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

April 21, 2021
Scientific Data

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

April 16, 2021
Human Molecular Genetics

Heterozygous variants in SPTBN1 cause intellectual disability and autism

April 13, 2021
American Journal of Medical Genetics, Part A

Variants in PRKAR1B cause a neurodevelopmental disorder with autism spectrum disorder, apraxia, and insensitivity to pain

April 8, 2021
Genetics in Medicine
A Description of Novel Variants and Review of Phenotypic Spectrum in UBA5-related Early Epileptic Encephalopathy

April 2, 2021
Cold Spring Harbor Molecular Case Studies

Functional and structural analysis of cytokine selective IL6ST defects that cause recessive hyper-IgE syndrome

March 23, 2021
Journal of Allergy and Clinical Immunology

Identification of rare and common regulatory variants in pluripotent cells using population-scale transcriptomics

March 4, 2021
Nature Genetics
Commonalities across computational workflows for uncovering explanatory variants in undiagnosed cases

February 12, 2021
Genetics in Medicine

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

February 9, 2021
Genetics

Machine Learning of Patient Characteristics to Predict Admission Outcomes in the Undiagnosed Diseases Network

February 1, 2021
JAMA Network Open

De novo TRIM8 variants impair its protein localization to nuclear bodies and cause developmental delay, epilepsy, and focal segmental glomerulosclerosis

January 25, 2021
Linkage-specific deubiquitylation by OTUD5 defines an embryonic pathway intolerant to genomic variation

January 20, 2021

Science Advances

Expansion of NEUROD2 phenotypes to include developmental delay without seizures

January 13, 2021

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

December 22, 2020

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

December 3, 2020

American Journal of Human Genetics

Histone H3.3 beyond cancer: Germline mutations in Histone 3 Family 3A and 3B cause a previously unidentified neurodegenerative disorder in 46 patients

December 2, 2020

Science Advances

An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids

November 26, 2020

Genetics in Medicine

BICRA, a SWI/SNF Complex Member, Is Associated with BAF-Disorder Related Phenotypes in Humans and Model Organisms

November 23, 2020
Discovery of a novel CHD7 CHARGE syndrome variant by integrated omics analyses

November 13, 2020

American Journal of Medical Genetics, Part A

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

November 7, 2020

Molecular Genetics & Genomic Medicine

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

November 5, 2020

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

November 3, 2020
Frontiers in Immunology

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

October 27, 2020
Journal of Genetic Counseling

Somatic Mutations in UBA1 and Severe Adult-Onset Autoinflammatory Disease

October 27, 2020
New England Journal of Medicine

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

October 23, 2020
**Genetics in Medicine**

**Novel NUDT2 variant causes intellectual disability and polyneuropathy**

October 15, 2020

*Annals of Clinical and Translational Neurology*

**Transcriptome-directed analysis for Mendelian disease diagnosis overcomes limitations of conventional genomic testing**

October 1, 2020

*Journal of Clinical Investigation*

**A relatively common homozygous TRAPPC4 splicing variant is associated with an early-infantile neurodegenerative syndrome**

September 8, 2020

*European Journal of Human Genetics*
Biallelic MADD variants cause a phenotypic spectrum ranging from developmental delay to a multisystem disorder

August 6, 2020

Brain

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

July 30, 2020

Molecular Genetics & Genomic Medicine

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

July 20, 2020

American Journal of Human Genetics

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

July 18, 2020
Journal of Inherited Metabolic Disease

Early infantile epileptic encephalopathy due to biallelic pathogenic variants in PIGQ: Report of seven new subjects and review of the literature

June 26, 2020

Journal of Inherited Metabolic Disease

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

June 16, 2020

PLoS Genetics

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

June 3, 2020

Human Molecular Genetics
Mutations in GET4 Disrupt the Transmembrane Domain Recognition Complex Pathway

May 12, 2020
Journal of Inherited Metabolic Disease

Phenotypic spectrum and transcriptomic profile associated with germline variants in TRAF7

May 7, 2020
Genetics in Medicine

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

May 5, 2020
Genetics in Medicine

De Novo Variants in CDK19 Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy

April 23, 2020
American Journal of Human Genetics
The Undiagnosed Diseases Program: Approach to Diagnosis

