

Resource Summary Report

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MARRVEL

RRID:SCR_016871

Type: Tool

Proper Citation

MARRVEL (RRID:SCR_016871)

Resource Information

URL: <http://marrvel.org/>

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Description: Web tool to search multiple public variant databases simultaneously and provide a unified interface to facilitate the search process. Used for integration of human and model organism genetic resources to facilitate functional annotation of the human genome. Used for analysis of human genes and variants by cross-disciplinary integration of records available in public databases to facilitate clinical diagnosis and basic research.

Abbreviations: MARRVEL

Synonyms: Model organism Aggregated Resources for Rare Variant ExpLoration

Resource Type: analysis service resource, data analysis service, data or information resource, database, production service resource, service resource

Defining Citation: [PMID:28502612](#)

Keywords: integration, database, model, genetic, resource, functional, annotation, genome, data, analysis, dataset, rare, variant, exploration, bio.tools

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Availability: Free, Public, Freely available

Resource Name: MARRVEL

Resource ID: SCR_016871

Alternate IDs: biotools:marrvel

Alternate URLs: <https://bio.tools/marrvel>

Record Creation Time: 20220129T080332+0000

Record Last Update: 20260905T132814+0000

Ratings and Alerts

No rating or validation information has been found for MARRVEL.

No alerts have been found for MARRVEL.

Data and Source Information

Source: [SciCrunch Registry](#)

Usage and Citation Metrics

We found 25 mentions in open access literature.

Listed below are recent publications. The full list is available at [PRECISE-TBI](#) .

Mace K, et al. (2026) Cross-species evidence for a developmental origin of adult hypersomnia with loss of synaptic adhesion molecules beat-1a/CADM2. Nature communications, 17(1), 1628.

Scala M, et al. (2026) ASAH2 deficiency affects sphingolipid homeostasis and neuromotor control, causing a progressive neurological disorder. HGG advances, 7(2), 100587.

Kaizuka T, et al. (2024) FAM81A is a postsynaptic protein that regulates the condensation of postsynaptic proteins via liquid-liquid phase separation. PLoS biology, 22(3), e3002006.

Casas-Tint?? S, et al. (2024) *Drosophila* as a Model for Human Disease: Insights into Rare and Ultra-Rare Diseases. *Insects*, 15(11).

Lu S, et al. (2022) Loss-of-function variants in *TIAM1* are associated with developmental delay, intellectual disability, and seizures. *American journal of human genetics*, 109(4), 571.

Vidali S, et al. (2021) Characterising a homozygous two-exon deletion in *UQCRH*: comparing human and mouse phenotypes. *EMBO molecular medicine*, 13(12), e14397.

Baldrige D, et al. (2021) Model organisms contribute to diagnosis and discovery in the undiagnosed diseases network: current state and a future vision. *Orphanet journal of rare diseases*, 16(1), 206.

Guillemyn B, et al. (2021) Loss of *TANGO1* Leads to Absence of Bone Mineralization. *JBMR plus*, 5(3), e10451.

Usmani MA, et al. (2021) De novo and bi-allelic variants in *AP1G1* cause neurodevelopmental disorder with developmental delay, intellectual disability, and epilepsy. *American journal of human genetics*, 108(7), 1330.

Ravenscroft TA, et al. (2021) Heterozygous loss-of-function variants significantly expand the phenotypes associated with loss of *GDF11*. *Genetics in medicine : official journal of the American College of Medical Genetics*, 23(10), 1889.

Genovesi ML, et al. (2021) *GDF5* mutation case report and a systematic review of molecular and clinical spectrum: Expanding current knowledge on genotype-phenotype correlations. *Bone*, 144, 115803.

Richard EM, et al. (2021) Bi-allelic variants in *SPATA5L1* lead to intellectual disability, spastic-dystonic cerebral palsy, epilepsy, and hearing loss. *American journal of human genetics*, 108(10), 2006.

Mitani T, et al. (2021) High prevalence of multilocus pathogenic variation in neurodevelopmental disorders in the Turkish population. *American journal of human genetics*, 108(10), 1981.

Griffin A, et al. (2021) Phenotypic analysis of catastrophic childhood epilepsy genes. *Communications biology*, 4(1), 680.

Chenouard V, et al. (2021) Advances in Genome Editing and Application to the Generation of Genetically Modified Rat Models. *Frontiers in genetics*, 12, 615491.

Chung HL, et al. (2020) De Novo Variants in *CDK19* Are Associated with a Syndrome Involving Intellectual Disability and Epileptic Encephalopathy. *American journal of human genetics*, 106(5), 717.

Mao D, et al. (2020) De novo *EIF2AK1* and *EIF2AK2* Variants Are Associated with Developmental Delay, Leukoencephalopathy, and Neurologic Decompensation. *American journal of human genetics*, 106(4), 570.

Graves HK, et al. (2020) A Genetic Screen for Genes That Impact Peroxisomes in *Drosophila* Identifies Candidate Genes for Human Disease. *G3* (Bethesda, Md.), 10(1), 69.

Chung HL, et al. (2020) Loss- or Gain-of-Function Mutations in ACOX1 Cause Axonal Loss via Different Mechanisms. *Neuron*, 106(4), 589.

Guo H, et al. (2019) Disruptive mutations in TANC2 define a neurodevelopmental syndrome associated with psychiatric disorders. *Nature communications*, 10(1), 4679.