

Resource Summary Report

Generated by NIF on Apr 27, 2025

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](https://pubmed.ncbi.nlm.nih.gov/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: 20250426T055858+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 1623 mentions in open access literature.

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

Castera L, et al. (2025) A European Survey to Identify Challenges in the Management of Metabolic Dysfunction-Associated Steatotic Liver Disease. Liver international : official journal of the International Association for the Study of the Liver, 45(2), e16224.

Assentato L, et al. (2025) The type of environment has a greater impact on the larval microbiota of Anopheles arabiensis than on the microbiota of their breeding water. FEMS microbiology ecology, 101(1).

Hendrycks W, et al. (2025) Deterministic and stochastic effects drive the gut microbial

diversity in cucurbit-feeding fruit flies (Diptera, Tephritidae). PloS one, 20(1), e0313447.

Yang S, et al. (2025) Expanded non-invasive prenatal testing offers better detection of fetal copy number variations but not chromosomal aneuploidies. PloS one, 20(1), e0312184.

Mao X, et al. (2025) A phenotype-based AI pipeline outperforms human experts in differentially diagnosing rare diseases using EHRs. NPJ digital medicine, 8(1), 68.

Kim JM, et al. (2025) Uncovering potential causal genes for undiagnosed congenital anomalies using an in-house pipeline for trio-based whole-genome sequencing. Human genomics, 19(1), 1.

Hutchins L, et al. (2025) Arthropods are kin: Operationalizing Indigenous data sovereignty to respectfully utilize genomic data from Indigenous lands. Molecular ecology resources, 25(2), e13822.

Bouزيد A, et al. (2025) Whole exome sequencing identifies ABHD14A and MRNIP as novel candidate genes for developmental language disorder. Scientific reports, 15(1), 367.

Fernando MATM, et al. (2025) Testing Phylogenetic Placement Accuracy of DNA Barcode Sequences on a Fish Backbone Tree: Implications of Backbone Tree Completeness and Species Representation. Ecology and evolution, 15(1), e70817.

Zeng Y, et al. (2025) Prenatal genetic detection in foetus with gallbladder size anomalies: cohort study and systematic review of the literature. Annals of medicine, 57(1), 2440638.

Zhao W, et al. (2025) GoFCards: an integrated database and analytic platform for gain of function variants in humans. Nucleic acids research, 53(D1), D976.

Zhu B, et al. (2025) A multi-omics spatial framework for host-microbiome dissection within the intestinal tissue microenvironment. Nature communications, 16(1), 1230.

García-Bodelón Á, et al. (2025) Predators in the Dark: Metabarcoding Reveals Arcellinida Communities Associated with Bat Guano, Endemic to Dinaric Karst in Croatia. Microbial ecology, 87(1), 166.

Hebert PDN, et al. (2025) Barcode 100K Specimens: In a Single Nanopore Run. Molecular ecology resources, 25(1), e14028.

Budzinski L, et al. (2025) Single-cell microbiota phenotyping reveals distinct disease and therapy-associated signatures in Crohn's disease. Gut microbes, 17(1), 2452250.

DeVilbiss SE, et al. (2025) From subsidies to stressors: Positively skewed ecological gradients alter biological responses to nutrients in streams. Ecological applications : a publication of the Ecological Society of America, 35(1), e3086.

Zhuang J, et al. (2024) Prenatal diagnosis and molecular cytogenetic characterization of fetuses with central nervous system anomalies using chromosomal microarray analysis: a seven-year single-center retrospective study. *Scientific reports*, 14(1), 2271.

Huang J, et al. (2024) Associations between genomic aberrations, increased nuchal translucency, and pregnancy outcomes: a comprehensive analysis of 2,272 singleton pregnancies in women under 35. *Frontiers in medicine*, 11, 1376319.

Kim M, et al. (2024) Prostate cancers with distinct transcriptional programs in Black and White men. *Genome medicine*, 16(1), 92.

Zhang W, et al. (2024) Commensal microbiome dysbiosis elicits interleukin-8 signaling to drive fibrotic skin disease. *PNAS nexus*, 3(7), pgae273.