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PPP1R12A-Related Urogenital and/or Brain Malformation Syndrome
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Smith-Magenis Syndrome

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El-Hattab-Alkuraya Syndrome

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MN1 C-Terminal Truncation Syndrome

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SCARB2-Related Action Myoclonus – Renal Failure Syndrome

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ANKRD26-Related Thrombocytopenia

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Calpainopathy

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SLC6A1-Related Neurodevelopmental Disorder

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- SLC26A2-Related Multiple Epiphyseal Dysplasia

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CHD8-Related Neurodevelopmental Disorder with Overgrowth

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- Canavan Disease

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- Fructose-1,6-Bisphosphatase Deficiency

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- KCNT1-Related Epilepsy

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- Multicentric Osteolysis Nodulosis and Arthropathy
- Progressive Pseudorheumatoid Dysplasia

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- Bryant-Li-Bhoj Neurodevelopmental Syndrome

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- Hypomyelination and Congenital Cataract

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- Friedreich Ataxia

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- Greig Cephalopolysyndactyly Syndrome
- Proteus Syndrome
- SLC25A19-Related Thiamine Metabolism Dysfunction

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Bikker, Hennie, PhD; University of Amsterdam
- Long QT Syndrome Overview
- Trichorhinophalangeal Syndrome

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- SHOX Deficiency Disorders

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- Huppke-Brendel Syndrome
- Isolated Sulfite Oxidase Deficiency

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Cold-Induced Sweating Syndrome including Crisponi Syndrome
Congenital Stromal Corneal Dystrophy

Bomont, Pascale, PhD; Institute for Neurosciences of Montpellier
GAN-Related Neurodegeneration

Bonafé, Luisa, MD, PhD; Lausanne University Hospital
Achondrogenesis Type 1B
Atelosteogenesis Type 2
Diastrophic Dysplasia
SLC26A2-Related Multiple Epiphyseal Dysplasia

Bonkovsky, Herbert, MD; Wake Forest University School of Medicine
Familial Porphyria Cutanea Tarda
Hepatoerythropoietic Porphyria

Bonne, Gisèle, PhD; Centre de Recherche en Myologie
Emery-Dreifuss Muscular Dystrophy

Bontoux, Luc, MD; Centre Hospitalier Universitaire d'Angers
SH3TC2-Related Hereditary Motor and Sensory Neuropathy

Boon, Elles MJ, PhD; Leiden University Medical Center
CADASIL

Boon, Laurence M, MD, PhD; Cliniques universitaires Saint-Luc
TEK-Related Venous Malformations

Boonstra, Nils-Erik, MD; Haukeland University Hospital
ADAMTSL4-Related Eye Disorders

Booth, Kevin, PhD; Indiana School of Medicine
GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss

Borck, Guntram, MD, PhD; University of Ulm
Kaufman Oculocerebrofacial Syndrome

Bornfeld, Norbert, MD; University of Essen
Retinoblastoma

Borroni, Barbara, MD; University of Brescia
Spinocerebellar Ataxia Type 38

Bostwick, Bret, MD; Baylor College of Medicine
CDK13-Related Disorder
PORCN-Related Developmental Disorders

Bouchard, Jean-Pierre, MD; Laval University
ARSACS
SYNE1 Deficiency

Boulinguiez, Alexis, PhD; Centre de Recherche en Myologie
Oculopharyngeal Muscular Dystrophy

Bourdeaut, Franck, MD, PhD; Paris Sciences et Lettres Research University
Rhabdoid Tumor Predisposition Syndrome

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

Boussion, Simon, MD; CHU de Lille
Thrombocytopenia Absent Radius Syndrome

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

Bower, Matthew A, MS; University of Minnesota Medical Center, Fairview
- PAX2-Related Disorder

Boyce, Alison M, MD; National Institute of Dental and Craniofacial Research
- Fibrous Dysplasia / McCune-Albright Syndrome

Boycott, Kym M, MD, PhD; Children's Hospital of Eastern Ontario
- Mandibulofacial Dysostosis with Microcephaly
- Microcephaly-Capillary Malformation Syndrome
- THOC6 Intellectual Disability Syndrome
- VLDLR Cerebellar Hypoplasia
- X-Linked Congenital Stationary Night Blindness

Boyd, Charles D, PhD; John A Burns School of Medicine, University of Hawaii
- Pseudoxanthoma Elasticum

Boyd, Kerry E, MD; Hamilton Health Services
- Smith-Magenis Syndrome

Brabbing-Goldstein, Dana, MD; Rabin Medical Center
- Kaufman Oculocerebrofacial Syndrome

Bradbury, Kimberley, BA, MA, MBBS; Great Ormond Street Hospital for Children
- BCL11A-Related Intellectual Disability

Braden, Ruth, PhD; University of Melbourne
- FOXP1 Syndrome

Brais, Bernard, M Phil, MD, PhD; McGill University
- ARSACS
- GAA-FGF14-Related Ataxia
- Oculopharyngeal Muscular Dystrophy

Bramswig, Nuria C, MD; Institute of Human Genetics
- UNC80 Deficiency

Brandi, Maria Luisa, MD, PhD; Italian Foundation for Research on Bone Diseases
- Multiple Endocrine Neoplasia Type 1

Brand, Randall, MD; University of Pittsburgh
- MUTYH Polyposis

Brandt, Vicky L; Baylor College of Medicine
- MECP2 Disorders
- Spinocerebellar Ataxia Type 1

Brashear, Allison, MD; Wake Forest School of Medicine
ATP1A3-Related Neurologic Disorders

Braverman, Nancy E, MD, MS; McGill University
Chondrodysplasia Punctata 1, X-Linked
Rhizomelic Chondrodysplasia Punctata Type 1
Zellweger Spectrum Disorder

Bredrup, Cecilie, MD, PhD; Haukeland University Hospital
Congenital Stromal Corneal Dystrophy

Bree, Alanna F, MD; Dermatology Specialists of Houston
TP63-Related Disorders

Brennan, Christine, PhD; University of Colorado
Smith-Magenis Syndrome

Bresin, Elena, MD; Istituto di Ricerche Farmacologiche Mario Negri–IRCCS
Genetic Atypical Hemolytic-Uremic Syndrome

Bressman, Susan B, MD; Columbia University Medical Center
DYT1 Early-Onset Isolated Dystonia

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

Brewer, Eileen D, MD; Baylor College of Medicine
Lowe Syndrome

Brice, Alexis, MD; Hôpital Pitié-Salpêtrière
Parkin Type of Early-Onset Parkinson Disease
Spastic Paraplegia 11

Brice, Glen W, RGN BSc (Hons); St George's, University of London
Lymphedema-Distichiasis Syndrome
Milroy Disease

Briere, Lauren, MS; Massachusetts General Hospital
GNAO1-Related Disorder

Briggs, Michael D, PhD; Newcastle University
COMP-Related Pseudoachondroplasia
Multiple Epiphyseal Dysplasia, Dominant

Brilliant, Murray H, PhD; University of Wisconsin School of Medicine
Oculocutaneous Albinism Type 4

Brisson, Jean-Denis, MD; Sherbrooke University
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum

Broadbridge, Elizabeth, BS; National Human Genome Research Institute
RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies

Brockmann, Knut, MD; University Medical Center
FOXG1 Syndrome

Brock, Pamela, MS; The Ohio State University Comprehensive Cancer Center
Multiple Endocrine Neoplasia Type 4

Brock, Stefanie, MD; Universitair Ziekenhuis Brussel
PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia

Bronicki, Lucas, PhD; Cincinnati Children's Hospital Medical Center
WAS-Related Disorders

Brooks, Brian P, MD, PhD; National Eye Institute
Oculocutaneous Albinism and Ocular Albinism Overview

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

Brower, Cheryl L, MSPH, RN; Puget Sound Blood Center
Hemophilia A
Hemophilia B

Brown, Jeremy M, MD; Addenbrooke's Hospital
CHMP2B Frontotemporal Dementia

Brown, W Ted, MD, PhD; Institute for Basic Research in Developmental Disabilities
Hutchinson-Gilford Progeria Syndrome

Bruckner, Anna L, MD; University of Colorado School of Medicine
Epidermolysis Bullosa Simplex

Bruel, Ange-Line, PhD; University of Burgundy
Oral-Facial-Digital Syndrome Type I

Brugada, Josep, MD, PhD; University of Barcelona
Brugada Syndrome

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

Brugada, Ramon, MD, PhD; University of Girona
Brugada Syndrome

Brunetti-Pierri, Nicola, MD; University of Naples "Federico II"
Chondrodysplasia Punctata 1, X-Linked
Myhre Syndrome

Brunkow, Mary E, PhD; Institute for Systems Biology

SOST-Related Sclerosing Bone Dysplasias

Brunner, Han, MD, PhD; Radboud University Medical Center
Autosomal Dominant Robinow Syndrome

Bruno, Claudio, MD, PhD; Gaslini Institute
Hypomyelination and Congenital Cataract

Brunzell, John D, MD; University of Washington School of Medicine
Familial Lipoprotein Lipase Deficiency

Brusco, Alfredo, PhD; University of Turin
Spinocerebellar Ataxia Type 28
Spinocerebellar Ataxia Type 38

Brussino, Alessandro, MD; University of Turin
Spinocerebellar Ataxia Type 28

Bryant, Laura, PhD; Children's Hospital of Philadelphia
Bryant-Li-Bhoj Neurodevelopmental Syndrome

Bryceson, Yenan, PhD; Karolinska Institutet
Familial Hemophagocytic Lymphohistiocytosis

Brzezinski, Jack, MD, PhD; Hospital for Sick Children
Wilms Tumor Predisposition

Brüggemann, Norbert, MD; University of Lübeck
Parkin Type of Early-Onset Parkinson Disease

Buck, Cassandra, MS; Massachusetts General Hospital
Isolated Gonadotropin-Releasing Hormone (GnRH) Deficiency

Buckley, Michael Francis, MB ChB, PhD; Prince of Wales Hospital
Hepatic Veno-Occlusive Disease with Immunodeficiency

Bull, Laura N, PhD; University of California, San Francisco
ATP8B1 Deficiency

Bupp, Caleb, MD; Spectrum Health
Bachmann-Bupp Syndrome

Burgmaier, Kathrin, MD; University of Cologne
Autosomal Recessive Polycystic Kidney Disease – PKHD1

Burkardt, Deepika, DO; National Human Genome Research Institute
HIST1H1E Syndrome

Burke, Wylie, MD, PhD; University of Washington
BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Burnett, John R, MB ChB, MD, PhD; University of Western Australia

Abetalipoproteinemia
APOB-Related Familial Hypobetalipoproteinemia
Chylomicron Retention Disease
Familial Combined Hypolipidemia
Familial Lipoprotein Lipase Deficiency
Tangier Disease

Burrage, Lindsay, MD, PhD; Baylor College of Medicine
TANGO2 Deficiency

Burr, Amanda, MS; Casey Eye Institute / Oregon Health & Science University
RPE65-Related Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy

Burrell, T Lindsey, PhD; Emory University School of Medicine
3q29 Recurrent Deletion

Burt, Randall W, MD; Huntsman Cancer Institute
APC-Associated Polyposis Conditions

Bushby, Katharine Mary, MB ChB, MD; University of Newcastle upon Tyne
Collagen VI-Related Dystrophies

Busra, Aynekin, PhD; University College London
WARS2 Deficiency

Butler-Browne, Gillian, PhD; Centre de Recherche en Myologie
Oculopharyngeal Muscular Dystrophy

Byers, Peter H, MD; University of Washington Medical Center
COL1A1/2 Osteogenesis Imperfecta
Vascular Ehlers-Danlos Syndrome

Böckenhauer, Detlef, MD, PhD; University Hospitals Leuven
X-Linked Hypophosphatemia

Bönnemann, Carsten G, MD; National Institute of Neurological Disorders and Stroke
Collagen VI-Related Dystrophies

C

Cabalza, Jessica L; George Washington University School of Medicine
Succinic Semialdehyde Dehydrogenase Deficiency

Cabanillas, Maria E, MD; University of Texas MD Anderson Cancer Center
CDC73-Related Disorders

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

Calame, Daniel, MD, PhD; Baylor College of Medicine

ENTPD1-Related Neurodevelopmental Disorder

Caldovic, Ljubica, PhD; Children's National Hospital
Ornithine Transcarbamylase Deficiency

Cali, Elisa, MD; UCL Queen Square Institute of Neurology
Riboflavin Transporter Deficiency

Callewaert, Bert L, MD, PhD; Ghent University Hospital
Arterial Tortuosity Syndrome
Congenital Contractural Arachnodactyly
ELN-Related Cutis Laxa
LTBP4-Related Cutis Laxa

Calvert, Guy, DPhil; Dup15q Alliance
Maternal 15q Duplication Syndrome

Camacho, Jose A, MD; University of California, Irvine
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome

Camaschella, Clara, MD; Istituto Scientifico San Raffaele
TFR2-Related Hemochromatosis

Campeau, Philippe M, MD; Sainte-Justine Hospital
KAT6B Disorders
Spondylometaphyseal Dysplasia, Corner Fracture Type
TBC1D24-Related Disorders

Campuzano, Oscar, PhD; University of Girona
Brugada Syndrome

Cao, Antonio, MD; Consiglio Nazionale delle Ricerche
Alpha-Thalassemia
Beta-Thalassemia

Capece, Tara; University of Pittsburgh
ZAP70-Related Combined Immunodeficiency

Capp, Philip K; George Washington University School of Medicine
Succinic Semialdehyde Dehydrogenase Deficiency

Caprioli, Jessica, Biol Sci D; IRCCS – Istituto di Ricerche Farmacologiche Mario Negri
Genetic Atypical Hemolytic-Uremic Syndrome

Caputi, Caterina, MD; Sapienza Università di Roma
PRICKLE1-Related Disorders

Carey, Catherine, MD; Torbay Hospital
Alstrom Syndrome

Cario, Holger, MD; University Medical Center Ulm
- Primary Familial and Congenital Polycythemia

Carlberg, Katie, MD; University of Washington
- Sickle Cell Disease

Carlo, Maria I, MD; Memorial Sloan Kettering Cancer Center
- BAP1 Tumor Predisposition Syndrome

Carmignac, Virginie, PhD; Université de Bourgogne
- Salih Myopathy

Caron, Nicholas S, PhD; University of British Columbia
- Huntington Disease

Carr, Ann, MS; Westat
- DICER1 Tumor Predisposition

Carrillo, Nuria, MD; National Human Genome Research Institute
- Disorders of Intracellular Cobalamin Metabolism
- GNE Myopathy
- Propionic Acidemia

Carson, Vincent J, MD; Clinic for Special Children
- Maple Syrup Urine Disease

Carter, Melissa T, MD, MSc; The Children's Hospital of Eastern Ontario
- Microcephaly-Capillary Malformation Syndrome
- NOTCH3-Related Lateral Meningocele Syndrome

Carvalho, Luciani Renata, MD, PhD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo
- PROP1-Related Combined Pituitary Hormone Deficiency

Carvill, Gemma, PhD; Northwestern University
- CHD2-Related Neurodevelopmental Disorders
- KCNT1-Related Epilepsy

Casari, Giorgio, PhD; San Raffaele University
- Spastic Paraplegia 7

Casey, Hannah L, BS; University of Chicago
- Spinocerebellar Ataxia Type 6

Cassidy, Suzanne B, MD; Children's Hospital of Philadelphia
- Prader-Willi Syndrome

Cathey, Sara S, MD; Greenwood Genetic Center
- GNPTAB-Related Disorders

Caudill, Marissa; University of Connecticut Health Center
Isolated Gonadotropin-Releasing Hormone (GnRH) Deficiency

Cavallin, Mara, MD; Paris Descartes University
Tubulinopathies Overview

Cebulla, Colleen, MD, PhD; The Ohio State University
BAP1 Tumor Predisposition Syndrome

Cecchi, Alana C, MS; University of Texas Health Science Center
Heritable Thoracic Aortic Disease Overview

Cederbaum, Stephen, MD; UCLA Medical Center
Arginase Deficiency

Chaix, Charlène; La Timone Children's Hospital
Trichohepatoenteric Syndrome

Chamberlain, Stormy, PhD; University of Connecticut Health Center
Maternal 15q Duplication Syndrome

Chamova, Teodora, MD, PhD; Medical University of Sofia
CTDP1-Related Congenital Cataracts, Facial Dysmorphism, and Neuropathy

Chander, Varuna, MS; Baylor College of Medicine
Xia-Gibbs Syndrome

Chandra, Sharat, MD; Cincinnati Children's Hospital Medical Center
WAS-Related Disorders

Chang, Irene J, MD; Seattle Children's Hospital Seattle, Washington
Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Chanprasert, Sirisak, MD; Baylor College of Medicine
TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Chansky, Howard A, MD; University of Washington
Hereditary Multiple Osteochondromas

Chan, Sophelia HS, MD; University of Hong Kong
CHKB-Related Muscular Dystrophy

Chao, Daniel L, MD, PhD; Shiley Eye Institute / UCSD
RPE65-Related Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy

Chapman, Kimberly A, MD, PhD; Children's National Health System
Urea Cycle Disorders Overview

Charles, Jane, MD; Medical University of South Carolina
Smith-Magenis Syndrome

Chaussenot, Annabelle, MD; Nice Hospital

CHCHD10-Related Disorders

Chelban, Viorica, MD; University College London Institute of Neurology
NKX6-2-Related Disorder

Chen, Dong-Hui, MD, PhD; University of Washington Medical Center
SAMD9L Ataxia-Pancytopenia Syndrome
Spinocerebellar Ataxia Type 14

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

Cheng, Shirley SW, MD; Hong Kong Children's Hospital
CTNNB1 Neurodevelopmental Disorder
FAM111A-Related Skeletal Dysplasias

Chen, I-Ping, DDS, MS; UConn Health
Craniometaphyseal Dysplasia, Autosomal Dominant

Chen, Karin, MD; University of Washington
X-Linked Severe Combined Immunodeficiency

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

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

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

Chen, Yuan-Tsong, MD, PhD; Duke University Medical Center
Glycogen Storage Disease Type I

Chen, Zhongbo, BM BCh, MA; UCL Queen Square Institute of Neurology and National Hospital for Neurology and Neurosurgery
Spinocerebellar Ataxia Type 11

Cheung, Sau Wai, MBA, PhD; Baylor College of Medicine
Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated

Chew, Ben H, MD, MSc; University of Columbia
Primary Hyperoxaluria Type 1

Chinnery, Patrick F, BMedSci, MBBS, PhD; University of Cambridge
Hereditary Myopathy with Early Respiratory Failure
Leber Hereditary Optic Neuropathy
Neuroferritinopathy
POLG-Related Disorders

Primary Mitochondrial Disorders Overview

Chiplunkar, Shwetha, MBBS; National Institute of Mental Health & Neurosciences
Huppke-Brendel Syndrome

Chiquet, Brett, DDS, PhD; UTHealth School of Dentistry at Houston
Nonsyndromic Tooth Agenesis Overview

Chitayat, David, MD; Mount Sinai Hospital
Autosomal Dominant Robinow Syndrome
PPP1R12A-Related Urogenital and/or Brain Malformation Syndrome
Warsaw Syndrome

Chmiel, James F, MD; Case Western Reserve University School of Medicine
Cystic Fibrosis

Chopra, Maya, MBBS; Boston Children's Hospital
ANKRD17-Related Neurodevelopmental Syndrome

Chou, Stella T, MD; Children's Hospital of Philadelphia
GATA1-Related Cytopenia

Chrestian, Nicolas, MD; CHU Laval–CHU de Québec
Hereditary Neuropathy with Liability to Pressure Palsies

Christiaans, Imke, MD, PhD; University Medical Center Groningen
Long QT Syndrome Overview

Christodoulou, John, MBBS, PhD; Murdoch Children's Research Institute
Fumarate Hydratase Deficiency
MECP2 Disorders
Phosphoribosylpyrophosphate Synthetase Deficiency
Phosphoribosylpyrophosphate Synthetase Superactivity
Squalene Synthase Deficiency

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

Chudley, Albert E, MD; University of Manitoba
FREM1 Autosomal Recessive Disorders
Ritscher-Schinzel Syndrome

Chung, Brian HY, MD; University of Hong Kong
CTNNB1 Neurodevelopmental Disorder
MN1 C-Terminal Truncation Syndrome

Chung, Wendy K, MD, PhD; Columbia University
16p11.2 Recurrent Deletion

Okur-Chung Neurodevelopmental Syndrome
PACS1 Neurodevelopmental Disorder
PPP2R5D-Related Neurodevelopmental Disorder

Churpek, Jane E, MD, MS; University of Wisconsin School of Medicine and Public Health
DDX41-Associated Familial Myelodysplastic Syndrome and Acute Myeloid Leukemia

Ciapaite, Jolita, PhD; University Medical Center Utrecht
PLPBP Deficiency

Cimino, Theora, BS; University of California San Francisco
Hereditary Coproporphyria

Cirino, Allison L, MS; Brigham and Women's Hospital
Hypertrophic Cardiomyopathy Overview

Clark, Dinah M, MS; Children's Hospital of Philadelphia
Cornelia de Lange Syndrome

Clarke, Lorne A, MD; University of British Columbia
Mucopolysaccharidosis Type I

Clarke, Shoa L, MD, PhD; Stanford University School of Medicine
Familial Hypercholesterolemia

Clayton, Peter T, MD; UCL Great Ormond Street Institute of Child Health
Hypermanganesemia with Dystonia 1
SLC39A14 Deficiency

Cleary, John Douglas, PhD; University of Florida
Spinocerebellar Ataxia Type 8

Clericuzio, Carol, MD; University of New Mexico Health Sciences Center
Poikiloderma with Neutropenia

Cleveland, Don W, PhD; University of California San Diego
ALS2-Related Disorder

Clinton, Catherine, MS; Boston Children's Hospital
Diamond-Blackfan Anemia

Coe, Bradley P, PhD; University of Washington School of Medicine
DYRK1A Syndrome

Coelho, Teresa, MD, PhD; Centro Hospitalar Universitário do Porto
LAMA2 Muscular Dystrophy

Cogal, Andrea G, BS; Mayo Clinic
Dent Disease
Primary Hyperoxaluria Type 1

Cohen, Bruce H, MD; Northeast Ohio Medical University
POLG-Related Disorders

Cohn, Daniel H, PhD; Cedars-Sinai Medical Center
COMP-Related Pseudoachondroplasia

Colding-Jørgensen, Eskild, MD; University of Copenhagen
Myotonia Congenita

Cole, Trevor RP, MB ChB; Birmingham Women's Hospital
Sotos Syndrome

Collins, Debra L, MS; University of Kansas Medical Center
Von Hippel-Lindau Syndrome

Collins, Francis S, MD, PhD; National Institutes of Health
Hutchinson-Gilford Progeria Syndrome

Collins, Michael T, MD; National Institute of Dental and Craniofacial Research
Fibrous Dysplasia / McCune-Albright Syndrome
Hyperphosphatemic Familial Tumoral Calcinosis

Colombo, Sophie, MS, PhD; Columbia University Irving Medical Center
GNB1 Encephalopathy

Coman, David, MBBS; Queensland Children's Hospital
Fumarate Hydratase Deficiency
Squalene Synthase Deficiency

Conant, Alex, BS; Children's National Health System
Pelizaeus-Merzbacher-Like Disease 1
TUBB4A-Related Leukodystrophy

Concannon, Patrick J, PhD; University of Virginia
Nijmegen Breakage Syndrome

Concolino, Daniela, MD; University "Magna Graecia"
Poikiloderma with Neutropenia

Conley, Mary Ellen, MD; University of Tennessee
X-Linked Agammaglobulinemia

Connell, Fiona; Guy's Hospital
Milroy Disease

Connor, Jessica A, MS; Counsyl, Inc
EPB42-Related Hereditary Spherocytosis
Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Conta, Jessie, MS; Seattle Children's Hospital

Mowat-Wilson Syndrome

Conway, Gerard S, MD; University College London
Perrault Syndrome

Conway, Robert, MD; Wayne State University
PIK3CA-Related Overgrowth Spectrum

Cook Shukla, Lola, MS; Indiana University School of Medicine
Parkinson Disease Overview

Cook, Edwin H, MD; University of Illinois at Chicago
3q29 Recurrent Deletion
Maternal 15q Duplication Syndrome

Cook, Jared F, MA; Wake Forest School of Medicine
ATP1A3-Related Neurologic Disorders

Cooper, Edward C, MD, PhD; Baylor College of Medicine
KCNQ2-Related Disorders
KCNQ3-Related Disorders

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
KCNQ3-Related Disorders

Coppola, Giovanni, MD; University of California Los Angeles
Primary Familial Brain Calcification

Coppola, Stephanie, BS; Jefferson Medical College
Krabbe Disease

Corces, Victor, PhD; Emory University School of Medicine
CTCF-Related Disorder

Cormier-Daire, Valérie, MD, PhD; Paris Cité University
Geleophysic Dysplasia
Weill-Marchesani Syndrome

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

Cortese, Andrea, MD, PhD; University College London
RFC1 CANVAS / Spectrum Disorder

Coryell, Jason, MS; Oregon Health and Science University
Pantothenate Kinase-Associated Neurodegeneration

Coucke, Paul, MD, PhD; Ghent University Hospital
Arterial Tortuosity Syndrome

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

Couillard-Despres, Sebastien, PhD; Paracelsus Medical University Salzburg
DCX-Related Disorders

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

Cowan, Jason, MS; University of Miami Miller School of Medicine
LMNA-Related Dilated Cardiomyopathy

Cox, Diane W, PhD; University of Alberta
Alpha-1 Antitrypsin Deficiency
Wilson Disease

Cragun, Deborah, MS; University of Tampa
Cytochrome P450 Oxidoreductase Deficiency

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

Crombez, Eric A, MD; Ultragenyx Pharmaceutical Inc
Arginase Deficiency

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

Crowley, Leon, MSc; King's College London
TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia

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

Crow, Yanick J, BMedSci, MBBS, PhD; Central Manchester Foundation Trust University Hospitals
Aicardi-Goutieres Syndrome

Cruts, Marc, PhD; University of Antwerp

C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia

Cruz Corchado, Johnny; University of Iowa
C3 Glomerulopathy

Cruz, Vincent, MD, MS; DDC Clinic – Center for Special Needs Children
GM3 Synthase Deficiency

Cubells, Joseph F, MD, PhD; Emory University School of Medicine
3q29 Recurrent Deletion

Cudrici, Cornelia, MD; National Heart, Lung and Blood Institute
TNF Receptor-Associated Periodic Fever Syndrome

Culver, Julie O Bars, MS; Fred Hutchinson Cancer Research Center
BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Culver, Kathy, MS; Sutter Pacific Medical Foundation/California Pacific Medical Center
Simpson-Golabi-Behmel Syndrome Type 1

Cunniff, Christopher M, MD; Weill Cornell Medical College
Bloom Syndrome
Smith-Lemli-Opitz Syndrome

Cunningham, Lea, MD; National Cancer Institute
RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies

Cunningham, Michael L, MD, PhD; University of Washington Schools of Medicine and Dentistry
Saethre-Chotzen Syndrome

Curiel, Julian, BS; Children's National Health System
TUBB4A-Related Leukodystrophy

Cutting, Garry R, MD; Johns Hopkins University
Cystic Fibrosis
Thanatophoric Dysplasia
Treacher Collins Syndrome

D

D'Adamo, Maria Cristina, PhD; University of Malta
Episodic Ataxia Type 1

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

Da Silva, Jorge Diogo, MD, PhD; Santo Antonio University Hospital Center
Ellis-van Creveld Syndrome

Dafsari, Hormos Salimi, MD; University of Cologne

DYNC1H1-Related Disorders
EPG5-Related Disorder

Dagli, Aditi I, MD; Orlando Health Arnold Palmer Hospital
Angelman Syndrome
Glycogen Storage Disease Type III
Glycogen Storage Disease Type VI

Daiger, Stephen P, PhD; University of Texas Health Science Center
Nonsyndromic Retinitis Pigmentosa Overview

Dai, Hongzheng, PhD; Baylor College of Medicine
FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion Syndrome
MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect

Dalal, Ashwin, MD; Centre for DNA Fingerprinting and Diagnostics
Progressive Pseudorheumatoid Dysplasia

Dale, David C, MD; University of Washington Medical Center
ELANE-Related Neutropenia

Daley, Shane M, MD; Mayo Clinic
Jervell and Lange-Nielsen Syndrome

Dalton, Joline C, MS; Institute of Human Genetics
Myotonic Dystrophy Type 2
Spinocerebellar Ataxia Type 8

Daly, Mary B, MD, PhD; Fox Chase Cancer Center
BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Danek, Adrian, MD; Ludwig-Maximilians-Universität
McLeod Neuroacanthocytosis Syndrome
VPS13A Disease

Danzi, Matt, PhD; University of Miami Miller School of Medicine
GAA-FGF14-Related Ataxia

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

Das, Soma, PhD; University of Chicago
PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia
X-Linked Myotubular Myopathy

Davenport, Sandra LH, MD; Sensory Genetics and Neurodevelopment
CHD7 Disorder

Davies, Alexandra K, PhD; University of Cambridge

AP-4-Associated Hereditary Spastic Paraplegia

Davis, Joie; NIH
STAT3 Hyper IgE Syndrome
X-Linked Severe Combined Immunodeficiency

Day, John W, MD, PhD; Stanford University
Myotonic Dystrophy Type 2
Spinocerebellar Ataxia Type 8

Dazzo, Emanuela, PhD; CNR Institute of Neuroscience
Autosomal Dominant Epilepsy with Auditory Features

