PhD; Medical University of Innsbruck

Periodontal Ehlers-Danlos Syndrome

Züchner, Stephan, MD, PhD; University of Miami Miller School of Medicine

GAA-FGF14-Related Ataxia

GDAP1-Related Hereditary Motor and Sensory Neuropathy

MFN2 Hereditary Motor and Sensory Neuropathy

PNPLA6 Disorders

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Živná, Martina, PhD; Charles University

Autosomal Dominant Tubulointerstitial Kidney Disease -- MUC1

Autosomal Dominant Tubulointerstitial Kidney Disease -- REN

Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD

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