Resource Summary Report

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Deciphering Developmental Disorders

RRID:SCR_006171
Type: Tool

Proper Citation

Deciphering Developmental Disorders (RRID:SCR_006171)

Resource Information

URL: http://www.ddduk.org/

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Description: The Deciphering Developmental Disorders (DDD) study aims to find out if using new genetic technologies can help doctors understand why patients get developmental disorders. To do this we have brought together doctors in the 23 NHS Regional Genetics Services throughout the UK and scientists at the Wellcome Trust Sanger Institute, a charitably funded research institute which played a world-leading role in sequencing (reading) the human genome. The DDD study involves experts in clinical, molecular and statistical genetics, as well as ethics and social science. It has a Scientific Advisory Board consisting of scientists, doctors, a lawyer and patient representative, and has received National ethical approval in the UK. Over the next few years, we are aiming to collect DNA and clinical information from 12,000 undiagnosed children in the UK with developmental disorders and their parents. The results of the DDD study will provide a unique, online catalogue of genetic changes linked to clinical features that will enable clinicians to diagnose developmental disorders. Furthermore, the study will enable the design of more efficient and cheaper diagnostic assays for relevant genetic testing to be offered to all such patients in the UK and so transform clinical practice for children with developmental disorders. Over time, the work will also improve understanding of how genetic changes cause developmental disorders and why the severity of the disease varies in individuals. The Sanger Institute will contribute to the DDD study by performing genetic analysis of DNA samples from patients with developmental disorders, and their parents, recruited into the study through the Regional Genetics Services. Using microarray technology and the latest DNA sequencing methods, research teams will probe genetic information to identify mutations (DNA errors or rearrangements) and establish if these mutations play a role in the developmental disorders observed in patients. The DDD initiative grew out of the groundbreaking DECIPHER database, a global partnership of clinical genetics centres set up in 2004, which allows researchers and clinicians to share clinical and genomic data from patients worldwide. The

DDD study aims to transform the power of DECIPHER as a diagnostic tool for use by clinicians. As well as improving patient care, the DDD team will empower researchers in the field by making the data generated securely available to other research teams around the world. By assembling a solid resource of high-quality, high-resolution and consistent genomic data, the leaders of the DDD study hope to extend the reach of DECIPHER across a broader spectrum of disorders than is currently possible.

Abbreviations: DDD

Synonyms: Deciphering Developmental Disorders (DDD)

Resource Type: research forum portal, portal, service resource, data or information resource, disease-related portal, material storage repository, storage service resource, topical portal, biospecimen repository

Defining Citation: PMID:21679367

Keywords: microarray, sequencing, child, genome, chromosome, dna sequencing, ethics, interview, dna, saliva, clinical, genetics, gene, diagnosis, phenotype, clinical data, FASEB list

Related Condition: Developmental disorder, Genetic disorder, Parent, Neurodevelopmental disorder, Congenital anomaly, Abnormal growth, Dysmorphic feature, Unusual behavioral phenotype

Funding: Wellcome Trust ;
Health Innovation Challenge Fund

Resource Name: Deciphering Developmental Disorders

Resource ID: SCR_006171

Alternate IDs: nlx_151673

Record Creation Time: 20220129T080234+0000

Record Last Update: 20250516T053817+0000

Ratings and Alerts

No rating or validation information has been found for Deciphering Developmental Disorders.

No alerts have been found for Deciphering Developmental Disorders.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 40 mentions in open access literature.

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

Li D, et al. (2024) Spliceosome malfunction causes neurodevelopmental disorders with overlapping features. The Journal of clinical investigation, 134(1).

Nappi M, et al. (2024) Constitutive opening of the Kv7.2 pore activation gate causes KCNQ2-developmental encephalopathy. Proceedings of the National Academy of Sciences of the United States of America, 121(49), e2412388121.

Pagnamenta AT, et al. (2024) The impact of inversions across 33,924 families with rare disease from a national genome sequencing project. American journal of human genetics, 111(6), 1140.

Ragoussis V, et al. (2022) Using data from the 100,000 Genomes Project to resolve conflicting interpretations of a recurrent TUBB2A mutation. Journal of medical genetics, 59(4), 366.

Wright CF, et al. (2021) Evaluating variants classified as pathogenic in ClinVar in the DDD Study. Genetics in medicine : official journal of the American College of Medical Genetics, 23(3), 571.

Kaiser VB, et al. (2021) Mutational bias in spermatogonia impacts the anatomy of regulatory sites in the human genome. Genome research, 31(11), 1994.

Martinez-Granero F, et al. (2021) Comparison of the diagnostic yield of aCGH and genome-wide sequencing across different neurodevelopmental disorders. NPJ genomic medicine, 6(1), 25.

Levy MA, et al. (2021) Deficiency of TET3 leads to a genome-wide DNA hypermethylation episignature in human whole blood. NPJ genomic medicine, 6(1), 92.

Voisin N, et al. (2021) Variants in the degron of AFF3 are associated with intellectual disability, mesomelic dysplasia, horseshoe kidney, and epileptic encephalopathy. American journal of human genetics, 108(5), 857.

Balasubramanian M, et al. (2021) Comprehensive study of 28 individuals with SIN3A-related disorder underscoring the associated mild cognitive and distinctive facial phenotype. European journal of human genetics : EJHG, 29(4), 625.

Satterstrom FK, et al. (2020) Large-Scale Exome Sequencing Study Implicates Both Developmental and Functional Changes in the Neurobiology of Autism. Cell, 180(3), 568.

Beck DB, et al. (2020) Delineation of a Human Mendelian Disorder of the DNA Demethylation Machinery: TET3 Deficiency. American journal of human genetics, 106(2), 234.

Williamson KA, et al. (2020) Recurrent heterozygous PAX6 missense variants cause severe bilateral microphthalmia via predictable effects on DNA-protein interaction. Genetics in medicine : official journal of the American College of Medical Genetics, 22(3), 598.

Alharatani R, et al. (2020) Novel truncating mutations in CTNND1 cause a dominant craniofacial and cardiac syndrome. Human molecular genetics, 29(11), 1900.

Pagnamenta AT, et al. (2019) Delineation of dominant and recessive forms of LZTR1-associated Noonan syndrome. Clinical genetics, 95(6), 693.

Turnpenny PD, et al. (2018) Missense Mutations of the Pro65 Residue of PCGF2 Cause a Recognizable Syndrome Associated with Craniofacial, Neurological, Cardiovascular, and Skeletal Features. American journal of human genetics, 103(5), 786.

Low KJ, et al. (2018) Phenotype of CNTNAP1: a study of patients demonstrating a specific severe congenital hypomyelinating neuropathy with survival beyond infancy. European journal of human genetics : EJHG, 26(6), 796.

Wright CF, et al. (2018) Making new genetic diagnoses with old data: iterative reanalysis and reporting from genome-wide data in 1,133 families with developmental disorders. Genetics in medicine : official journal of the American College of Medical Genetics, 20(10), 1216.

Pagnamenta AT, et al. (2018) A homozygous variant disrupting the PIGH start-codon is associated with developmental delay, epilepsy, and microcephaly. Human mutation, 39(6), 822.

Bengani H, et al. (2017) Clinical and molecular consequences of disease-associated de novo mutations in SATB2. Genetics in medicine : official journal of the American College of Medical Genetics, 19(8), 900.