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Macrocephaly and developmental delay caused by missense variants in RAB5C

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HNRNPC haploinsufficiency affects alternative splicing of intellectual disability-associated genes and causes a neurodevelopmental disorder

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Heterozygous rare variants in NR2F2 cause a recognizable multiple congenital anomaly syndrome with developmental delays

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Monoallelic variation in DHX9, the gene encoding the DExH-box helicase DHX9, underlies neurodevelopment disorders and Charcot-Marie-Tooth disease

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American Journal of Human Genetics

A de novo missense variant in EZH1 associated with developmental delay exhibits functional deficits in Drosophila melanogaster

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Dominant negative variants in IKZF2 cause ICHAD syndrome, a new disorder characterised by immunodysregulation, craniofacial anomalies, hearing impairment, athelia and developmental delay

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Dominant-negative variant in SLC1A4 causes an autosomal dominant epilepsy syndrome

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De Novo Variants in MRTFB have gain of function activity in Drosophila and are associated with a novel neurodevelopmental phenotype with dysmorphic features

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H4C5 missense variant leads to a neurodevelopmental phenotype overlapping with Angelman syndrome

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Bi-allelic SNAPC4 variants dysregulate global alternative splicing and lead to neuroregression and progressive spastic paraparesis

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Full-length isoform sequencing for resolving the molecular basis of Charcot-Marie-Tooth 2A

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bioRxiv

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Exome/Genome Sequencing in Undiagnosed Syndromes

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A seed sequence variant in miR-145-5p causes multisystem smooth muscle dysfunction syndrome

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MYH2-associated myopathy caused by a novel splice-site variant

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Expansion of the clinical and molecular spectrum of WWOX-related epileptic encephalopathy
A recurrent single-exon deletion in TBCK might be under-recognized in patients with infantile hypotonia and psychomotor delay

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Gain-of-function mutations in ALPK1 cause an NF-κB-mediated autoinflammatory disease: functional assessment, clinical phenotyping and disease course of patients with ROSAH syndrome

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Variant-specific changes in RAC3 function disrupt corticogenesis in neurodevelopmental phenotypes

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Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis

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A homozygous splice variant in ATP5PO, disrupts mitochondrial complex V function and causes Leigh syndrome in two unrelated families

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A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder

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A novel, de novo intronic variant in POGZ causes White-Sutton syndrome

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Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development

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PRUNE1 c.933G>A synonymous variant induces exon 7 skipping, disrupts the DHHA2 domain, and leads to an atypical NMIHBA syndrome presentation: Case report and review of the literature

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A dominant negative variant of RAB5B disrupts maturation of surfactant protein B and surfactant protein C

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Proceedings of the National Academy of Sciences of the United States of America

PUS7 deficiency in human patients causes profound neurodevelopmental phenotype by dysregulating protein translation

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Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity

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Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling

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Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder

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Bilateral choanal stenosis in auriculocondylar syndrome caused by a PLCB4 variant

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The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder

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Gain-of-Function Mutations in RPA1 Cause a Syndrome with Short Telomeres and Somatic Genetic Rescue

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PPP3CA truncating variants clustered in the regulatory domain cause early-onset refractory epilepsy

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A novel de novo intronic variant in ITPR1 causes Gillespie syndrome

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American Journal of Medical Genetics, Part A
Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy

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Finding commonalities in rare diseases through the Undiagnosed Diseases Network

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Journal of the American Medical Informatics Association

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Scientific Data

Rare deleterious de novo missense variants in RNF2/RING2 are associated with a neurodevelopmental disorder with unique clinical features

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Heterozygous variants in SPTBN1 cause intellectual disability and autism

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American Journal of Medical Genetics, Part A

Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain

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A Description of Novel Variants and Review of Phenotypic Spectrum in UBA5-related Early Epileptic Encephalopathy

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Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases

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Machine Learning of Patient Characteristics to Predict Admission Outcomes in the Undiagnosed Diseases Network

