Kagami Ogata syndrome: a small deletion refines critical region for imprinting

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NPJ Genomic Medicine

A syndromic neurodevelopmental disorder caused by rare variants in PPFIA3

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American Journal of Human Genetics

Novel molecular mechanism in Malan syndrome uncovered through genome sequencing reanalysis, exon-level Array, and RNA sequencing

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High-dimensional immunophenotyping reveals immune cell aberrations in patients with undiagnosed inflammatory and autoimmune diseases

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Case report: ocular manifestations of a gain-of-function mutation in CLCN6, a newly diagnosed disease

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Dominant negative variants in KIF5B cause osteogenesis imperfecta via down regulation of mTOR signaling

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Biallelic CRELD1 variants cause a multisystem syndrome including neurodevelopmental phenotypes, cardiac dysrhythmias, and frequent infections

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Genetics in Medicine : Official Journal of the American College of Medical Genetics
Rare de novo gain-of-function missense variants in DOT1L are associated with developmental delay and congenital anomalies

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Probable digenic inheritance of Diamond-Blackfan anemia

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Nature Communications

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De novo missense variants in ZBTB47 are associated with developmental delays, hypotonia, seizures, gait abnormalities, and variable movement abnormalities

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Parental perspectives of episodic irritability in an ultra-rare genetic disorder associated with NACC1

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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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A humanized Caenorhabditis elegans model of Hereditary Spastic Paraplegia-associated variants in kinesin light chain KLC4

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

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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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Proceedings of the National Academy of Sciences of the United States of America
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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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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Loss-of-function variants in MYCBP2 cause neurobehavioural phenotypes and corpus callosum defects

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

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Loss of Neuron Navigator 2 Impairs Brain and Cerebellar Development

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A dominant negative variant of RAB5B disrupts maturation of surfactant protein B and surfactant protein C

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Proceedings of the National Academy of Sciences of the United States of America

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

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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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American Journal of Human Genetics

Bilateral choanal stenosis in auriculocondylar syndrome caused by a PLCB4 variant

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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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Functional analysis of a de novo variant in the neurodevelopment and generalized epilepsy disease gene NBEA

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TNPO2 variants associate with human developmental delays, neurologic deficits, and dysmorphic features and alter TNPO2 activity in Drosophila

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Pathogenic SPTBN1 variants cause an autosomal dominant neurodevelopmental syndrome

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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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PPP3CA truncating variants clustered in the regulatory domain cause early-onset refractory epilepsy

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Missense and truncating variants in CHD5 in a dominant neurodevelopmental disorder with intellectual disability, behavioral disturbances, and epilepsy

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Finding commonalities in rare diseases through the Undiagnosed Diseases Network

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

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Scientific Data

Rare deleterious de novo missense variants in RNF2/RING2 are associated with a neurodevelopmental disorder with unique clinical features

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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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Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases

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Genetics

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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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Expansion of NEUROD2 phenotypes to include developmental delay without seizures

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American Journal of Medical Genetics, Part A

Missense variants in CTNNB1 can be associated with vitreoretinopathy-Seven new cases of CTNNB1-associated neurodevelopmental disorder including a previously unreported retinal phenotype

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A Recurrent Gain-of-Function Mutation in CLCN6, Encoding the ClC-6 Cl-/H +-Exchanger, Causes Early-Onset Neurodegeneration

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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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DYRK1A pathogenic variants in two patients with syndromic intellectual disability and a review of the literature

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

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Family genetic result communication in rare and undiagnosed disease communities: understanding the practice

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Somatic Mutations in UBA1 and Severe Adult-Onset Autoinflammatory Disease

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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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European Journal of Human Genetics
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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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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De novo mutations in TOMM70, a receptor of the mitochondrial import translocase, cause neurological impairment

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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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American Journal of Human Genetics
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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Journal of Experimental Medicine

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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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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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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De novo and inherited variants in ZNF292 underlie a neurodevelopmental disorder with features of autism spectrum disorder

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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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American Journal of Human Genetics

Yield of whole exome sequencing in undiagnosed patients facing insurance coverage barriers to genetic testing

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Journal of Genetic Counseling

Using MARRVEL v1.2 for Bioinformatics Analysis of Human Genes and Variant Pathogenicity

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Postgraduate Medical Journal

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Journal of Visualized Experiments

