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A defect in mitochondrial fatty acid synthesis impairs iron metabolism and causes elevated ceramide levels

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A medical odyssey of a 72-year-old man with Charcot-Marie-Tooth disease type 2 newly diagnosed with biallelic variants in SORD gene causing sorbitol dehydrogenase deficiency

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Macrocephaly and developmental delay caused by missense variants in RAB5C

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HNRNPC haploinsufficiency affects alternative splicing of intellectual disability-associated genes and causes a neurodevelopmental disorder

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Heterozygous rare variants in NR2F2 cause a recognizable multiple congenital anomaly syndrome with developmental delays

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Monoallelic variation in DHX9, the gene encoding the DExH-box helicase DHX9, underlies neurodevelopment disorders and Charcot-Marie-Tooth disease

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A de novo missense variant in EZH1 associated with developmental delay exhibits functional deficits in Drosophila melanogaster

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Dominant negative variants in IKZF2 cause ICHAD syndrome, a new disorder characterised by immunodysregulation, craniofacial anomalies, hearing impairment, athelia and developmental delay

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Dominant-negative variant in SLC1A4 causes an autosomal dominant epilepsy syndrome

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De Novo Variants in MRTFB have gain of function activity in Drosophila and are associated with a novel neurodevelopmental phenotype with dysmorphic features

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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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Bi-allelic ATG4D variants are associated with a neurodevelopmental disorder characterized by speech and motor impairment

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Full-length isoform sequencing for resolving the molecular basis of Charcot-Marie-Tooth 2A

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SPTSSA variants alter sphingolipid synthesis and cause a complex hereditary spastic paraplegia

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**Continuing a search for a diagnosis: the impact of adolescence and family dynamics**
MYH2-associated myopathy caused by a novel splice-site variant

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Expansion of the clinical and molecular spectrum of WWOX-related epileptic encephalopathy
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A concurrent dual analysis of genomic data augments diagnoses: experiences of two clinical sites in the Undiagnosed Diseases Network

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Whole-Genome and Long-Read Sequencing Identify a Novel Mechanism in RFC1 Resulting in CANVAS Syndrome

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Gain-of-function mutations in ALPK1 cause an NF-κB-mediated autoinflammatory disease: functional assessment, clinical phenotyping and disease course of patients with ROSAH syndrome

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Personalized structural biology reveals the molecular mechanisms underlying heterogeneous epileptic phenotypes caused by de novo KCNC2 variants

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

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Endocannabinoid dysfunction in neurological disease: neuro-ocular DAGLA-related syndrome (NODRS)

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Phenotypic and genetic spectrum of ATP6V1A encephalopathy: a disorder of lysosomal homeostasis

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

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Patients as Partners in Rare Disease Diagnosis and Research

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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
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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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Finding commonalities in rare diseases through the Undiagnosed Diseases Network

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A Description of Novel Variants and Review of Phenotypic Spectrum in UBA5-related Early Epileptic Encephalopathy

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Novel NUDT2 variant causes intellectual disability and polyneuropathy

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Mutations in GET4 Disrupt the Transmembrane Domain Recognition Complex Pathway

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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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Defining the clinical phenotype of Saul-Wilson syndrome

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GATAD2B-associated Neurodevelopmental Disorder (GAND): Clinical and Molecular Insights Into a
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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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The Genome Empowerment Scale (GEmS): An Assessment of Parental Empowerment in Families with Undiagnosed Disease

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

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