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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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Finding commonalities in rare diseases through the Undiagnosed Diseases Network

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Heterozygous variants in SPTBN1 cause intellectual disability and autism

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Expansion of NEUROD2 phenotypes to include developmental delay without seizures

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Yippee Like 3 (ypel3) Is a Novel Gene Required for Myelinating and Perineurial Glia Development

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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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De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder

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Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis

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

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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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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 deamylase CCP1 causes infantile-onset neurodegeneration

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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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Further Evidence for the Involvement of EFL1 in a Shwachman-Diamond-like Syndrome and Expansion of the Phenotypic Features

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

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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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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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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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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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MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome

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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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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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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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Molecular Genetics and Metabolism

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