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Clinical sites of the Undiagnosed Diseases Network: unique contributions to genomic medicine and science

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

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Defining the clinical phenotype of Saul-Wilson syndrome

GATAD2B-associated Neurodevelopmental Disorder (GAND): Clinical and Molecular Insights Into a NuRD-related Disorder

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

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Navigating MARRVEL, a Web-Based Tool that Integrates Human Genomics and Model Organism Genetics Information

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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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Developing a genomics rotation: Practical training around variant interpretation for genetic counseling students

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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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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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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
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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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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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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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Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially

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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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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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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

July 4, 2016
Early intervention in genetic disorders is crucial for improving outcomes and quality of life. The NIH Undiagnosed Diseases Program and Network aims to identify and understand the underlying causes of unexplained conditions. This initiative has significant implications for modern medicine, potentially leading to new treatments and breakthroughs in genetic research.

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

This national extension of the Undiagnosed Diseases Network further expands the reach of this initiative, offering hope to individuals with elusive health conditions. The journal articles referenced provide valuable insights into the impact and potential applications of this groundbreaking work.