De Baere, Elfride, MD, PhD; Ghent University Hospital
Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome

de Boer, Irene, MD; Leiden University Medical Center
Retinal Vasculopathy with Cerebral Leukoencephalopathy and Systemic Manifestations

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
- GNB5-Related Neurodevelopmental Disorder

de Vernejoul, Marie-Christine, MD, PhD; Hopital Lariboisiere
- CLCN7-Related Osteopetrosis

De Vivo, Darryl, MD; Columbia University
- Glucose Transporter Type 1 Deficiency Syndrome
- Pyruvate Carboxylase Deficiency

De Voer, Richarda M, PhD; Radboud University Medical Center
- NTHL1 Tumor Syndrome

de Vries, Bert BA, MD, PhD; Radboud University Medical Center
- 15q13.3 Recurrent Deletion
- DYRK1A Syndrome
- Gabriele-de Vries Syndrome
- Koolen-de Vries Syndrome
- TRIO-Related Neurodevelopmental Disorder
- WAC-Related Intellectual Disability

Deardorff, Matthew A, MD, PhD; Children's Hospital Los Angeles
- Coffin-Siris Syndrome
- Cornelia de Lange Syndrome
- IMAGe Syndrome
- WDR26-Related Intellectual Disability

DeFelice, Magee, MD; Nemours Children's Health
- RNU4atac-opathy

Dehner, Louis P; Washington University Medical Center
- DICER1 Tumor Predisposition

del Gaudio, Daniela, PhD; University of Chicago
- Maturity-Onset Diabetes of the Young Overview

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

Amyotrophic Lateral Sclerosis Overview

Arrhythmogenic Right Ventricular Cardiomyopathy Overview

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

Autosomal Recessive Polycystic Kidney Disease – PKHD1

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

Perrault Syndrome

Demarest, Scott, MD; University of Colorado

CDKL5 Deficiency Disorder

SLC6A1-Related Neurodevelopmental Disorder

Demirdas, Serwet, MD, PhD; Erasmus Medical Center

TNXB-Related Classical-Like Ehlers-Danlos Syndrome

Dempsey, Melissa A, MS; Parkview Health

Allan-Herndon-Dudley Syndrome

Chondrodysplasia Punctata 2, X-Linked

Demuth, Ilja, PhD; Charité – Universitätsmedizin Berlin

Nijmegen Breakage Syndrome

Deneubourg, Celine, MSc; King's College London

EPG5-Related Disorder

Depienne, Christel, PhD; Hôpital Pitié-Salpêtrière

CLCN2-Related Leukoencephalopathy

Congenital Mirror Movements

Spastic Paraplegia 4

Der Kaloustian, Vazken M, MD; McGill University

Hidrotic Ectodermal Dysplasia 2

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

Glycogen Storage Disease Type III

Derry, W Brent, PhD; The Hospital for Sick Children

Familial Cerebral Cavernous Malformation

Desai, Jigar, PhD; Boston Children's Hospital

Congenital Fibrosis of the Extraocular Muscles Overview

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

Congenital Erythropoietic Porphyria
Erythropoietic Protoporphyria, Autosomal Recessive
Fabry Disease
HEXA Disorders
X-Linked Protoporphyria

Deuitch, Natalie, MS; National Human Genome Research Institute
RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies
TNF Receptor-Associated Periodic Fever Syndrome

Deutschländer, Angela B, MD; Mayo Clinic
DYT-GNAL
VPS35-Related Parkinson Disease

Devinsky, Orrin, MD; New York University Langone Medical Center
Maternal 15q Duplication Syndrome

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

Dhamija, Radhika, MD; Mayo Clinic
FGFR1-Related Hartsfield Syndrome
LZTR1- and SMARCB1-Related Schwannomatosis

Di Donato, Nataliya, MD, PhD; Universitätsklinikum Carl Gustav Carus
Baraitser-Winter Cerebrofrontofacial Syndrome

Di Giovanni, Giuseppe, PhD; Istituto Euro-Mediterraneo di Scienza e Tecnologia
Episodic Ataxia Type 1

Di Gregorio, Eleonora, PhD; Città della Salute e della Scienza University Hospital
Spinocerebellar Ataxia Type 38

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

Di Paola, Jorge, MD; Washington University School of Medicine
ETV6 Thrombocytopenia and Predisposition to Leukemia

Dias, Cristina, MD, PhD; King's College London
BCL11A-Related Intellectual Disability

Dias, Renuka, PhD; Birmingham Women's and Children's Hospital
WFS1 Spectrum Disorder

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

Dib-Hajj, Sulayman D, PhD; Yale University School of Medicine
SCN9A Neuropathic Pain Syndromes

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

Dickerson, Kathryn E, MD; University of Texas Southwestern Medical Center
Monosomy 7 Predisposition Syndromes Overview

Dietz, Harry C, MD; Johns Hopkins University School of Medicine
FBN1-Related Marfan Syndrome
Loeys-Dietz Syndrome

DiGiovanna, John J, MD; National Cancer Institute
Xeroderma Pigmentosum

Digweed, Martin, PhD; Charité – Universitätsmedizin Berlin
Nijmegen Breakage Syndrome

DiMauro, Salvatore, MD; Columbia University Medical Center
MELAS
MERRF
Mitochondrial DNA Deletion Syndromes

Dimmock, David P, MD; Children's Hospital of Wisconsin
Deoxyguanosine Kinase Deficiency

Dindot, Scott, PhD; Texas A&M University
Maternal 15q Duplication Syndrome

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

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

DiStasio, Andrew T, PhD; Nationwide Children's Hospital
ATP7A-Related Copper Transport Disorders

Disteche, Christine M, PhD; University of Washington
Y Chromosome Infertility

DiStefano, Charlotte, PhD; University of California Los Angeles
Maternal 15q Duplication Syndrome

Dobson-Stone, Carol, DPhil; University of Sydney
McLeod Neuroacanthocytosis Syndrome
VPS13A Disease

Dobyns, William B, MD; University of Minnesota

ATP6V0A2-Related Cutis Laxa
Encephalocraniocutaneous Lipomatosis
PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia
PIK3CA-Related Overgrowth Spectrum

Docherty, Louise Esther, BSc (Hons), PhD; Salisbury NHS Foundation Trust
Diabetes Mellitus, 6q24-Related Transient Neonatal

Doherty, Dan, MD, PhD; University of Washington
MN1 C-Terminal Truncation Syndrome

Doherty, Emily S, MD; Carilion Clinic
Muenke Syndrome

Doimo, Mara, PhD; University of Padova
Primary Coenzyme Q10 Deficiency Overview

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

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

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

Donkervoort, Sandra, MS; National Institute of Neurological Disorders and Stroke
Amyotrophic Lateral Sclerosis Overview
Collagen VI-Related Dystrophies
Inclusion Body Myopathy with Paget Disease of Bone and/or Frontotemporal Dementia

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

Doros, Leslie, MD; Children's National Medical Center
DICER1 Tumor Predisposition

Dorsey, Adrianne M; Children's National Medical Center
Succinic Semialdehyde Dehydrogenase Deficiency

Dotti, Maria Teresa, MD; University of Siena
Cerebrotendinous Xanthomatosis

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

Douzgou, Sofia, MD, PhD; Haukeland University Hospital
PPP2R1A-Related Neurodevelopmental Disorder

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

X-Linked Myotubular Myopathy

Downs, Jenny, MSc, PhD; Telethon Kids Institute
CDKL5 Deficiency Disorder

Drillings, Ian; Children's National Medical Center
Succinic Semialdehyde Dehydrogenase Deficiency

Driscoll, Daniel J, MD, PhD; University of Florida College of Medicine
Angelman Syndrome
Prader-Willi Syndrome

Drunat, Séverine, PharmD, PhD; APHP-Robert Debré University Hospital
ASPM Primary Microcephaly
Baraitser-Winter Cerebrofrontofacial Syndrome
WDR62 Primary Microcephaly

du Souich, Christèle, MSc; University of British Columbia
NSDHL-Related Disorders
UNC80 Deficiency

Dua Puri, Ratna, MD; Sir Ganga Ram Hospital
Primrose Syndrome

Dubois, Anna, BSc, MBChB; Royal Victoria Infirmary
CYLD Cutaneous Syndrome

Dugan, Stefanie N, MS; BloodCenter of Wisconsin
ANKRD26-Related Thrombocytopenia

Duis, Jessica, MD, MS; Children's Hospital Colorado
Schinzel-Giedion Syndrome
SETBP1 Haploinsufficiency Disorder

Duker, Angela, MS; Nemours Children's Health
Microcephalic Osteodysplastic Primordial Dwarfism Type II
Rhizomelic Chondrodysplasia Punctata Type 1
RNU4atac-opathy

Duley, John A, PhD; University of Queensland and Mater Research
Phosphoribosylpyrophosphate Synthetase Deficiency
Phosphoribosylpyrophosphate Synthetase Superactivity

Dulski, Jaroslaw, MD, PhD; Medical University of Gdansk
CSF1R-Related Disorder
DCTN1-Related Neurodegeneration
VPS35-Related Parkinson Disease

Dumitrescu, Alexandra M, MD, PhD; University of Chicago Medical Center
Allan-Herndon-Dudley Syndrome

Dunn, Clinton P, MD; University of Washington
X-Linked Hyper IgM Syndrome

Dunoyer, Margaux, MD; Assistance Publique-Hôpitaux de Paris
Congenital Mirror Movements

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

Dunø, Morten, PhD; Rigshospitalet
Myotonia Congenita

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

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

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

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

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

Durr, Alexandra, MD, PhD; Hôpital de la Pitié-Salpêtrière
Parkin Type of Early-Onset Parkinson Disease
Spastic Paraplegia 11
Spastic Paraplegia 4
Spinocerebellar Ataxia Type 28

Dye, Thomas J, MD; Cincinnati Children's Hospital Medical Center
DLG4-Related Synaptopathy

Dykens, Elisabeth, PhD; University of California, Los Angeles
Smith-Magenis Syndrome

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

Délot, Emmanuèle C, PhD; Children's National Hospital
Nonsyndromic 46,XX Testicular Disorders/Differences of Sex Development

E

Earl, Rachel, PhD; University of Washington
CHD8-Related Neurodevelopmental Disorder with Overgrowth

Ebens, Christen, MD, MPH; University of Minnesota
Fanconi Anemia

Ebrahimi-Fakhari, Darius, MD, PhD; Harvard Medical School
AP-4-Associated Hereditary Spastic Paraplegia
EPG5-Related Disorder
PRRT2-Associated Paroxysmal Movement Disorders
Spastic Paraplegia 15
TECPR2-Related Hereditary Sensory and Autonomic Neuropathy with Intellectual Disability

Eccles, Michael R, PhD; University of Otago
PAX2-Related Disorder

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

Edward, Deepak P, MD; University of Illinois at Chicago
Primary Congenital Glaucoma

Edwards, Corwin Q, MD; University of Utah School of Medicine
HFE-Related Hemochromatosis

Efthymiou, Stephanie, MSc, PhD; University College London
GRIA2-Related Neurodevelopmental Disorder
WARS2 Deficiency

Eggens, Veerle RC, MSc; University of Amsterdam
EXOSC3 Pontocerebellar Hypoplasia
TSEN54 Pontocerebellar Hypoplasia

Eichler, Evan E, PhD; University of Washington School of Medicine
CHD8-Related Neurodevelopmental Disorder with Overgrowth
DYRK1A Syndrome

Eichler, Florian S, MD; Massachusetts General Hospital
Canavan Disease

Ejaz, Resham, MD; McMaster Children's Hospital
NOTCH3-Related Lateral Meningocele Syndrome

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

Glycogen Storage Disease Type I

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

Asparagine Synthetase Deficiency

Deoxyguanosine Kinase Deficiency

El-Hattab-Alkuraya Syndrome

FARS2 Deficiency

FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion Syndrome

Glycogen Storage Disease Type IV

MELAS

Mitochondrial DNA Maintenance Defects Overview

MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect

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

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

Systemic Primary Carnitine Deficiency

TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated

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

LRRK2 Parkinson Disease

Elizondo, Leah I; Baylor College of Medicine

Schimke Immunoosseous Dysplasia

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

Ritscher-Schinzel Syndrome

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

Pseudohypoaldosteronism Type II

Elsas, Louis J, MD; University of Miami

Classic Galactosemia and Clinical Variant Galactosemia

Elsea, Sarah H, PhD; Baylor College of Medicine

Aromatic L-Amino Acid Decarboxylase Deficiency

MBD5 Haploinsufficiency

Smith-Magenis Syndrome

Else, Tobias, MD; University of Michigan

Hereditary Paraganglioma-Pheochromocytoma Syndromes

POT1 Tumor Predisposition

Elsharkawi, Ibrahim, MD; Washington University

Pitt-Hopkins Syndrome

Emanuel, Beverly S, PhD; Children's Hospital of Philadelphia
22q11.2 Deletion Syndrome
Emanuel Syndrome

Eng, Charis, MD, PhD; Cleveland Clinic
Multiple Endocrine Neoplasia Type 2
PTEN Hamartoma Tumor Syndrome

Engelborghs, Sebastiaan, MD, PhD; University of Antwerp
C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia

Engelen, Marc, MD, PhD; Amsterdam University Medical Centers
Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation

England, Jade, MD, MSc; University of Montreal
Spondylometaphyseal Dysplasia, Corner Fracture Type

Engle, Elizabeth C, MD; Children's Hospital Boston
Congenital Fibrosis of the Extraocular Muscles Overview
Duane Syndrome

Enns, Gregory M, MB ChB; Stanford University
Carnitine-Acylcarnitine Translocase Deficiency
MERRF
NGLY1-Related Congenital Disorder of Deglycosylation

Erez, Ayelet, MD, PhD; Baylor College of Medicine
Argininosuccinate Lyase Deficiency

Erro, Roberto, MD, PhD; University of Salerno
Familial Paroxysmal Nonkinesigenic Dyskinesia

Erwin, Angelika, MD, PhD; Cleveland Clinic
Congenital Erythropoietic Porphyria

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

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
NF2-Related Schwannomatosis

Evans, Kelly N, MD; Seattle Children's Hospital
- Apert Syndrome
- FGFR Craniosynostosis Syndromes Overview

Everett, Lorraine A; National Human Genome Research Institute
- Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

Evidente, Virgilio Gerald H, MD; Movement Disorders Center of Arizona
- X-Linked Dystonia-Parkinsonism Syndrome

Ewbank, Clifton, MD; University of California San Francisco
- Fumarate Hydratase Deficiency

Eymard-Pierre, Eleonore, PhD; Unité mixte de recherche 384 et Fédération de génétique humaine Auvergne
- ALS2-Related Disorder

F

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

Fahim, Abigail T, MD, PhD; University of Michigan Medical School
- Nonsyndromic Retinitis Pigmentosa Overview

Fahrner, Jill A, MD, PhD; Johns Hopkins School of Medicine
- TET3-Related Beck-Fahrner Syndrome

Falchetti, Alberto, MD; University Hospital of Careggi
- Multiple Endocrine Neoplasia Type 1

Falik Zaccai, Tzipora C, MD; Galilee Medical Center
- INSR-Related Severe Insulin Resistance Syndrome

Falk, Marni J, MD; Children's Hospital of Philadelphia
- Cohen Syndrome
- FGFR Craniosynostosis Syndromes Overview
- Mitochondrial DNA Deletion Syndromes
- Primary Pyruvate Dehydrogenase Complex Deficiency Overview

Fallatah, Wedad, MD; McGill University
- Rhizomelic Chondrodysplasia Punctata Type 1

Fam, Hok Khim, BSc; University of British Columbia
- Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive

Fanin, Marina, PhD; University of Padova
- Calpainopathy

Fanto, Manolis, PhD; King's College London

EPG5-Related Disorder

Fan, Yuting, MD; St Luke's Hospital
Y Chromosome Infertility

Faqeih, Eissa, MD; King Fahad Medical City
FARS2 Deficiency

Farlow, Janice, BA, BS; Indiana University School of Medicine
Parkinson Disease Overview

Farndon, Peter A, MD; Birmingham Women's Hospital
Nevoid Basal Cell Carcinoma Syndrome

Farrer, Matthew, PhD; University of British Columbia
LRRK2 Parkinson Disease

Fasham, James, BSc, MBChB, PhD; University of Exeter Medical School
HYAL2 Deficiency

Fatemi, Ali, MD; Kennedy Krieger Institute
X-Linked Adrenoleukodystrophy

Fauth, Christine, MD; Medical University of Innsbruck
FKBP14 Kyphoscoliotic Ehlers-Danlos Syndrome

Fecarotta, Simona, MD; Federico II University
Lysinuric Protein Intolerance

Fechner, Patricia Y, MD; University of Washington
Nonsyndromic Disorders of Testicular Development Overview

Federico, Antonio, MD; University of Siena
Cerebrotendinous Xanthomatosis

Feldman, Gerald L, MD, PhD; Wayne State University School of Medicine
BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Feldman, Howard H, MD; University of British Columbia
GRN Frontotemporal Dementia

Felton, Thomas, MS; McLendon Clinical Laboratories
DRPLA

Ferreira, Carlos R, MD; National Human Genome Research Institute
Barth Syndrome
Generalized Arterial Calcification of Infancy
Osteoglophonic Dysplasia
Primrose Syndrome
Prolidase Deficiency

Saul-Wilson Syndrome

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

Field, Amanda, MPH; ResourcePath
DICER1 Tumor Predisposition

Figlewicz, Denise A, PhD; University of Michigan Medical School
Facioscapulohumeral Muscular Dystrophy

Filipovich, Alexandra H, MD; Cincinnati Children's Hospital
Familial Hemophagocytic Lymphohistiocytosis
WAS-Related Disorders
X-Linked Hyper IgM Syndrome
X-Linked Lymphoproliferative Disease

Finanger, Erika, MD; Oregon Health and Science University
Spinal Muscular Atrophy

Fink, John K, MD; University of Michigan
Hereditary Spastic Paraplegia Overview

Finucane, Brenda M, MS; Geisinger Health System
17q12 Recurrent Deletion Syndrome
Maternal 15q Duplication Syndrome
Smith-Magenis Syndrome

Fishbein, Lauren, MD, PhD; University of Colorado School of Medicine
Hereditary Paraganglioma-Pheochromocytoma Syndromes

Fisher, Simon E, DPhil; Max Planck Institute for Psycholinguistics
FOXP2-Related Speech and Language Disorder

Fish, Jennifer, PhD; University of Massachusetts Lowell
SATB2-Associated Syndrome

Fiskerstrand, Torunn, MD, PhD; University of Bergen
ADAMTSL4-Related Eye Disorders

Fitzgibbon, Jude, PhD; Queen Mary University of London
CEBPA-Associated Familial Acute Myeloid Leukemia (AML)

FitzPatrick, David R, MD; University of Edinburgh
SOX2 Disorder

Flanagan, Maeve, BA; Weill Cornell Medical College
Bloom Syndrome

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

Collagen VI-Related Dystrophies

Fleming, Judith C, PhD
Thiamine-Responsive Megaloblastic Anemia Syndrome

Flemming, Kelly D, MD; Mayo Clinic
Familial Cerebral Cavernous Malformation

Florenzano, Pablo, MD; National Institute of Dental and Craniofacial Research
Fibrous Dysplasia / McCune-Albright Syndrome

Fluharty, Arvan L, PhD; University of California, Los Angeles
Arylsulfatase A Deficiency

Fogli, Anne, PhD; UMR INSERM 1103
Childhood Ataxia with Central Nervous System Hypomyelination / Vanishing White Matter

Foley, A Reghan, MD; National Institute of Neurological Disorders and Stroke
Collagen VI-Related Dystrophies

Fong, Jamie C, MS; University of California, San Francisco
Holt-Oram Syndrome

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

Foroud, Tatiana, PhD; Indiana University School of Medicine
Parkinson Disease Overview

Forsythe, Elizabeth, BMedSci, MBBS; University College London
Bardet-Biedl Syndrome Overview

Forsyth, RaeLynn, MD; Johns Hopkins University School of Medicine
Bardet-Biedl Syndrome Overview

Forwood, Caitlin, BSc (Hons), MBBS (Hons); Sydney Children's Hospitals Network
CLCN4-Related Neurodevelopmental Disorder

Foss, Kimberly, MS; Seattle Children's Hospital
PPP2R5D-Related Neurodevelopmental Disorder

Foster, Rebecca, PhD; St Louis Children's Hospital
Smith-Magenis Syndrome

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

Fox, Mark H, MD; University of Iowa
PRICKLE1-Related Disorders

Franco, Brunella, MD; University of Naples Federico II
Microphthalmia with Linear Skin Defects Syndrome

Oral-Facial-Digital Syndrome Type I

Francomano, Clair A, MD; National Institutes of Health
Achondroplasia
Hypochondroplasia

Franques, Jérôme, MD; Assistance Publique – Hôpitaux de Marseille
Hypokalemic Periodic Paralysis

Frantzen, Carlijn, MD; University Medical Center Groningen
Von Hippel-Lindau Syndrome

Frazier, Marsha L, PhD; UT MD Anderson Cancer Center
Peutz-Jeghers Syndrome

Freeman, Alexandra F, MD; National Institutes of Health
STAT3 Hyper IgE Syndrome

French, Tegan; Murdoch Children's Research Institute
Thanatophoric Dysplasia

Frey, Beat M, MD; Swiss Red Cross
McLeod Neuroacanthocytosis Syndrome

Fridovich-Keil, Judith L, PhD; Emory University School of Medicine
Duarte Variant Galactosemia
Epimerase Deficiency Galactosemia

Friedman, Jan M, MD, PhD; University of British Columbia
Neurofibromatosis 1

Friedman, Jennifer, MD; University of California San Diego
ADCY5 Dyskinesia
Sepiapterin Reductase Deficiency

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

Friez, Michael J, PhD; Greenwood Genetic Center
GNPTAB-Related Disorders

Frontali, Marina, MD; Italian National Research Council
DRPLA

Frühwald, Michael C, MD, PhD; University Medical Center Augsburg
Rhabdoid Tumor Predisposition Syndrome

Fu, Jiao, MD; University of Chicago Medical Center
Allan-Herndon-Dudley Syndrome

Fujimura, Frank K, PhD; GMP Genetics, Inc

Hemophilia A
Hemophilia B

Fukutake, Toshio, MD, PhD; Kameda Medical Center
HTRA1 Disorder

Fung, Jasmine LF; University of Hong Kong
MN1 C-Terminal Truncation Syndrome

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

G

Gabriele, Michele, MS, PhD; European Institute of Oncology
Gabriele-de Vries Syndrome

Gadea, Randi, MS; Children's Mercy Hospitals and Clinics
Ayme-Gripp Syndrome

Gafni, Rachel I, MD; National Institute of Dental and Craniofacial Research
Hyperphosphatemic Familial Tumoral Calcinosis

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

Galanello, Renzo, MD; Ospedale Regionale Microcitemie
Alpha-Thalassemia
Beta-Thalassemia

Gallagher, Emily R, MD, MPH; University of Washington
Saethre-Chotzen Syndrome

Gallie, Brenda L, MD; Hospital for Sick Children
Retinoblastoma

Gallus, Gian Nicola, Dsci; University of Siena
Cerebrotendinous Xanthomatosis

Gamache, Pierre-Luc, MD, PhD; CHU de Québec
SYNE1 Deficiency

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

3q29 Recurrent Deletion
Duarte Variant Galactosemia

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

Ganetzky, Rebecca, MD; Children's Hospital of Philadelphia
Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency
Primary Pyruvate Dehydrogenase Complex Deficiency Overview
TRMU Deficiency

Gangishetti, Prasanna K, MBBS; Mount Sinai School of Medicine
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

Garber, Judy, MD, MPH; Dana Farber Cancer Institute
Li-Fraumeni Syndrome

Garber, Kathryn, PhD; Emory University School of Medicine
CTCF-Related Disorder

Garbern, James Y, MD, PhD; University of Rochester Medical Center
PLP1 Disorders

Garcia, Christine Kim, MD, PhD; Columbia University Medical Center
Pulmonary Fibrosis Predisposition Overview

García-Murias, María, MS; Centro de Investigación Biomédica en red de Enfermedades Raras
Spinocerebellar Ataxia Type 36

Garden, Gwenn A, MD, PhD; University of Washington
Spinocerebellar Ataxia Type 7

Gardner, Kathy Lou, MD; University of Pittsburgh
Familial Hemiplegic Migraine

Gardner, RJM, MB ChB; University of Otago
Spinocerebellar Ataxia Type 20

Garland, Emily M, PhD; Vanderbilt University Medical Center
Dopamine Beta-Hydroxylase Deficiency

Gasior, Maciej, MD, PhD; George Washington University School of Medicine
Succinic Semialdehyde Dehydrogenase Deficiency

Gassner, Christoph, PhD; Swiss Red Cross
McLeod Neuroacanthocytosis Syndrome

Gatti, Richard A, MD; David Geffen School of Medicine at UCLA
Ataxia-Telangiectasia
Nijmegen Breakage Syndrome

Gaudette, Mara, MS; Northwestern University Medical School
Amyotrophic Lateral Sclerosis Overview

Gaughan, Sommer, RD; Children's Hospital Colorado
Hereditary Fructose Intolerance

Gauvreau, Claudie, MD, MSc; Laval University
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum

Gazdagh, Gabriella, MD; University Hospital Southampton
TRIO-Related Neurodevelopmental Disorder

Gazda, Hanna T, MD, PhD; Harvard Medical School
Diamond-Blackfan Anemia

Gazzerro, Elisabetta, MD; Gaslini Institute
Hypomyelination and Congenital Cataract

Gear, Russell, MB ChB; Victorian Clinical Genetics Services
Osteopathia Striata with Cranial Sclerosis

Geberhiwot, Tarekegn, MD; University Hospitals
Alstrom Syndrome

Gelb, Bruce D, MD; Icahn School of Medicine at Mount Sinai
Char Syndrome
Noonan Syndrome with Multiple Lentigines

German, James, MD; Weill Cornell Medical College
Bloom Syndrome

Gertler, Tracy, MD, PhD; Children's Hospital of Chicago
KCNT1-Related Epilepsy

Geschwind, Daniel H, MD, PhD; University of California, Los Angeles
Primary Familial Brain Calcification

Ghali, Neeti, MBChB, MD; London North West University Healthcare NHS Trust
TNXB-Related Classical-Like Ehlers-Danlos Syndrome

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

Ghosh, Rajarshi, PhD; National Institute of Allergy and Infectious Diseases
GARS-Associated Axonal Neuropathy

Gibbs, Richard A, PhD; Baylor College of Medicine
Xia-Gibbs Syndrome

Gibson, K Michael, PhD; Washington State University Spokane
Succinic Semialdehyde Dehydrogenase Deficiency

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

Gibson, William Thomas, MD, PhD; BC Children's Hospital
- EED-Related Overgrowth
- SETD1B-Related Neurodevelopmental Disorder

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
- Von Hippel-Lindau Syndrome

Gillis, David, MD; Hadassah Hebrew University Hospital
- Familial Hyperinsulinism

Gimpel, Charlotte, MD; Heidelberg University
- Autosomal Recessive Polycystic Kidney Disease – PKHD1

Ginevic, Ilona, RD; Icahn School of Medicine at Mount Sinai
- Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency / Trifunctional Protein Deficiency

Giovanni, Monica A, MS; Geisinger Health System
- Lysosomal Acid Lipase Deficiency

Girirajan, Santhosh, MBBS, PhD; Pennsylvania State University
- 16p12.2 Recurrent Deletion
- Smith-Magenis Syndrome

Girisha, Katta Mohan, MD, PhD; Manipal Academy of Higher Education
- EBF3 Neurodevelopmental Disorder
- EXOC6B-Related Spondyloepimetaphyseal Dysplasia with Joint Laxity
- ISCA1-Related Multiple Mitochondrial Dysfunctions Syndrome
- Multicentric Osteolysis Nodulosis and Arthropathy
- Progressive Pseudorheumatoid Dysplasia

Giunta, Cecilia, PhD; University Children's Hospital
- FKBP14 Kyphoscoliotic Ehlers-Danlos Syndrome

Giusti, Francesca, MD, PhD; Villa Donatello Hospital
- Multiple Endocrine Neoplasia Type 1

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

Familial Hyperinsulinism

Glassford, Megan, MMSc; Emory University School of Medicine
3q29 Recurrent Deletion

Glass, Ian, MBChB, MD; University of Washington
Joubert Syndrome
SHOX Deficiency Disorders

Glinton, Kevin, MD, PhD; Baylor College of Medicine
TANGO2 Deficiency

Gochuico, Bernadette R, MD; National Human Genome Research Institute
Hermansky-Pudlak Syndrome

Godfrey, Maurice, PhD; University of Nebraska Medical Center
Congenital Contractural Arachnodactyly

Gogineni, Kishore Sai, MSc; Kasturba Medical College, Manipal
LPIN2-Related Majeed Syndrome

Golabi, Mahin, MD, MPH; San Francisco General Hospital
Simpson-Golabi-Behmel Syndrome Type 1

Golas, Gretchen A, MS, RN; National Institutes of Health
Chediak-Higashi Syndrome

Gold-Von Simson, Gabrielle, BA, MD; NYU Medical Center
NTRK1 Congenital Insensitivity to Pain with Anhidrosis

Goldberg, Ethan M, MD, PhD; Children's Hospital of Philadelphia
SCN3A-Related Neurodevelopmental Disorder

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

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

Goldin, Ehud, PhD; National Institutes of Health
Mucolipidosis IV

Goldman, I David, MD; Albert Einstein College of Medicine
FOLR1-Related Cerebral Folate Transport Deficiency
Hereditary Folate Malabsorption

Goldstein, Amy, MD; Children's Hospital of Philadelphia
Mitochondrial DNA Deletion Syndromes