Navigating MARRVEL, a Web-Based Tool that Integrates Human Genomics and Model Organism Genetics Information
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Journal of Visualized Experiments
De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia

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American Journal of Human Genetics

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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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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Molecular Genetics & Genomic Medicine

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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American Journal of Medical Genetics, Part A

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

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American Journal of Medical Genetics, Part A

A Patient with Sjogren's Syndrome and Subsequent Diagnosis of Inclusion Body Myositis and Light-Chain Amyloidosis

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Journal of General Internal Medicine
Elucidation of the phenotypic spectrum and genetic landscape in primary and secondary microcephaly

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Genetics in Medicine

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

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Human Mutation

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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Genetics in Medicine

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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Journal of Genetic Counseling

Microdeletions excluding YWHAE and PAFAH1B1 cause a unique leukencephalopathy: further delineation of the 17p13.3 microdeletion spectrum

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Genetics in Medicine

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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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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Genetics in Medicine

Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation

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American Journal of Human Genetics
Loss of tubulin deglutamylase 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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New England Journal of Medicine

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

Cog4 is required for protrusion and extension of the epithelium in the developing semicircular canals

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Mechanisms of Development
Phenotypic expansion in DDX3X - a common cause of intellectual disability in females

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Annals of Clinical and Translational Neurology

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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BMC Health Services Research

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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American Journal of Human Genetics

Atypical Alexander disease with dystonia, retinopathy, and a brain mass mimicking astrocytoma

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Neurology Genetics

Further Evidence for the Involvement of EFL1 in a Shwachman-Diamond-like Syndrome and Expansion of the Phenotypic Features

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Cold Spring Harbor Molecular Case Studies
De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features

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American Journal of Human Genetics

A comprehensive iterative approach is highly effective in diagnosing individuals who are exome negative

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Genetics in Medicine

5,10-methenyltetrahydrofolate synthetase deficiency causes a neurometabolic disorder associated with microcephaly, epilepsy, and cerebral hypomyelination

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Molecular Genetics and Metabolism
Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome

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American Journal of Human Genetics

Functional variants in TBX2 are associated with a syndromic cardiovascular and skeletal developmental disorder

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Human Molecular Genetics

Understanding Adult Participant and Parent Empowerment Prior to Evaluation in the Undiagnosed Diseases Network

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Journal of Genetic Counseling

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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American Journal of Human Genetics

Clinical heterogeneity of mitochondrial NAD kinase deficiency caused by a NADK2 start loss variant

February 1, 2018
American Journal of Medical Genetics, Part A

Phenotypic heterogeneity of ZMPSTE24 deficiency

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American Journal of Medical Genetics, Part A
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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Journal of Pediatrics

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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American Journal of Human Genetics
Implementing the Single Institutional Review Board Model: Lessons from the Undiagnosed Diseases Network

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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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Genetics in Medicine

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Phenotypic and molecular characterisation of CDK13-related congenital heart defects, dysmorphic facial features and intellectual developmental disorders

August 14, 2017
Genome Medicine

Clinically severe CACNA1A alleles affect synaptic function and neurodegeneration differentially

July 24, 2017
PLoS Genetics

MARRVEL: Integration of Human and Model Organism Genetic Resources to Facilitate Functional Annotation of the Human Genome

May 11, 2017
American Journal of Human Genetics

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

May 2, 2017
Journal of Neurogenetics

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

April 17, 2017
Orphanet Journal of Rare Diseases

The Undiagnosed Diseases Network: Accelerating Discovery about Health and Disease

February 2, 2017
American Journal of Human Genetics

A Recurrent De Novo Variant in NACC1 Causes a Syndrome Characterized by Infantile Epilepsy, Cataracts, and Profound Developmental Delay

January 26, 2017
American Journal of Human Genetics
A Syndromic Neurodevelopmental Disorder Caused by De Novo Variants in EBF3

January 5, 2017
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

November 11, 2016
Cold Spring Harbor Molecular Case Studies

De Novo Truncating Variants in ASXL2 Are Associated with a Unique and Recognizable Clinical Phenotype

November 6, 2016
American Journal of Human Genetics

A recurrent p.Arg92Trp variant in steroidogenic factor-1 (NR5A1) can act as a molecular switch in human sex development

July 4, 2016
The NIH Undiagnosed Diseases Program and Network: Applications to modern medicine

January 22, 2016

Molecular Genetics and Metabolism

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

November 3, 2015

The Journal of the American Medical Association