Resource Summary Report

Generated by NIF on May 25, 2025

AutismKB

RRID:SCR_006937
Type: Tool

Proper Citation

AutismKB (RRID:SCR_006937)

Resource Information

URL: http://autismkb.cbi.pku.edu.cn/

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Description: Genetic factors contribute significantly to ASD. AutismKB is an evidence-based knowledgebase of Autism spectrum disorder (ASD) genetics. The current version contains 2193 genes (99 syndromic autism related genes and 2135 non-syndromic autism related genes), 4617 Copy Number Variations (CNVs) and 158 linkage regions associated with ASD by one or more of the following six experimental methods: # Genome-Wide Association Studies (GWAS); # Genome-wide CNV studies; # Linkage analysis; # Low-scale genetic association studies; # Expression profiling; # Other low-scale gene studies. Based on a scoring and ranking system, 99 syndromic autism related genes and 383 non-syndromic autism related genes (434 genes in total) were designated as having high confidence. Autism spectrum disorder (ASD) is a heterogeneous neurodevelopmental disorder with a prevalence of 1.0-2.6%. The three core symptoms of ASD are: # impairments in reciprocal social interaction; # communication impairments; # presence of restricted, repetitive and stereotyped patterns of behavior, interests and activities.

Abbreviations: AutismKB

Synonyms: Autism Knowledgebase

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

Defining Citation: PMID:22139918

Keywords: gene, copy number variation, linkage region, genome-wide association study, family-based association study, case-control association study, expression profile, blast,

syndromic, non-syndromic, snp, vntr, bio.tools, FASEB list

Related Condition: Autism spectrum disorder, Autism

Funding: Merck ;
Johnson and Johnson ;
Natural Science Foundation of China 31025014;
Natural Science Foundation of China 2011CBA01102

Resource Name: AutismKB

Resource ID: SCR_006937

Alternate IDs: biotools:autismkb, nlx_151318

Alternate URLs: https://bio.tools/autismkb

Record Creation Time: 20220129T080238+0000

Record Last Update: 20250525T030958+0000

Ratings and Alerts

No rating or validation information has been found for AutismKB.

No alerts have been found for AutismKB.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 33 mentions in open access literature.

Listed below are recent publications. The full list is available at NIF.

Kasitipradit K, et al. (2025) Sex-specific effects of prenatal bisphenol A exposure on transcriptome-interactome profiles of autism candidate genes in neural stem cells from offspring hippocampus. Scientific reports, 15(1), 2882.

Cucinotta F, et al. (2023) Diagnostic yield and clinical impact of chromosomal microarray analysis in autism spectrum disorder. Molecular genetics & genomic medicine, 11(8), e2182.

Qiu S, et al. (2023) Nexus between genome-wide copy number variations and autism spectrum disorder in Northeast Han Chinese population. BMC psychiatry, 23(1), 96.

Chehbani F, et al. (2022) Yield of array-CGH analysis in Tunisian children with autism spectrum disorder. Molecular genetics & genomic medicine, 10(8), e1939.

Shen L, et al. (2021) Comparative analysis of the autism?related variants between different autistic children in a family pedigree. Molecular medicine reports, 24(4).

Pang W, et al. (2021) Untangle the Multi-Facet Functions of Auts2 as an Entry Point to Understand Neurodevelopmental Disorders. Frontiers in psychiatry, 12, 580433.

Mullegama SV, et al. (2021) Transcriptome analysis of MBD5-associated neurodevelopmental disorder (MAND) neural progenitor cells reveals dysregulation of autism-associated genes. Scientific reports, 11(1), 11295.

Lee S, et al. (2021) Gene Dosage- and Age-Dependent Differential Transcriptomic Changes in the Prefrontal Cortex of Shank2-Mutant Mice. Frontiers in molecular neuroscience, 14, 683196.

Gao H, et al. (2021) Drug repositioning based on network-specific core genes identifies potential drugs for the treatment of autism spectrum disorder in children. Computational and structural biotechnology journal, 19, 3908.

Al-Mubarak BR, et al. (2020) Whole exome sequencing in ADHD trios from single and multi-incident families implicates new candidate genes and highlights polygenic transmission. European journal of human genetics : EJHG, 28(8), 1098.

Bitar T, et al. (2019) Identification of rare copy number variations reveals PJA2, APCS, SYNPO, and TAC1 as novel candidate genes in Autism Spectrum Disorders. Molecular genetics & genomic medicine, 7(8), e786.

Zhou WZ, et al. (2019) Targeted resequencing of 358 candidate genes for autism spectrum disorder in a Chinese cohort reveals diagnostic potential and genotype-phenotype correlations. Human mutation, 40(6), 801.

Pichitpunpong C, et al. (2019) Phenotypic subgrouping and multi-omics analyses reveal reduced diazepam-binding inhibitor (DBI) protein levels in autism spectrum disorder with severe language impairment. PloS one, 14(3), e0214198.

Thongkorn S, et al. (2019) Sex Differences in the Effects of Prenatal Bisphenol A Exposure on Genes Associated with Autism Spectrum Disorder in the Hippocampus. Scientific reports, 9(1), 3038.

Kasem E, et al. (2018) Neurexins and neuropsychiatric disorders. Neuroscience research, 127, 53.

Chen C, et al. (2018) IDGenetics: a comprehensive database for genes and mutations of intellectual disability related disorders. Neuroscience letters, 685, 96.

Wang P, et al. (2018) Enriched expression of genes associated with autism spectrum

disorders in human inhibitory neurons. Translational psychiatry, 8(1), 13.

Yang C, et al. (2018) AutismKB 2.0: a knowledgebase for the genetic evidence of autism spectrum disorder. Database : the journal of biological databases and curation, 2018.

Wang P, et al. (2017) CRISPR/Cas9-mediated heterozygous knockout of the autism gene CHD8 and characterization of its transcriptional networks in cerebral organoids derived from iPS cells. Molecular autism, 8, 11.

Ahmad M, et al. (2017) Topoisomerase 3? is the major topoisomerase for mRNAs and linked to neurodevelopment and mental dysfunction. Nucleic acids research, 45(5), 2704.