Goldstein, David B, PhD; Columbia University Irving Medical Center
GNB1 Encephalopathy

Goldstein, Jennifer L, MS, PhD; University of North Carolina-Chapel Hill
Glycogen Storage Disease Type I
Phosphorylase Kinase Deficiency

Gomes, Alicia, MS; University of Alabama
LZTR1- and SMARCB1-Related Schwannomatosis

Gomez-Ospina, Natalia, MD, PhD; Stanford University
Arylsulfatase A Deficiency

Gomez, Christopher M, MD, PhD; University of Chicago
Spinocerebellar Ataxia Type 6

Gonzalez-Gandolfi, Christina X; Children's Hospital of Philadelphia
Simpson-Golabi-Behmel Syndrome Type 1

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

Goodman, Stephen I, MD; University of Colorado
Glutaric Acidemia Type I

Goodnight, Scott H, MD; Oregon Health and Science University
Factor V Leiden Thrombophilia

Goodspeed, Kimberly, MD; University of Texas Southwestern Medical Center
Aspartylglucosaminuria
SLC6A1-Related Neurodevelopmental Disorder

Gordillo, Miriam, PhD; Mount Sinai School of Medicine
ESCO2 Spectrum Disorder

Gordon, Christopher T, PhD; Institut Imagine
ANKRD17-Related Neurodevelopmental Syndrome
MN1 C-Terminal Truncation Syndrome

Gordon, Kristiana, MD; St George's University Hospitals
Milroy Disease

Gordon, Leslie B, MD, PhD; Alpert Medical School of Brown University
Hutchinson-Gilford Progeria Syndrome

Gorell, Emily, DO, MS; University of Cincinnati College of Medicine
Epidermolysis Bullosa with Pyloric Atresia

Gorman, Gráinne S, MBBCh, PhD; Newcastle University
RRM2B Mitochondrial DNA Maintenance Defects

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

Gosky, Michael, BS, MS; University of Pittsburgh
LMNB1-Related Autosomal Dominant Leukodystrophy

Gospe, Sidney M, MD, PhD; University of Washington
Hypermanganesemia with Dystonia 1
Pyridoxine-Dependent Epilepsy - ALDH7A1

Gossye, Helena, MD; University of Antwerp
C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia

Gottlieb, Bruce, PhD; McGill University
Androgen Insensitivity Syndrome

Gottlob, Irene, MD; University of Leicester
FRMD7-Related Infantile Nystagmus

Goudy, Steven, MD; Vanderbilt University
IRF6-Related Disorders

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

Gouw, Launce G-C, MD, PhD; University of Utah School of Medicine
Spinocerebellar Ataxia Type 7

Govindaraj, Periyasamy, PhD; National Institute of Mental Health & Neurosciences
Huppke-Brendel Syndrome

Graham, Brett, MD, PhD; Indiana University School of Medicine
TANGO2 Deficiency

Graham, John M, MD, ScD; Cedars-Sinai Medical Center
Bohring-Opitz Syndrome
KCNK9 Imprinting Syndrome
PIK3CA-Related Overgrowth Spectrum

Graham, Rona K, PhD; University of Sherbrooke
Huntington Disease

Grand, Katheryn L, MS; Children's Hospital of Philadelphia
WDR26-Related Intellectual Disability

Grange, Dorothy Katherine, MD; Washington University School of Medicine
Cantú syndrome
Hypohidrotic Ectodermal Dysplasia

Greally, Marie T, MD, MSc; Our Lady's Children's Hospital, Crumlin
Shprintzen-Goldberg Syndrome

Greenbaum, Larry A, MD, PhD; Emory University

Hereditary Distal Renal Tubular Acidosis

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

Hereditary Paraganglioma-Pheochromocytoma Syndromes

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

Hidrotic Ectodermal Dysplasia 2

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

Phenylalanine Hydroxylase Deficiency

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

Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

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

ALK-Related Neuroblastic Tumor Susceptibility

Green, Glenn Edward, MD; University of Michigan

Branchiootorenal Spectrum Disorder

Genetic Hearing Loss Overview

Jervell and Lange-Nielsen Syndrome

Green, Peter HR, MD; Columbia University Medical Center

Celiac Disease

Gregersen, Pernille Axél, MD; Aarhus University Hospital

Type II Collagen Disorders Overview

Gregory, Allison, MS; Oregon Health & Science University

Beta-Propeller Protein-Associated Neurodegeneration

Fatty Acid Hydroxylase-Associated Neurodegeneration

MECR-Related Neurologic Disorder

Mitochondrial Membrane Protein-Associated Neurodegeneration

Neurodegeneration with Brain Iron Accumulation Disorders Overview

Pantothenate Kinase-Associated Neurodegeneration

PLA2G6-Associated Neurodegeneration

SLC39A14 Deficiency

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

DRPLA

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

Ayme-Gripp Syndrome

HRAS-Related Costello Syndrome

NOTCH3-Related Lateral Meningocele Syndrome

Grishchuk, Yulia, PhD; Harvard Medical School

Mucolipidosis IV

Groffen, Alexander J, PhD; Amsterdam University Medical Center
Long QT Syndrome Overview

Gropman, Andrea L, MD; Children's National Health System
Holoprosencephaly Overview
Smith-Magenis Syndrome
Urea Cycle Disorders Overview

Gros-Louis, François, PhD; Université Laval
SYNE1 Deficiency

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

Grzeschik, Karl-Heinz, PhD; Philipps-Universität
NSDHL-Related Disorders

Grünert, Sarah C, MD; Medical Center-University of Freiburg
Glycogen Storage Disease Type III

Guella, Ilaria, PhD; University of British Columbia
LRRK2 Parkinson Disease

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

Gunay-Aygun, Meral, MD; Johns Hopkins University School of Medicine
Alstrom Syndrome
Bardet-Biedl Syndrome Overview
Costeff Syndrome

Guo, Rose, DO; Cone Health
1q21.1 Recurrent Microdeletion

Gupta, Karn, MD; Wake Forest University School of Medicine
Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD

Gupta, Rajat, MD; Birmingham Women's and Children's Hospital
WFS1 Spectrum Disorder

Gupta, Sweta, MD; Indiana Hemophilia & Thrombosis Center
Complete Plasminogen Activator Inhibitor 1 Deficiency

Gurrola, Jose G, MD; University of Iowa
OTOF-Related Deafness

Guttmacher, Alan E, MD; National Institutes of Health
Hereditary Hemorrhagic Telangiectasia

Günes, Nilay, MD; Istanbul University
Trichorhinophalangeal Syndrome

H

Haack, Tobias, MD; Helmholtz Zentrum & Technische Universität München
Beta-Propeller Protein-Associated Neurodegeneration

Haan, J, MD, PhD; Leiden University Medical Center
CADASIL

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

Hackman, Peter, PhD; University of Helsinki
Salih Myopathy
Udd Distal Myopathy - Tibial Muscular Dystrophy

Hack, Remco, MD; Leiden University Medical Center
CADASIL

Hahn, Angelika F, MD; Western University
Cold-Induced Sweating Syndrome including Crisponi Syndrome

Haigh, Brendan, PhD; University of British Columbia
Huntington Disease

Hain, Heather S, MS, PhD; Children's Hospital of Philadelphia
22q11.2 Deletion Syndrome

Hainque, Bernard, PhD; Assistance Publique - Hôpitaux de Paris
Hypokalemic Periodic Paralysis

Hakim, Alan, BA, MA, MBBChir; Harley Street Clinic
Hypermobile Ehlers-Danlos Syndrome

Hakola, Panu, MD, PhD; Niuvanniemi Hospital
Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy

Haldeman-Englert, Chad R, MD; Fullerton Genetics Center
1q21.1 Recurrent Microdeletion
Branchiooculofacial Syndrome
Disorders of GNAS Inactivation
FGFR Craniosynostosis Syndromes Overview

Halpern, Gabrielle J, ChB, MB; Rabin Medical Center
Familial Dysautonomia
Familial Mediterranean Fever

Haltia, Matti J, MD, PhD; University of Helsinki

Polycystic Lipomembranous Osteodysplasia with Sclerosing Leukoencephalopathy

Hamdan, Fadi F, PhD; CHU Ste-Justine Research Center
SYNGAP1-Related Intellectual Disability

Hamersma, Herman, MD; Flora Clinic
SOST-Related Sclerosing Bone Dysplasias

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
Kabuki Syndrome

Hannig, Vickie L, MS; Vanderbilt Medical Center
X-Linked Spondyloepiphyseal Dysplasia Tarda

Hansen-Kiss, Emily, MA, MS; UTHealth School of Dentistry at Houston
Nonsyndromic Tooth Agenesis Overview

Hansen, C David, MD; University of Utah
Pachyonychia Congenita

Hanson, Ellen, PhD; Boston Children's Hospital
16p11.2 Recurrent Deletion

Hanson, Nancy, MS; University of Washington
Werner Syndrome

Harbison, Madeleine D, MD; Icahn School of Medicine at Mount Sinai
Silver-Russell Syndrome

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

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

TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia

Harris, Anne K, MPH; Childen's Minnesota
DICER1 Tumor Predisposition

Harris, James C, MD; Johns Hopkins University School of Medicine
HPRT1 Disorders

Harris, Peter C, PhD; Mayo Clinic
Dent Disease
Polycystic Kidney Disease, Autosomal Dominant
Primary Hyperoxaluria Type 1
Primary Hyperoxaluria Type 3

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
Rhabdoid Tumor Predisposition Syndrome

Hathaway, Evan R, MS; Children's Hospital of Philadelphia
Simpson-Golabi-Behmel Syndrome Type 1

Haviland, Isabel, MD; Boston Children's Hospital
CDKL5 Deficiency Disorder

Hayashi, Masahiro, MD, PhD; Yamagata University
Oculocutaneous Albinism Type 4

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

Hayflick, Susan J, MD; Oregon Health & Science University
Beta-Propeller Protein-Associated Neurodegeneration
Fatty Acid Hydroxylase-Associated Neurodegeneration

MECR-Related Neurologic Disorder
Mitochondrial Membrane Protein-Associated Neurodegeneration
Neurodegeneration with Brain Iron Accumulation Disorders Overview
Pantothenate Kinase-Associated Neurodegeneration
PLA2G6-Associated Neurodegeneration
SLC39A14 Deficiency

Hedera, Peter, MD, PhD; University of Louisville
Hereditary Spastic Paraplegia Overview
Spastic Paraplegia 3A

Hefner, Margaret A, MS; Saint Louis University School of Medicine
CHD7 Disorder

Hegele, Robert A, MD; Western University
Abetalipoproteinemia
APOB-Related Familial Hypobetalipoproteinemia
Chylomicron Retention Disease
Familial Combined Hypolipidemia
Familial Lipoprotein Lipase Deficiency
Tangier Disease

Hehr, Ute, MD; Center for Human Genetics
DCX-Related Disorders

Heidlebaugh, Alexis R, ScM; Geisinger Health System
CHD8-Related Neurodevelopmental Disorder with Overgrowth

Heiman, Meadow, MS; Indiana Hemophilia & Thrombosis Center
Complete Plasminogen Activator Inhibitor 1 Deficiency

Heimer, Gali, MD, PhD; Sheba Medical Center, Sackler School of Medicine
MECR-Related Neurologic Disorder
TECPR2-Related Hereditary Sensory and Autonomic Neuropathy with Intellectual Disability

Helbig, Ingo, MD; Children's Hospital of Philadelphia
CHD2-Related Neurodevelopmental Disorders

Helbig, Katherine L, MS; Children's Hospital of Philadelphia
SCN3A-Related Neurodevelopmental Disorder

Helsmoortel, Céline, MSc; University of Antwerp
ADNP-Related Disorder

He, Miao, PhD; University of Pennsylvania
Epimerase Deficiency Galactosemia

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

Peters Plus Syndrome
TBC1D24-Related Disorders
Trichorhinophalangeal Syndrome

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

Henry, Marie-Louise, MS; University of Michigan
POT1 Tumor Predisposition

Heppner, Jonathan, PhD; University of British Columbia
Mucopolysaccharidosis Type I

Herbert, Mrudu, MD, MPH; University of Kentucky
Phosphorylase Kinase Deficiency

Herman, Gail Ellen, MD, PhD; Nationwide Children's Hospital
Chondrodysplasia Punctata 2, X-Linked
X-Linked Myotubular Myopathy

Herman, Isabella, MD, PhD; Baylor College of Medicine
ENTPD1-Related Neurodevelopmental Disorder

Hermann, Andreas, MD, PhD; University Medical Center Rostock
VPS13A Disease

Hershberger, Ray E, MD; The Ohio State University
Dilated Cardiomyopathy Overview
LMNA-Related Dilated Cardiomyopathy

Hershfield, Michael, MD; Duke University Medical Center
Adenosine Deaminase Deficiency

Hershkovitz, Tova, MD; The Genetics Institute - Rambam Health Care Campus
GLYT1 Encephalopathy

Heutink, Peter, PhD; Vrije Universiteit Medical Center
MAPT-Related Frontotemporal Dementia

Hicks, Debbie, PhD; University of Newcastle upon Tyne
Collagen VI-Related Dystrophies

High, Frances A, MD, PhD; MassGeneral Hospital for Children
Congenital Diaphragmatic Hernia Overview

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

Hildebrandt, Friedhelm, MD; Boston Children's Hospital
Sphingosine Phosphate Lyase Insufficiency Syndrome

Hill, D Ashley, MD; George Washington University School of Medicine
DICER1 Tumor Predisposition

Hines, Melissa, MD; St Jude Children's Research Hospital
X-Linked Lymphoproliferative Disease

Hing, Anne V, MD; Seattle Children's Hospital
Apert Syndrome

Hingorani, Melanie, MA, MBBS, MD; Moorfields Eye Hospital
PAX6-Related Aniridia

Hipp, Heather, MD; Emory University School of Medicine
FMR1 Disorders

Hirano, Michio, MD; Columbia University Medical Center
MELAS
MERRF
Mitochondrial DNA Deletion Syndromes
Mitochondrial Neurogastrointestinal Encephalopathy Disease

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

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

Hirst, Jennifer, PhD; University of Cambridge
AP-4-Associated Hereditary Spastic Paraplegia

Hisama, Fuki M, MD; University of Washington
ADCY5 Dyskinesia
SCN9A Neuropathic Pain Syndromes
Werner Syndrome

Hisaund, Michael, BSc; University of Leicester
FRMD7-Related Infantile Nystagmus

Hoang, Stephanie, MSc; University of Alberta Hospital
Choroideremia
X-Linked Congenital Retinoschisis
X-Linked Congenital Stationary Night Blindness

Hobbs, William, MD, PhD; University of Washington
Sickle Cell Disease

Hobson, Grace M, PhD; Nemours Alfred I duPont Hospital for Children
- Pelizaeus-Merzbacher-Like Disease 1
- PLP1 Disorders

Ho, Carolyn, MD; Brigham and Women's Hospital
- Hypertrophic Cardiomyopathy Overview

Hodge, Jennelle, PhD; Cedars-Sinai Medical Center
- 17q12 Recurrent Duplication

Hoffman, Erin P, MS; Littleton Adventist Hospital
- Lysosomal Acid Lipase Deficiency

Hogarth, Penelope, MD; Oregon Health & Science University
- Beta-Propeller Protein-Associated Neurodegeneration
- MECR-Related Neurologic Disorder
- Mitochondrial Membrane Protein-Associated Neurodegeneration
- PLA2G6-Associated Neurodegeneration

Ho, Gladys; The Children's Hospital at Westmead
- MECP2 Disorders

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

Holder, J Lloyd, MD, PhD; Texas Children's Hospital
- SYNGAP1-Related Intellectual Disability

Holland, Steven M, MD; National Institute of Allergy and Infectious Diseases - NIH
- Chronic Granulomatous Disease
- STAT3 Hyper IgE Syndrome

Holm, Ida E, DMSc, MD; Aarhus University
- CHMP2B Frontotemporal Dementia

Hong, Kui, MD, PhD; Heart Institute of Nanchang University
- Brugada Syndrome

Hoogerbrugge, Nicoline, MD, PhD; Radboud University Medical Center
- NTHL1 Tumor Syndrome

Hooper, Amanda J, PhD; University of Western Australia
- Abetalipoproteinemia
- APOB-Related Familial Hypobetalipoproteinemia
- Chylomicron Retention Disease
- Familial Combined Hypolipidemia
- Familial Lipoprotein Lipase Deficiency

Tangier Disease

Hoover-Fong, Julie E, MD, PhD; Johns Hopkins University
Autosomal Dominant TRPV4 Disorders
Nail-Patella Syndrome

Hopfer, Suellen, MS, PhD; Pennsylvania State University
Primary Familial Brain Calcification

Hopkin, Robert J, MD; University of Cincinnati College of Medicine
Cytochrome P450 Oxidoreductase Deficiency

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

Hopp, Katharina, PhD; Mayo Clinic - Minnesota
Dent Disease

Horsthemke, Bernhard, PhD; University of Essen
Retinoblastoma

Horvath, Anelia, PhD; National Institutes of Health
Carney Complex

Hosoi, Yasushi, MD; Hamamatsu University School of Medicine
Aceruloplasminemia

Ho, Stephanie KL, MD; Clinical Genetic Service, Department of Health
CTNNB1 Neurodevelopmental Disorder
Strømme Syndrome

Houck, Kimberly, MD; Baylor College of Medicine
TANGO2 Deficiency

Houge, Gunnar, MD, PhD; Haukeland University Hospital
PPP2R1A-Related Neurodevelopmental Disorder

Houlden, Henry, MD, PhD; University College London
DRPLA
GAA-FGF14-Related Ataxia
GRIA2-Related Neurodevelopmental Disorder
NKX6-2-Related Disorder
RFC1 CANVAS / Spectrum Disorder
Riboflavin Transporter Deficiency
Spinocerebellar Ataxia Type 11
WARS2 Deficiency

Howard, Heidi C, PhD; Radboud University Medical Center

Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum

Howard, Vanessa C, MSN, RN; St Jude Children's Research Hospital
X-Linked Agammaglobulinemia

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

Hoyme, H Eugene, MD; Sanford Children's Hospital
Hyaline Fibromatosis Syndrome

Hoyos-Martinez, Alfonso, MD; Baylor College of Medicine
TANGO2 Deficiency

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

Hsu, Amy P; National Institute of Allergy and Infectious Diseases
STAT3 Hyper IgE Syndrome

Hudgins, Louanne, MD; Stanford University
Kabuki Syndrome

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

Hufnagel, Robert B, MD, PhD; National Eye Institute - NIH
PNPLA6 Disorders

Hughes, Derralynn A, DPhil, MA; University College Medical School
Fabry Disease
Gaucher Disease

Huizing, Marjan, PhD; National Human Genome Research Institute
Costeff Syndrome
GNE Myopathy
Hermansky-Pudlak Syndrome

Hull, Judith L, MS; Memorial Sloan-Kettering Cancer Center
BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Hull, Peter R, MD, PhD; Dalhousie University
Pachyonychia Congenita

Hulton, Sally-Anne, MD; Birmingham Children's Hospital
Primary Hyperoxaluria Type 2

Hume, Stacey, PhD; University of Alberta
Choroideremia

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

CDC73-Related Disorders

Hundley, Rachel, PhD; Boston Children's Hospital
16p11.2 Recurrent Deletion

Hunt, David, MBBS, PhD; Princess Anne Hospital
PURA-Related Neurodevelopmental Disorders

Hunter, Alasdair GW, MD; Children's Hospital of Eastern Ontario
RPS6KA3-Related Intellectual Disability

Hunter, David G, MD, PhD; Children's Hospital Boston
Congenital Fibrosis of the Extraocular Muscles Overview
Duane Syndrome

Hunter, Elizabeth, BS; UTHealth School of Dentistry at Houston
Nonsyndromic Tooth Agenesis Overview

Hunter, Jessica Ezzell, PhD; Kaiser Permanente Northwest
FMR1 Disorders

Hunter, JM, PhD; Nationwide Children's Hospital
Spinal Muscular Atrophy, X-Linked Infantile

Huntsman, David G, MD; University of British Columbia
Hereditary Diffuse Gastric Cancer

Hupertz, Vera, MD; Cleveland Clinic
Alpha-1 Antitrypsin Deficiency

Huq, Mahbubul, MD, PhD; University of British Columbia
Huntington Disease

Hurley, R Morrison, MD; British Columbia's Children's and Women's Hospital
Primary Hyperoxaluria Type 1

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

Huryn, Laryssa A, MD; National Eye Institute
DICER1 Tumor Predisposition

Huston, Haley, BS; Bloodworks Northwest
Hemophilia A
Hemophilia B

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

Hyde, Samuel M, MMSc; University of Texas MD Anderson Cancer Center
CDC73-Related Disorders

Häberle, Johannes, MD; University Children's Hospital Zurich
Carbonic Anhydrase VA Deficiency

I

Iacovazzo, Donato, MD; Queen Mary University of London
X-Linked Acrogigantism

Idkowiak, Jan, MD, PhD; University of Birmingham
Cytochrome P450 Oxidoreductase Deficiency

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

Ikeda, Marc D, MD; Children's Hospital of Pittsburgh
ZAP70-Related Combined Immunodeficiency

Ikeda, Shu-ichi, MD, PhD; Shinshu University Hospital
Hereditary Transthyretin Amyloidosis

Ikeda, Yoshio, MD, PhD; Institute of Genetic Medicine
Spinocerebellar Ataxia Type 8

Indo, Yasuhiro, MD, PhD; Kumamoto University Hospital
NTRK1 Congenital Insensitivity to Pain with Anhidrosis

Infante, Elena, MS; University of Pittsburgh
MUTYH Polyposis

Innes, A Micheil, BSc, MD; University of Calgary
Au-Kline Syndrome
SHORT Syndrome
THOC6 Intellectual Disability Syndrome
TRPM3-Related Neurodevelopmental Disorder

Innis, Jeffrey W, MD, PhD; University of Michigan
Hand-Foot-Genital Syndrome

Introne, Wendy J, MD; National Human Genome Research Institute
Alkaptonuria
Chediak-Higashi Syndrome
Hermansky-Pudlak Syndrome

Irons, Mira, MD; Children's Hospital Boston
Smith-Lemli-Opitz Syndrome

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

Isaacs, Adrian M, DPhil; University College London

CHMP2B Frontotemporal Dementia

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

Ito, Daisuke, MD, PhD; Keio University
BSCL2-Related Neurologic Disorders / Seipinopathy

Iwasa, Yoichiro, MD, PhD; University of Iowa
Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

J

Jabs, Ethylin Wang, MD; Icahn School of Medicine at Mount Sinai
ESCO2 Spectrum Disorder
STAC3 Disorder
Treacher Collins Syndrome

Jackson, Andrew, PhD; University of Edinburgh
Microcephalic Osteodysplastic Primordial Dwarfism Type II
RNU4atac-opathy

Jackson, Michelle A, MS; University of Texas MD Anderson Cancer Center
CDC73-Related Disorders

James, Aaron; University of California San Francisco
Simpson-Golabi-Behmel Syndrome Type 1

James, Paula, MD; Queen's University
von Willebrand Disease

Jankovic, Joseph, MD; Baylor College of Medicine
NKX2-1-Related Disorders

Jansen, Anna C, MD, PhD; Universitair Ziekenhuis Brussel
PAFAH1B1-Related Lissencephaly / Subcortical Band Heterotopia
Progressive Myoclonus Epilepsy, Lafora Type

Janssens, Veerle, PhD; KU Leuven
PPP2R1A-Related Neurodevelopmental Disorder

Jasperson, Kory W; University of Utah
APC-Associated Polyposis Conditions

Jeffery, Steve, PhD; St George's University of London
Lymphedema-Distichiasis Syndrome
Milroy Disease

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

Familial Hemiplegic Migraine

Jeste, Shafali Spurling, MD; Children's Hospital Los Angeles
Maternal 15q Duplication Syndrome

Jethva, Reena, MD; St Christopher's Hospital for Children
Short-Chain Acyl-CoA Dehydrogenase Deficiency

Jewett, Tamison, MD; Wake Forest University School of Medicine
1q21.1 Recurrent Microdeletion

Jinnah, Hyder A, MD, PhD; Emory University
HPRT1 Disorders

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

Johannesen, Katrine, MD, PhD; Rigshospitalet
SLC6A1-Related Neurodevelopmental Disorder

Johann, Pascal, MD; German Cancer Consortium (DKTK)
Rhabdoid Tumor Predisposition Syndrome

Johannsen, Peter, MD, PhD; Rigshospitalet
CHMP2B Frontotemporal Dementia

Johnson-Kerner, Bethany, MD, PhD; University of California, San Francisco
DDX3X-Related Neurodevelopmental Disorder

Johnson, Eric W, PhD; Barrow Neurological Institute
Familial Cerebral Cavernous Malformation

Johnson, Judith, MS; Cincinnati Children's Hospital Medical Center
Autoimmune Lymphoproliferative Syndrome
Familial Hemophagocytic Lymphohistiocytosis
WAS-Related Disorders
X-Linked Hyper IgM Syndrome
X-Linked Lymphoproliferative Disease

Johnson, Kyle P, MD; Oregon Health and Science University
Smith-Magenis Syndrome

Johnson, Maribel J, MA, RN; Puget Sound Blood Center
Hemophilia A
Hemophilia B

Johnson, Rebecca H, MD; Seattle Children's Hospital
ALK-Related Neuroblastic Tumor Susceptibility

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

Greig Cephalopolysyndactyly Syndrome

Jones, Mary-Kayt N, BA; University of Iowa Hospitals and Clinics
GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss

Jordan, Elizabeth, MMSc; The Ohio State University
Dilated Cardiomyopathy Overview
LMNA-Related Dilated Cardiomyopathy

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

Josephson, Neil C, MD; Seattle Genetics
Hemophilia A
Hemophilia B

Joshi, Nishtha; Baylor College of Medicine
KCNQ2-Related Disorders
KCNQ3-Related Disorders

Jungbluth, Heinz, MD, PhD; King's College London
DYNC1H1-Related Disorders
EPG5-Related Disorder

Jung, Hans H, MD; University Hospital Zurich
McLeod Neuroacanthocytosis Syndrome

Jurgens, Julie A, PhD; Boston Children's Hospital
Congenital Fibrosis of the Extraocular Muscles Overview

Jurkat-Rott, Karin, MD, PhD; Ulm University
Hyperkalemic Periodic Paralysis

Jägle, Herbert, MD; University of Regensburg
Achromatopsia

K

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

Kacena, Melissa A, PhD; Indiana University School of Medicine
GATA1-Related Cytopenia

Kahle, Kristopher T, MD, PhD; Massachusetts General Hospital
Muenke Syndrome
Pseudohypoaldosteronism Type II

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

GBE1 Adult Polyglucosan Body Disease

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

CTDP1-Related Congenital Cataracts, Facial Dysmorphism, and Neuropathy

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

ATP7A-Related Copper Transport Disorders

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

EPB42-Related Hereditary Spherocytosis

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

Beckwith-Wiedemann Syndrome

Simpson-Golabi-Behmel Syndrome Type 1

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

CLCN4-Related Neurodevelopmental Disorder

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

Alagille Syndrome

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

PLP1 Disorders

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

DICER1 Tumor Predisposition

FH Tumor Predisposition Syndrome

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

ARSACS

Ataxia-Telangiectasia

Kang, Jingqiong, PhD; Vanderbilt University Medical Center

SLC6A1-Related Neurodevelopmental Disorder

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

Cherubism

Kantarci, Sibel, PhD; Quest Diagnostics Nichols Institute

Donnai-Barrow Syndrome

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

Periodontal Ehlers-Danlos Syndrome

Kaplan, Julie D, MD; Cleveland Clinic Foundation

MYRF-Related Cardiac Urogenital Syndrome

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

Cystic Fibrosis

Thanatophoric Dysplasia

Karpati, George, MD; Montreal Neurological Institute
GNE Myopathy

Kashtan, Clifford E, MD; University of Minnesota
Alport Syndrome

Kaspar, Roger L, PhD; Transderm, Inc
Pachyonychia Congenita

Katsanis, Sara Huston, MS; Northwestern University
Treacher Collins Syndrome

Kaurah, Pardeep, MSc, PhD; University of British Columbia
Hereditary Diffuse Gastric Cancer

Kaur, Parneet, MSc; Manipal Academy of Higher Education
ISCA1-Related Multiple Mitochondrial Dysfunctions Syndrome

Kaur, Simranpreet, M Phil, MSc; University of Melbourne
MECP2 Disorders

Kaya, Namik, PhD; King Faisal Specialist Hospital & Research Center
ISCA2-Related Mitochondrial Disorder
NKX6-2-Related Disorder

Kaylor, Julie, MS; Arkansas Children's Hospital
SATB2-Associated Syndrome

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

Keats, Bronya JB, PhD; Louisiana State University Health Sciences Center
Usher Syndrome Type I
Usher Syndrome Type II

Keegan, Catherine E, MD, PhD; University of Michigan
Nonsyndromic Disorders of Testicular Development Overview

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

Kelley, Philip M, PhD; Boys Town National Research Hospital
OTOF-Related Deafness

Kelly, Edward J, PhD; University of Washington
Bietti Crystalline Dystrophy

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

Keppler-Noreuil, Kim, MD; University of Wisconsin School of Medicine and Public Health
- Cranioectodermal Dysplasia
- PIK3CA-Related Overgrowth Spectrum

Kerrigan, John F, MD; Phoenix Children's Hospital
- Fumarate Hydratase Deficiency

Khaikin, Yannay, BSc; The Hospital for Sick Children
- STXBP1 Encephalopathy with Epilepsy

