

Resource Summary Report

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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: database, data or information resource

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: 20260725T191145+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 1797 mentions in open access literature.

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

Ye H, et al. (2026) Associations between gut microbiota and personality traits: insights from a captive common marmoset (*Callithrix jacchus*) colony. *Microbiology spectrum*, 14(1), e0044325.

Liu X, et al. (2025) Increasing Stemness Drives Prostate Cancer Progression, Plasticity, Therapy Resistance and Poor Patient Survival. *bioRxiv* : the preprint server for biology.

Pan P, et al. (2025) The Genetics of 241 Fetuses With Talipes Equinovarus: A 8-Year Monocentric Retrospective Study. *Molecular genetics & genomic medicine*, 13(2), e70076.

Contrò G, et al. (2025) CDK13-Related Disorder: Novel Insights From A Series of 27 Cases and Recommendations for Clinical Management. *Clinical genetics*, 108(2), 146.

Wang X, et al. (2025) The phenotypic and genetic spectrum of AKT3-related neurodevelopmental condition. *Scientific reports*, 15(1), 7484.

Ogawa C, et al. (2025) Diversity of Subgroup of *Fibrobacter succinogenes* in the Rumen and Feces of Cattle and Sheep. *Animal science journal = Nihon chikusan Gakkaiho*, 96(1), e70043.

Twigg SRF, et al. (2025) The power of mouse models in the diagnostic odyssey of patients with rare congenital anomalies. *Mammalian genome : official journal of the International Mammalian Genome Society*, 36(2), 354.

Kalanithy JC, et al. (2025) TFAP2E is implicated in central nervous system, orofacial and maxillofacial anomalies. *Journal of medical genetics*, 62(2), 126.

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.

Jiang M, et al. (2025) Sequential prenatal diagnosis of fetal skeletal dysplasia: A cohort study. *Acta obstetrica et gynecologica Scandinavica*, 104(5), 860.

Bennett AJ, et al. (2025) Molecular epidemiology of *Eimeria* spp. parasites and the faecal microbiome of Indiana bats (*Myotis sodalis*): a non-invasive, multiplex metabarcode survey of an endangered species. *Microbial genomics*, 11(2).

Rosas HD, et al. (2025) Gut dysbiosis in Down syndrome: A potentially unexplored culprit for early Alzheimer's disease. *Alzheimer's & dementia : the journal of the Alzheimer's Association*, 21(6), e70330.

Fanoro OT, et al. (2025) Metabolic profiling and antibacterial activity of secondary metabolites extracted from the endophytic bacteria of *Combretum erythrophyllum*. *Scientific reports*, 15(1), 14739.

Rodriguez-Cruz UE, et al. (2025) Running against the clock: exploring microbial diversity in an extremely endangered microbial oasis in the Chihuahuan Desert. *FEMS microbiology ecology*, 101(5).

Arbesfeld JA, et al. (2025) Mapping MAVE data for use in human genomics applications. *Genome biology*, 26(1), 179.

Francioli D, et al. (2025) Microbial inoculants modulate the rhizosphere microbiome, alleviate plant stress responses, and enhance maize growth at field scale. *Genome biology*, 26(1), 148.

Kim GJ, et al. (2025) Exome sequencing of patients with syndromic tall stature reveals four novel candidate genes. *Endocrine connections*, 14(7).

Shumeri E, et al. (2025) Digenesis in Charcot-Marie-Tooth Disease: Impact of Combined Mutations in the MFN2 and GDAP1 Genes. *Journal of the peripheral nervous system : JPNS*, 30(3), e70044.

Sun R, et al. (2025) A recurrent ABCC2 c.2439 + 5G > A variant disturbs mRNA splicing and causes Dubin-Johnson syndrome. *BMC medical genomics*, 18(1), 118.

Kardish MR, et al. (2025) More Than a Stick in the Mud: Eelgrass Leaf and Root Bacterial Communities Are Distinct From Those on Physical Mimics. *Environmental microbiology reports*, 17(3), e70086.