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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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Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7

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

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

Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency
Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing

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Navigating MARRVEL, a Web-Based Tool that Integrates Human Genomics and Model Organism
De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia

The Undiagnosed Diseases Network as a tool for graduate medical education

Partial loss of USP9X function leads to a male neurodevelopmental and behavioural disorder converging on TGFβ signalling
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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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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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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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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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Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants

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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

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

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

Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research
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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A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay

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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

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