Khan, Sadiya S, MD; Northwestern University Feinberg School of Medicine
- Complete Plasminogen Activator Inhibitor 1 Deficiency

Khaytin, Ilya, MD, PhD; Northwestern University
- Congenital Central Hypoventilation Syndrome

Kibrom, Sara, MD; Stanford University School of Medicine
- SCARB2-Related Action Myoclonus – Renal Failure Syndrome

Kidd, Kendrah, MS; Wake Forest University School of Medicine
- Autosomal Dominant Tubulointerstitial Kidney Disease -- MUC1
- Autosomal Dominant Tubulointerstitial Kidney Disease -- REN
- Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD

Kimberling, William J, PhD; Boys Town National Research Hospital
- Usher Syndrome Type I
- Usher Syndrome Type II

Kimonis, Virginia, MD; University of California Irvine Medical Center
- Inclusion Body Myopathy with Paget Disease of Bone and/or Frontotemporal Dementia

Kinsley, Lisa, MS; Northwestern University Feinberg School of Medicine
- Amyotrophic Lateral Sclerosis Overview

Kirk, Jessica, BS; Yale University School of Medicine
- GATA1-Related Cytopenia

Kirmani, Salman, MBBS; Aga Khan University
- Hereditary Paraganglioma-Pheochromocytoma Syndromes

Kirschner, Lawrence, MD, PhD; The Ohio State University Comprehensive Cancer Center
- Multiple Endocrine Neoplasia Type 4

Kishnani, Priya S, MD; Duke University Medical Center
- Glycogen Storage Disease Type I
- Phosphorylase Kinase Deficiency

Kish, Stephen, PhD; University of Toronto
- Tyrosine Hydroxylase Deficiency

Klaiman, Cheryl, PhD; Emory University School of Medicine
- 3q29 Recurrent Deletion

Klar, Aharon, MD; Hebrew University
- INSR-Related Severe Insulin Resistance Syndrome

Klasson, Timothy D, BSc; University Medical Center Utrecht
- Von Hippel-Lindau Syndrome

Kleefstra, Tjitske, MD, PhD; Radboud University Medical Center
- DDX3X-Related Neurodevelopmental Disorder
- Kleefstra Syndrome

Klein-Tasman, Bonita P, PhD; University of Wisconsin-Milwaukee
- 7q11.23 Duplication Syndrome

Klein, Christine, MD; University of Lübeck
- Hereditary Dystonia Overview
- Parkin Type of Early-Onset Parkinson Disease
- PINK1 Type of Young-Onset Parkinson Disease
- PRRT2-Associated Paroxysmal Movement Disorders

Klein, Christopher J, MD; Mayo Clinic
- DNMT1-Related Disorder
- GBE1 Adult Polyglucosan Body Disease

Klein, Pierre, MSc; Université Pierre et Marie Curie
- Oculopharyngeal Muscular Dystrophy

Klein, Roger D, JD, MD; Cleveland Clinic
- CEBPA-Associated Familial Acute Myeloid Leukemia (AML)
- Hereditary Paraganglioma-Pheochromocytoma Syndromes

Klein, Steven D, MD, PhD; Children's Hospital of Philadelphia
- Simpson-Golabi-Behmel Syndrome Type 1

Kleta, Robert, MD, PhD; National Institutes of Health
- Cystinosis
- Free Sialic Acid Storage Disorders

Kline, Antonie D, MD; Greater Baltimore Medical Center
- Au-Kline Syndrome

Klopstock, Thomas, MD; University Hospital, LMU Munich
- Mitochondrial Membrane Protein-Associated Neurodegeneration

Kmiec, Tomasz, MD; Children's Memorial Health Institute
- Mitochondrial Membrane Protein-Associated Neurodegeneration

Kmoch, Stanislav, PhD; Charles University
- Autosomal Dominant Tubulointerstitial Kidney Disease -- MUC1
- Autosomal Dominant Tubulointerstitial Kidney Disease -- REN
- Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD
- Rotor Syndrome

Knappskog, Per Morten, PhD; Haukeland University Hospital
- ADAMTSL4-Related Eye Disorders
- Cold-Induced Sweating Syndrome including Crisponi Syndrome
- Congenital Stromal Corneal Dystrophy

Knight Johnson, Amy, MS; University of Chicago
- Maturity-Onset Diabetes of the Young Overview

Knisely, AS, MD; Medizinische Universität Graz
- ATP8B1 Deficiency
- Rotor Syndrome

Knoers, Nine, MD, PhD; University Medical Center Groningen
- Cranioectodermal Dysplasia
- Hereditary Nephrogenic Diabetes Insipidus
- Nephronophthisis-Related Ciliopathies

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

Knowles, Michael R, MD; University of North Carolina at Chapel Hill
- Primary Ciliary Dyskinesia

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

Koenekoop, Robert K, MD, PhD; McGill University Health Center
- Usher Syndrome Type I
- Usher Syndrome Type II

Koene, Saskia, MD, PhD; Leiden University Medical Center
- FOXP1 Syndrome

Koenig, Mary Kay, MD; McGovern Medical School
- Tuberous Sclerosis Complex

Koenig, Michel, MD, PhD; Université de Montpellier
- Ataxia with Oculomotor Apraxia Type 2

Kohlhase, Jürgen, MD; SYNLAB Center for Human Genetics
- SALL4-Related Disorders

Townes-Brocks Syndrome

Kohlmann, Wendy, MS; University of Michigan Health System
Lynch Syndrome

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

Kolevzon, Alexander, MD; Icahn School of Medicine at Mount Sinai
FOXP1 Syndrome

Konkle, Barbara A, MD; University of Washington
Hemophilia A
Hemophilia B

Konno, Takuya, MD, PhD; Niigata University
DCTN1-Related Neurodegeneration

Koolen, David A, MD, PhD; Radboud University Medical Center
Koolen-de Vries Syndrome

Kool, Marcel, PhD; German Cancer Consortium (DKTK)
Rhabdoid Tumor Predisposition Syndrome

Kooy, Frank, PhD; University of Antwerp
ADNP-Related Disorder

Korbonits, Márta, MD, PhD; Queen Mary University of London
AIP Familial Isolated Pituitary Adenomas
X-Linked Acrogigantism

Kordes, Uwe, MD; University Medical Center Hamburg – Eppendorf
Rhabdoid Tumor Predisposition Syndrome

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

Kornak, Uwe, MD, PhD; Universitätsmedizin Göttingen
ATP6V0A2-Related Cutis Laxa
CLCN7-Related Osteopetrosis

Koskiniemi, Marja-Leena, MD, PhD; University of Helsinki
Progressive Myoclonic Epilepsy Type 1

Kostic, Ana, PhD; Icahn School of Medicine at Mount Sinai
FOXP1 Syndrome

Kowdley, Kris V, MD; Virginia Mason Medical Center
HFE-Related Hemochromatosis

Koy, Anne, MD; University of Cologne

GNAO1-Related Disorder

Kraemer, Kenneth H, MD; National Cancer Institute
Xeroderma Pigmentosum

Krahn, Katherine Nash, MS; University of Iowa
IRF6-Related Disorders

Krajewski, Karen, MS; Wayne State University School of Medicine
PLP1 Disorders

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

Krantz, Ian D, MD; The Children's Hospital of Philadelphia
Alagille Syndrome
Cornelia de Lange Syndrome

Krasnewich, Donna M, MD, PhD; National Institutes of Health
Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview
PMM2-CDG

Krause, Amanda, MBBCh, PhD; University of the Witwatersrand
Huntington Disease-Like 2

Krey, Ilona, MD; Univeristy of Leipzig Medical Center
GRIN2D-Related Developmental and Epileptic Encephalopathy

Kriek, Marjolein, MD, PhD; Leiden University Medical Center
Peters Plus Syndrome

Kronn, David F, MD; New York Medical College
Hereditary Folate Malabsorption

Kruer, Michael C, MD; University of South Dakota Sanford School of Medicine
Fatty Acid Hydroxylase-Associated Neurodegeneration

Kruszka, Paul, MD, MPH; GeneDx
Holoprosencephaly Overview
Muenke Syndrome
Weiss-Kruszka Syndrome

Kucine, Nicole, MD, MS; Weill Cornell Medicine
Bloom Syndrome

Kuhlenbäumer, Gregor, MD, PhD; University of Kiel
GAN-Related Neurodegeneration

Kuiper, Roland P, PhD; Princess Máxima Center for Pediatric Oncology
NTHL1 Tumor Syndrome

Kujovich, Jody L, MD; Oregon Health and Science University
- Factor V Leiden Thrombophilia
- Prothrombin Thrombophilia

Kumar, Ajith V, MD; Great Ormond Street Hospital
- AIP Familial Isolated Pituitary Adenomas

Kumar, Anil; University of Leicester
- FRMD7-Related Infantile Nystagmus

Kumaran, Neruban, BSc, MBBS; University College London
- Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview

Kumble, Smitha, MBBS; Murdoch Children's Research Institute
- Chondrodysplasia Punctata 2, X-Linked

Kupfer, Gary, MD; Yale University School of Medicine
- Fanconi Anemia

Kurahashi, Hirokazu, MD, PhD; Aichi Medical University
- Autosomal Dominant Sleep-Related Hypermotor (Hyperkinetic) Epilepsy

Kurian, Manju Ann, MBBChir, PhD; UCL Great Ormond Street Institute of Child Health
- Aromatic L-Amino Acid Decarboxylase Deficiency
- Beta-Propeller Protein-Associated Neurodegeneration
- DNAJC6 Parkinson Disease
- KMT2B-Related Dystonia
- PLA2G6-Associated Neurodegeneration
- SLC12A5-Related Epilepsy of Infancy with Migrating Focal Seizures
- SLC39A14 Deficiency
- SLC6A3-Related Dopamine Transported Deficiency Syndrome

Kurolap, Alina, MSc, RN; The Genetics Institute - Rambam Health Care Campus
- GLYT1 Encephalopathy

Kurth, Ingo, MD; Uniklinik RWTH Aachen
- Hereditary Sensory and Autonomic Neuropathy Type II

Kurtz-Nelson, Evangeline C, PhD; University of Washington
- CHD8-Related Neurodevelopmental Disorder with Overgrowth

Kurtzman, Tracey L; University of Arizona Colleges of Medicine and Science
- Smith-Lemli-Opitz Syndrome

Kushner, Jessica D, MS; Oregon Health and Science University
- Dilated Cardiomyopathy Overview

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

Snyder-Robinson Syndrome

Kutsche, Kerstin, PhD; University Medical Center Hamburg-Eppendorf
CASK Disorders
EBF3 Neurodevelopmental Disorder

Kälviäinen, Reetta, MD, PhD; Kuopio University Hospital
Progressive Myoclonic Epilepsy Type 1

Kölker, Stefan, MD; University Hospital Heidelberg
Classic Isovaleric Acidemia

König, Arne, MD; Philipps-Universität
NSDHL-Related Disorders

Küry, Sébastien, DVM, PhD; Service de Génétique médicale – CHU
Hereditary Fibrosing Poikiloderma with Tendon Contractures, Myopathy, and Pulmonary Fibrosis

L

La Spada, Albert R, MD, PhD; University of California Irvine School of Medicine
Spinal and Bulbar Muscular Atrophy
Spinocerebellar Ataxia Type 7

Labrador, Emma, RN; Connecticut Children's Medical Center
Glycogen Storage Disease Type VI

Lacau St Guily, Jean, MD; Centre de Recherche en Myologie
Oculopharyngeal Muscular Dystrophy

Lacbawan, Felicitas L, MD; Cleveland Clinic
Alpha-1 Antitrypsin Deficiency

Lachlan, Katherine, MD; Southampton University
CHD4 Neurodevelopmental Disorder

Lai, Jennifer, MD; University of California San Francisco
Hereditary Coproporphyria

Laing, Nigel G, PhD; University of Western Australia
Laing Distal Myopathy

Lai, Yi-Chen, MD; Baylor College of Medicine
TANGO2 Deficiency

Laje, Gonzalo, MD; National Institute of Mental Health
Smith-Magenis Syndrome

Lalani, Seema R, MD; Baylor College of Medicine
CHD7 Disorder

TANGO2 Deficiency

Lal, Dennis, PhD; Cleveland Clinic
SLC6A1-Related Neurodevelopmental Disorder

Lam, Christina, MD; University of Washington
NGLY1-Related Congenital Disorder of Deglycosylation
PMM2-CDG

Lamont, Phillipa, PhD; Royal Perth Hospital
Laing Distal Myopathy

Lampe, Anne Katrin, MD; Western General Hospital
Collagen VI-Related Dystrophies

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

Lange, Dirk, PhD; University of British Columbia
Primary Hyperoxaluria Type 1

Lange, Lara M, MD; University of Lübeck
PINK1 Type of Young-Onset Parkinson Disease

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

Langer, Katherine, BA; Weill Cornell Medicine
Bloom Syndrome

Lanpher, Brendan C, MD; Mayo Clinic
Urea Cycle Disorders Overview

Larizza, Lidia, MD; IRCSS Istituto Auxologico Italiano
Baller-Gerold Syndrome
Poikiloderma with Neutropenia

Larkin, Allyson, MD; Children's Hospital of Pittsburgh
ZAP70-Related Combined Immunodeficiency

Larsen Haidle, Joy, MS; North Memorial Hospital
Juvenile Polyposis Syndrome

Larson, Austin, MD; University of Colorado
Glutaric Acidemia Type I

LaRusch, Jessica, PhD; Ariel Precision Medicine
Pancreatitis Overview
PRSS1-Related Hereditary Pancreatitis

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

Maternal 15q Duplication Syndrome

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

Laurent, Michaël R, MD, PhD; University Hospitals Leuven
X-Linked Hypophosphatemia

Law, Linda, MBA, MD; Advicenne
Hereditary Distal Renal Tubular Acidosis

Lawlor, Michael W, MD, PhD; Medical College of Wisconsin
X-Linked Myotubular Myopathy

Lawrence, Jessica, MSc; Alberta Health Services
Bestrophinopathies

Layman, Lawrence C, MD; Medical College of Georgia
PROP1-Related Combined Pituitary Hormone Deficiency

Le Goff, Carine, PhD; INSERM U781
Geleophysic Dysplasia

Leachman, Sancy A, MD, PhD; University of Utah
Pachyonychia Congenita

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

LeBlanc, Shannon, MBBS; University of Melbourne
Pycnodysostosis

Lebwohl, Benjamin, MD, MS; Columbia University
Celiac Disease

Ledbetter, David H, PhD; Geisinger Health System
17q12 Recurrent Deletion Syndrome
CHD8-Related Neurodevelopmental Disorder with Overgrowth

Lee, Bo Hoon, MD; University of Rochester Medical Center
PURA-Related Neurodevelopmental Disorders

Lee, Brendan H, MD, PhD; Baylor College of Medicine
Argininosuccinate Lyase Deficiency
Cleidocranial Dysplasia Spectrum Disorder
KAT6B Disorders

Lee, Chung U, MD; Stanford University
Carnitine-Acylcarnitine Translocase Deficiency
MERRF

Lee, Kristen N, MD; University of Michigan
Citrullinemia Type I

Lee, Mianne, MSc; Univeristy of Hong Kong
CTNNB1 Neurodevelopmental Disorder
MN1 C-Terminal Truncation Syndrome

Lee, Soohee, PhD; The New York Blood Center
McLeod Neuroacanthocytosis Syndrome

Lee, Thomas, MD, MSc; University of Ottawa
Bestrophinopathies

Legare, Janet M, MD; University of Wisconsin
Achondroplasia

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

LeGuern, Eric, MD, PhD; Hôpital de la Pitié-Salpêtrière
SH3TC2-Related Hereditary Motor and Sensory Neuropathy

Lehesjoki, Anna-Elina, MD, PhD; University of Helsinki
Marinesco-Sjogren Syndrome
Progressive Myoclonic Epilepsy Type 1

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

Lehman, Christopher, MS; Geisinger Health System
16p11.2 Recurrent Deletion

Lehmann-Horn, Frank, MD, MS, PhD; Ulm University
Hyperkalemic Periodic Paralysis
Hypokalemic Periodic Paralysis

Lehmberg, Kai, MD; University Medical Center Eppendorf
Familial Hemophagocytic Lymphohistiocytosis

Leiding, Jennifer W, MD; Johns Hopkins University
Chronic Granulomatous Disease

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

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

Lemire, Gabrielle, MD; Children's Hospital of Eastern Ontario Research Institute
KAT6B Disorders

THOC6 Intellectual Disability Syndrome

Lemke, Johannes R, MD; University of Leipzig Medical Center
GRIN1-Related Neurodevelopmental Disorder
GRIN2B-Related Neurodevelopmental Disorder
GRIN2D-Related Developmental and Epileptic Encephalopathy

Lemmers, Richard JLF, PhD; Leiden University Medical Center
Facioscapulohumeral Muscular Dystrophy

Lemmink, Henny, PhD; University Medical Center
Hereditary Nephrogenic Diabetes Insipidus

Lentz, Jennifer J, PhD; Louisiana State University Health Sciences Center
Usher Syndrome Type I
Usher Syndrome Type II

Leonard, Helen, MBChB, MPH; Telethon Kids Institute
CDKL5 Deficiency Disorder

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

Leroy, Bart P, MD, PhD; Ghent University Hospital & Ghent University
Adult Refsum Disease

Leroy, Jules G, MD, PhD; Ghent University Hospital
GNPTAB-Related Disorders

Leslie, Elizabeth J, PhD; Emory University
IRF6-Related Disorders

Leslie, Nancy D, MD; Cincinnati Children's Hospital Medical Center
Pompe Disease
Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Lesnik Oberstein, Saskia AJ, MD, PhD; Leiden University Medical Center
CADASIL
Peters Plus Syndrome

Letra, Ariadne, DDS, MS, PhD; UTHealth School of Dentistry at Houston
Nonsyndromic Tooth Agenesis Overview

Leturcq, France, MD; Hôpital Cochin
Emery-Dreifuss Muscular Dystrophy

Leung, Alva, BS; California Pacific Medical Center
Simpson-Golabi-Behmel Syndrome Type 1

Leung, Lai Ting, MD; Clinical Genetic Service, Department of Health

Strømme Syndrome

Leuzzi, Vincenzo, MD; Sapienza Università di Roma
PRICKLE1-Related Disorders

Levade, Thierry, MD, PhD; Centre de Recherches en Cancérologie de Toulouse
ASAH1-Related Disorders

Leventer, Richard J, BMedSci, MBBS, PhD; Royal Children's Hospital
PURA-Related Neurodevelopmental Disorders

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

Levy-Lahad, Ephrat, MD; Sharre Zedek Medical Center
BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Levy, Amanda M, MSc; Rigshospitalet
DLG4-Related Synaptopathy

Levy, Harvey L, MD; Boston Children's Hospital
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency

Levy, Howard P, MD, PhD; Johns Hopkins University School of Medicine
Hypermobile Ehlers-Danlos Syndrome

Levy, Rebecca V, BM BCh, MSc; Montefiore Medical Center
17q12 Recurrent Deletion Syndrome

Lewandowska, Magdalena, MD; Indiana Hemophilia & Thrombosis Center
Complete Plasminogen Activator Inhibitor 1 Deficiency

Lewis, David B, MD; Stanford University School of Medicine
Schimke Immunoosseous Dysplasia

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

Lichter-Konecki, Uta, MD, PhD; UPMC Children's Hospital of Pittsburgh
Ornithine Transcarbamylase Deficiency
Urea Cycle Disorders Overview

Li, Chumei, MD, PhD; McMaster University
FREM1 Autosomal Recessive Disorders

Liebau, Max, MD; University of Cologne
Autosomal Recessive Polycystic Kidney Disease – PKHD1

Lieske, John C, MD; Mayo Clinic
Dent Disease
Primary Hyperoxaluria Type 1

Primary Hyperoxaluria Type 3

Li, Frederick, MD; Dana Farber Cancer Institute
Li-Fraumeni Syndrome

Li, Hong, MD, PhD; Emory University School of Medicine
CTCF-Related Disorder

Lilien, Marc, MD, PhD; University Medical Center Utrecht
Nephronophthisis-Related Ciliopathies

Li, Longchuan, PhD; Emory University School of Medicine
3q29 Recurrent Deletion

Lim, Albert Z, MBBS (Hons); Newcastle University
RRM2B Mitochondrial DNA Maintenance Defects

Lin, Angela E, MD; MassGeneral Hospital for Children
Branchiooculofacial Syndrome
Cranioectodermal Dysplasia
HRAS-Related Costello Syndrome
MN1 C-Terminal Truncation Syndrome
Myhre Syndrome

Lindhurst, Marjorie J, PhD; National Institutes of Health
SLC25A19-Related Thiamine Metabolism Dysfunction

Lindor, Noralane M, MD; Mayo Clinic
Myhre Syndrome

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

Lines, Matthew, MD, MSc; University of Calgary
Mandibulofacial Dysostosis with Microcephaly
TRPM3-Related Neurodevelopmental Disorder

Links, Thera P, MD, PhD; University Medical Center Groningen
Von Hippel-Lindau Syndrome

Lin, Xi, MD, PhD; University of Texas Medical Branch
Spinocerebellar Ataxia Type 1

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

Lipska-Zietkiewicz, Beata S, MD, PhD; Medical University of Gdansk
Genetic Steroid-Resistant Nephrotic Syndrome Overview
WT1 Disorder

Liu, Lawrence U, MD; Icahn School of Medicine at Mount Sinai
- Familial Porphyria Cutanea Tarda
- Hepatoerythropoietic Porphyria

Liu, Paul, MD, PhD; National Human Genome Research Institute
- RUNX1 Familial Platelet Disorder with Associated Myeloid Malignancies

Lloyd, Ricardo V, MD, PhD; Mayo Clinic
- Hereditary Paraganglioma-Pheochromocytoma Syndromes

Lochmüller, Hanns, MD; Children's Hospital of Eastern Ontario Research Institute
- Congenital Myasthenic Syndromes Overview

Loeys, Bart L, MD, PhD; Antwerp University Hospital
- EFEMP2-Related Cutis Laxa
- FBLN5-Related Cutis Laxa
- Loeys-Dietz Syndrome

Lohmann, Dietmar R, MD; University Hospital Essen
- Retinoblastoma

Lohmann, Katja, PhD; University of Lübeck
- Hereditary Dystonia Overview

Lohr, Jamie, MD; University of Minnesota Amplatz Children's Hospital
- Autosomal Dominant Robinow Syndrome

Lo, Ivan FM, MD; Clinical Genetic Service, Department of Health
- CTNNB1 Neurodevelopmental Disorder
- FAM111A-Related Skeletal Dysplasias
- Strømme Syndrome

Longoni, Mauro, MD; Harvard Medical School
- Congenital Diaphragmatic Hernia Overview
- Donnai-Barrow Syndrome

Loomes, Kathleen M, MD; University of Pennsylvania
- Alagille Syndrome

Lopez, Christina, BS; California Pacific Medical Center
- Simpson-Golabi-Behmel Syndrome Type 1

LoPiccolo, Mary Kate, MD; Icahn School of Medicine at Mount Sinai
- Long-Chain Hydroxyacyl-CoA Dehydrogenase Deficiency / Trifunctional Protein Deficiency

Lory, Philippe, PhD; Université de Montpellier 1 et 2
- Hypokalemic Periodic Paralysis

Lossie, Amy C; University of Florida College of Medicine

Angelman Syndrome

Lossos, Alexander, MD; Hadassah-Hebrew University Medical Center
GBE1 Adult Polyglucosan Body Disease

Lotze, Timothy, MD; Baylor College of Medicine
GARS-Associated Axonal Neuropathy

Louie, Raymond J, PhD; Greenwood Genetic Center
IPEX Syndrome

Lou, Shu, MD; University of British Columbia
Schimke Immunoosseous Dysplasia

Loyd, James E, MD; Vanderbilt University Medical Center
Heritable Pulmonary Arterial Hypertension Overview

Lubarr, Naomi, MD; Beth Israel Medical Center
DYT1 Early-Onset Isolated Dystonia

Luchsinger, Kadi, BS; Dup15q Alliance
Maternal 15q Duplication Syndrome

Lucia, Alejandro, MD, PhD; European University of Madrid
Glycogen Storage Disease Type V

Lucking, Christoph, MD; Ludwig-Maximilians-University
Parkin Type of Early-Onset Parkinson Disease

Lucky, Anne W, MD; Cincinnati Children's Hospital
Dystrophic Epidermolysis Bullosa
Epidermolysis Bullosa Simplex
Epidermolysis Bullosa with Pyloric Atresia
Junctional Epidermolysis Bullosa

Luk, Ho-ming, MD; Hong Kong Children's Hospital
CTNNB1 Neurodevelopmental Disorder
FAM111A-Related Skeletal Dysplasias
Strømme Syndrome

Lupski, James R, MD, PhD; Baylor College of Medicine
Smith-Magenis Syndrome

Lusk, Laina, MMSc; Children's Hospital of Philadelphia
Maternal 15q Duplication Syndrome
PACS1 Neurodevelopmental Disorder

Lyman, Benjamin, DO; LSU Health Science Center
Cystic Fibrosis

Lynch, Henry, MD; Creighton University Hereditary Cancer Center
MUTYH Polyposis

Lyons, Michael J, MD; Greenwood Genetic Center
MED12-Related Disorders

Lücke, Thomas, MD, PhD; Hannover Medical School
Schimke Immunoosseous Dysplasia

Lüdecke, Hermann-Josef, PhD; Universitätsklinikum Düsseldorf
TXNL4A-Related Craniofacial Disorders

M

Maas, Saskia, MD; University of Amsterdam
Trichorhinophalangeal Syndrome

Maccollin, Mia M, MD; Harvard Medical School/Massachusetts General Hospital
NF2-Related Schwannomatosis

Macdermott, Seamus, MD; Torbay Hospital
Alstrom Syndrome

MacDonald, Ian M, MDCM; University of Alberta
Bestrophinopathies
Choroideremia
Weill-Marchesani Syndrome
X-Linked Congenital Retinoschisis
X-Linked Congenital Stationary Night Blindness

MacDonald, Stella K, BSc; Children's Hospital of Eastern Ontario Research Institute
Mandibulofacial Dysostosis with Microcephaly
VLDLR Cerebellar Hypoplasia

MacFarland, Suzanne P, MD; Children's Hospital of Philadelphia
Juvenile Polyposis Syndrome

MacFarlane, Julie, MS; Xenon Pharmaceuticals Inc
Juvenile Hemochromatosis

Machol, Keren, MD, PhD; Baylor College of Medicine
Cleidocranial Dysplasia Spectrum Disorder

Machowicz, Rafal, MD; Medical University of Warsaw
Familial Hemophagocytic Lymphohistiocytosis

Mackay, Deborah JG, MA, PhD; Salisbury NHS Foundation Trust
Diabetes Mellitus, 6q24-Related Transient Neonatal

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

DLG4-Related Synaptopathy

MacLeod, Erin, PhD, RD; Children's National Hospital
Ornithine Transcarbamylase Deficiency

MacLeod, Heather, MS; University of Chicago
Arrhythmogenic Right Ventricular Cardiomyopathy Overview

Maconachie, Gail, BSc, PhD; University of Leicester
FRMD7-Related Infantile Nystagmus

Madhok, Sehajvir, MD; Columbia University Irving Medical Center
HNRNPH2-Related Neurodevelopmental Disorder

Magenis, Ellen, MD; Oregon Health and Science University
Smith-Magenis Syndrome

Magoulas, Pilar L, MS; Baylor College of Medicine
Glycogen Storage Disease Type IV

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

Mah, Dean Y, MD, MSc; University of Alberta
Bestrophinopathies

Maher, Eamonn R, MD; University of Cambridge
PLA2G6-Associated Neurodegeneration

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

Maillard, Camille, PhD; Université de Paris
Tubulinopathies Overview

Makaryan, Vahagn, MD; University of Washington Medical Center
ELANE-Related Neutropenia

Mak, Christopher CY, MB ChB, PhD; University of Hong Kong
MN1 C-Terminal Truncation Syndrome

Malfait, Fransiska, MD, PhD; Ghent University Hospital
Classic Ehlers-Danlos Syndrome

Malicdan, May Christine V, MD, PhD; National Human Genome Research Institute
Chediak-Higashi Syndrome
GNE Myopathy
Hermansky-Pudlak Syndrome

Malm, Dag, MD, PhD; Tromsø Center of Internal Medicine
Alpha-Mannosidosis

Mancini, Cecilia, PhD; University of Turin
Spinocerebellar Ataxia Type 28

Mandell, Roseann; Massachusetts General Hospital
Fumarate Hydratase Deficiency

Mannens, Marcel MAM, PhD; University of Amsterdam
Long QT Syndrome Overview

Manohar, Vaishnavi, MT; King's College London
TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia

Manole, Andreea, BSc, PhD; UCL Queen Square Institute of Neurology and National Hospital for Neurology and Neurosurgery
Riboflavin Transporter Deficiency

Manoli, Irini, MD, PhD; National Human Genome Research Institute
Isolated Methylmalonic Acidemia
STAC3 Disorder

Mansour, Sahar; St George's, University of London
Lymphedema-Distichiasis Syndrome
Milroy Disease

Mao, Rong, MD; University of Utah
Legius Syndrome

Marafi, Dana, MD, MSc; Kuwait University
El-Hattab-Alkuraya Syndrome

Marazita, Mary L, PhD; University of Pittsburgh
Congenital Central Hypoventilation Syndrome

Marbach, Felix, MD; Heidelberg University
Schaaf-Yang Syndrome

Marcelis, Carlo LM, MD; Radboud University Nijmegen Medical Center
Feingold Syndrome 1

