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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Genomics Research with Undiagnosed Children: Ethical Challenges at the Boundaries of Research and Clinical Care

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The contribution of mosaicism to genetic diseases and de novo pathogenic variants

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

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Biallelic variants in Ribonuclease Inhibitor (RNH1), an inflammasome modulator, are associated with a distinctive subtype of acute necrotizing encephalopathy

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Participation in a national diagnostic research study: assessing the patient experience

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Bi-allelic variants in INTS11 are associated with a complex neurological disorder

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Unraveling non-participation in genomic research: A complex interplay of barriers, facilitators, and sociocultural factors

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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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A Case Study of Dysfunctional Nicotinamide Metabolism in a 20-Year-Old Male

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Undiagnosed diseases: Needs and opportunities in 20 countries participating in the Undiagnosed Diseases Network International

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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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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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A humanized Caenorhabditis elegans model of Hereditary Spastic Paraplegia-associated variants in
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MYH2-associated myopathy caused by a novel splice-site variant

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Neuromuscular Disorders

A de novo hexokinase 1 (HK1) variant presenting as Boucher-Neuhäuser syndrome

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American Journal of Medical Genetics

Expansion of the clinical and molecular spectrum of WWOX-related epileptic encephalopathy

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A recurrent single-exon deletion in TBCK might be under-recognized in patients with infantile hypotonia and psychomotor delay

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Human Mutation

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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Loss-of-function variants in MYCBP2 cause neurobehavioural phenotypes and corpus callosum defects

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The recurrent de novo c.2011C>T missense variant in MTSS2 causes syndromic intellectual disability

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Beyond race: Recruitment of diverse participants in clinical genomics research for rare disease

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Wide range of phenotypic severity in individuals with late truncations unique to the predominant CDKL5 transcript in the brain

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Repeat expansions nested within tandem CNVs: A unique structural change in GLS exemplifies the diagnostic challenges of non-coding pathogenic variation

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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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Adults with lysosomal storage diseases in the undiagnosed diseases network

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

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Seminars in Medical Genetics, Part C of the American Journal of Medical Genetics (AJMG)

Genome sequencing reveals novel noncoding variants in PLA2G6 and LMNB1 causing progressive neurologic disease

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Lord of the fruit flies: an interview with Hugo Bellen

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ModelMatcher: A scientist-centric online platform to facilitate collaborations between stakeholders of rare and undiagnosed disease research

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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 NMHBA 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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What Has the Undiagnosed Diseases Network Taught Us About the Clinical Applications of Genomic Testing?

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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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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 and social cognition deficit but no seizures

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Identifying digenic disease genes via machine learning in the Undiagnosed Diseases Network

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COPB2 loss of function causes a coatopathy with osteoporosis and developmental delay
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TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila
One is the loneliest number: genotypic matchmaking using the electronic health record

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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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Phenotypic expansion of CACNA1C-associated disorders to include isolated neurological manifestations
Clinical application of a scale to assess genomic healthcare empowerment (GEmS): Process and illustrative case examples

Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of GDF11

"Doctors can read about it, they can know about it, but they've never lived with it": How parents use social media throughout the diagnostic odyssey
TSPEAR variants are primarily associated with ectodermal dysplasia and tooth agenesis but not hearing loss: A novel cohort study

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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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Model organisms contribute to diagnosis and discovery in the Undiagnosed Diseases Network: current state and a future vision

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Detection of a mosaic CDKL5 deletion and inversion by optical genome mapping ends an exhaustive diagnostic odyssey

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

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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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A fish with no sex: gonadal and adrenal functions partition between zebrafish NR5A1 co-orthologs

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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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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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Discovery of a novel CHD7 CHARGE syndrome variant by integrated omics analyses

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KMT2B-related disorders: expansion of the phenotypic spectrum and long-term efficacy of deep brain stimulation

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MPEG1/Perforin-2 Haploinsufficiency Associated Polymicrobial Skin Infections and Considerations for Interferon-γ Therapy

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Novel NUDT2 variant causes intellectual disability and polyneuropathy

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

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Predominant and novel de novo variants in 29 individuals with ALG13 deficiency: Clinical description, biomarker status, biochemical analysis and treatment suggestions

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

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Mutations in GET4 Disrupt the Transmembrane Domain Recognition Complex Pathway

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Alternative transcripts in variant interpretation: the potential for missed diagnoses and misdiagnoses

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The Undiagnosed Diseases Program: Approach to Diagnosis

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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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Truncating variants in UBAP1 associated with childhood-onset nonsyndromic hereditary spastic paraplegia

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VarSight: prioritizing clinically reported variants with binary classification algorithms

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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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The Genome Empowerment Scale (GEmS): An Assessment of Parental Empowerment in Families with Undiagnosed Disease

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Genomics in medicine: a novel elective rotation for internal medicine residents

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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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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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Lysosomal storage and albinism due to effects of a de novo CLCN7 variant on lysosomal acidification

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Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic splice-site and
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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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A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis

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SLC35A2-CDG: Functional Characterization, Expanded Molecular, Clinical, and Biochemical Phenotypes of 30 Unreported Individuals

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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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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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