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

**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 Agency:** Wellcome Trust, Health Innovation Challenge Fund

**Resource Name:** Deciphering Developmental Disorders

**Resource ID:** SCR_006171

**Alternate IDs:** nlx_151673

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**Ratings and Alerts**

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

No alerts have been found for Deciphering Developmental Disorders.

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**Data and Source Information**

Source: [SciCrunch Registry](https://www.scribbr.com)

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**Usage and Citation Metrics**

We found 37 mentions in open access literature.
Listed below are recent publications. The full list is available at RRID.

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.


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), 571-575.


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.

Ragoussis V, et al. (2021) Using data from the 100,000 Genomes Project to resolve conflicting interpretations of a recurrent mutation. Journal of medical genetics.