Marchuk, Douglas, PhD; Duke University
Familial Cerebral Cavernous Malformation

Marconi, Roberto, MD; Ospedale Misericordia
Spastic Paraplegia 7

Marcucci, Guido, MD; Ohio State University
CEBPA-Associated Familial Acute Myeloid Leukemia (AML)

Margolis, Russell L, MD; Johns Hopkins University School of Medicine
Huntington Disease-Like 2

Marini, Francesca, PhD; Italian Foundation for Research on Bone Diseases
Multiple Endocrine Neoplasia Type 1

Markovitz, Rebecca, MD, PhD; Baylor College of Medicine
GARS-Associated Axonal Neuropathy

Mark, Paul, MD; Michigan State University
Congenital NAD Deficiency Disorder

Marquard, Jessica, MS; Cleveland Clinic
Multiple Endocrine Neoplasia Type 2

Marquardt, Thorsten, Dr. med.; Universitätsklinikum Münster
SLC39A8-CDG

Marras, Connie, MD, PhD; University of Toronto
Hereditary Dystonia Overview

Marshall, Jan D, MS; The Jackson Laboratory
Alstrom Syndrome

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

Marsh, Eric D, MD, PhD; Children's Hospital of Philadelphia
CDKL5 Deficiency Disorder

Marsh, Rebecca A, MD; Cincinnati Children's Hospital
Familial Hemophagocytic Lymphohistiocytosis
X-Linked Lymphoproliferative Disease

Martin, Christa L, PhD; Geisinger Health System
17q12 Recurrent Deletion Syndrome
CHD8-Related Neurodevelopmental Disorder with Overgrowth
PACS1 Neurodevelopmental Disorder

Martin, Donna M, MD, PhD; University of Michigan
CHD7 Disorder

Martin, George M, MD; University of Washington
Werner Syndrome

Martin, Jack W, MD; The University of Texas MD Anderson Cancer Center
CDC73-Related Disorders

Martin, Rick A, MD; Saint Louis University
Mucopolysaccharidosis Type II

Martín, Bertha, PhD; University of Iowa
C3 Glomerulopathy

Martín, Miguel A, PhD; Hospital 12 de Octubre Research Institute
Glycogen Storage Disease Type V

Marzin, Pauline, MD; Paris Cité University
Geleophysic Dysplasia
Weill-Marchesani Syndrome

Mastrangelo, Mario, MD, PhD; Sapienza Università di Roma
PRICKLE1-Related Disorders

Mastrianni, James A, MD, PhD; University of Chicago
Genetic Prion Disease

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

Matern, Dietrich, MD, PhD; Mayo Clinic College of Medicine
Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Mathews, Jennifer, MS; University of North Carolina at Chapel Hill
Angelman Syndrome

Mathieu, Jean, MD; Complexe Hospitalier de la Sagamie
ARSACS

Matilla-Dueñas, Antoni, BSc, MSc, PhD; Germans Trias i Pujol Research Institute
Spinocerebellar Ataxia Type 37

Matsumoto, Naomichi, MD, PhD; Yokohama City University Graduate School of Medicine
Coffin-Siris Syndrome

Matsuura, Tohru, MD; Jichi Medical University
Spinocerebellar Ataxia Type 10

Mattison, Kari; Mayo Clinic - Minnesota
Dent Disease

Matton, Charlotte, MSc; Ghent University
Blepharophimosis, Ptosis, and Epicanthus Inversus Syndrome

Mavrogiannis, Lampros A, DPhil, MSc; St James's University Hospital
Enlarged Parietal Foramina

Mazzanti, Andrea, MD, PhD; ICS Maugeri
Catecholaminergic Polymorphic Ventricular Tachycardia

Mazzeu, Juliana, PhD; University of Brasília
Autosomal Dominant Robinow Syndrome

McCarthy, Liam, MD; Birmingham Women's and Children's Hospital
WFS1 Spectrum Disorder

McCormick, Elizabeth M, MS; Children's Hospital of Philadelphia
Primary Pyruvate Dehydrogenase Complex Deficiency Overview

McCormick, Sally PA, PhD; University of Otago
Tangier Disease

McCray, Brett A, MD, PhD; Johns Hopkins University School of Medicine
Autosomal Dominant TRPV4 Disorders

McDaniel, D Olga, PhD; University of Mississippi Medical Center
Spinocerebellar Ataxia Type 3

McDermott, Deborah A, MS; Consultant, Human Genetics and Genetic Counseling
Holt-Oram Syndrome

McDonald-McGinn, Donna M, MS; Children's Hospital of Philadelphia
22q11.2 Deletion Syndrome

McDonald, Jamie, MS; University of Utah Medical Center
Hereditary Hemorrhagic Telangiectasia

McDonell, Laura M, MD, PhD; Dalhousie University
Microcephaly-Capillary Malformation Syndrome

McFarland, Robert, MA, MBBS, PhD; Newcastle University
RRM2B Mitochondrial DNA Maintenance Defects

McFarquhar, Ashley, BSc; McGill University
Spondylometaphyseal Dysplasia, Corner Fracture Type

McGarrity, Thomas J, MD; Milton S Hershey Medical Center
Peutz-Jeghers Syndrome

McGovern, Margaret M, MD, PhD; Stony Brook University School of Medicine
Acid Sphingomyelinase Deficiency

McIntosh, Iain, PhD; American University of the Caribbean
Nail-Patella Syndrome

McLean, WH Irwin, DSc; University of Dundee
Pachyonychia Congenita

McMullin, Mary Frances, MD; Queens University
Primary Familial and Congenital Polycythemia

McNally, Elizabeth, MD, PhD; University of Chicago
Arrhythmogenic Right Ventricular Cardiomyopathy Overview

McNiven, Vanda, MD, MSc; McMaster Children's Hospital
Au-Kline Syndrome

McTaggart, Kerry, MSc; National Institutes of Health

Choroideremia

McTague, Amy, MBChB; UCL Great Ormond Street Institute of Child Health
SLC12A5-Related Epilepsy of Infancy with Migrating Focal Seizures

Medin, Jeffrey A, PhD; Medical College of Wisconsin
ASAH1-Related Disorders

Medne, Livija, MS; Children's Hospital of Philadelphia
Emanuel Syndrome

Mefford, Heather C, MD, PhD; St Jude Children's Research Hospital
15q13.3 Recurrent Deletion
17q12 Recurrent Duplication
CHD2-Related Neurodevelopmental Disorders
SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders

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

Mehta, Parinda A, MD; Cincinnati Children's Hospital Medical Center
Fanconi Anemia

Meijer, Inge A, MD, PhD; Université de Montréal
Spastic Paraplegia 8
VPS13D Movement Disorder

Meisler, Miriam H, PhD; University of Michigan
SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders

Mele, Caterina, Biol Sci D; Istituto di Ricerche Farmacologiche Mario Negri–IRCCS
Genetic Atypical Hemolytic-Uremic Syndrome

Mellerio, Jemima, BSc, MBBS, MD; Guy's and St Thomas' NHS Foundation Trust
Hidrotic Ectodermal Dysplasia 2

Mellgren, Anne Elisabeth Christensen, MD, PhD; University of Bergen
ADAMTSL4-Related Eye Disorders

Melo de Gusmao, Claudio, MD; Boston Children's Hospital
DRPLA

Meltzer, Meira Rina, MA, MS; National Institutes of Health
Choroideremia
X-Linked Congenital Retinoschisis

Mendonca, Berenice B, MD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo
PROP1-Related Combined Pituitary Hormone Deficiency

Mendoza-Londono, Roberto, MD, MS; The Hospital for Sick Children and University of Toronto

Caffey Disease
Cleidocranial Dysplasia Spectrum Disorder
MBD5 Haploinsufficiency

Menezes, Simone, BS; UTHealth School of Dentistry at Houston
Nonsyndromic Tooth Agenesis Overview

Menichelli, Danilo, MD; Sapienza University of Rome
Factor V Leiden Thrombophilia

Mercier, Sandra, MD, PhD; Service de Génétique médicale – CHU
Hereditary Fibrosing Poikiloderma with Tendon Contractures, Myopathy, and Pulmonary Fibrosis

Mercimek-Andrews, Saadet, MD, PhD; University of Alberta
Creatine Deficiency Disorders
STXBP1 Encephalopathy with Epilepsy

Merideth, Melissa A, MD, MPH; National Human Genome Research Institute
DICER1 Tumor Predisposition

Merkens, Louise S, PhD; Oregon Health & Science University
Sitosterolemia

Merla, Giuseppe; University of Naples Federico II
GNB5-Related Neurodevelopmental Disorder

Meroni, Germana, PhD; University of Trieste
MID1-Related Opitz G/BBB Syndrome

Merritt, J Lawrence, MD; Seattle Children's Hospital
Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Mervis, Carolyn B, PhD; University of Louisville
7q11.23 Duplication Syndrome

Mesika, Aviv; Bar-Ilan University
INSR-Related Severe Insulin Resistance Syndrome

Messiaen, Ludwine, MD; University of Alabama
LZTR1- and SMARCB1-Related Schwannomatosis

Messinger, Yoav, MD; Children's Hospitals and Clinics of Minnesota
DICER1 Tumor Predisposition

Meyer, Esther, PhD; UCL Great Ormond Street Institute of Child Health
SLC39A14 Deficiency

Meyer, Lauren, MD, PhD; University of Washington
X-Linked Lymphoproliferative Disease

Miceli, Francesco, PhD; University of Naples Federico II

KCNQ2-Related Disorders
KCNQ3-Related Disorders

Michaelides, Michel, BSc, MBBS, MD; University College London
Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview

Michael, Julianne, MS; Spectrum Health
Bachmann-Bupp Syndrome

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

Michaud, Jacques L, MD; CHU Ste-Justine Research Center
SYNGAP1-Related Intellectual Disability

Michelucci, Roberto, MD, PhD; Bellaria Hospital
Autosomal Dominant Epilepsy with Auditory Features

Milewicz, Dianna M, MD, PhD; University of Texas Health Science Center
Heritable Thoracic Aortic Disease Overview

Miller, Daniel Guthrie, MD, PhD; Seattle Children's Hospital
Facioscapulohumeral Muscular Dystrophy
FGFR Craniosynostosis Syndromes Overview

Miller, David T, MD, PhD; Boston Children's Hospital / Harvard Medical School
16p11.2 Recurrent Deletion
Dystrophinopathies

Miller, Ian O, MD; Miami Children's Hospital
SCN1A-Related Seizure Disorders

Miller, Jennifer L, MD, MS; University of Florida College of Medicine
Prader-Willi Syndrome

Miller, Timothy M, MD, PhD; Washington University School of Medicine
TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia

Miller, Vanessa Rangel, MS; Emory University
Mowat-Wilson Syndrome

Milliner, Dawn S, MD; Mayo Clinic
Dent Disease
Primary Hyperoxaluria Type 1
Primary Hyperoxaluria Type 3

Mills, Philippa B, PhD; UCL Great Ormond Street Institute of Child Health
Hypermanganesemia with Dystonia 1
PNPO Deficiency

SLC39A14 Deficiency

Milstone, Leonard M, MD; Yale University
Pachyonychia Congenita

Miltenberger-Miltenyi, Gabriel, MD; Universidade de Lisboa
VPS13A Disease

Milunsky, Jeff Mark, MD; Center for Human Genetics, Inc
Branchiooculofacial Syndrome
Waardenburg Syndrome Type I

Minetti, Carlo, MD; Gaslini Institute
Hypomyelination and Congenital Cataract

Min, Rogier, PhD; Amsterdam University Medical Center
CLCN2-Related Leukoencephalopathy
Megalencephalic Leukoencephalopathy with Subcortical Cysts

Min, Sang Hee, MD; Albert Einstein College of Medicine
Hereditary Folate Malabsorption

Miraglia del Giudice, Emanuele, MD; Second University of Naples
KCNQ2-Related Disorders
KCNQ3-Related Disorders

Mirzaa, Ghayda, MD; Seattle Children's Research Institute
Microcephaly-Capillary Malformation Syndrome
MPPH Syndrome
PIK3CA-Related Overgrowth Spectrum
PPP2R5D-Related Neurodevelopmental Disorder

Misko, Albert, MD, PhD; Massachusetts General Hospital / Harvard Medical School
Molybdenum Cofactor Deficiency
Mucolipidosis IV

Mitchell, Amanda, MS; Oregon Health Sciences University
Bietti Crystalline Dystrophy

Mitchell, Elyse, MS; Mayo Clinic
17q12 Recurrent Duplication

Mitchell, John J, MD; McGill University
Phenylalanine Hydroxylase Deficiency

Mitchel, Marissa W, MS; Geisinger Health System
16p11.2 Recurrent Deletion
17q12 Recurrent Deletion Syndrome

CHD8-Related Neurodevelopmental Disorder with Overgrowth

Mittaz-Crettol, Lauréane, PhD; Lausanne University Hospital
Achondrogenesis Type 1B
Atelosteogenesis Type 2
Diastrophic Dysplasia
SLC26A2-Related Multiple Epiphyseal Dysplasia

Miyajima, Hiroaki, MD; Hamamatsu University School of Medicine
Aceruloplasminemia

Miyake, Christina Y, MD, MS; Baylor College of Medicine
TANGO2 Deficiency

Moeschler, John, MD, MS; Geisel School of Medicine/Dartmouth College
16p12.2 Recurrent Deletion

Mohassel, Payam, MD; National Institute of Neurological Disorders and Stroke
Collagen VI-Related Dystrophies

Mohnach, Lauren, MS; University of Michigan
Nonsyndromic Disorders of Testicular Development Overview

Moi, Paolo, MD; Ospedale Regionale per le Microcitemie
Alpha-Thalassemia

Monaco, Anthony P, MD, PhD; Tufts University
VPS13A Disease

Moog, Ute, MD; Heidelberg University
CASK Disorders
Encephalocraniocutaneous Lipomatosis

Moore, Anthony T, BM BCh, MA; University of California San Francisco
Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview
PAX6-Related Aniridia

Moosajee, Mariya, BSc, MBBS, PhD; Moorfields Eye Hospital
PAX6-Related Aniridia

Morales Corado, J Andres, MBBS; Columbia University
Carnitine-Acylcarnitine Translocase Deficiency

Morales, Ana, MS; The Ohio State University
Dilated Cardiomyopathy Overview
LMNA-Related Dilated Cardiomyopathy

Moran, Rocio T, MD; The MetroHealth System
Stickler Syndrome

Morava, Eva, MD, PhD; Mayo Clinic
SERAC1 Deficiency

Moreau, Lisa, MS; Dana Farber Cancer Institute
Fanconi Anemia

Moreira Marques, Juliana, PhD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo
PROP1-Related Combined Pituitary Hormone Deficiency

Moreira, Maria-Ceu, MSc, PhD; Professor of Molecular Biology and Genetics
Ataxia with Oculomotor Apraxia Type 2

Morel Swols, Dayna, MS; University of Miami Miller School of Medicine
KBG Syndrome

Moreno-De-Luca, Daniel, BSc, MD; Brown University
17q12 Recurrent Deletion Syndrome

Moretti-Ferreira, Danilo, PhD; Sao Paulo State University
Oral-Facial-Digital Syndrome Type I

Morgan, Angela, PhD; University of Melbourne
FOXP1 Syndrome
FOXP2-Related Speech and Language Disorder
Koolen-de Vries Syndrome
SETBP1 Haploinsufficiency Disorder

Morimoto, Marie, PhD; National Human Genome Research Institute
Chediak-Higashi Syndrome
Schimke Immunoosseous Dysplasia

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
ATP8B1 Deficiency

Morris, Colleen A, MD; University of Nevada, Reno School of Medicine
7q11.23 Duplication Syndrome
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
- Multiple Epiphyseal Dysplasia, Dominant
- Stickler Syndrome
- X-Linked Hypophosphatemia

Mortimer, Peter, MD; St George's, University of London
- Lymphedema-Distichiasis Syndrome
- Milroy Disease

Morton, D Holmes, MD; Clinic for Special Children
- Maple Syrup Urine Disease

Moser, Ann B, BA; Kennedy Krieger Institute
- Rhizomelic Chondrodysplasia Punctata Type 1
- X-Linked Adrenoleukodystrophy
- Zellweger Spectrum Disorder

Moser, Hugo W, MD; Kennedy Krieger Institute
- X-Linked Adrenoleukodystrophy
- Zellweger Spectrum Disorder

Moskowitz, Samuel M, MD; Massachusetts General Hospital
- Cystic Fibrosis

Motulsky, Arno G, MD; University of Washington
- HFE-Related Hemochromatosis

Moufawad El Achkar, Christelle, MD; Harvard Medical School
- PRRT2-Associated Paroxysmal Movement Disorders

Mroczek, Magdalena, MD; University College London
- WARS2 Deficiency

Mucha, Bettina E, MD; Sainte-Justine Hospital
- TBC1D24-Related Disorders

Muenke, Maximilian, MD; American College of Medical Genetics and Genomics
- Holoprosencephaly Overview
- Muenke Syndrome

Mullegama, Sureni V, PhD; Sam Houston State University
- MBD5 Haploinsufficiency

Mulle, Jennifer Gladys, MHSc, PhD; Rutgers University
- 3q29 Recurrent Deletion

Munns, Craig, MBBS, PhD; The Children's Hospital at Westmead
SHOX Deficiency Disorders

Muram-Zborovski, Talia, MD; University of Utah
Legius Syndrome

Muraresku, Colleen, MS; Children's Hospital of Philadelphia
TRMU Deficiency

Murdock, David, MD; Baylor College of Medicine
Xia-Gibbs Syndrome

Murphy, Melissa M, PhD; Emory University School of Medicine
3q29 Recurrent Deletion

Murray, Jeffrey C, MD; University of Iowa
IRF6-Related Disorders

Murray, Michael F, MD; Geisinger Health System
Lysosomal Acid Lipase Deficiency

Murray, Mitzi L, MA, MD; University of Washington
Vascular Ehlers-Danlos Syndrome

Myers, Kasiani, MD; Cincinnati Children's Hospital Medical Center
Shwachman-Diamond Syndrome

Myers, Kenneth A, MD, PhD; University of Melbourne
GRIN2A-Related Speech Disorders and Epilepsy

Myers, Scott M, MD; Geisinger Health System
17q12 Recurrent Deletion Syndrome
CHD8-Related Neurodevelopmental Disorder with Overgrowth

Mymin, David, MBBCh; University of Manitoba
Sitosterolemia

Myrie, Semone B, PhD; University of Manitoba
Sitosterolemia

Mäkitie, Outi, MD, PhD; University of Helsinki and Helsinki University Hospital
Cartilage-Hair Hypoplasia - Anauxetic Dysplasia Spectrum Disorders

Méneret, Aurélie, MD, PhD; Institut du Cerveau et de la Moelle épinière
Congenital Mirror Movements

Möller, Birk; University of Cologne
DYNC1H1-Related Disorders

Müller, Juliane S, PhD; University of Cambridge
Congenital Myasthenic Syndromes Overview

Münchau, Alexander, MD; University of Lübeck
Hereditary Dystonia Overview

Mütze, Ulrike, MD; University Hospital Heidelberg
Classic Isovaleric Acidemia

N

Nabais Sá, Maria J, MD, MPH, PhD; Radboud University Medical Center
Gabriele-de Vries Syndrome

Nagamani, Sandesh C Sreenath, MBBS, MD; Baylor College of Medicine
Argininosuccinate Lyase Deficiency

Nagappa, Madhu, MD; National Institute of Mental Health & Neurosciences
Huppke-Brendel Syndrome
Isolated Sulfite Oxidase Deficiency

Nagaraj, Chinmayee B, MS; Cincinnati Children's Hospital
Autoimmune Lymphoproliferative Syndrome
WAS-Related Disorders

Nagy, Amanda, MD; Massachusetts General Hospital
Canavan Disease

Nagy, Sara, MD, MSc; University College London
WARS2 Deficiency

Nahhas, Norah, MD; Children's National Health System
LMNB1-Related Autosomal Dominant Leukodystrophy
Pelizaeus-Merzbacher-Like Disease 1
TUBB4A-Related Leukodystrophy

Naidu, Sakkubai, MD; Kennedy Krieger Institute
Alexander Disease

Nakamura, Katsuya, MD, PhD; Shinshu University School of Medicine
Hereditary Transthyretin Amyloidosis

Nakano, Taizo A, MD; Children's Hospital Colorado
Monosomy 7 Predisposition Syndromes Overview

Nakaya Fletcher, Shelley, BS; BloodWorks Northwest
Hemophilia A
Hemophilia B

Namavar, Yasmin, MSc; University of Amsterdam
TSEN54 Pontocerebellar Hypoplasia

Nance, Martha A, MD; Hennepin County Medical Center

Cockayne Syndrome

Napolitano, Carlo, MD, PhD; ICS Maugeri
CACNA1C-Related Disorders
Catecholaminergic Polymorphic Ventricular Tachycardia

Narayanan, Dhanya Lakshmi, MD; Kasturba Medical College, Manipal
EBF3 Neurodevelopmental Disorder
ISCA1-Related Multiple Mitochondrial Dysfunctions Syndrome
LPIN2-Related Majeed Syndrome

Narayan, Srinivas B, PhD; Children's Hospital of Philadelphia
Carnitine Palmitoyltransferase 1A Deficiency

Narumi, Satoshi, MD, PhD; National Research Institute for Child Health and Development
MIRAGE Syndrome

Nash, David R, MD; University of Pittsburgh
ZAP70-Related Combined Immunodeficiency

Nasir, Ramzi, MD, MPH; Boston Children's Hospital
16p11.2 Recurrent Deletion

Nattakom, Mary, BA; Columbia University
PPP2R5D-Related Neurodevelopmental Disorder

Navas, Placido, PhD; Universidad Pablo de Olavide
Primary Coenzyme Q10 Deficiency Overview

Naylor, Rochelle, MD; University of Chicago
Maturity-Onset Diabetes of the Young Overview

Need, Anna, PhD; Imperial College London
NGLY1-Related Congenital Disorder of Deglycosylation

Neilan, Edward G, MD, PhD; Harvard Medical School
Cockayne Syndrome

Neiman, Aaron, PhD; Stony Brook University
VPS13A Disease

Neira-Fresneda, Juanita, MD; Baylor College of Medicine
Potocki-Lupski Syndrome

Nelson, Adam, MBBS; Cincinnati Children's Hospital Medical Center
Shwachman-Diamond Syndrome

Nelson, David L, PhD; Baylor College of Medicine
Incontinentia Pigmenti

Nemes, Karolina, MD, PhD; University Medical Center Augsburg

Rhabdoid Tumor Predisposition Syndrome

Nemeth, Andrea H, DPhil; Churchill Hospital and Institute of Molecular Medicine
Hereditary Dystonia Overview

Nesterova, Galina, MD; NIH
Cystinosis

Netchine, Irene, MD, PhD; Hôpital Armand Trousseau
Silver-Russell Syndrome

Neufeld, Ellis J, MD, PhD
Thiamine-Responsive Megaloblastic Anemia Syndrome

Neuhaus, Emily, PhD; University of Washington
CHD8-Related Neurodevelopmental Disorder with Overgrowth

Neuser, Sonja, MD; University of Leipzig Medical Center
TECPR2-Related Hereditary Sensory and Autonomic Neuropathy with Intellectual Disability

Newman, William G, MA, PhD; University of Manchester and Manchester University NHS Foundation Trust
Perrault Syndrome
Urofacial Syndrome

New, Maria I, PhD; Mount Sinai School of Medicine
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

Nguyen, Matthew Huu, MD; University of New South Wales
CLCN4-Related Neurodevelopmental Disorder

Nichols, Colin G, PhD; Washington University School of Medicine
Cantú syndrome

Nichols, Kim E, MD; St Jude Children's Research Hospital
Familial Hemophagocytic Lymphohistiocytosis
Li-Fraumeni Syndrome
X-Linked Lymphoproliferative Disease

Nicholson, Garth A, MBBS, PhD; University of Sydney
SPTLC1-Related Hereditary Sensory Neuropathy

Nicklas, Janice A, PhD; University of Vermont
HPRT1 Disorders

Nicole, Sophie, PhD; Groupe Hospitalier Pitié-Salpêtrière
Hypokalemic Periodic Paralysis

Nicoli, Elena-Raluca, PharmR, PhD; National Human Genome Research Institute
Chediak-Higashi Syndrome

Nielsen, Jørgen E, MD, PhD; Rigshospitalet

CHMP2B Frontotemporal Dementia

Nielsen, Maartje, MD, PhD; Leiden University Medical Center
MUTYH Polyposis
NTHL1 Tumor Syndrome

Nielsen, Troels T, MSc, PhD; Rigshospitalet
CHMP2B Frontotemporal Dementia

Niewisch, Marena R, MD; National Cancer Institute, NIH
Dyskeratosis Congenita and Related Telomere Biology Disorders

Niinikoski, Harri, MD, PhD; University of Turku
Lysinuric Protein Intolerance

Nikkel, Sarah M, MD; BC Children's Hospital
Floating-Harbor Syndrome
Hypochondroplasia

Nillesen, Willy M, BSc; Radboud University Medical Center
Kleefstra Syndrome

Nilssen, Øivind, PhD; University of Tromsø
Alpha-Mannosidosis

Nimkarn, Saroj, MD; Bumrungrad Hospital
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

Nisbet, Alex F, BS; Children's Hospital of Philadelphia
Simpson-Golabi-Behmel Syndrome Type 1

Nishi, Mirian Yumie, PhD; Hospital das Clínicas da Faculdade de Medicina da Universidade de São Paulo
PROP1-Related Combined Pituitary Hormone Deficiency

Nishino, Ichizo, MD, PhD; National Institute of Neuroscience
CHKB-Related Muscular Dystrophy

Nishio, Shin-ya, PhD; Shinshu University School of Medicine
Nonsyndromic Hearing Loss and Deafness, Mitochondrial

Nobile, Carlo; CNR Institute of Neuroscience
Autosomal Dominant Epilepsy with Auditory Features

Noonan, Kristin M, MD; The Medical College of Wisconsin
Donnai-Barrow Syndrome

Noon, Sarah Elizabeth, MS; The Children's Hospital of Philadelphia
Cornelia de Lange Syndrome

Noreau, Anne, MSc; University of Montreal
SYNE1 Deficiency

Noris, Marina, PhD; Istituto di Ricerche Farmacologiche Mario Negri–IRCCS
Genetic Atypical Hemolytic-Uremic Syndrome

Northrup, Hope, MD; McGovern Medical School
Mucopolysaccharidosis Type III
Tuberous Sclerosis Complex

Nowaczyk, Malgorzata JM, MD; McMaster University
Floating-Harbor Syndrome
Smith-Lemli-Opitz Syndrome

Nozaki, Hiroaki, MD, PhD; Niigata University
HTRA1 Disorder

Nunes Guimaraes, Izolda, PhD; Sao Paulo State University
Oral-Facial-Digital Syndrome Type I

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

Nunes, Virginia, PhD; University of Barcelona
Lysinuric Protein Intolerance

Nussbaum, Robert L, MD; Invitae Corporation
Lowe Syndrome

Nyhan, William L, MD, PhD; University of California San Diego
HPRT1 Disorders

O

O'Brien, Kevin J, RN; National Human Genome Research Institute
Hermansky-Pudlak Syndrome

O'Donnell-Luria, Anne, MD, PhD; Boston Children's Hospital
KMT2E-Related Neurodevelopmental Disorder

O'Ferrall, Erin, MD, MSc; Montreal Neurological Institute
GNE Myopathy

O'Neill, J Patrick, PhD; University of Vermont
HPRT1 Disorders

O'Toole, Edel, MD, PhD; University of London
Pachyonychia Congenita

Ocansey, Sharon, BSc, MBBS, MSc; St George's University Hospital NHS Foundation Trust
EZH2-Related Overgrowth

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

NTRK1 Congenital Insensitivity to Pain with Anhidrosis

Oetgen, Matthew, MD; Children's National Medical Center
Mucopolysaccharidosis Type IVA

Oglesbee, Devin, PhD; Mayo Clinic
Short-Chain Acyl-CoA Dehydrogenase Deficiency

Oishi, Kimihiko, MD; Jikei University School of Medicine
Thiamine-Responsive Megaloblastic Anemia Syndrome

Okialda, Krystle A, BS; University of Washington
Bietti Crystalline Dystrophy

Okur, Volkan, MD; New York Genome Center
Okur-Chung Neurodevelopmental Syndrome

Oliveira, Joao, MD, PhD; Federal University of Pernambuco
Primary Familial Brain Calcification

Oliveira, Jorge, MSc, PhD; Universidade do Porto
LAMA2 Muscular Dystrophy

Oliver Petit, Isabelle, MD; Children's Hospital
Allan-Herndon-Dudley Syndrome

Olney, Ann Haskins, MD; University of Nebraska Medical Center
SLC25A24 Fontaine Progeroid Syndrome

Olson, Heather, MD, MS; Boston Children's Hospital
CDKL5 Deficiency Disorder

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

Ombrello, Amanda, MD; National Human Genome Research Institute
TNF Receptor-Associated Periodic Fever Syndrome

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

Onodera, Osamu, MD, PhD; Niigata University
HTRA1 Disorder
Spinocerebellar Ataxia Type 17

Opal, Puneet, MD, PhD; Northwestern University Feinberg School of Medicine
GAN-Related Neurodegeneration
Spinocerebellar Ataxia Type 1

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

Congenital Deafness with Labyrinthine Aplasia, Microtia, and Microdontia
Deafness and Myopia Syndrome

Origa, Raffaella, MD; Ospedale Pediatrico Microcitemico Antonio Cao
Alpha-Thalassemia
Beta-Thalassemia

