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De Novo Variants in MRTFB have gain of function activity in Drosophila and are associated with a
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H4C5 missense variant leads to a neurodevelopmental phenotype overlapping with Angelman syndrome

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Bi-allelic SNAPC4 variants dysregulate global alternative splicing and lead to neuroregression and progressive spastic paraparesis

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TMEM161B regulates cerebral cortical gyration, Sonic Hedgehog signaling, and ciliary structure in the developing central nervous system

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A seed sequence variant in miR-145-5p causes multisystem smooth muscle dysfunction syndrome

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Continuing a search for a diagnosis: the impact of adolescence and family dynamics

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MYH2-associated myopathy caused by a novel splice-site variant

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A de novo hexokinase 1 (HK1) variant presenting as Boucher-Neuhäuser syndrome
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Loss-of-function variants in MYCBP2 cause neurobehavioural phenotypes and corpus callosum defects

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Variant-specific changes in RAC3 function disrupt corticogenesis in neurodevelopmental phenotypes

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A homozygous splice variant in ATP5PO, disrupts mitochondrial complex V function and causes Leigh syndrome in two unrelated families

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A novel DPH5-related diphthamide-deficiency syndrome causing embryonic lethality or profound neurodevelopmental disorder

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The microRNA processor DROSHA is a candidate gene for a severe progressive neurological disorder

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A novel, de novo intronic variant in POGZ causes White-Sutton syndrome

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Systematic analysis of physical examination characteristics of 94 individuals with Joubert syndrome: Keys to suspecting the diagnosis

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Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development

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PRUNE1 c.933G>A synonymous variant induces exon 7 skipping, disrupts the DHHA2 domain, and leads to an atypical NMIHBA syndrome presentation: Case report and review of the literature

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A dominant negative variant of RAB5B disrupts maturation of surfactant protein B and surfactant protein C

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PUS7 deficiency in human patients causes profound neurodevelopmental phenotype by dysregulating protein translation

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Bi-allelic variants in neuronal cell adhesion molecule cause a neurodevelopmental disorder characterized by developmental delay, hypotonia, neuropathy/spasticity

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Loss of IRF2BPL impairs neuronal maintenance through excess Wnt signaling

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Rare germline heterozygous missense variants in BRCA1-associated protein 1, BAP1, cause a syndromic neurodevelopmental disorder

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Bilateral choanal stenosis in auriculocondylar syndrome caused by a PLCB4 variant

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The clinical and molecular spectrum of QRICH1 associated neurodevelopmental disorder

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Gain-of-Function Mutations in RPA1 Cause a Syndrome with Short Telomeres and Somatic Genetic Rescue

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De novo variants in CACNA1E found in patients with intellectual disability, developmental regression and social cognition deficit but no seizures

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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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Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome

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Pathogenic MAST3 variants in the STK domain are associated with epilepsy
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TSPEAR variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study

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PPP3CA truncating variants clustered in the regulatory domain cause early-onset refractory epilepsy

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A novel de novo intronic variant in ITPR1 causes Gillespie syndrome

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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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A resource of lipidomics and metabolomics data from individuals with undiagnosed diseases

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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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Functional and structural analysis of cytokine selective IL6ST defects that cause recessive hyper-IgE syndrome

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Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics

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Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases

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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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Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation

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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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Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients

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An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids

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BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms

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Family genetic result communication in rare and undiagnosed disease communities: understanding the practice

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Somatic Mutations in UBA1 and Severe Adult-Onset Autoinflammatory Disease

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Novel NUDT2 variant causes intellectual disability and polyneuropathy

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Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing

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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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Missed diagnoses: Clinically relevant lessons learned through medical mysteries solved by the Undiagnosed Diseases Network

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De Novo Variants in the ATPase Module of MORC2 Cause a Neurodevelopmental Disorder with Growth Retardation and Variable Craniofacial Dysmorphism

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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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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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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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VarSight: prioritizing clinically reported variants with binary classification algorithms

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

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The Undiagnosed Diseases Network as a tool for graduate medical education

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Partial loss of USP9X function leads to a male neurodevelopmental and behavioural disorder converging on TGFβ signalling

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The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases

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**Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy**

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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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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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A toolkit for genetics providers in follow-up of patients with non-diagnostic exome sequencing

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Extracutaneous manifestations in phacomatosis cesioflammea and cesiomarmorata: Case series and literature review

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

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Kilquist Syndrome: A Novel Syndromic Hearing Loss Disorder Caused by Homozygous Deletion of SLC12A2

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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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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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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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Phenotypic expansion in DDX3X - a common cause of intellectual disability in females

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novoCaller: A Bayesian network approach for de novo variant calling from pedigree and population sequence data

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Characteristics of undiagnosed diseases network applicants: implications for referring providers

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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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**De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features**

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

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5,10-methenyltetrahydrofolate synthetase deficiency causes a neurometabolic disorder associated with microcephaly, epilepsy, and cerebral hypomyelination

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