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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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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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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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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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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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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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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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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 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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Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts

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

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A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis

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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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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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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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American Journal of Human Genetics

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

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American Journal of Human Genetics

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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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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Characteristics of undiagnosed diseases network applicants: implications for referring providers

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

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

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5,10-methy1tetrahydrofolate 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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Human Molecular Genetics
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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American Journal of Medical Genetics, Part A
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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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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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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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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