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

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Novel NUDT2 variant causes intellectual disability and polyneuropathy

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

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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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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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Journal of Inherited Metabolic Disease

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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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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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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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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De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder
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Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing

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De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental
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Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis
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IgG4-related disease: association with a rare gene variant expressed in cytotoxic T cells

A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing
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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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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Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region

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

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**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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Loss of tubulin deglutamylase CCP1 causes infantile-onset neurodegeneration

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Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease

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

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

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

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IRF2BPL is Associated with Neurological Phenotypes

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

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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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**Phenotypic heterogeneity of ZMPSTE24 deficiency**

January 17, 2018

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**Genotype-phenotype correlations in individuals with pathogenic RERE variants**

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

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

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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

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American Journal of Human Genetics

A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3

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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

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

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

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

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

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