Orrell, Richard W, BSc, MD; University College London
ALS2-Related Disorder

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

Orten, Dana J, PhD; Boys Town National Research Hospital
Usher Syndrome Type II

Orthmann-Murphy, Jennifer, MD, PhD; Johns Hopkins University
Pelizaeus-Merzbacher-Like Disease 1

Ortiz-Gonzalez, Xilma, MD, PhD; Children's Hospital of Philadelphia
SPTBN4 Disorder

Osborne, Jenae, MS; University of Michigan
POT1 Tumor Predisposition

Osborne, Lucy R, PhD; University of Toronto
7q11.23 Duplication Syndrome

Oshima, Junko, MD, PhD; University of Washington
Werner Syndrome

Ostergaard, Elsebet, MD, PhD; National University Hospital Rigshospitalet
SUCLA2-Related Mitochondrial DNA Depletion Syndrome, Encephalomyopathic Form with Methylmalonic Aciduria

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

Ostrer, Harry, MD; New York University School of Medicine
Nonsyndromic Disorders of Testicular Development Overview

Ostrowski, Philip J, MD; St George's University Hospitals NHS Foundation Trust
Tatton-Brown-Rahman Syndrome

Oswald, Gretchen L, MS; Johns Hopkins Medical Center
Chondrodysplasia Punctata 1, X-Linked

Othman, Amna A, MBBCh; National Human Genome Research Institute
Osteoglophonic Dysplasia

Ottman, Ruth, PhD; Columbia University

Autosomal Dominant Epilepsy with Auditory Features

Ozelius, Laurie, PhD; Massachusetts General Hospital
ATP1A3-Related Neurologic Disorders
DYT1 Early-Onset Isolated Dystonia
SGCE Myoclonus-Dystonia

P

Paciorkowski, Alex R, MD; University of Rochester Medical Center
PURA-Related Neurodevelopmental Disorders

Padiath, Quasar, MBBS, PhD; University of Pittsburgh
LMNB1-Related Autosomal Dominant Leukodystrophy

Pagon, Roberta A, MD; University of Washington
Nonsyndromic Retinitis Pigmentosa Overview
Spinocerebellar Ataxia Type 7

Paisey, Richard B, MD; Torbay Hospital
Alstrom Syndrome

Pais, Lynn, MS, MSc; Boston Children's Hospital
KMT2E-Related Neurodevelopmental Disorder

Pallais, J Carl, MD, MPH; Massachusetts General Hospital
Isolated Gonadotropin-Releasing Hormone (GnRH) Deficiency

Palmer, Elizabeth, MBBS, PhD; University of New South Wales
ATN1-Related Neurodevelopmental Disorder
CLCN4-Related Neurodevelopmental Disorder

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

Pal, Tuya, MD; Vanderbilt-Ingram Cancer Center
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
Parkinson Disease Overview

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

Papapoulos, Socrates, MD, PhD; Leiden University Medical Center
SOST-Related Sclerosing Bone Dysplasias

Pappas, John G, MD, MS; NYU Grossman School of Medicine
SETD2 Neurodevelopmental Disorders

Paquis-Flucklinger, Véronique, MD, PhD; Nice Hospital
CHCHD10-Related Disorders

Parboosingh, Jillian S, PhD; University of Calgary
VLDLR Cerebellar Hypoplasia

Parente Freixo, João, MD; Universidade do Porto
LAMA2 Muscular Dystrophy

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

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

Parker, Alasdair Patrick John, MA, MBBS, MD; Addenbrooke's Hospital
Congenital Insensitivity to Pain Overview

Parker, Charles J, MD; University of Utah School of Medicine
HFE-Related Hemochromatosis

Parkin, Kimberly; Massachusetts General Hospital
Pitt-Hopkins Syndrome

Park, Julien H, Dr. med.; Universitätsklinikum Münster
SLC39A8-CDG

Park, Julie R, MD; Seattle Children's Hospital
ALK-Related Neuroblastic Tumor Susceptibility

Parks, Sharie B, PhD; Oregon Health and Science University
Dilated Cardiomyopathy Overview

Parodi, Livia, PhD; Sorbonne Université – Pitié-Salpêtrière University Hospital
Spastic Paraplegia 4

Pascotto, Antonio, MD; Second University of Naples
KCNQ2-Related Disorders

Pascual, Juan M, MD, PhD; University of Texas Southwestern Medical Center
Glucose Transporter Type 1 Deficiency Syndrome

Pasini, Elena, MD; Bellaria Hospital

Autosomal Dominant Epilepsy with Auditory Features

Passarge, Eberhard, MD; University of Essen
Retinoblastoma

Passemard, Sandrine, MD, PhD; APHP-Robert Debré University Hospital
ASPM Primary Microcephaly
WDR62 Primary Microcephaly

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
NKX2-1-Related Disorders

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

Patel, Swati G, MD, MS; University of Colorado Anschutz Medical Center
APC-Associated Polyposis Conditions

Patterson, Marc, MD; Mayo Clinic
Niemann-Pick Disease Type C

Patwari, Pallavi P, MD; Children's Memorial Hospital
Congenital Central Hypoventilation Syndrome

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

Paulson, Henry, MD, PhD; University of Michigan Medical School
Spinocerebellar Ataxia Type 3

Pearl, Phillip L, MD; George Washington University School of Medicine
Succinic Semialdehyde Dehydrogenase Deficiency

Pearson, Deborah A, PhD; McGovern Medical School
Tuberous Sclerosis Complex

Pearson, Toni S, MBBS, MD; Nationwide Children's Hospital
Aromatic L-Amino Acid Decarboxylase Deficiency

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

MYH9-Related Disease

Peikert, Kevin, MD; University Medical Center Rostock
McLeod Neuroacanthocytosis Syndrome
VPS13A Disease

Pellerin, David, MD, MSc; University College London
GAA-FGF14-Related Ataxia

Pelzer, Nadine, MD, PhD; Leiden University Medical Center
Retinal Vasculopathy with Cerebral Leukoencephalopathy and Systemic Manifestations

Pena, Izabella, PhD; Massachusetts Institute of Technology
PLPBP Deficiency

Pencheva, Bojana, MMSc; Emory University School of Medicine
ETV6 Thrombocytopenia and Predisposition to Leukemia

Pendyal, Surekha, MEd, MSc, RD; Duke University Medical Center
Glycogen Storage Disease Type I

Pennesi, Mark E, MD, PhD; Casey Eye Institute - Oregon Health & Science University
Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview
RPE65-Related Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy

Penttilä, Sini, PhD; Fimlab Laboratories
ANO5 Muscle Disease

Pepin, Melanie G, MS; University of Washington Medical Center
COL1A1/2 Osteogenesis Imperfecta
Vascular Ehlers-Danlos Syndrome

Percy, Melanie, PhD; Belfast City Hospital
Primary Familial and Congenital Polycythemia

Perez Botero, Juliana, MD; BloodCenter of Wisconsin
ANKRD26-Related Thrombocytopenia

Perlman, Susan, MD; David Geffen School of Medicine at UCLA
Ataxia-Telangiectasia
Hereditary Ataxia Overview

Peron, Angela, MD; San Paolo Hospital, Università degli Studi di Milano
BCL11A-Related Intellectual Disability
Snyder-Robinson Syndrome

Perrier, Nancy D, MD; University of Texas MD Anderson Cancer Center
CDC73-Related Disorders

Perrino, Melissa R, MD; St Jude Children's Research Hospital

ALK-Related Neuroblastic Tumor Susceptibility

Perry, Monique, MD; Clinical Center - National Institutes of Health
Alkaptonuria

Pescosolido, Matthew F, BA; Brown University
Christianson Syndrome

Pessia, Mauro, PhD; Istituto Euro-Mediterraneo di Scienza e Tecnologia
Episodic Ataxia Type 1

Petit, Florence, MD, PhD; CHU de Lille
Thrombocytopenia Absent Radius Syndrome

Petrovic, Aleksandra, MD; University of Washington
X-Linked Severe Combined Immunodeficiency

Petrucelli, Nancie, MS; Karmanos Cancer Institute, Cancer Genetic Counseling Service
BRCA1- and BRCA2-Associated Hereditary Breast and Ovarian Cancer

Pfautsch, Miranda; Dr Kiran C Patel College of Medicine - Tampa Bay Campus
Spinal Muscular Atrophy, X-Linked Infantile

Pfeffer, Gerald, MD, PhD; University of Calgary
Hereditary Myopathy with Early Respiratory Failure

Pfendner, Ellen G, PhD; GeneDx, Inc
Dystrophic Epidermolysis Bullosa
Epidermolysis Bullosa Simplex
Epidermolysis Bullosa with Pyloric Atresia
Junctional Epidermolysis Bullosa

Phelan, Katy, PhD; Florida Cancer Specialists & Research Institute
Phelan-McDermid Syndrome-SHANK3 Related

Phillips, Ian R, PhD; Queen Mary, University of London
Primary Trimethylaminuria

Phillips, John, PhD; University of Utah School of Medicine
Familial Porphyria Cutanea Tarda
Hepatoerythropoietic Porphyria

Phillips, John A, MD; Vanderbilt University Medical Center
Heritable Pulmonary Arterial Hypertension Overview
PROP1-Related Combined Pituitary Hormone Deficiency

Phillips, Lindsay, PhD; University of Calgary
Au-Kline Syndrome

Phornphutkul, Chanika, MD; Brown University

Alkaptonuria

Phowthongkum, Prasit, MD; University of Washington
SAMD9L Ataxia-Pancytopenia Syndrome

Piard, Juliette, MD; CHRU de Besançon – Hôpital Saint-Jacques
Baller-Gerold Syndrome

Picker, Jonathan D, MBChB, PhD; Boston Children's Hospital
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency

Pieke-Dahl, Sandra, PhD; Ohio State University, Department of Psychology
Usher Syndrome Type I
Usher Syndrome Type II

Pierre, Germaine, MBBS, MSc; Bristol Royal Hospital for Children
Barth Syndrome

Pierson, Christopher R, MD, PhD; Nationwide Children's Hospital
X-Linked Myotubular Myopathy

Pignatelli, Pasquale, MD, PhD; Sapienza University of Rome
Factor V Leiden Thrombophilia

Pilarski, Robert, MS, MSW; The Ohio State University
BAP1 Tumor Predisposition Syndrome
PTEN Hamartoma Tumor Syndrome

Pilz, Daniela T, MD, PhD; University Hospital of Wales
Baraitser-Winter Cerebrofrontofacial Syndrome

Pinsky, Leonard, MD; McGill University
Androgen Insensitivity Syndrome

Piperno, Alberto, MD; University of Milano-Bicocca
Juvenile Hemochromatosis

Pithukpakorn, Manop, MD; Mahidol University
Birt-Hogg-Dube Syndrome
FH Tumor Predisposition Syndrome

Pitteloud, Nelly, MD; Massachusetts General Hospital
Isolated Gonadotropin-Releasing Hormone (GnRH) Deficiency

Pitt, James, PhD; Murdoch Children's Research Institute
Squalene Synthase Deficiency

Pizzo, Lucilla, MS; Pennsylvania State University
16p12.2 Recurrent Deletion

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

COL4A1-Related Disorders

Platzer, Konrad, MD; University of Leipzig Medical Center
GRIN1-Related Neurodevelopmental Disorder
GRIN2B-Related Neurodevelopmental Disorder
GRIN2D-Related Developmental and Epileptic Encephalopathy

Plecko, Barbara, MD; Medical University of Graz
PNPO Deficiency

Plitt, Gilman, MD; Cleveland Clinic
Multiple Endocrine Neoplasia Type 2

Plon, Sharon E, MD, PhD; Baylor College of Medicine
Rothmund-Thomson Syndrome

Plotkin, Scott, MD, PhD; Harvard Medical School and Massachusetts General Hospital
LZTR1- and SMARCB1-Related Schwannomatosis

Pober, Barbara R, MD; MassGeneral Hospital for Children
Congenital Diaphragmatic Hernia Overview
Donnai-Barrow Syndrome

Poke, Gemma, BSc, MBBS; University of Otago
GNB5-Related Neurodevelopmental Disorder

Porter, Christopher C, MD; Emory University School of Medicine
ETV6 Thrombocytopenia and Predisposition to Leukemia

Portigal, Cheryl L, MSc; University of British Columbia
Mucopolysaccharidosis Type I

Potocki, Lorraine, MD; Baylor College of Medicine
GARS-Associated Axonal Neuropathy
Potocki-Lupski Syndrome
Smith-Magenis Syndrome

Powell, Lawrie, MD, PhD; University of Queensland
HFE-Related Hemochromatosis

Prabhu, Sandeep, MD; University of Oklahoma
MBTPS1-Related Spondyloepimetaphyseal Dysplasia with Elevated Lysosomal Enzymes

Prada, Carlos, MD; Ann & Robert H Lurie Children's Hospital of Chicago
DLG4-Related Synaptopathy

Prades, Silvia, PhD; Ataxia UK
DRPLA

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

Sphingosine Phosphate Lyase Insufficiency Syndrome

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

Prasov, Lev, MD, PhD; University of Michigan Medical School
MYRF-Related Cardiac Urogenital Syndrome

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

Preston, Matthew K, MD; University of Washington
Facioscapulohumeral Muscular Dystrophy

Priori, Silvia G, MD, PhD; ICS Maugeri
CACNA1C-Related Disorders
Catecholaminergic Polymorphic Ventricular Tachycardia

Prior, Thomas W, PhD; Case Western Reserve University
Spinal Muscular Atrophy

Prokisch, Holger, PhD; Technische Universität München
Mitochondrial Membrane Protein-Associated Neurodegeneration

Pronk, JC, PhD; Vrije Universiteit Medical Center
Megalencephalic Leukoencephalopathy with Subcortical Cysts

Proudlock, Frank A, BSc, MSc, PhD; University of Leicester
FRMD7-Related Infantile Nystagmus

Ptacek, Louis J, MD; University of California at San Francisco
Spinocerebellar Ataxia Type 7

Puck, Jennifer M, MD; University of California San Francisco
STAT3 Hyper IgE Syndrome
X-Linked Severe Combined Immunodeficiency

Puffenberger, Erik G, PhD; Clinic for Special Children
Maple Syrup Urine Disease

Pulst, Stefan M, MD; University of Utah
Spinocerebellar Ataxia Type 13
Spinocerebellar Ataxia Type 2

Putnam, Andrea, MS; New York Weill Cornell Center
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

Puzriakova, Arina, BSc, MSc; UCL Queen Square Institute of Neurology and National Hospital for Neurology and Neurosurgery

Spinocerebellar Ataxia Type 11

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

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

Q

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

LAMA2 Muscular Dystrophy

Quinonez, Shane C, MD; University of Michigan

Citrullinemia Type I

Dihydrolipoamide Dehydrogenase Deficiency

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

R

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

Mucolipidosis III Gamma

Rabin, Rachel, MS; NYU Grossman School of Medicine

SETD2 Neurodevelopmental Disorders

Rahman, Joyeeta, BSc; UCL Great Ormond Street Institute of Child Health

Mitochondrial DNA-Associated Leigh Syndrome Spectrum

Rahman, Nazneen, BM BCh, PhD; Institute of Cancer Research

EZH2-Related Overgrowth

Sotos Syndrome

Rahman, Shamima, PhD; UCL Great Ormond Street Institute of Child Health

Mitochondrial DNA-Associated Leigh Syndrome Spectrum

Nuclear Gene-Encoded Leigh Syndrome Spectrum Overview

Rai, Karan, BS; The Ohio State University

BAP1 Tumor Predisposition Syndrome

Raininko, Raili, MD, PhD; Uppsala University
LMNB1-Related Autosomal Dominant Leukodystrophy

Rajan, Neil, MBBS, PhD; Royal Victoria Infirmary
CYLD Cutaneous Syndrome

Rajasekaran, Surender, MD, MPH; Spectrum Health
Bachmann-Bupp Syndrome

Ramnitz, Mary Scott, MD; National Institute of Dental and Craniofacial Research
Hyperphosphatemic Familial Tumoral Calcinosis

Ramos, Eliana Marisa, PhD; University of California Los Angeles
Primary Familial Brain Calcification

Rampoldi, Luca, PhD; Ospedale San Raffaele
VPS13A Disease

Rana, Huma Q, MD; Dana-Farber Cancer Institute
FH Tumor Predisposition Syndrome

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

Ranum, Laura PW, PhD; University of Florida
Myotonic Dystrophy Type 2
Spinocerebellar Ataxia Type 8

Rappold, Gudrun A, PhD; Heidelberg University
FOXP1 Syndrome
SHOX Deficiency Disorders

Raskind, Wendy H, MD, PhD; University of Washington Medical Center
ADCY5 Dyskinesia
GATA1-Related Cytopenia
Hereditary Multiple Osteochondromas
SAMD9L Ataxia-Pancytopenia Syndrome
Spinocerebellar Ataxia Type 14

Ratisoontorn, Chootima, DDS, PhD; Chulalongkorn University
Saethre-Chotzen Syndrome

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

Rawlings, David J, MD; University of Washington

X-Linked Severe Combined Immunodeficiency

Rawls-Castillo, Brandy, MS, RD; Baylor College of Medicine

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

STRC-Related Autosomal Recessive Hearing Loss

Reding, Madeline Q, MPH, MS; Mayo Clinic

NDP-Related Retinopathies

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

McLeod Neuroacanthocytosis Syndrome

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

Succinic Semialdehyde Dehydrogenase Deficiency

Rees, Mark I, PhD; Swansea University

Hereditary Hyperekplexia Overview

Refetoff, Samuel, MD; University of Chicago Medical Center

Allan-Herndon-Dudley Syndrome

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

Heritable Thoracic Aortic Disease Overview

Regier, Debra S, MD, PhD; Rare Disease Institute - Children's National Hospital

GLB1-Related Disorders

Mucopolysaccharidosis Type IVA

Phenylalanine Hydroxylase Deficiency

Rehder, Catherine, PhD; Duke University Medical Center
Phosphorylase Kinase Deficiency

Reichenberger, Ernst, PhD; UConnHealth
Craniometaphyseal Dysplasia, Autosomal Dominant

Reijnders, Margot RF, MD; Radboud Unversity Medical Center
PURA-Related Neurodevelopmental Disorders

Reilly, Mary M, MBBCh, MD; University College London
RFC1 CANVAS / Spectrum Disorder

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

Reischl-Hajiabadi, Anna, MD; University Hospital Heidelberg
Classic Isovaleric Acidemia

Reiter, Lawrence T, PhD; University of Tennessee Health Science Center
Maternal 15q Duplication Syndrome

Remuzzi, Giuseppe, MD; Istituto di Ricerche Farmacologiche Mario Negri–IRCCS
Genetic Atypical Hemolytic-Uremic Syndrome

Renaud, Mathilde, MD, PhD; CHRU de Nancy
GAA-FGF14-Related Ataxia

Rendtorff, Nanna Dahl, PhD; University Hospital / Kennedy Center
WFS1 Spectrum Disorder

Revah-Politi, Anya, MS; Columbia University Irving Medical Center
GNB1 Encephalopathy

Riazi, Sheila, MD; University of Toronto
Malignant Hyperthermia Susceptibility

Richard, Gabriele, MD; GeneDx, Inc
Autosomal Recessive Congenital Ichthyosis

Richard, Pascale, PharmD, PhD; APHP, Sorbonne Université
Oculopharyngeal Muscular Dystrophy

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

Richter, Andrea, PhD; Sainte-Justine Hospital, University of Montreal
ARSACS

Richter, Mary K; National Foundation for Ectodermal Dysplasias

Hypohidrotic Ectodermal Dysplasia

Rich, Thereasa A, MS; University of Texas MD Anderson Cancer Center
CDC73-Related Disorders

Rinaldo, Piero, MD, PhD; Mayo Clinic College of Medicine
Medium-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Rioseco-Camacho, Natalia, BA; University of California, Irvine
Hyperornithinemia-Hyperammonemia-Homocitrullinuria Syndrome

Robbins, Emily; George Washington University School of Medicine
Succinic Semialdehyde Dehydrogenase Deficiency

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

Roberts, Eve, MD; University of Toronto
Wilson Disease

Robertson, David, MD; Vanderbilt University
Dopamine Beta-Hydroxylase Deficiency

Robertson, Nic, PhD; University of Edinburgh
RNU4atac-opathy

Robertson, Stephen, DPhil; University of Otago
FLNB Disorders
X-Linked Otopalatodigital Spectrum Disorders

Robin, Nathaniel H, MD; University of Alabama at Birmingham
FGFR Craniosynostosis Syndromes Overview
Stickler Syndrome

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

Rodan, Lance, MD; Boston Children's Hospital
KMT2E-Related Neurodevelopmental Disorder

Roetto, Antonella, PhD; University of Torino
TFR2-Related Hemochromatosis

Rogers, R Curtis, MD; RCR Genetics
Phelan-McDermid Syndrome-SHANK3 Related
RPS6KA3-Related Intellectual Disability

Rohrbach, Marianne, MD, PhD; University Children's Hospital
FKBP14 Kyphoscoliotic Ehlers-Danlos Syndrome

Rohrer, Jonathan, PhD; UCL Queen Square Institute of Neurology

MAPT-Related Frontotemporal Dementia

Roifman, Maian, MD; University of Toronto
Autosomal Dominant Robinow Syndrome

Rolle, Myron, MD; Massachusetts General Hospital
Muenke Syndrome

Rommens, Johanna M, PhD; The Hospital for Sick Children, University of Toronto
Shwachman-Diamond Syndrome

Ronco, Pierre M, MD, PhD; Hôpital Tenon / Université Pierre et Marie Curie
COL4A1-Related Disorders

Roos, Peter, MD, PhD; Rigshospitalet
CHMP2B Frontotemporal Dementia

Roscioli, Tony, MBBS, PhD; Sydney Children's Hospital
Hepatic Veno-Occlusive Disease with Immunodeficiency

Rosenberg, Henry, MD; Malignant Hyperthermia Association of the United States
Malignant Hyperthermia Susceptibility

Rosenberg, Stacy Lyn, MD; Children's Hospital of Pittsburgh
ZAP70-Related Combined Immunodeficiency

Rosenfeld, Jill, MS; Baylor College of Medicine
16p12.2 Recurrent Deletion

Ross, Alison J, PhD; University College London
Bardet-Biedl Syndrome Overview

Rossetti, Sandro, MD; Mayo Clinic
Dent Disease

Rossi, Alessandro, MD; University of Naples "Federico II"
Glycogen Storage Disease Type III

Rossi, Andrea, MD; G Gaslini Pediatric Institute and University of Genova
Hypomyelination and Congenital Cataract

Rossi, Christopher T, MD; Children's National Medical Center
DICER1 Tumor Predisposition

Rossignol, Francis, MD; National Human Genome Research Institute
Prolidase Deficiency

Rosso, Sonia M, MD, PhD; Havenziekenhuis
MAPT-Related Frontotemporal Dementia

Ross, Owen A, PhD; Mayo Clinic
LRRK2 Parkinson Disease

VPS35-Related Parkinson Disease

Roston, Alexandra, MD; University of British Columbia
SETD1B-Related Neurodevelopmental Disorder

Rothermel, Caroline E, BA; National Human Genome Research Institute
GLB1-Related Disorders

Roth, Fanny, PhD; Centre de Recherche en Myologie
Oculopharyngeal Muscular Dystrophy

Rouleau, Guy A, MD, PhD; McGill University
Hereditary Motor and Sensory Neuropathy with Agenesis of the Corpus Callosum
Oculopharyngeal Muscular Dystrophy
Spastic Paraplegia 8
SYNE1 Deficiency

Roullet, Jean-Baptiste, PhD; Washington State University
Succinic Semialdehyde Dehydrogenase Deficiency

Rouzier, Cécile, MD, PhD; Nice Hospital
CHCHD10-Related Disorders

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

Ruaud, Lyse, MD, PhD; APHP-Robert Debré University Hospital
WDR62 Primary Microcephaly

Rudnick, Sean, MD; Wake Forest University School of Medicine
Familial Porphyria Cutanea Tarda
Hepatoerythropoietic Porphyria

Ruivenkamp, Claudia, PhD; Leiden University Medical Center
Peters Plus Syndrome

Rumbos Siurana, Elisa, MSc; University College London
GRIA2-Related Neurodevelopmental Disorder

Rumsby, Gill, PhD; University College London Hospitals
Primary Hyperoxaluria Type 2

Ruppe, Mary D, MD; The Methodist Hospital
X-Linked Hypophosphatemia

Russell, Bianca, MD; Cincinnati Childen's Hospital
Bohring-Opitz Syndrome

Russell, Meaghan K, MPH; MassGeneral Hospital for Children
Congenital Diaphragmatic Hernia Overview

Russman, Barry S, MD; Oregon Health and Science University
Spinal Muscular Atrophy

Rutkowski, Anne, MD; Cure CMD
LAMA2 Muscular Dystrophy

Rutkowski, Timothy P, PhD; Emory University School of Medicine
3q29 Recurrent Deletion

Rutledge, S Lane, MD; University of Alabama at Birmingham
Nonketotic Hyperglycinemia

Rutten, Julie, MD, PhD; Leiden University Medical Center
CADASIL

Ryan, Brigid, PhD; University of Auckland
MAPT-Related Frontotemporal Dementia

Ryan, Shannon, MSc; Montreal Children's Hospital
Phenylalanine Hydroxylase Deficiency

Rydning, Siri Lynne, MD; University of Oslo
Spastic Paraplegia 4

Récan-Budiartha, Dominique, MD; Hopital Cochin
Emery-Dreifuss Muscular Dystrophy

Rødahl, Eyvind, MD, PhD; University of Bergen
ADAMTSL4-Related Eye Disorders
Congenital Stromal Corneal Dystrophy

S

Saal, Howard M, MD; Cincinnati Children's Hospital Medical Center
IRF6-Related Disorders
Silver-Russell Syndrome

Saba, Julie, MD, PhD; UCSF / Benioff Children's Hospital
Sphingosine Phosphate Lyase Insufficiency Syndrome

Sabet Rasekh, Parisa, MD; Children's National Health System
LMNB1-Related Autosomal Dominant Leukodystrophy

Sacharow, Stephanie J, MD; Boston Children's Hospital
Homocystinuria Caused by Cystathionine Beta-Synthase Deficiency
Spinal Muscular Atrophy, X-Linked Infantile

Sadleir, Lynette Grant, MB ChB; University of Otago
GNB5-Related Neurodevelopmental Disorder

Saenz-Ayala, Sofia, MD; Cincinnati Children's Hospital Medical Center
Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Saffari, Afshin, MD; Harvard Medical School
EPG5-Related Disorder

Saheki, Takeyori, MD, PhD; Kagoshima University Graduate School of Medical and Dental Sciences
Citrin Deficiency

Sahota, Amrik, PhD; Rutgers University
Adenine Phosphoribosyltransferase Deficiency

Saito, Kayoko, MD, PhD; Tokyo Women's Medical University School of Medicine
Fukuyama Congenital Muscular Dystrophy

Sajorda, Brian J, BA; Children's Hospital of Philadelphia
Simpson-Golabi-Behmel Syndrome Type 1

Sako, Shuhei, MD; Jikei University School of Medicine
Thiamine-Responsive Megaloblastic Anemia Syndrome

Saldaris, Jacinta, PhD; Telethon Kids Institute
CDKL5 Deficiency Disorder

Salgado, Carlos, MD; Stanford University School of Medicine
Schimke Immunoosseous Dysplasia

Salih, Mustafa A, Dr Med Sci, MBBS, MD; King Saud University
Salih Myopathy
SH3TC2-Related Hereditary Motor and Sensory Neuropathy
Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive

Salomons, Gajja S, PhD; Amsterdam University Medical Centers
Creatine Deficiency Disorders
Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation

Salpea, Paraskevi, PhD; National Institutes of Health
Carney Complex

Salpietro, Vincenzo, MD; University College London
GRIA2-Related Neurodevelopmental Disorder

Salter, Claire, BMedSci, MBBS; University of Exeter Medical School
PI4KA-Related Disorder

Salviati, Leonardo, MD, PhD; University of Padova
Primary Coenzyme Q10 Deficiency Overview

Sambuughin, Nyamkhishig, PhD; Uniformed Services University
Malignant Hyperthermia Susceptibility

Sampaio Moura, Natalia, BS; National Human Genome Research Institute
Adenosine Deaminase 2 Deficiency

Samson, Ricardo A, MD; Children's Heart Center Nevada
Jervell and Lange-Nielsen Syndrome

Sanchez Russo, Rossana, MD; Emory University School of Medicine
3q29 Recurrent Deletion

Sandoval, Claudio, MD; New York Medical College
Hereditary Folate Malabsorption

Sands, Tristan T, MD, PhD; Columbia University Irving Medical Center
GNB1 Encephalopathy

Santani, Avni B, PhD; Children's Hospital of Philadelphia
Carnitine Palmitoyltransferase 1A Deficiency

Santen, Gijs, MD, PhD; Leiden University Medical Center
ARID1B-Related Disorder
Coffin-Siris Syndrome

Santos, Fernando, MD, PhD; University of Oviedo
Hereditary Distal Renal Tubular Acidosis

Santos, Manuela, MD; Centro Hospitalar Universitário do Porto
LAMA2 Muscular Dystrophy

Sanz, Maureen M, PhD; Molloy College
Bloom Syndrome

Sapp, Julie C, ScM; National Institutes of Health
Proteus Syndrome

Sardh, Eliane, MD, PhD; Karolinska University Hospital
Acute Intermittent Porphyria

Sarquella-Brugada, Georgia, MD, PhD; Hospital Sant Joan de Deu
Brugada Syndrome

Sarret, Catherine, MD, PhD; Centre Hospitalier Universitaire de Clermont-Ferrand
Allan-Herndon-Dudley Syndrome

