Resource Summary Report

Generated by PRECISE-TBI on Sep 7, 2026

DECIPHER

RRID:SCR_006552
Type: Tool

Proper Citation

DECIPHER (RRID:SCR_006552)

Resource Information

URL: http://decipher.sanger.ac.uk/

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Description: Interactive database which incorporates a suite of tools designed to aid the interpretation of submicroscopic chromosomal imbalance. Used to enhance clinical diagnosis by retrieving information from bioinformatics resources relevant to the imbalance found in the patient. Contributing to the DECIPHER database is a Consortium, comprising an international community of academic departments of clinical genetics. Each center maintains control of its own patient data (which are password protected within the center""s own DECIPHER project) until patient consent is given to allow anonymous genomic and phenotypic data to become freely viewable within Ensembl and other genome browsers. Once data are shared, consortium members are able to gain access to the patient report and contact each other to discuss patients of mutual interest, thus facilitating the delineation of new microdeletion and microduplication syndromes.

Abbreviations: DECIPHER

Synonyms: Database of Chromosomal Imbalance and Phenotype in Humans using Ensembl Resources, DECIPHER: Database of Chromosomal Imbalance and Phenotype in Humans using Ensembl Resources, Database of Chromosomal Imbalance Phenotype in Humans using Ensembl Resources, Decipher

Resource Type: data or information resource, database

Defining Citation: PMID:19344873

Keywords: chromosomal imbalance, phenotype, chromosome, gene, genome, deletion, duplication, copy number, genotype, polymorphism, FASEB list

Related Condition: Developmental disorder, Microdeletion Syndrome, Overgrowth syndrome, Microduplication syndrome, Deletion syndrome, Duplication syndrome, Wolf-Hirschhorn Syndrome, Williams-Beuren Syndrome, Smith-Magenis Syndrome, Etc

Funding: Wellcome Trust WT077008

Availability: Acknowledgement required

Resource Name: DECIPHER

Resource ID: SCR_006552

Alternate IDs: nlx_151653, OMICS_00265

Record Creation Time: 20220129T080236+0000

Record Last Update: 20260905T133135+0000

Ratings and Alerts

No rating or validation information has been found for DECIPHER.

No alerts have been found for DECIPHER.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 1991 mentions in open access literature.

Listed below are recent publications. The full list is available at PRECISE-TBI .

Mishra S, et al. (2026) Decoding Genetic Risk Factors in Recurrent Pregnancy Loss: An Integrative Chromosomal and Bioinformatics Approach. Biochemical genetics.

Low KJ, et al. (2026) The LMSz method - an automatable scalable approach to constructing gene-specific growth charts in rare disorders. European journal of human genetics : EJHG, 34(3), 348.

Papadopoulou S, et al. (2026) Shared stream-lake patterns in diversity, rRNA-based activity and community assembly of bacteria and microeukaryotes under distinct hydrological regimes. FEMS microbiology ecology, 102(3).

Cho G, et al. (2026) Bayesian Pairwise Compositional Lotka-Volterra Modeling Infers Potential Rhizosphere Microbial Suppressors of Ralstonia pseudosolanacearum. The plant pathology journal, 42(1), 50.

Magistrado D, et al. (2026) The Aedes aegypti bacterial microbiota is robust to infection with the obligate microsporidian parasite Edhazardia aedis. bioRxiv : the preprint server for biology.

Low KJ, et al. (2026) How do clinician and parent-reported data differ? An analysis of similarity and difference in the datasets from a cross-syndrome genetics cohort study

(GenROC). Journal of medical genetics, 63(4), 259.

Du C, et al. (2026) The clinical value of invasive prenatal diagnosis in fetuses with isolated aberrant right subclavian artery: a retrospective study. BMC pregnancy and childbirth, 26(1).

Zhang Y, et al. (2026) Prenatal diagnosis of distal Xq28 duplication syndrome: case reports and literature review. Molecular cytogenetics, 19(1).

Jost C, et al. (2026) De novo heterozygous variants of the RSF1 gene are responsible for a syndromic neurodevelopmental disorder. European journal of human genetics : EJHG, 34(4), 554.

Zhang L, et al. (2026) Prenatal diagnosis and follow-up of a child with mosaic tetrasomy 9p without obvious abnormal clinical manifestations. Molecular cytogenetics, 19(1).

Saia F, et al. (2026) Genetic architecture and clinical features of Tourette syndrome in a child and adolescent cohort: an explorative clinical exome-based study. Frontiers in psychiatry, 17, 1744145.

Zheng Z, et al. (2026) Application of chromosomal microarray analysis for prenatal diagnosis in 315 ultrasonically abnormal fetuses. Frontiers in genetics, 17, 1744468.

Yang Y, et al. (2026) Gut Microbiome Alterations in Canine Idiopathic Epilepsy: A Pairwise Case-Control Study. bioRxiv : the preprint server for biology.

Andersen S, et al. (2026) Oncostatin M receptor deficiency as a novel candidate genetic cause of autosomal recessive hyper-IgE syndrome. Journal of human immunity, 2(3), e20250119.

Álvarez-Rodríguez B, et al. (2026) Comparative analysis of deep mutational scanning datasets in enteroviruses A and B identifies functional divergence and therapeutic targets. Nature ecology & evolution, 10(3), 467.

Iordanescu II, et al. (2026) Prenatal Diagnosis of Sex Chromosome Aneuploidies: A Retrospective Study Using QF-PCR, SNP-Based Chromosomal Microarray Analysis, and NIPT. Genes, 17(2).

Zhang Z, et al. (2026) Enhanced genetic diagnosis in early pregnancy loss: an integrated approach using CNV-Seq and STR genotyping. Reproductive biology and endocrinology : RB&E, 24(1).

Glassford M, et al. (2026) Familial p.(Ala73Thr) Variant in GNB2 Associated With Mild Neurodevelopmental Features and Pilocytic Astrocytoma. Clinical case reports, 14(6), e72913.

Bodkhe R, et al. (2026) Environmental microbial extracts for longitudinal studies of gut microbiome assembly and maintenance. bioRxiv : the preprint server for biology.

Chirmade S, et al. (2026) GWAS SVatalog: a visualization tool to aid fine-mapping of GWAS loci with structural variations. Heredity, 135(3), 199.