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De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis

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American Journal of Human Genetics

Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation

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Expansion of NEUROD2 phenotypes to include developmental delay without seizures

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American Journal of Medical Genetics, Part A

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A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Cl-/H +-Exchanger, Causes Early-Onset Neurodegeneration

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Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing

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A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome

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Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder

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De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism

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Predominant and novel de novo variants in 29 individuals with ALG13 deficiency: Clinical description, biomarker status, biochemical analysis and treatment suggestions

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Journal of Inherited Metabolic Disease

Early infantile epileptic encephalopathy due to biallelic pathogenic variants in PIGQ: Report of seven new subjects and review of the literature

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Yippee Like 3 (ypel3) Is a Novel Gene Required for Myelinating and Perineurial Glia Development

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PLoS Genetics

De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment

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Mutations in GET4 Disrupt the Transmembrane Domain Recognition Complex Pathway

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Journal of Inherited Metabolic Disease

Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7

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Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses

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Genetics in Medicine

De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy

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American Journal of Human Genetics

The Undiagnosed Diseases Program: Approach to Diagnosis

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Translational Science of Rare Diseases
Dominant-negative Mutations in Human IL6ST Underlie hyper-IgE Syndrome

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Limitations of Exome Sequencing in Detecting Rare and Undiagnosed Diseases

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American Journal of Medical Genetics, Part A

De Novo EIF2AK1 and EIF2AK2 Variants Are Associated With Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation

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Loss-Or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms

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Neuron
Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency

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Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing

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Defining the clinical phenotype of Saul-Wilson syndrome

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GATAD2B-associated Neurodevelopmental Disorder (GAND): Clinical and Molecular Insights Into a
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De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder

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De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Collosum, Axon, Cardiac, Ocular, and Genital Defects

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American Journal of Human Genetics
Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing

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Journal of Genetic Counseling

Using MARRVEL v1.2 for Bioinformatics Analysis of Human Genes and Variant Pathogenicity

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In Vivo Functional Study of Disease-associated Rare Human Variants Using Drosophila

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Journal of Visualized Experiments

Navigating MARRVEL, a Web-Based Tool that Integrates Human Genomics and Model Organism Genetics Information

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Journal of Visualized Experiments

De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia

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American Journal of Human Genetics
The Undiagnosed Diseases Network as a tool for graduate medical education

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Partial loss of USP9X function leads to a male neurodevelopmental and behavioural disorder converging on TGFβ signalling

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The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases

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Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy

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Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts

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Lysosomal storage and albinism due to effects of a de novo CLCN7 variant on lysosomal acidification

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American Journal of Human Genetics

Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis

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Human Mutation

Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic splice-site and
Charcot-Marie-Tooth phenotype with early onset symptoms

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IgG4-related disease: association with a rare gene variant expressed in cytotoxic T cells

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Molecular Genetics & Genomic Medicine

A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing

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Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review

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Reported environmental exposures are inversely associated with obtaining a genetic diagnosis in the Undiagnosed Diseases Network

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American Journal of Medical Genetics, Part A

A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis

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Journal of General Internal Medicine

Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly

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Genetics in Medicine
SLC35A2-CDG: Functional Characterization, Expanded Molecular, Clinical, and Biochemical Phenotypes of 30 Unreported Individuals

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Human Mutation

Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes

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Kilquist Syndrome: A Novel Syndromic Hearing Loss Disorder Caused by Homozygous Deletion of SLC12A2

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Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies

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Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region

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Epilepsia

Cases from the Undiagnosed Diseases Network: The continued value of counseling skills in a new genomic era

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Journal of Genetic Counseling
Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukoencephalopathy: further delineation of the 17p13.3 microdeletion spectrum

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Genetics in Medicine

Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay

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American Journal of Human Genetics