Sas, David J, DO; Mayo Clinic
Primary Hyperoxaluria Type 1
Primary Hyperoxaluria Type 3

Sassa, Shigeru, MD, PhD; The Rockefeller University
Acute Intermittent Porphyria

Sattler, Elke C, MD; Ludwig Maximilian University of Munich

Birt-Hogg-Dube Syndrome

Saulnier, Celine A, PhD; Neurodevelopmental Assessment & Consulting Services

3q29 Recurrent Deletion

Saul, Robert A, MD; Greenwood Genetics Center

FMR1 Disorders

Saunders-Pullman, Rachel, MD, MPH; Icahn School of Medicine at Mount Sinai / Mount Sinai Beth Israel

LRRK2 Parkinson Disease

SGCE Myoclonus-Dystonia

Saunders, Carol J, PhD; University of Missouri

Pitt-Hopkins Syndrome

Sauvé, Yves, PhD; University of Alberta

X-Linked Congenital Stationary Night Blindness

Savage, Sharon A, MD; National Cancer Institute, NIH

Dyskeratosis Congenita and Related Telomere Biology Disorders

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

Cystic Fibrosis

Savarese, Marco, PhD; University of Helsinki

Salih Myopathy

Savarirayan, Ravi, MBBS, MD; University of Melbourne

Chondrodysplasia Punctata 2, X-Linked

Fibrodysplasia Ossificans Progressiva

Osteopathia Striata with Cranial Sclerosis

Pycnodysostosis

Schmid Metaphyseal Chondrodysplasia

Thanatophoric Dysplasia

Type II Collagen Disorders Overview

Savoia, Anna, PhD; Institute for Maternal and Child Health - IRCCS Burlo Garofolo

MYH9-Related Disease

Scaglia, Fernando, MD; Baylor College of Medicine

Deoxyguanosine Kinase Deficiency

MELAS

Mitochondrial DNA Maintenance Defects Overview

MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect

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

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

TANGO2 Deficiency

TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Scarpa, Maurizio, MD, PhD; Horst Schmidt Klinik

Mucopolysaccharidosis Type II

Schaaf, Christian P, MD, PhD; Heidelberg University

15q13.3 Recurrent Deletion

NR2F1-Related Neurodevelopmental Disorder

Schaaf-Yang Syndrome

Xq28 Duplication Syndrome, Int22h1/Int22h2 Mediated

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

Genetic Hearing Loss Overview

Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

Schaefer, Franz, MD; Heidelberg University

Autosomal Recessive Polycystic Kidney Disease – PKHD1

Schanen, N Carolyn, MD, PhD; Dup15q Alliance

Maternal 15q Duplication Syndrome

Scharenberg, Andrew M, MD; University of Washington

X-Linked Severe Combined Immunodeficiency

Scharer, Gunter, MD, PhD; Denver Genetic Laboratories, University of Colorado

Nonketotic Hyperglycinemia

Scheffer, Ingrid E, MBBS, PhD; University of Melbourne

Autosomal Dominant Sleep-Related Hypermotor (Hyperkinetic) Epilepsy

FOXP2-Related Speech and Language Disorder

GRIN2A-Related Speech Disorders and Epilepsy

Scheper, Gert C, PhD; Vrije Universiteit Medical Center

Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation

Megalencephalic Leukoencephalopathy with Subcortical Cysts

Scherer, Gerd, PhD; University of Freiburg

Campomelic Dysplasia

Scheuerle, Angela E, MD; University of Texas Southwestern Medical Center

Incontinentia Pigmenti

Schiffmann, Raphael, MD, MHSc; Baylor Scott & White Research Institute

Childhood Ataxia with Central Nervous System Hypomyelination / Vanishing White Matter

Mucolipidosis IV

Schilsky, Michael, MD; Yale University School of Medicine
Wilson Disease

Schimke, R Neil, MD; University of Kansas Medical Center
Von Hippel-Lindau Syndrome

Schimmenti, Lisa A, MD; Mayo Clinic
Myhre Syndrome
NDP-Related Retinopathies
PAX2-Related Disorder

Schindler, Alice, MS; National Institute of Neurological Disorders and Stroke
Autosomal Dominant TRPV4 Disorders

Schinkel, Alfred, PhD; Netherlands Cancer Institute
Rotor Syndrome

Schirwani, Schaida, MB ChB, MSc; Sheffield Children's NHS Foundation Trust
ASXL3-Related Disorder

Schlade-Bartusiak, Kamilla, PhD; University of Alberta
Alpha-1 Antitrypsin Deficiency

Schlechter, Catherine, MS; Casey Eye Institute - Oregon Health Sciences University
Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview

Schlis, Krysta, MD; Children's Hospital & Research Center Oakland
Sickle Cell Disease

Schlotawa, Lars, MD; University Medical Center Göttingen
Multiple Sulfatase Deficiency

Schmale, Gregory A, MD; University of Washington
Hereditary Multiple Osteochondromas

Schmitz, Matthias, MD; University Medical School
Genetic Prion Disease

Schneider, Katherine A, MPH; Dana Farber Cancer Institute
Li-Fraumeni Syndrome

Schneider, Susanne A, MD, PhD; Ludwig Maximilian University
PINK1 Type of Young-Onset Parkinson Disease

Schneppenheim, Reinhard, MD, PhD; University Medical Center Hamburg – Eppendorf
Rhabdoid Tumor Predisposition Syndrome

Schon, Katherine Rose, MA, MB ChB; Addenbrooke's Hospital
Congenital Insensitivity to Pain Overview

Schoser, Benedikt, MD; Ludwig-Maximilians University

Myotonic Dystrophy Type 2

Schreiber, John M, MD; Children's National Hospital
SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders

Schreuder, Andrea B, MD, PhD; University Medical Center Groningen
Glycogen Storage Disease Type III

Schrier Vergano, Samantha A, MD; Children's Hospital of the King's Daughters
ARID1B-Related Disorder
Coffin-Siris Syndrome
IMAGe Syndrome

Schroer, Richard, MD; Greenwood Genetic Center
Epimerase Deficiency Galactosemia

Schuchman, Edward H, PhD; Icahn School of Medicine at Mount Sinai
Acid Sphingomyelinase Deficiency

Schuelke, Markus, MD; Charité - Universitätsmedizin Berlin
Ataxia with Vitamin E Deficiency

Schultz, Kris Ann P, MD; Children's Minnesota
DICER1 Tumor Predisposition
FH Tumor Predisposition Syndrome

Schulz, Ansgar, MD; Universitätsklinik für Kinder- und Jugendmedizin
CLCN7-Related Osteopetrosis

Schulze, Jeanine, MS; Indiana University School of Medicine
Parkinson Disease Overview

Schutte, Brian C, PhD; Michigan State University
IRF6-Related Disorders

Schwartz, Charles E, MS, PhD; Greenwood Genetic Center
RPS6KA3-Related Intellectual Disability
Snyder-Robinson Syndrome

Schwartz, David A, MD; University of Colorado Denver
Pulmonary Fibrosis Predisposition Overview

Schwartz, Mary E; PC Project
Pachyonychia Congenita

Schwartz, Stuart, PhD; Laboratory Corporation of America
Prader-Willi Syndrome

Schwarz, Guenter; Cologne University
Molybdenum Cofactor Deficiency

Scott, C Ronald, MD; University of Washington
Tyrosinemia Type I

Scott, Daryl A, MD, PhD; University of Utah
GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss
Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct
RERE-Related Disorders

Scriver, Charles R, MD; McGill University
Phenylalanine Hydroxylase Deficiency

Scruggs, Brittni A, MD, PhD; Mayo Clinic
NDP-Related Retinopathies

Seabra, Miguel C, MD, PhD; Imperial College School of Medicine
Choroideremia

Sebastio, Gianfranco, MD; Federico II University of Naples
Lysinuric Protein Intolerance

Seckington, Rebecca; University of Queensland School of Medicine
HFE-Related Hemochromatosis

Sedel, Frederic, MD, PhD; MedDay Pharmaceuticals
CLCN2-Related Leukoencephalopathy

Sekijima, Yoshiki, MD, PhD; Shinshu University School of Medicine
Hereditary Transthyretin Amyloidosis

Selber, Paulo, MD; Royal Children's Hospital Melbourne
PURA-Related Neurodevelopmental Disorders

Seminara, Stephanie, MD; Massachusetts General Hospital
Isolated Gonadotropin-Releasing Hormone (GnRH) Deficiency

Senaratne, T Niroshini, PhD; University of California, Los Angeles
NFIA-Related Disorder

Sentner, Christiaan P, MD; University Medical Center Groningen
Glycogen Storage Disease Type III

Sequerra Amram Cohen, Ana, PhD; Icahn School of Medicine at Mount Sinai
EED-Related Overgrowth

Sereda, Christina, MSc; National Institutes of Health
Choroideremia

Seront, Emmanuel, MD, PhD; Cliniques universitaires Saint-Luc
TEK-Related Venous Malformations

Sethi, Sanjeev, MD, PhD; Mayo Clinic

C3 Glomerulopathy

Shah, Hitesh, MS; Manipal Academy of Higher Education
Multicentric Osteolysis Nodulosis and Arthropathy
Progressive Pseudorheumatoid Dysplasia

Shakkottai, Vikram, MD, PhD; University of Michigan Medical School
Spinocerebellar Ataxia Type 3

Shapiro, Amy D, MD; Indiana Hemophilia & Thrombosis Center
Complete Plasminogen Activator Inhibitor 1 Deficiency

Sharer, J Daniel, PhD; University of Alabama at Birmingham
Duarte Variant Galactosemia

Sharpe, Lindsay T, MA, PhD; Institute of Ophthalmology
Achromatopsia

Shashi, Vandana, MD; Duke University
NGLY1-Related Congenital Disorder of Deglycosylation

Shaw, Adam, BMed, MD; Guy's & Saint Thomas' Hospitals
Trichorhinophalangeal Syndrome

Shaw, Chris, BS; University of Washington
ADCY5 Dyskinesia

Shchelochkov, Oleg A, MD; National Institutes of Health
Propionic Acidemia

Shearer, A Eliot, MD, PhD; Harvard Medical School
Genetic Hearing Loss Overview
OTOF-Related Deafness
STRC-Related Autosomal Recessive Hearing Loss

Sheen, Volney L, MD, PhD; Harvard Medical School
FLNA Deficiency

Sheffield, Val C, MD, PhD; University of Iowa College of Medicine
GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss
Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

Shelton, Celeste, MS, PhD; Ariel Precision Medicine
Pancreatitis Overview
PRSS1-Related Hereditary Pancreatitis

Shen, Yiping, PhD; Boston Children's Hospital
16p11.2 Recurrent Deletion

Shephard, Elizabeth A, MSc, PhD; University College London

Primary Trimethylaminuria

Sheppard, Sarah E, MD, PhD; Eunice Kennedy Shriver National Institute of Child Health and Human Development

Wiedemann-Steiner Syndrome

Sheridan, Eamonn, MD; University of Leeds

RAB18 Deficiency

Sherr, Elliott H, MD, PhD; University of California, San Francisco

DDX3X-Related Neurodevelopmental Disorder

RERE-Related Disorders

Shieh, Joseph TC, MD, PhD; University of California San Francisco

Hyaline Fibromatosis Syndrome

Shih, Vivian E, MD; Massachusetts General Hospital

Fumarate Hydratase Deficiency

Shiloh-Malawsky, Yael, MD; University of North Carolina at Chapel Hill

DRPLA

Shimamura, Akiko, MD, PhD; Dana Farber Cancer Institute

Fanconi Anemia

Shirvan, Leila, BA; National Human Genome Research Institute

HEXA Disorders

Shneider, Benjamin L, MD; University of Pittsburgh Medical Center

ATP8B1 Deficiency

Shoffner, John M, MD; Medical Neurogenetics, LLC

Mitochondrial Neurogastrointestinal Encephalopathy Disease

Shohat, Mordechai, MD; Maccabi Genetic Institute

Familial Dysautonomia

Familial Mediterranean Fever

Sholto-Douglas-Vernon, Carolyn, PhD; St George's University of London

Milroy Disease

Shooner, Kerry, MS; Cincinnati Children's Hospital Medical Center

Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Shukla, Anju, MD; Manipal Academy of Higher Education

ISCA1-Related Multiple Mitochondrial Dysfunctions Syndrome

Multicentric Osteolysis Nodulosis and Arthropathy

Progressive Pseudorheumatoid Dysplasia

Shultz, Sarah, PhD; Emory University School of Medicine

3q29 Recurrent Deletion

Shuman, Cheryl, MS; University of Toronto
Beckwith-Wiedemann Syndrome

Siddique, Nailah, MSN, RN; Northwestern University Feinberg School of Medicine
Amyotrophic Lateral Sclerosis Overview

Siddique, Teepu, DSc (Hon), MD; Northwestern University Feinberg Medical School
Amyotrophic Lateral Sclerosis Overview

Siebert, Reiner, MD; Ulm University & Ulm University Medical Center
Rhabdoid Tumor Predisposition Syndrome

Sieff, Colin, MBBCh; Boston Children's Hospital and Dana Farber Cancer Institute
Diamond-Blackfan Anemia

Sieni, Elena, MD; Meyer Children's University Hospital
Familial Hemophagocytic Lymphohistiocytosis

Sieving, Paul A, MD, PhD; University of California, Davis
X-Linked Congenital Retinoschisis

Silber, Sherman J, MD; Saint Luke's Hospital
Y Chromosome Infertility

Simons, Cas, PhD; University of Queensland
TUBB4A-Related Leukodystrophy

Simpson, Kara L, MS; Children's National Hospital
Ornithine Transcarbamylase Deficiency
Urea Cycle Disorders Overview

Sims, Katherine B, MD; Massachusetts General Hospital / Harvard Medical School
NDP-Related Retinopathies

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

Singer, Kaitlyn, MS; Geisinger Health System
16p11.2 Recurrent Deletion

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

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

Singh, Swati, MSc; Manipal Academy of Higher Education
EXOC6B-Related Spondyloepimetaphyseal Dysplasia with Joint Laxity

Sinnreich, Michael, MD, PhD; Montreal Neurological Institute

GNE Myopathy

Siper, Paige, PhD; Icahn School of Medicine at Mount Sinai
FOXP1 Syndrome

Sisodiya, Sanjay, MD, PhD; University College London
TBC1D24-Related Disorders

Sivakumar, Kumaraswamy, MD; St Joseph's Hospital and Medical Center
GARS-Associated Axonal Neuropathy

Skefos, Catherine M, MA, MS; The University of Texas MD Anderson Cancer Center
CDC73-Related Disorders

Skinner, Jonathan Robert, MB ChB, MD; University of Auckland
GNB5-Related Neurodevelopmental Disorder

Skraban, Cara M, MD; Children's Hospital of Philadelphia
WDR26-Related Intellectual Disability

Slattery, Susan M, MD, MS; Northwestern University
Congenital Central Hypoventilation Syndrome

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

Smith, Rebecca, MS; Geisinger Health System
16p11.2 Recurrent Deletion

Smith, Richard JH, MD; University of Iowa Hospitals and Clinics
Branchiootorenal Spectrum Disorder
C3 Glomerulopathy
DFNA2 Nonsyndromic Hearing Loss
Genetic Hearing Loss Overview
GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss
OTOF-Related Deafness
Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

Smith, Simone, MSc; Geisinger Health System
PACS1 Neurodevelopmental Disorder

Smith, Stephanie C, MS; University of Mississippi Medical Center
Spinocerebellar Ataxia Type 3

Sniderman King, Lisa, MSc; Genzyme Corporation
Tyrosinemia Type I

Snijders Blok, Lot, MD; Radboud University Medcal Center
DDX3X-Related Neurodevelopmental Disorder

Snow-Bailey, Karen, PhD; Auckland City Hospital
Multiple Endocrine Neoplasia Type 2

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

Soares, Ana Rita, MD; Santo Antonio University Hospital Center
Ellis-van Creveld Syndrome

Sobacchi, Cristina, MS; Humanitas Clinical and Research Center

CLCN7-Related Osteopetrosis

Sobeih, Magdi M, MD, PhD; Boston Children's Hospital
16p11.2 Recurrent Deletion

Sobrido, Maria J, MD, PhD; Fundación Pública Galega de Medicina Xenómica-SERGAS
Primary Familial Brain Calcification
Spinocerebellar Ataxia Type 36

So, Jodi Y, BA; Stanford School of Medicine
Epidermolysis Bullosa Simplex

Soldovieri, Maria Virginia, PhD; University of Molise
KCNQ2-Related Disorders
KCNQ3-Related Disorders

Soler-Alfonso, Claudia, MD; Baylor College of Medicine
TANGO2 Deficiency

Solomon, Benjamin D, MD; National Institutes of Health
Holoprosencephaly Overview

Solomon, Beth, MS; National Institutes of Health
Smith-Magenis Syndrome

Solomon, Cindy, MS; Myriad Genetic Laboratories, Inc
APC-Associated Polyposis Conditions

Solomon, Sheila, MS; GeneDx, Inc
Pancreatitis Overview
PRSS1-Related Hereditary Pancreatitis

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

Sotero de Menezes, Marcio A, MD; Swedish Neuroscience Institute
SCN1A-Related Seizure Disorders

Spacey, Sian, MD; University of British Columbia
Familial Paroxysmal Nonkinesigenic Dyskinesia

Sparks, Susan E, MD, PhD; Sanofi Genzyme
Congenital Disorders of N-Linked Glycosylation and Multiple Pathway Overview
LAMA2 Muscular Dystrophy
PMM2-CDG

Spence, Sarah J, MD, PhD; Children's Hospital Boston
Maternal 15q Duplication Syndrome

Sperandeo, Maria Pia, PhD; Federico II University

Lysinuric Protein Intolerance

Spiegel, Ronen, MD; Emek Medical Center
Mucolipidosis III Gamma

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

Splawski, Igor, PhD; Assistant, Children's Hospital Boston
CACNA1C-Related Disorders

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

Squires, James E, MD, MS; UPMC Children's Hospital of Pittsburgh
ATP8B1 Deficiency
Pediatric Genetic Cholestatic Liver Disease Overview

Sreedharan, Jemeen, BSc, MBBS, PhD; King's College London
TARDBP-Related Amyotrophic Lateral Sclerosis-Frontotemporal Dementia

Srivastava, Siddharth, MD; Boston Children's Hospital
Alexander Disease
SETBP1 Haploinsufficiency Disorder

Stanich, Peter P, MD; The Ohio State University Wexner Medical Center
APC-Associated Polyposis Conditions

Stanley, Charles A, MD; University of Pennsylvania
Permanent Neonatal Diabetes Mellitus

Starr, Lois J, MD, PhD; University of Nebraska Medical Center
Myhre Syndrome
SLC25A24 Fontaine Progeroid Syndrome

Statland, Jeffrey M, MD; University of Kansas Medical Center
Andersen-Tawil Syndrome

Staudt, Martin, MD; LMU Munich
FOXG1 Syndrome

Steeds, Rick, MD; University Hospitals
Alstrom Syndrome

Steeves, Marcie, MS; MassGeneral Hospital for Children
Pitt-Hopkins Syndrome

Steinberg, Steven J, PhD; ARUP Laboratories
Rhizomelic Chondrodysplasia Punctata Type 1
X-Linked Adrenoleukodystrophy

Zellweger Spectrum Disorder

Steiner, Robert D, MD; University of Wisconsin School of Medicine and Public Health
COL1A1/2 Osteogenesis Imperfecta
Sitosterolemia

Stein, Jennifer L, MS; Cleveland Clinic Genomic Medicine Institute
PTEN Hamartoma Tumor Syndrome

Steinlein, Ortrud K, MD, PhD; Ludwig Maximilian University of Munich
Birt-Hogg-Dube Syndrome

Steinman, Kyle J, MD; Seattle Children's Hospital
16p11.2 Recurrent Deletion

Steinmann, Beat, MD; University Children's Hospital
PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome

Stephens, Karen, PhD; University of Washington Medical Center
Epidermolysis Bullosa Simplex

Stephenson, John BP; Royal Hospital for Sick Children
Aicardi-Goutieres Syndrome

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

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

Stevanin, Giovanni, PhD; Hôpital de la Pitié-Salpêtrière
Spastic Paraplegia 11

Stevens, Cathy A, MD; University of Tennessee College of Medicine
Rubinstein-Taybi Syndrome

Stevenson, David A, MD; Stanford University
Capillary Malformation-Arteriovenous Malformation Syndrome
Hereditary Hemorrhagic Telangiectasia
Legius Syndrome

Stevenson, Roger E, MD; Greenwood Genetic Center
Alpha-Thalassemia X-Linked Intellectual Disability Syndrome
Snyder-Robinson Syndrome

Stewart, Blythe; University of Edinburgh
MYRF-Related Cardiac Urogenital Syndrome

Stewart, Douglas R, MD; National Cancer Institute
DICER1 Tumor Predisposition

Stojinski, Carol, MD, MS; Drexel University College of Medicine
Mitochondrial Short-Chain Enoyl-CoA Hydratase 1 Deficiency

Stojkovic, Tanya, MD; Institut de Myologie
Oculopharyngeal Muscular Dystrophy

Stokman, Marijn, MD, PhD; Radboud University Medical Centre
Nephronophthisis-Related Ciliopathies

Stolle, Catherine A, PhD; Children's Hospital of Philadelphia
Von Hippel-Lindau Syndrome

Stoller, James K, MD, MS; Cleveland Clinic
Alpha-1 Antitrypsin Deficiency

Stone, Jeremy T, BSc; Mayo Clinic
LRRK2 Parkinson Disease

Storey, Elsdon, DPhil; Monash University
Spinocerebellar Ataxia Type 20

Stover, Niamh B, MS; Banner Health
Bietti Crystalline Dystrophy

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

Stratton, Pamela, MD; National Institute of Neurological Disorders and Stroke
DICER1 Tumor Predisposition

Strauss, Arnold W, MD; Cincinnati Children's Hospital Medical Center
Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Strauss, Kevin A, MD; Clinic for Special Children
Maple Syrup Urine Disease

Strober, Jonathan, MD; UCSF / Benioff Children's Hospital
Sphingosine Phosphate Lyase Insufficiency Syndrome

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

Stumpel, Connie, MD, PhD; Academic Hospital Maastricht
L1 Syndrome

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
CHST3-Related Skeletal Dysplasia
Diastrophic Dysplasia
SLC26A2-Related Multiple Epiphyseal Dysplasia

Sutton, V Reid, MD; Baylor College of Medicine
Aicardi Syndrome
PORCN-Related Developmental Disorders
TP63-Related Disorders
White-Sutton Syndrome

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

Alkaptonuria

Suzuki, Tamio, MD, PhD; Yamagata University
Oculocutaneous Albinism Type 4

Sveden, Abigail, MS; Boston Children's Hospital
ANKRD17-Related Neurodevelopmental Syndrome

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

Sweadner, Kathleen J, PhD; Massachusetts General Hospital
ATP1A3-Related Neurologic Disorders

Sweeney, Elizabeth, MB ChB, MD; Liverpool Women's Hospital
Nail-Patella Syndrome

Sweeney, William E, MS; Medical College and Children's Health System of Wisconsin
Autosomal Recessive Polycystic Kidney Disease – PKHD1

Sweetser, David A, MD, PhD; Massachusetts General Hospital
GNAO1-Related Disorder
Pitt-Hopkins Syndrome

Swoboda, Kathryn J, MD; Massachusetts General Hospital
ATP1A3-Related Neurologic Disorders
Tyrosine Hydroxylase Deficiency

Sybert, Virginia P, MD; University of Washington
Epidermolysis Bullosa Simplex

Symoens, Sofie, PhD; Ghent University Hospital
Classic Ehlers-Danlos Syndrome

Synofzik, Matthis, MD; Center for Neurology & Hertie Institute for Clinical Brain Research
ARSACS
GAA-FGF14-Related Ataxia
PNPLA6 Disorders

Syx, Delfien, PhD; Ghent University Hospital
Classic Ehlers-Danlos Syndrome

T

Tabarki, Brahim, MD; Prince Sultan Military Medical City
Biotin-Thiamine-Responsive Basal Ganglia Disease
SLC25A19-Related Thiamine Metabolism Dysfunction

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

Hypokalemic Periodic Paralysis

Taglialatela, Maurizio, MD, PhD; University of Naples Federico II
KCNQ2-Related Disorders
KCNQ3-Related Disorders

Tait, Jonathan F, MD, PhD; University of Washington School of Medicine
Cystic Fibrosis
HFE-Related Hemochromatosis

Takahashi, Hitoshi, MD, PhD; Niigata University
Spinocerebellar Ataxia Type 17

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

Takasaki, Kaoru, MD; Children's Hospital of Philadelphia
GATA1-Related Cytopenia

Takashima, Hiroshi, MD, PhD; Kagoshima University Graduate School of Medical and Dental Sciences
Spinocerebellar Ataxia with Axonal Neuropathy, Autosomal Recessive

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
Huppke-Brendel Syndrome
Isolated Sulfite Oxidase Deficiency

Tamary, Hannah, MD; Schneider Children's Medical Center of Israel
Alpha-Thalassemia
Congenital Dyserythropoietic Anemia Type I

Tamburro, Joan, DO; Cleveland Clinic
Isolated and Classic Cutis Marmorata Telangiectatica Congenita

Tamura, Deborah, RN; National Cancer Institute
Xeroderma Pigmentosum

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

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 Gesú Children's Hospital IRCSS
Noonan Syndrome with Multiple Lentigines

Tatton-Brown, Katrina, BM BCh, MD; St George's University of London
EZH2-Related Overgrowth
HIST1H1E Syndrome
Sotos Syndrome
Tatton-Brown-Rahman Syndrome

Tawana, Kiran, MB ChB, PhD; Queen Mary University of London
CEBPA-Associated Familial Acute Myeloid Leukemia (AML)

Tawil, Rabi, MD; University of Rochester Medical Center
Andersen-Tawil Syndrome
Facioscapulohumeral Muscular Dystrophy

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

Taylor, Cora M, PhD; Geisinger Health System
16p11.2 Recurrent Deletion
CHD8-Related Neurodevelopmental Disorder with Overgrowth
PACS1 Neurodevelopmental Disorder

Taylor, Kelly A, MS; Vanderbilt Medical Center
ATP8B1 Deficiency

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

Taylor, Robert W, PhD; Newcastle University

RRM2B Mitochondrial DNA Maintenance Defects

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
Holoprosencephaly Overview

Tekin, Mustafa, MD; John P Hussman Institute for Human Genomics
Congenital Deafness with Labyrinthine Aplasia, Microtia, and Microdontia
Deafness and Myopia Syndrome
KBG Syndrome

Temple, Isabel Karen, MD; Southampton University Hospitals NHS Trust
Diabetes Mellitus, 6q24-Related Transient Neonatal

Teng, Joyce, MD, PhD; Stanford School of Medicine
Epidermolysis Bullosa Simplex

Terry, Sharon F, MA; PXE International
Pseudoxanthoma Elasticum

Terwindt, Gisela M, MD, PhD; Leiden University Medical Center
CADASIL
Retinal Vasculopathy with Cerebral Leukoencephalopathy and Systemic Manifestations

Testa, Giuseppe, MA, MD, PhD; European Institute of Oncology
Gabriele-de Vries Syndrome

Thabet, Farah, MD; Fattouma Bourguiba University Hospital
SLC25A19-Related Thiamine Metabolism Dysfunction

Thauvin-Robinet, Christel, MD, PhD; University of Burgundy
Oral-Facial-Digital Syndrome Type I

Thibert, Ronald L, DO, MSPH; Massachusetts General Hospital
Maternal 15q Duplication Syndrome
Pitt-Hopkins Syndrome

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
- Dihydrolipoamide Dehydrogenase Deficiency

Thomas, Julian, MD; Children's Hospital Orange County
- DDX3X-Related Neurodevelopmental Disorder

Thomas, Mervyn G, MBChB, PhD; University of Leicester
- FRMD7-Related Infantile Nystagmus
- Oculocutaneous Albinism and Ocular Albinism Overview

Thomas, Rhys, PhD; Newcastle University
- Hereditary Hyperekplexia Overview

Thomas, Shery; University of Leicester
- FRMD7-Related Infantile Nystagmus

Thompson, Arthur R, MD, PhD; University of Washington
- Hemophilia A
- Hemophilia B

Thompson, Reid, MD; Johns Hopkins University
- Barth Syndrome

Thorburn, David R, PhD; Royal Children's Hospital and University of Melbourne
- Mitochondrial DNA-Associated Leigh Syndrome Spectrum
- Nuclear Gene-Encoded Leigh Syndrome Spectrum Overview

Thorpe, Ryan K, MD; University of Iowa
- OTOF-Related Deafness

Thunell, Stig, MD, PhD; Karolinska Institute
- Acute Intermittent Porphyria

Tifft, Cynthia J, MD, PhD; National Human Genome Research Institute
- GLB1-Related Disorders
- GM2 Activator Deficiency
- HEXA Disorders
- Sandhoff Disease

Tijssen, Marina AJ, MD; University Medical Center Groningen
- Hereditary Hyperekplexia Overview

Tiller, George E, MD, PhD; Kaiser Permanente
- Hypochondroplasia
- X-Linked Spondyloepiphyseal Dysplasia Tarda

Timmerman, Vincent, PhD; Institute Born Bunge

GAN-Related Neurodegeneration

Timothy, Katherine W, BS; Harvard Medical School
CACNA1C-Related Disorders

Tinkle, Brad T, MD, PhD; Advocate Children's Hospital
Pompe Disease
Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

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

Tkachenko, Nataliya, MD; Santo Antonio University Hospital Center
Ellis-van Creveld Syndrome