April 13, 2020
Translational Science of Rare Diseases

Dominant-negative Mutations in Human IL6ST Underlie hyper-IgE Syndrome

March 24, 2020
Journal of Experimental Medicine

Limitations of Exome Sequencing in Detecting Rare and Undiagnosed Diseases

March 19, 2020
American Journal of Medical Genetics, Part A

De Novo EIF2AK1 and EIF2AK2 Variants Are Associated With Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation
March 16, 2020
American Journal of Human Genetics

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

March 9, 2020
Neuron

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

February 10, 2020
Molecular Genetics and Metabolism

**Myopathy associated with homozygous PYROXD1 pathogenic variants detected by genome sequencing**

February 9, 2020
Neuropathology
Defining the clinical phenotype of Saul-Wilson syndrome

January 17, 2020
Genetics in Medicine

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

January 17, 2020
Genetics in Medicine

Statistically-driven Metabolite and Lipid Profiling of Patients from the Undiagnosed Diseases Network

November 19, 2019
Analytical Chemistry

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

November 14, 2019
Truncating variants in UBAP1 associated with childhood-onset nonsyndromic hereditary spastic paraplegia

November 7, 2019

Human Mutation

VarSight: prioritizing clinically reported variants with binary classification algorithms

October 15, 2019

BMC Bioinformatics

Diagnostic utility of transcriptome sequencing for rare Mendelian diseases

October 14, 2019

Genetics in Medicine
De Novo Pathogenic Variants in N-cadherin Cause a Syndromic Neurodevelopmental Disorder with Corpus Collosum, Axon, Cardiac, Ocular, and Genital Defects

October 3, 2019
American Journal of Human Genetics

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

September 3, 2019
Journal of Genetic Counseling

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

September 1, 2019
Current Protocols in Bioinformatics

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

August 25, 2019
Clinical Genetics

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

August 22, 2019
Postgraduate Medical Journal

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

August 20, 2019
Journal of Visualized Experiments

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

August 15, 2019
Journal of Visualized Experiments
De Novo Variants in WDR37 Are Associated with Epilepsy, Colobomas, Dysmorphism, Developmental Delay, Intellectual Disability, and Cerebellar Hypoplasia

July 18, 2019
American Journal of Human Genetics

The Undiagnosed Diseases Network as a tool for graduate medical education

July 10, 2019
American Journal of Medicine

Partial loss of USP9X function leads to a male neurodevelopmental and behavioural disorder converging on TGFβ signalling

June 29, 2019
Biological Psychiatry

The fruit fly at the interface of diagnosis and pathogenic mechanisms of rare and common human diseases

June 22, 2019
Magnetic Resonance Imaging characteristics in case of TOR1AIP1 muscular dystrophy

June 21, 2019
Clinical Imaging

Identification of rare-disease genes using blood transcriptome sequencing and large control cohorts

June 3, 2019
Nature Medicine

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

May 30, 2019
American Journal of Human Genetics
Heterozygous variants in MYBPC1 are associated with an expanded neuromuscular phenotype beyond arthrogryposis

May 5, 2019
Human Mutation

Whole genome sequencing reveals novel IGHMBP2 variant leading to unique cryptic splice-site and Charcot-Marie-Tooth phenotype with early onset symptoms

April 25, 2019
Molecular Genetics & Genomic Medicine

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

April 16, 2019
Molecular Genetics & Genomic Medicine

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

April 9, 2019
Journal of Genetic Counseling
Extracutaneous manifestations in phacomatosis cescioflammea and cesiomarmorata: Case series and literature review

March 28, 2019

American Journal of Medical Genetics, Part A

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

March 23, 2019

American Journal of Medical Genetics, Part A

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

March 18, 2019

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

March 7, 2019
Genetics in Medicine

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

February 28, 2019
Human Mutation

Bi-allelic Variants in TONSL Cause SPONASTRIME Dysplasia and a Spectrum of Skeletal Dysplasia Phenotypes

February 11, 2019
American Journal of Human Genetics

Kilquist Syndrome: A Novel Syndromic Hearing Loss Disorder Caused by Homozygous Deletion of SLC12A2