Recurrent mosaic MTOR c.5930C>T (p.Thr1977Ile) variant causing megalencephaly, asymmetric polymicrogyria, and cutaneous pigmentary mosaicism: Case report and review of the literature

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American Journal of Medical Genetics, Part A

Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism

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ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis

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Genetics in Medicine

Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation

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American Journal of Human Genetics

Loss of tubulin de glutamylase CCP1 causes infantile-onset neurodegeneration

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EMBO Journal
Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease

October 10, 2018
New England Journal of Medicine

A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation

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American Journal of Human Genetics

Cog4 is required for protrusion and extension of the epithelium in the developing semicircular canals

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Mechanisms of Development

Phenotypic expansion in DDX3X - a common cause of intellectual disability in females

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Annals of Clinical and Translational Neurology
novoCaller: A Bayesian network approach for de novo variant calling from pedigree and population sequence data

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Bioinformatics

Characteristics of undiagnosed diseases network applicants: implications for referring providers

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BMC Health Services Research

An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian disease gene discovery

August 13, 2018
npj Genomic Medicine
IRF2BPL Is Associated with Neurological Phenotypes

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American Journal of Human Genetics

Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma

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Neurology Genetics

Further Evidence for the Involvement of EFL1 in a Shwachman-Diamond-like Syndrome and Expansion of the Phenotypic Features

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Cold Spring Harbor Molecular Case Studies

De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features

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American Journal of Human Genetics
A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative

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Genetics in Medicine

5,10-methenyltetrahydrofolate synthetase deficiency causes a neurometabolic disorder associated with microcephaly, epilepsy, and cerebral hypomyelination

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Molecular Genetics and Metabolism

Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome

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American Journal of Human Genetics
Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder

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Human Molecular Genetics

Understanding Adult Participant and Parent Empowerment Prior to Evaluation in the Undiagnosed Diseases Network

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Journal of Genetic Counseling

Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants

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Genetics in Medicine

Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder

February 22, 2018
Clinical heterogeneity of mitochondrial NAD kinase deficiency caused by a NADK2 start loss variant

February 1, 2018
American Journal of Medical Genetics, Part A

Phenotypic heterogeneity of ZMPSTE24 deficiency

January 17, 2018
American Journal of Medical Genetics, Part A

Genotype-phenotype correlations in individuals with pathogenic RERE variants

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Human Mutation
A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network

January 11, 2018

Journal of Pediatrics

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Psychosocial Profiles of Parents of Children with Undiagnosed Diseases: Managing Well or Just Managing?

January 2, 2018

Journal of Genetic Counseling

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De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability

November 2, 2017

American Journal of Human Genetics

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Implementing the Single Institutional Review Board Model: Lessons from the Undiagnosed Diseases Network

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Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases

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Genetics in Medicine

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Genetics

Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders

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Genome Medicine
Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially

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PloS Genetics

MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome

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American Journal of Human Genetics

Expanding the phenotypic spectrum of GABRG2 variants: a recurrent GABRG2 missense variant associated with a severe phenotype

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Journal of Neurogenetics

A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network

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The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease

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American Journal of Human Genetics

A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay

January 26, 2017
American Journal of Human Genetics

A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3

January 5, 2017
American Journal of Human Genetics
Exome Sequencing Identifies De Novo Pathogenic Variants in FBN1 and TRPS1 in a Patient with a Complex Connective Tissue Phenotype

November 11, 2016
Cold Spring Harbor Molecular Case Studies

De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype

November 6, 2016
American Journal of Human Genetics

A recurrent p.Arg92Trp variant in steroidogenic factor-1 (NR5A1) can act as a molecular switch in human sex development

July 4, 2016
Human Molecular Genetics

The NIH Undiagnosed Diseases Program and Network: Applications to modern medicine

January 22, 2016
Molecular Genetics and Metabolism

The Undiagnosed Diseases Network of the National Institutes of Health: A National Extension

November 3, 2015

The Journal of the American Medical Association