Parental perspectives of episodic irritability in an ultra-rare genetic disorder associated with NACC1

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Orphanet Journal of Rare Diseases

A defect in mitochondrial fatty acid synthesis impairs iron metabolism and causes elevated ceramide levels

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Nature Metabolism

A medical odyssey of a 72-year-old man with Charcot-Marie-Tooth disease type 2 newly diagnosed with biallelic variants in SORD gene causing sorbitol dehydrogenase deficiency

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American Journal of Medical Genetics Part A

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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A humanized Caenorhabditis elegans model of Hereditary Spastic Paraplegia-associated variants in kinesin light chain KLC4

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Disease Models and Mechanisms

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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Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care

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

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Biallelic variants in Ribonuclease Inhibitor (RNH1), an inflammasome modulator, are associated with a distinctive subtype of acute necrotizing encephalopathy

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Participation in a national diagnostic research study: assessing the patient experience

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Bi-allelic variants in INTS11 are associated with a complex neurological disorder

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Unraveling non-participation in genomic research: A complex interplay of barriers, facilitators, and sociocultural factors

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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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A Case Study of Dysfunctional Nicotinamide Metabolism in a 20-Year-Old Male

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Bi-allelic ATG4D variants are associated with a neurodevelopmental disorder characterized by speech and motor impairment

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Nature Publishing Journal in Genomic Medicine

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

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SPTSSA variants alter sphingolipid synthesis and cause a complex hereditary spastic paraplegia

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Exome/Genome Sequencing in Undiagnosed Syndromes

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Annual Review of Medicine

TMEM161B regulates cerebral cortical gyration, Sonic Hedgehog signaling, and ciliary structure in the developing central nervous system

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

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

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Continuing a search for a diagnosis: the impact of adolescence and family dynamics

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

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Neuromuscular Disorders

A de novo hexokinase 1 (HK1) variant presenting as Boucher-Neuhäuser syndrome

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American Journal of Medical Genetics

Expansion of the clinical and molecular spectrum of WWOX-related epileptic encephalopathy

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American Journal of Medical Genetics

A recurrent single-exon deletion in TBCK might be under-recognized in patients with infantile hypotonia and psychomotor delay

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A concurrent dual analysis of genomic data augments diagnoses: experiences of two clinical sites in the Undiagnosed Diseases Network

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Whole-Genome and Long-Read Sequencing Identify a Novel Mechanism in RFC1 Resulting in CANVAS Syndrome

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Loss-of-function variants in MYCBP2 cause neurobehavioural phenotypes and corpus callosum defects

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The recurrent de novo c.2011C>T missense variant in MTSS2 causes syndromic intellectual disability

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Beyond race: Recruitment of diverse participants in clinical genomics research for rare disease

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AnFiSA: An open-source computational platform for the analysis of sequencing data for rare genetic disease

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Wide range of phenotypic severity in individuals with late truncations unique to the predominant CDKL5 transcript in the brain

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Repeat expansions nested within tandem CNVs: A unique structural change in GLS exemplifies the diagnostic challenges of non-coding pathogenic variation

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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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Personalized structural biology reveals the molecular mechanisms underlying heterogeneous epileptic phenotypes caused by de novo KCNC2 variants

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

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Adults with lysosomal storage diseases in the undiagnosed diseases network

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Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome (NODRS)

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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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Journal of Inherited Metabolic Disease

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

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The microRNA processor DROSHA is a candidate gene for a severe progressive neurological disorder

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

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Systematic analysis of physical examination characteristics of 94 individuals with Joubert syndrome: Keys to suspecting the diagnosis

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Seminars in Medical Genetics, Part C of the American Journal of Medical Genetics (AJMG)

Genome sequencing reveals novel noncoding variants in PLA2G6 and LMNB1 causing progressive neurologic disease

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Lord of the fruit flies: an interview with Hugo Bellen

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Disease Models & Mechanisms

ModelMatcher: A scientist-centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research

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Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development

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The Cerebellum

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

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What Has the Undiagnosed Diseases Network Taught Us About the Clinical Applications of Genomic Testing?

