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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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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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Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy

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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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Expansion of NEUROD2 phenotypes to include developmental delay without seizures

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

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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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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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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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Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing

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

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

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

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

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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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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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Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation

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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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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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Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder

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

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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

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

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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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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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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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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

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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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