February 10, 2019
**Human Mutation**

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

February 1, 2019

Journal of Genetic Counseling

---

**Pathogenic variants in USP7 cause a neurodevelopmental disorder with speech delays, altered behavior, and neurologic anomalies**

January 25, 2019

Genetics in Medicine

---

**Spectrum of neurodevelopmental disease associated with the GNAO1 guanosine triphosphate-binding region**

January 25, 2019

Epilepsia
Cases from the Undiagnosed Diseases Network: The continued value of counseling skills in a new genomic era

January 24, 2019
Journal of Genetic Counseling

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

December 20, 2018
Genetics in Medicine

Expanding the Spectrum of BAF-Related Disorders: De Novo Variants in SMARCC2 Cause a Syndrome with Intellectual Disability and Developmental Delay

December 20, 2018
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

December 19, 2018
Bi-allelic TMEM94 Truncating Variants Are Associated with Neurodevelopmental Delay, Congenital Heart Defects, and Distinct Facial Dysmorphism

December 6, 2018
American Journal of Human Genetics

ClinPhen extracts and prioritizes patient phenotypes directly from medical records to expedite genetic disease diagnosis

December 5, 2018
Genetics in Medicine

Pathogenic Variants in Fucokinase Cause a Congenital Disorder of Glycosylation

November 29, 2018
American Journal of Human Genetics
Loss of tubulin deglutamylase CCP1 causes infantile-onset neurodegeneration

November 14, 2018
EMBO Journal

Effect of Genetic Diagnosis on Patients with Previously Undiagnosed Disease

October 10, 2018
New England Journal of Medicine

A Recurrent De Novo Heterozygous COG4 Substitution Leads to Saul-Wilson Syndrome, Disrupted Vesicular Trafficking, and Altered Proteoglycan Glycosylation

October 4, 2018
American Journal of Human Genetics

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

October 1, 2018
Mechanisms of Development
Phenotypic expansion in DDX3X - a common cause of intellectual disability in females

September 15, 2018
Annals of Clinical and Translational Neurology

novoCaller: A Bayesian network approach for de novo variant calling from pedigree and population sequence data

August 30, 2018
Bioinformatics

Characteristics of undiagnosed diseases network applicants: implications for referring providers

August 22, 2018
BMC Health Services Research

An integrated clinical program and crowdsourcing strategy for genomic sequencing and Mendelian
disease gene discovery

August 13, 2018
npj Genomic Medicine

IRF2BPL Is Associated with Neurological Phenotypes

July 26, 2018
American Journal of Human Genetics

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

July 20, 2018
Neurology Genetics

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

July 3, 2018
Cold Spring Harbor Molecular Case Studies
De Novo Missense Variants in TRAF7 Cause Developmental Delay, Congenital Anomalies, and Dysmorphic Features

June 28, 2018
American Journal of Human Genetics

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

June 15, 2018
Genetics in Medicine

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

June 15, 2018
Molecular Genetics and Metabolism
Heterozygous Truncating Variants in POMP Escape Nonsense-Mediated Decay and Cause a Unique Immune Dysregulatory Syndrome

May 24, 2018
American Journal of Human Genetics

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

May 2, 2018
Human Molecular Genetics

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

March 1, 2018
Journal of Genetic Counseling

Autosomal recessive Noonan syndrome associated with biallelic LZTR1 variants

February 22, 2018
Biallelic Mutations in ATP5F1D, which Encodes a Subunit of ATP Synthase, Cause a Metabolic Disorder

February 22, 2018
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

January 17, 2018
American Journal of Medical Genetics, Part A
Genotype-phenotype correlations in individuals with pathogenic RERE variants

January 13, 2018

Human Mutation

A New Approach to Rare Diseases of Children: The Undiagnosed Diseases Network

January 11, 2018

Journal of Pediatrics

Psychosocial Profiles of Parents of Children with Undiagnosed Diseases: Managing Well or Just Managing?

January 2, 2018

Journal of Genetic Counseling

De Novo Mutations in Protein Kinase Genes CAMK2A and CAMK2B Cause Intellectual Disability

November 2, 2017

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

October 23, 2017
Clinical and Translational Science

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

September 14, 2017
Genetics in Medicine

Model Organisms Facilitate Rare Disease Diagnosis and Therapeutic Research

September 1, 2017
Genetics
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
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