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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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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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GATAD2B-associated Neurodevelopmental Disorder (GAND): Clinical and Molecular Insights Into a NuRD-related Disorder
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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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Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis

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

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

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

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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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Loss of tubulin deglutamylase CCP1 causes infantile-onset neurodegeneration

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

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Understanding Adult Participant and Parent Empowerment Prior to Evaluation in 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
Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research

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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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