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

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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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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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Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7

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

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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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American Journal of Medical Genetics, Part A
De Novo EIF2AK1 and EIF2AK2 Variants Are Associated With Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation

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American Journal of Human Genetics

Loss- Or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms

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Neuron

Homozygous splice-variants in human ARV1 cause GPI-anchor synthesis deficiency

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GATAD2B-associated Neurodevelopmental Disorder (GAND): Clinical and Molecular Insights Into a
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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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**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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Journal of Genetic Counseling

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

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In Vivo Functional Study of Disease-associated Rare Human Variants Using Drosophila

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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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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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IgG4-related disease: association with a rare gene variant expressed in cytotoxic T cells

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

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

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

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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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Epilepsia

**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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*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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American Journal of Human Genetics

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 deacetylase CCP1 causes infantile-onset neurodegeneration

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EMBO Journal

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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American Journal of Human Genetics
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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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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De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and
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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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**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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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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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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Implementing the Single Institutional Review Board Model: Lessons from the Undiagnosed Diseases
Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases

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Genetics in Medicine

Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research

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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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Genome Medicine
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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Expanding the phenotypic spectrum of GABRG2 variants: a recurrent GABRG2 missense variant associated with a severe phenotype

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Journal of Neurogenetics
A window into living with an undiagnosed disease: illness narratives from the Undiagnosed Diseases Network

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Orphanet Journal of Rare Diseases

The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease

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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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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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The NIH Undiagnosed Diseases Program and Network: Applications to modern medicine

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The Undiagnosed Diseases Network of the National Institutes of Health: A National Extension

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