

# Resource Summary Report

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## MARRVEL

RRID:SCR\_016871

Type: Tool

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### Proper Citation

MARRVEL (RRID:SCR\_016871)

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### 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:** service resource, data or information resource, data analysis service, database, production service resource, analysis 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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NCRR R24 RR032668;  
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NHGRI U01 HG007709;  
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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:** 20260729T044425+0000

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## Ratings and Alerts

No rating or validation information has been found for MARRVEL.

No alerts have been found for MARRVEL.

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## Data and Source Information

**Source:** [SciCrunch Registry](#)

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## Usage and Citation Metrics

We found 22 mentions in open access literature.

**Listed below are recent publications.** The full list is available at [nidm-terms](#) .

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.

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

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

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.

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.

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

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.

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.

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

Ansar M, et al. (2019) Bi-allelic Variants in IQSEC1 Cause Intellectual Disability, Developmental Delay, and Short Stature. American journal of human genetics, 105(5), 907.

Wang J, et al. (2019) Loss of Oxidation Resistance 1, OXR1, Is Associated with an Autosomal-Recessive Neurological Disease with Cerebellar Atrophy and Lysosomal Dysfunction. American journal of human genetics, 105(6), 1237.

Thurmond J, et al. (2019) FlyBase 2.0: the next generation. Nucleic acids research, 47(D1), D759.