

# Resource Summary Report

Generated by SPARC SAWG on Sep 17, 2026

## AutDB

RRID:SCR\_001872

Type: Tool

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### Proper Citation

AutDB (RRID:SCR\_001872)

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### Resource Information

**URL:** <https://gene.sfari.org/database/human-gene/>

**Proper Citation:** AutDB (RRID:SCR\_001872)

**Description:** Curated public database for autism research built on information extracted from the studies on molecular genetics and biology of Autism Spectrum Disorders (ASD). The genetic information includes data from linkage and association studies, cytogenetic abnormalities, and specific mutations associated with ASD. New gene submissions are welcome. Modules: \* Human Gene: thoroughly annotated list of genes that have been studied in the context of autism, with information on the genes themselves, relevant references from the literature, and the nature of the evidence. Uniquely, SFARI Gene incorporates information on both common and rare variants. \* Animal Model: information about lines of genetically modified mice that represent potential models of autism. This information includes the nature of the targeting construct, the background strain and, most importantly, a thorough summary of the phenotypic features of the mice that are most relevant to autism. \* Protein Interaction (PIN): compilation of all known direct protein interactions for those gene products implicated in autism. It presents both graphical and tabular views of interactomes, highlighting connections between autism candidate genes. Each protein interaction is manually verified by consultation with the primary reference. \* Copy Number Variant (CNV): a parallel resource providing genetic information about all known copy number variants linked to autism. \* Gene Scoring: includes a "score" for each autism candidate gene, based on an assessment of the strength of human genetic evidence.

**Abbreviations:** AutDB

**Synonyms:** AutDB - An Interface to Autism Research, Simons Foundation Autism Research Initiative Gene: Autism Database, SFARI Gene: AutDB, SFARI Gene, AutDB: a Genetic Database for Autism Spectrum Disorders

**Resource Type:** data or information resource, data repository, database, service resource, storage service resource

**Defining Citation:** [PMID:19015121](#)

**Keywords:** duplication, gene, genetic syndrome, genetic variation, allelic, autism, autism spectrum disorder, deletion, molecular function, molecular genetics, single-gene disruption, genetic association, genetic variation, allelic variant, copy number variant, cytogenetic, disruption, idiopathic asd, monogenic, mutation, polymorphism, human, animal model, mouse, protein interaction, sfari gene, phenotype, protein interaction, gene scoring, systems biology

**Related Condition:** Autism Spectrum Disorder, Autism

**Funding:** MindSpec: Informatics for Neurodevelopmental Conditions

**Availability:** Free, Freely available

**Resource Name:** AutDB

**Resource ID:** SCR\_001872

**Alternate IDs:** nif-0000-02587

**Alternate URLs:** <http://www.mindspec.org/products/autdb/>, <https://gene.sfari.org/autdb/>

**Old URLs:** <http://autism.mindspec.org/autdb/>

**Record Creation Time:** 20220129T080210+0000

**Record Last Update:** 20260912T195533+0000

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## Ratings and Alerts

No rating or validation information has been found for AutDB.

No alerts have been found for AutDB.

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## Data and Source Information

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

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## Usage and Citation Metrics

We found 64 mentions in open access literature.

**Listed below are recent publications.** The full list is available at [SPARC SAWG](#) .

Boros BD, et al. (2026) Antecedent enhancer activity predicts future susceptibility to seizures in mice. Nature communications, 17(1), 300.

Sterben SP, et al. (2026) Microglia Mitochondria Support Neuronal Maturation via Metabolic and Transcriptional Reprogramming in Human 3D In Vitro Brain Model. Advanced science (Weinheim, Baden-Wurttemberg, Germany), 13(25), e08815.

Marin-Llobet A, et al. (2026) Case Report: MECP2 and SH3KBP1 variants associated with autism spectrum disorder and immune dysregulation. *Frontiers in immunology*, 17, 1814088.

Gill C, et al. (2026) Convergence and divergence of genes informed by common and rare variants of autism spectrum disorders in tissue-specific pathways and gene networks. *Translational psychiatry*, 16(1).

Pérez-Sisqués L, et al. (2026) Autism-like phenotypes and increased NMDAR2D expression in mice with KDM5B histone lysine demethylase deficiency. *Science advances*, 12(21), eadq6577.

Zhao X, et al. (2025) Regulatory Genomic Circuitry of Brain Age by Integrative Functional Genomic Analyses. *Genomics, proteomics & bioinformatics*, 23(5).

Boros BD, et al. (2025) Prior epigenetic status predicts future susceptibility to seizures in mice. *bioRxiv : the preprint server for biology*.

Lertpeerapan P, et al. (2025) Sex-dependent epigenetic disruption of YY1 binding by prenatal BPA exposure downregulates *Matr3* and alters *Agap1* splicing in the offspring hippocampus. *Biology of sex differences*, 16(1), 63.

Sharma M, et al. (2025) Genetic crosstalk of autism spectrum disorders and epilepsy: an insight into the presynapse. *Frontiers in neurology*, 16, 1677134.

Mirabella F, et al. (2025) Glycosylation Pathways Targeted by Deregulated miRNAs in Autism Spectrum Disorder. *International journal of molecular sciences*, 26(2).

Aparicio JG, et al. (2025) Trio exome sequencing of an optic nerve hypoplasia cohort reveals evidence for polygenic architecture. *Ophthalmic genetics*, 46(6), 581.

Zhao X, et al. (2025) Unveiling genetic architecture of white matter microstructure through unsupervised deep representation learning of fractional anisotropy maps. *Research square*.

Dahawi M, et al. (2024) Genetic heterogeneity in familial forms of genetic generalized epilepsy: from mono- to oligogenism. *Human genomics*, 18(1), 130.

LeMaster C, et al. (2024) Mapping structural variants to rare disease genes using long-read whole genome sequencing and trait-relevant polygenic scores. *medRxiv : the preprint server for health sciences*.

Kereszturi É, et al. (2024) Database-assisted screening of autism spectrum disorder related gene set. *Molecular brain*, 17(1), 55.

Ben-Mahmoud A, et al. (2024) Genome Sequencing Identifies 13 Novel Candidate Risk Genes for Autism Spectrum Disorder in a Qatari Cohort. *International journal of molecular sciences*, 25(21).

Bar O, et al. (2024) Reanalysis of Trio Whole-Genome Sequencing Data Doubles the Yield in Autism Spectrum Disorder: De Novo Variants Present in Half. *International journal of molecular sciences*, 25(2).

López-Tobón A, et al. (2023) GTF2I dosage regulates neuronal differentiation and social behavior in 7q11.23 neurodevelopmental disorders. *Science advances*, 9(48), eadh2726.

More RP, et al. (2023) Identifying rare genetic variants in 21 highly multiplex autism families: the role of diagnosis and autistic traits. *Molecular psychiatry*, 28(5), 2148.

Shah S, et al. (2023) Brain Gene Co-Expression Network Analysis Identifies 22q13 Region Genes Associated with Autism, Intellectual Disability, Seizures, Language Impairment, and Hypotonia. *Genes*, 14(11).