Todd, Peter K, MD, PhD; University of Michigan School of Medicine
FMR1 Disorders

Tokuda, Takahiko, MD, PhD; Kyoto Prefectural University Hospital
Hereditary Transthyretin Amyloidosis

Tolar, Jakub, MD, PhD; University of Minnesota
Fanconi Anemia

Tonduti, Davide, MD, PhD; V Buzzi Children's Hospital
Allan-Herndon-Dudley Syndrome

Toone, Jennifer, BSc; Children's and Women's Health Centre of British Columbia
Nonketotic Hyperglycinemia

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

Toriello, Helga V, PhD; Michigan State University
Oral-Facial-Digital Syndrome Type I
Thrombocytopenia Absent Radius Syndrome

Toro, Camilo, MD; National Human Genome Research Institute
Chediak-Higashi Syndrome
GM2 Activator Deficiency
HEXA Disorders
Sandhoff Disease
SCARB2-Related Action Myoclonus – Renal Failure Syndrome

Toro, Jorge R, MD; National Institutes of Health
Birt-Hogg-Dube Syndrome
FH Tumor Predisposition Syndrome

Torres, Vicente E, MD, PhD; Mayo Clinic

Polycystic Kidney Disease, Autosomal Dominant

Toyoshima, Yasuko, MD, PhD; Agano Hospital
Spinocerebellar Ataxia Type 17

Traboulsi, Elias I, MD, MEd; Cleveland Clinic Lerner College of Medicine
Cohen Syndrome
Isolated and Classic Cutis Marmorata Telangiectatica Congenita

Trahms, Cristine, MS, RD; University of Washington
Tyrosinemia Type I

Tran-Fadulu, Van, MS; University of Texas Medical School at Houston
Heritable Thoracic Aortic Disease Overview

Tranebjærg, Lisbeth, MD, PhD; University of Copenhagen
Deafness-Dystonia-Optic Neuronopathy Syndrome
Jervell and Lange-Nielsen Syndrome
WFS1 Spectrum Disorder

Trevisson, Eva, MD, PhD; University of Padova
Primary Coenzyme Q10 Deficiency Overview

Trifiro, Mark A, MD; McGill University
Androgen Insensitivity Syndrome

Trinh, Joanne, BSc; University of British Columbia
LRRK2 Parkinson Disease

Trollet, Capucine, PhD; Centre de Recherche en Myologie
Oculopharyngeal Muscular Dystrophy

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

Trzupek, Karmen M, MS; InformedDNA
Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview
Usher Syndrome Type I
Usher Syndrome Type II

Tsang, Mandy HY, MMSc; University of Hong Kong
CTNNB1 Neurodevelopmental Disorder

Tsilou, Ekaterini, MD; Center for Biologics Evaluation and Research, US FDA
Weill-Marchesani Syndrome

Tsuji, Shoji, MD, PhD; University of Tokyo Graduate School of Medicine
DRPLA
Spinocerebellar Ataxia Type 17

Tsunogai, Toshiki, MD; Jikei University School of Medicine
Thiamine-Responsive Megaloblastic Anemia Syndrome

Tuchman, Mendel, MD; Children's National Medical Center
Urea Cycle Disorders Overview

Turner, Joyce T, MS; Children's National Hospital
Wilms Tumor Predisposition

Turner, Stefanie, MS; 17q12 Foundation
17q12 Recurrent Deletion Syndrome

Turnpenny, Peter D, BSc, ChB, MB; Royal Devon University Healthcare NHS Foundation Trust
Spondylocostal Dysostosis, Autosomal Recessive

Tuschl, Karin, MD, PhD; UCL Great Ormond Street Institute of Child Health
Hypermanganesemia with Dystonia 1
SLC39A14 Deficiency

Tuupanen, Sari, PhD; Blueprint Genetics
X-Linked Congenital Stationary Night Blindness

Tümer, Zeynep, DMSc, MD, PhD; Rigshospitalet
DLG4-Related Synaptopathy

Tüysüz, Beyhan, MD, PhD; Istanbul University
Trichorhinophalangeal Syndrome

U

Udd, Bjarne, MD, PhD; Tampere University Hospital
ANO5 Muscle Disease
Salih Myopathy
Udd Distal Myopathy - Tibial Muscular Dystrophy

Uhlig, Holm; NIHR Oxford Biomedical Research Centre
PI4KA-Related Disorder

Uitto, Jouni, MD, PhD; Thomas Jefferson University
Kindler Syndrome
Lipoid Proteinosis
Pseudoxanthoma Elasticum

Umair, Muhammad, PhD; King Saud Bin Abdulaziz University for Health Sciences
EMC10-Related Neurodevelopmental Disorder

Unger, Sheila, MD; University of Lausanne
Achondrogenesis Type 1B
Atelosteogenesis Type 2

Campomelic Dysplasia
CHST3-Related Skeletal Dysplasia
Diastrophic Dysplasia
SLC26A2-Related Multiple Epiphyseal Dysplasia

Upadia, Jariya, MD; Tulane University School of Medicine
Cystic Fibrosis

Urban, Zsolt, PhD; University of Pittsburgh
EFEMP2-Related Cutis Laxa
ELN-Related Cutis Laxa
LTBP4-Related Cutis Laxa

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

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

Usami, Shin-ichi, MD, PhD; Shinshu University School of Medicine
Nonsyndromic Hearing Loss and Deafness, Mitochondrial

Uyanik, Gökhan, MD; Hanusch Hospital
CASK Disorders
DCX-Related Disorders

V

Vahidnezhad, Hassan, MSc, PhD; Pasteur Institute of Iran
Kindler Syndrome
Lipoid Proteinosis

Vakkilainen, Svetlana, MD, PhD; University of Helsinki and Helsinki University Hospital
Cartilage-Hair Hypoplasia - Anauxetic Dysplasia Spectrum Disorders

Valdmanis, Paul N, PhD; University of Washington
Spastic Paraplegia 8

Valencia, C Alexander, PhD; Cincinnati Children's Hospital Medical Center
Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency

Valentin, Ilia, MD; Heidelberg University
NR2F1-Related Neurodevelopmental Disorder

Valeriani, Emanuele, MD; Sapienza University of Rome
Factor V Leiden Thrombophilia

Valle, Laura, PhD; Catalan Institute of Oncology
Lynch Syndrome

Valverde de Morales, Hannah Gabriela, MD; Integra Genetics
- CTCF-Related Disorder

van Belzen, Martine, PhD; Leiden University Medical Center
- Peters Plus Syndrome

van Bokhoven, Hans, PhD; Radboud University Medical Center
- TP63-Related Disorders

van Bon, Bregje WM, MD, PhD; Radboud University Medical Center
- 15q13.3 Recurrent Deletion
- DYRK1A Syndrome
- Schinzel-Giedion Syndrome
- SETBP1 Haploinsufficiency Disorder

Van Broeckhoven, Christine, DSc, PhD; University of Antwerp
- C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia

Van Camp, Guy, PhD; University of Antwerp
- Genetic Hearing Loss Overview
- GJB2-Related Autosomal Recessive Nonsyndromic Hearing Loss
- Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

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

Van den Veyver, Ignatia Barbara, MD; Baylor College of Medicine
- Aicardi Syndrome
- PORCN-Related Developmental Disorders

van der Crabben, Saskia N, MD, PhD; Amsterdam UMC
- Serine Deficiency Disorders

van der Knaap, Marjo S, MD, PhD; Amsterdam University Medical Centers
- Childhood Ataxia with Central Nervous System Hypomyelination / Vanishing White Matter
- CLCN2-Related Leukoencephalopathy
- Hypomyelination and Congenital Cataract
- Leukoencephalopathy with Brain Stem and Spinal Cord Involvement and Lactate Elevation
- Megalencephalic Leukoencephalopathy with Subcortical Cysts
- TUBB4A-Related Leukodystrophy

van der Maarel, Silvere M, MD; Leiden University Medical Center
- Facioscapulohumeral Muscular Dystrophy

van der Sluijs, Pleuntje J, MSc; Leiden University Medical Center
- ARID1B-Related Disorder

van der Zee, Julie, PhD; University of Antwerp

C9orf72-Related Amyotrophic Lateral Sclerosis and/or Frontotemporal Dementia

Van Dijck, Anke, MD, PhD; University of Antwerp
ADNP-Related Disorder

van Dijk, Fleur S, MD, PhD; London North West University Healthcare NHS Trust
Periodontal Ehlers-Danlos Syndrome
TNXB-Related Classical-Like Ehlers-Danlos Syndrome

van Dijk, Tessa, MD, PhD; Leiden University Medical Center
EXOSC3 Pontocerebellar Hypoplasia
TSEN54 Pontocerebellar Hypoplasia

Van Esch, Hilde, MD, PhD; University Hospitals Leuven
MECP2 Duplication Syndrome

Van Hauwe, Peter; University of Antwerp
Pendred Syndrome / Nonsyndromic Enlarged Vestibular Aqueduct

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
PLPBP Deficiency

van Leeuwaarde, 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
ATP6V0A2-Related Cutis Laxa
Baller-Gerold Syndrome
Berardinelli-Seip Congenital Lipodystrophy
FBLN5-Related Cutis Laxa

van Nesselrooij, Bernadette, MD, PhD; University Medical Center Utrecht
Von Hippel-Lindau Syndrome

van Os, Nienke, MD, PhD; Radboud University Medical Center
Ataxia-Telangiectasia

van Ravenswaaij-Arts, Conny M, MD, PhD; University Medical Center Groningen
CHD7 Disorder

van Spaendonk, Rosalina ML, PhD; Amsterdam University Medical Center
PLP1 Disorders

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

van Swieten, John C, MD, PhD; Erasmus Medical Center
MAPT-Related Frontotemporal Dementia

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

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
Pelizaeus-Merzbacher-Like Disease 1
POLR3-Related Leukodystrophy
TUBB4A-Related Leukodystrophy

Vandeweyer, Geert, PhD; Antwerp University Hospital
ADNP-Related Disorder

VanSickle, Elizabeth, MS; Spectrum Health
Bachmann-Bupp Syndrome

Vargas, Mauricio, MD, PhD; Oregon Health Sciences University
Bietti Crystalline Dystrophy

Varon, Raymonda, PhD; Charité – Universitätsmedizin Berlin
Nijmegen Breakage Syndrome

Varvagiannis, Konstantinos, MD; Cyprus Institute of Neurology & Genetics
TRIO-Related Neurodevelopmental Disorder
WAC-Related Intellectual Disability

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
SLC25A24 Fontaine Progeroid Syndrome

Velayos Baeza, Antonio, PhD; Wellcome Trust Centre for Human Genetics
VPS13A Disease

Velez-Bartolomei, Frances, MD; Stanford University
MERRF

Velleman, Shelley L, PhD; University of Vermont
7q11.23 Duplication Syndrome

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
Long QT Syndrome Overview

Viskochil, David H, MD, PhD; University of Utah
BCL11A-Related Intellectual Disability
Legius Syndrome

Vissers, Lisenka ELM, PhD; Radboud University Medical Center
Squalene Synthase Deficiency
TRIO-Related Neurodevelopmental Disorder
WAC-Related Intellectual Disability

Vissing, John, DMSc, MD; Rigshospitalet
Myotonia Congenita

Vnencak-Jones, Cindy, PhD; GC Vanderbilt University Medical Center
PROP1-Related Combined Pituitary Hormone Deficiency

Vockley, Jerry, MD, PhD; University of Pittsburgh
Short-Chain Acyl-CoA Dehydrogenase Deficiency

Vogel-Farley, Vanessa, BA; Dup15q Alliance
Maternal 15q Duplication Syndrome

Volpini, Victor, MD, PhD; Bellvitge Biomedical Research Institute
Spinocerebellar Ataxia Type 37

Vos, Yvonne J, PhD; University Medical Center Groningen
L1 Syndrome

W

Wagner, Klaus, MD, PhD; Medical University Graz
BSCL2-Related Neurologic Disorders / Seipinopathy

Wagner, Victoria F, MS; McGovern Medical School
Mucopolysaccharidosis Type III

Wagnon, Jacy L, PhD; University of Michigan
SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders

Waguespack, Steven G, MD; University of Texas MD Anderson Cancer Center
CDC73-Related Disorders

Wakefield, Emily, MS; Cincinnati Children's Hospital Medical Center
X-Linked Lymphoproliferative Disease

Waldman, Amy; Children's Hospital of Philadelphia
Alexander Disease

Walker, Elaine F, PhD; Emory University
3q29 Recurrent Deletion

Walker, Ruth H, MBBS, MD, PhD; Mount Sinai School of Medicine
McLeod Neuroacanthocytosis Syndrome
VPS13A Disease

Walkovich, Kelly, MD; University of Michigan Medical School
ZAP70-Related Combined Immunodeficiency

Wallace, Stephanie E, MD; Seattle Children's Hospital
Camurati-Engelmann Disease

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

Walsh, Christopher A, MD, PhD; Harvard Medical School
FLNA Deficiency

Wanders, Ronald JA, PhD; Academic Medical Center, University of Amsterdam
Adult Refsum Disease

Wang, Bruce, MD; University of California San Francisco
Hereditary Coproporphyria

Wang, Dong, MD; Emory University Affiliated Hospital
Glucose Transporter Type 1 Deficiency Syndrome
Pyruvate Carboxylase Deficiency

Wang, Heng, MD, PhD; DDC Clinic – Center for Special Needs Children

Cohen Syndrome
GM3 Synthase Deficiency
Prolidase Deficiency

Wang, Hsiao-Lin, PhD; Emory University School of Medicine
CTCF-Related Disorder

Wang, Hua, MD, PhD; Loma Linda University
MBTPS1-Related Spondyloepimetaphyseal Dysplasia with Elevated Lysosomal Enzymes

Wang, Jing, MD; Baylor College of Medicine
TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Wang, Julia, BS; Baylor College of Medicine
MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect
TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Wang, Leo H, MD, PhD; University of Washington
Facioscapulohumeral Muscular Dystrophy

Wangler, Michael F, MD; Baylor College of Medicine
ACTG2 Visceral Myopathy
Xia-Gibbs Syndrome

Wang, Lisa L, MD; Baylor College of Medicine
Baller-Gerold Syndrome
Poikiloderma with Neutropenia
Rothmund-Thomson Syndrome

Wang, Raymond, MD; University of California, Irvine
Mucopolysaccharidosis Type VII

Wang, Zhao, MD; Beijing Friendship Hospital
Familial Hemophagocytic Lymphohistiocytosis

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

Warby, Simon C, PhD; University of Montreal
Huntington Disease

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

Wasserstein, Melissa P, MD; Albert Einstein College of Medicine
Acid Sphingomyelinase Deficiency
Free Sialic Acid Storage Disorders
Krabbe Disease

Wassif, Christopher A, PhD; National Institute of Child Health and Human Development
Smith-Lemli-Opitz Syndrome

Waterham, Hans R, PhD; Academic Medical Center, University of Amsterdam
Adult Refsum Disease
Squalene Synthase Deficiency

Waters, Aoife M, BCh, MB BAO, MSc; University College London
Bardet-Biedl Syndrome Overview

Waters, Michael F, MD, MS, PhD; Barrow Neurological Institute
Spinocerebellar Ataxia Type 13

Wattendorf, Daniel J, MD; National Institutes of Health
Xeroderma Pigmentosum

Watts, Giles, PhD; University of East Anglia
Inclusion Body Myopathy with Paget Disease of Bone and/or Frontotemporal Dementia

Waxman, Stephen G, MD, PhD; Yale University School of Medicine
SCN9A Neuropathic Pain Syndromes

Weaver, K Nicole, MD; Cincinnati Children's Hospital Medical Center
HRAS-Related Costello Syndrome
Sphingosine Phosphate Lyase Insufficiency Syndrome

Weaver, W Curtis, BS; Geisinger Health System
16p11.2 Recurrent Deletion

Webb, Bryn D, MD, PhD; Icahn School of Medicine at Mount Sinai
STAC3 Disorder

Weber, Frank, MD, PhD; German Air Force Center of Aviation Medicine
Hyperkalemic Periodic Paralysis
Hypokalemic Periodic Paralysis

Weckhuysen, Sarah, MD, PhD; University of Antwerp
DEPDC5-Related Epilepsy
KCNQ2-Related Disorders
KCNQ3-Related Disorders

Weemaes, Corry, MD, PhD; Radboud University Medical Center
Ataxia-Telangiectasia

Weese-Mayer, Debra E, MD; Children's Memorial Hospital
Congenital Central Hypoventilation Syndrome

Wei, Chongjuan, PhD; University of Texas MD Anderson Cancer Center
Peutz-Jeghers Syndrome

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
Hereditary Fructose Intolerance

Weiss, Karin, MD; Technion-Israel Institute of Technology
CHD4 Neurodevelopmental Disorder

Weiss, Karl Heinz, MD; Salem Medical Center
Wilson Disease

Weiss, Marjan M, MD, PhD; Leiden University Medical Center
Peters Plus Syndrome

Weiss, Mitchell J, MD, PhD; St Jude Children's Research Hospital
GATA1-Related Cytopenia

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

Weksberg, Rosanna, MD, PhD; The Hospital for Sick Children
Beckwith-Wiedemann Syndrome

Weleber, Richard G, MD; Casey Eye Institute of Oregon Health Sciences University
Bietti Crystalline Dystrophy
Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview
Nonsyndromic Retinitis Pigmentosa Overview

Welsh, Holly I, MS; University of Missouri
Pitt-Hopkins Syndrome

Wenger, David A, PhD; Thomas Jefferson University Hospital
Krabbe Disease

Wenger, Olivia K, MD; New Leaf Center
HYAL2 Deficiency

Wenger, Tara L, MD, PhD; Seattle Children's Hospital
Apert Syndrome
FGFR Craniosynostosis Syndromes Overview

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

Wenstrup, Richard, MD; Cincinnati Children's Hospital Medical Center
Classic Ehlers-Danlos Syndrome
PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome

Westbroek, Wendy, PhD; National Institutes of Health
- Chediak-Higashi Syndrome

Weston, Michael D, MA; Boys Town National Research Hospital
- Usher Syndrome Type I
- Usher Syndrome Type II

Wevers, Ron A, PhD; Radboud University Nijmegen Medical Center
- CLPB Deficiency
- SERAC1 Deficiency
- Squalene Synthase Deficiency

Whatley, Sharon D, MSc, PhD; University Hospital of Wales
- Acute Intermittent Porphyria

Whitcomb, David C, MD, PhD; University of Pittsburgh & University of Pittsburgh Medical Center
- Pancreatitis Overview
- PRSS1-Related Hereditary Pancreatitis

White-Brown, Alexandre M, MSc; Children's Hospital of Eastern Ontario
- DLG4-Related Synaptopathy

White, Colin T, MD; British Columbia's Children's and Women's Hospital
- Primary Hyperoxaluria Type 1

White, Janson, PhD; Invitae Corporation
- White-Sutton Syndrome

White, Stormi; Emory University School of Medicine
- 3q29 Recurrent Deletion

White, Susan M, MD; Victorian Clinical Genetics Service
- Floating-Harbor Syndrome

Whitman, Mary C, MD, PhD; Harvard Medical School
- Congenital Fibrosis of the Extraocular Muscles Overview
- Duane Syndrome

Whitton, Chloe, BMed, MD; University of New South Wales
- ATN1-Related Neurodevelopmental Disorder

Wider, Christian, MD; Centre Hospitalier Universitaire Vaudois
- DCTN1-Related Neurodegeneration

Wieczorek, Dagmar, MD; Universitätsklinikum Düsseldorf
- Coffin-Siris Syndrome
- TXNL4A-Related Craniofacial Disorders

Wierenga, Andrea, PhD; SIVOTEC Bioinformatics

MBTPS1-Related Spondyloepimetaphyseal Dysplasia with Elevated Lysosomal Enzymes

Wierenga, Klaas, MD; Mayo Clinic Florida
MBTPS1-Related Spondyloepimetaphyseal Dysplasia with Elevated Lysosomal Enzymes
SPTBN4 Disorder

Wieser, Thomas, MD; Fachkrankenhaus Jerichow
Carnitine Palmitoyltransferase II Deficiency

Wiesner, Georgia L, MD, MS; Children's Hospital of Philadelphia
Multiple Endocrine Neoplasia Type 2

Wilcox, William R, MD, PhD; Emory University School of Medicine
Camurati-Engelmann Disease

Wildin, Robert S, MD; Oregon Health Sciences University
Hereditary Nephrogenic Diabetes Insipidus

Wilkie, Andrew OM, MA; University of Oxford
Enlarged Parietal Foramina

Wilkin, Douglas J; Federal Bureau of Investigation
Achondroplasia

Willemsen, Michèl, MD, PhD; Radboud University Medical Center
Ataxia-Telangiectasia

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

Williams, Denise, MD; Birmingham Women's and Children's Hospital
Alstrom Syndrome
WFS1 Spectrum Disorder

Williams, Gretchen, BS; Children's Hospitals and Clinics of Minnesota
DICER1 Tumor Predisposition

Williamson, Kathleen A, PhD; University of Edinburgh
SOX2 Disorder

Wilson, Frederick H, MD, PhD; Harvard Medical School
Pseudohypoaldosteronism Type II

Winkler, Jürgen, MD; University Hospital Erlangen
DCX-Related Disorders

Winter, Robin M; Institute of Child Health
Three M Syndrome

Wise, Anastasia L, PhD; National Jewish Health and University of Colorado Denver
Pulmonary Fibrosis Predisposition Overview

Wissinger, Bernd, MSc, PhD; University of Tübingen
Achromatopsia

Wiwattanadittakul, Natrujee, MD; Chiang Mai University
Succinic Semialdehyde Dehydrogenase Deficiency

Wlodarski, Marcin, MD, PhD; St Jude Children's Research Hospital
Monosomy 7 Predisposition Syndromes Overview

Wojcieszek, Joanne, MD; Indiana University School of Medicine
Parkinson Disease Overview

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

Wolfe, Lynne, ARNP, MS; National Human Genome Research Institute
NGLY1-Related Congenital Disorder of Deglycosylation
Short-Chain Acyl-CoA Dehydrogenase Deficiency

Wolf, Nicole I, MD, PhD; Amsterdam University Medical Centers
Hypomyelination and Congenital Cataract
PI4KA-Related Disorder
PLP1 Disorders

Wollnik, Bernd, MD; University of Cologne
Coffin-Siris Syndrome

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

Wong, Lee-Jun C, PhD; Baylor College of Medicine
Deoxyguanosine Kinase Deficiency
FARS2 Deficiency
FBXL4-Related Encephalomyopathic Mitochondrial DNA Depletion Syndrome
Mitochondrial DNA Maintenance Defects Overview
MPV17-Related Hepatocerebral Mitochondrial DNA Maintenance Defect
TK2-Related Mitochondrial DNA Maintenance Defect, Myopathic Form

Wong, Melanie, MBBS, PhD; Children's Hospital Westmead
Hepatic Veno-Occlusive Disease with Immunodeficiency

Woods, Christopher Geoffrey, MB ChB; University of Cambridge
Congenital Insensitivity to Pain Overview

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

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

CLPB Deficiency
SERAC1 Deficiency

Wright, Benjamin, MD; University Hospitals Birmingham
WFS1 Spectrum Disorder

Wright, Galen EB, PhD; University of British Columbia
Huntington Disease

Wright, J Timothy, DDS, MS; University of North Carolina
Hypohidrotic Ectodermal Dysplasia

Wright, Michael J, ChB, MB, MSc; Institute of Human Genetics
COMP-Related Pseudoachondroplasia
Multiple Epiphyseal Dysplasia, Dominant

Wszolek, Zbigniew K, MD; Mayo Clinic
CSF1R-Related Disorder
DCTN1-Related Neurodegeneration
DYT-GNAL
VPS35-Related Parkinson Disease

Wu, Bai-Lin, M Med, PhD; Boston Children's Hospital
16p11.2 Recurrent Deletion

Wuyts, Wim, PhD; University and University Hospital of Antwerp
Hereditary Multiple Osteochondromas

X

Xia, Maya, BA; COMBINEDBrain
SCN8A-Related Epilepsy and/or Neurodevelopmental Disorders

Xiao, Changrui, MD; University of California Irvine
GM2 Activator Deficiency
Sandhoff Disease
SCARB2-Related Action Myoclonus – Renal Failure Syndrome

Xin, Baozhong, PhD; DDC Clinic – Center for Special Needs Children
GM3 Synthase Deficiency

Xu, Manlong, MD, PhD; University of Alberta
Choroideremia

Y

Yamada, Koki, MD, PhD; Children's Hospital Boston
Congenital Fibrosis of the Extraocular Muscles Overview

Yamada, Mitsunori, MD, PhD; Shinshu University School of Medicine
Spinocerebellar Ataxia Type 17

Yamanaka, Koji, MD, PhD; RIKEN Brain Science Institute
ALS2-Related Disorder

Yang, Jiandong, PhD; George Washington University School of Medicine
DICER1 Tumor Predisposition

Yang, Paul, MD, PhD; Casey Eye Institute - Oregon Health Sciences University
Bietti Crystalline Dystrophy
Leber Congenital Amaurosis / Early-Onset Severe Retinal Dystrophy Overview

Yang, Yaping, PhD; AiLife Diagnostics
TANGO2 Deficiency

Yap, Kai Lee, PhD; Northwestern University
Congenital Central Hypoventilation Syndrome

Yates, T Michael, MBChB; University of Edinburgh
SOX2 Disorder

Yau, Mabel, MD; Mount Sinai School of Medicine
21-Hydroxylase-Deficient Congenital Adrenal Hyperplasia

Yehia, Lamis, PhD; Cleveland Clinic
PTEN Hamartoma Tumor Syndrome

Yen, Timothy, MD; University of Colorado Anschutz Medical Center
APC-Associated Polyposis Conditions

Yeowell, Heather N, PhD; Duke University Medical Center
PLOD1-Related Kyphoscoliotic Ehlers-Danlos Syndrome

Yntema, Helger G, PhD; Radboud University Medical Center
Kleefstra Syndrome

Yonker, Lael, MD; MassGeneral Hospital for Children
Pitt-Hopkins Syndrome

Yoshida, Kunihiro, MD, PhD; Shinshu University Hospital
Hereditary Transthyretin Amyloidosis

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

Young, Danielle O, MS; Kimball Genetics, Inc
Celiac Disease

Young, Elizabeth, BSc, PhD; Royal Devon and Exeter Healthcare NHS Trust
Spondylocostal Dysostosis, Autosomal Recessive

Young, William F, MD, MSc; Mayo Clinic
Hereditary Paraganglioma-Pheochromocytoma Syndromes

Youssefian, Leila, MSc, PhD; Thomas Jefferson University
Kindler Syndrome
Lipoid Proteinosis

Yu-Wai-Man, Patrick, BMedSci, MBBS, PhD; University of Cambridge
Leber Hereditary Optic Neuropathy
NR2F1-Related Neurodevelopmental Disorder

Yuan, Bo, PhD; Baylor College of Medicine
Potocki-Lupski Syndrome

Z

Zackai, Elaine H, MD; Children's Hospital of Philadelphia
22q11.2 Deletion Syndrome
Emanuel Syndrome

Zadeh, Neda, MD; CHOC Children's Hospital
KCNK9 Imprinting Syndrome

Zaki, Maha S, MD, PhD; National Research Centre
UNC80 Deficiency

Zandee, Wouter, MD, PhD; University Medical Center Groningen
Von Hippel-Lindau Syndrome

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

Zarate, Yuri A, MD; University of Arkansas for Medical Sciences
SATB2-Associated Syndrome

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

Zbuk, Kevin M, MD; Cleveland Clinic Genomic Medicine Institute
PTEN Hamartoma Tumor Syndrome

Zelley, Kristin, MS; Children's Hospital of Philadelphia
Li-Fraumeni Syndrome

Zerr, Inga, MD; University Medical School
Genetic Prion Disease

Zhai, Yi, MD, PhD; University of Alberta
Choroideremia

Zhang, Kejian, MBA, MD; GoBroad Healthcare Group
- Autoimmune Lymphoproliferative Syndrome
- Familial Hemophagocytic Lymphohistiocytosis
- Very Long-Chain Acyl-Coenzyme A Dehydrogenase Deficiency
- WAS-Related Disorders
- X-Linked Hyper IgM Syndrome
- X-Linked Lymphoproliferative Disease

Ziegler, John B, MBBS, MD; Sydney Children's Hospital
- Hepatic Veno-Occlusive Disease with Immunodeficiency

Ziegler, Shira G, MD, PhD; Johns Hopkins University School of Medicine
- Generalized Arterial Calcification of Infancy

Zipfel, Peter F, PhD; Hans Knoll Institute
- C3 Glomerulopathy

Zippin, Jonathan, MD, PhD; Joan and Sanford I Weill Medical College of Cornell University
- Oculocutaneous Albinism and Ocular Albinism Overview

Zobor, Ditta, MD, PhD; University of Tübingen
- Achromatopsia

Zoghbi, Huda Y, MD; Baylor College of Medicine
- MECP2 Disorders
- Spinocerebellar Ataxia Type 1

Zschocke, Johannes, MD, PhD; Medical University of Innsbruck
- Periodontal Ehlers-Danlos Syndrome

Züchner, Stephan, MD, PhD; University of Miami Miller School of Medicine
- GAA-FGF14-Related Ataxia
- GDAP1-Related Hereditary Motor and Sensory Neuropathy
- MFN2 Hereditary Motor and Sensory Neuropathy
- PNPLA6 Disorders

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Živná, Martina, PhD; Charles University
- Autosomal Dominant Tubulointerstitial Kidney Disease -- MUC1
- Autosomal Dominant Tubulointerstitial Kidney Disease -- REN
- Autosomal Dominant Tubulointerstitial Kidney Disease -- UMOD

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