

**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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**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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**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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**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 TGF $\beta$  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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**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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**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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**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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**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 PFAFH1B1 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 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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**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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**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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**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**

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**Looking beyond the exome: a phenotype-first approach to molecular diagnostic resolution in rare and undiagnosed diseases**

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**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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**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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**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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**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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