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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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Patients as Partners in Rare Disease Diagnosis and Research

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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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De novo variants in CACNA1E found in patients with intellectual disability, developmental regression

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Variable clinical severity in TANGO2 deficiency: Case series and literature review

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Identifying digenic disease genes via machine learning in the Undiagnosed Diseases Network

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COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay

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Genetic counselor roles in the Undiagnosed Diseases Network research study: clinical care, collaboration, and curation

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Functional analysis of a de novo variant in the neurodevelopment and generalized epilepsy disease gene NBEA

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TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila

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Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome

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Pathogenic MAST3 variants in the STK domain are associated with epilepsy

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Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations

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Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples

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Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11

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"Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey

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TSPEAR variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study

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Rare disease patient matchmaking: development and outcomes of an internet case-finding strategy in the Undiagnosed Diseases Network

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

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Model organisms contribute to diagnosis and discovery in the Undiagnosed Diseases Network: current state and a future vision

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Progressive cerebellar atrophy in a patient with complex II and III deficiency and a novel deleterious variant in SDHA: A Counseling Conundrum

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Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey

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A novel de novo intronic variant in ITPR1 causes Gillespie syndrome

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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

A resource of lipidomics and metabolomics data from individuals with undiagnosed diseases

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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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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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Functional and structural analysis of cytokine selective IL6ST defects that cause recessive hyper-IgE syndrome

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Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics

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Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases

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A fish with no sex: gonadal and adrenal functions partition between zebrafish NR5A1 co-orthologs

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

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JAMA Network Open

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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Missense variants in CTNNB1 can be associated with vitreoretinopathy-Seven new cases of CTNNB1-associated neurodevelopmental disorder including a previously unreported retinal phenotype

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Molecular Genetics & Genomic Medicine

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

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Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients

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An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids

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BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms

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Discovery of a novel CHD7 CHARGE syndrome variant by integrated omics analyses

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DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature

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KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation

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MPEG1/Perforin-2 Haploinsufficiency Associated Polymicrobial Skin Infections and Considerations for Interferon-γ Therapy

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Family genetic result communication in rare and undiagnosed disease communities: understanding the practice

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Somatic Mutations in UBA1 and Severe Adult-Onset Autoinflammatory Disease

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Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science

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Novel NUDT2 variant causes intellectual disability and polyneuropathy

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Annals of Clinical and Translational Neurology

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

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Journal of Clinical Investigation

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

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European Journal of Human Genetics

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

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Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network

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

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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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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

NuRD-related Disorder

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Statistically-driven Metabolite and Lipid Profiling of Patients from the Undiagnosed Diseases Network

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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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Truncating variants in UBAP1 associated with childhood-onset nonsyndromic hereditary spastic paraplegia

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VarSight: prioritizing clinically reported variants with binary classification algorithms

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Diagnostic utility of transcriptome sequencing for rare Mendelian diseases

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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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Current Protocols in Bioinformatics

The Genome Empowerment Scale (GEmS): An Assessment of Parental Empowerment in Families with Undiagnosed Disease

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Genomics in medicine: a novel elective rotation for internal medicine residents

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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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Nature Medicine

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

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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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American Journal of Medical Genetics, Part A

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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American Journal of Human Genetics

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

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Human Mutation

Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students

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

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Genetics in Medicine

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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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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American Journal of Human Genetics

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

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Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation

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American Journal of Human Genetics

Loss of tubulin deglutamylase CCP1 causes infantile-onset neurodegeneration

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EMBO Journal

Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease

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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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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

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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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A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative

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

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

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Clinical heterogeneity of mitochondrial NAD kinase deficiency caused by a NADK2 start loss variant

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American Journal of Medical Genetics, Part A

Phenotypic heterogeneity of ZMPSTE24 deficiency

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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

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Journal of Pediatrics

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

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

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American Journal of Human Genetics

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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Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research

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

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Orphanet Journal of Rare Diseases

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

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American Journal of Human Genetics

A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3

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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
