

Searching DECIPHER

DECIPHER has an advanced search facility that allows querying the database using a combination of terms including phenotypes, gene symbols, chromosomal locations/bands and other fields. Please follow this short guide to help you make effective queries against the DECIPHER database.

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The Search Box

DECIPHER search box is available on the top of every DECIPHER web page.



Clicking on the magnifying glass will open up a page that has some search examples.

Search Examples

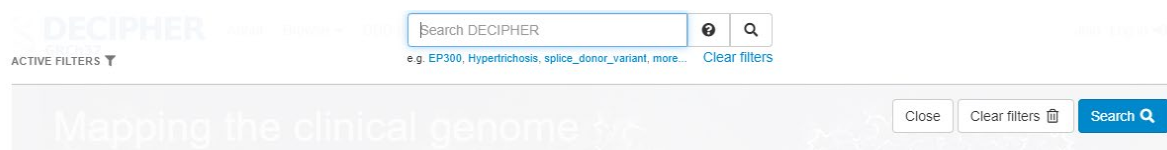
- [Hypertrichosis](#) - by phenotype
- [hp:0001831](#) - by HPO Identifier. Prefix the HPO ID you wish to search with **hp:**
- [6:157099063-157531913](#) - by position
- [17p11.2](#) - by band (band will be converted to position)
- [EP300](#) - by gene
- [Benign](#) - by **pathogenicity**
- [Biparental](#) - by **inheritance**
- [splice_donor_variant](#) - by **consequence**
- [255882](#) - by DECIPHER patient ID
- [dendritic spines](#) - plain text search in syndrome names, descriptions, etc.

You can combine multiple terms, by separating them with a comma and a space, for example:

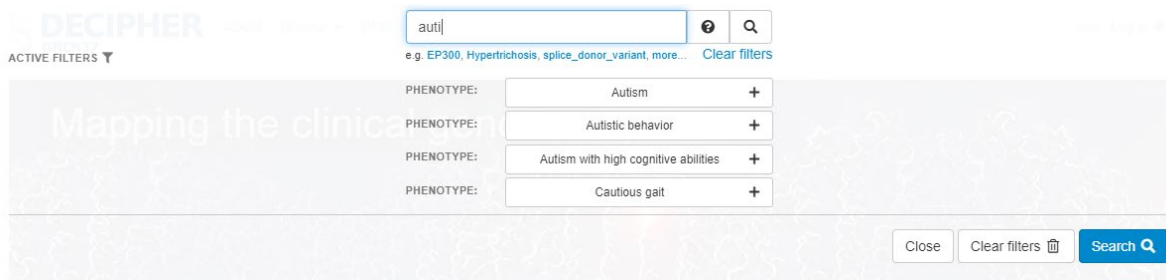
- [17p11.2, Dyscalculia](#) - Combine band and phenotype
- [Arachnodactyly, High palate](#) - Search for patients with more than one phenotype

Simple Searching

The moment you place your cursor inside the search box, a new panel opens up as below:



Begin typing your query text, for example the gene name or phenotype term you are interested in. As you type the term, the system will begin to suggest terms from our database. (Note that suggested search terms do not necessarily have associated patient records).



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ACTIVE FILTERS ▼

Search: 🔍 🔍

e.g. EP300, Hypertrichosis, splice_donor_variant, more... [Clear filters](#)

PHENOTYPE: +

PHENOTYPE: +

PHENOTYPE: +

PHENOTYPE: +

Close

Choose the search term of your choice with your mouse and click on “Search” to perform your search.

Search results from the DECIPHER database that match the query term are shown as below. For more information on any result, clicking on the DECIPHER Patient ID will open the patient record.

Search results for "phenotype:Autism" ([Refine Search](#))

Patients: 1682

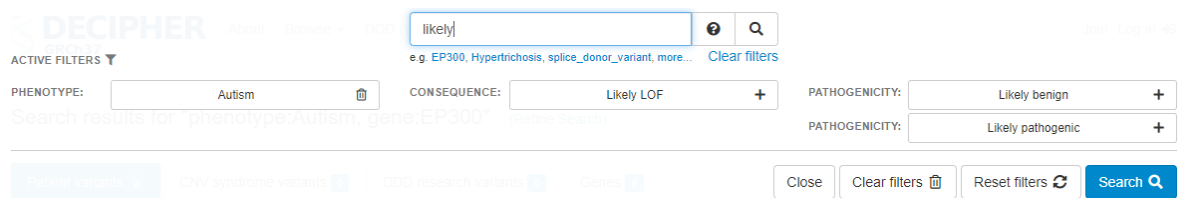
Patients: 1 to 10 of 1682

DECIPHER Patient ID	Sex	Phenotype(s)	Variants	Contact
106	46XX	Abnormal eyebrow morphology, Autism, Bulbous nose, Deeply set eye, Hypotelorism, Intellectual disability, Localized hirsutism, Microcephaly, Microtia, Narrow mouth, Proportionate short stature, Seizures	1	<input type="button" value="✉"/>
126	46XX	Autism, Intellectual disability, Joint laxity, Muscular hypotonia, Strabismus	1	<input type="button" value="✉"/>
132	46XY	Autism, Intellectual disability, Muscular hypotonia	1	<input type="button" value="✉"/>

Refining search results

If your search has returned more (or fewer) results than you expected, you can change the search terms by clicking on “Refine Search”.

This opens up the search box again with your existing filters in place, and allows additional search terms to be added to or removed from the search, for example an additional phenotype, gene or variant pathogenicity. As before, as you type the term, the system will begin to suggest terms from our database.



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ACTIVE FILTERS ▼

Search: 🔍 🔍

e.g. EP300, Hypertrichosis, splice_donor_variant, more... [Clear filters](#)

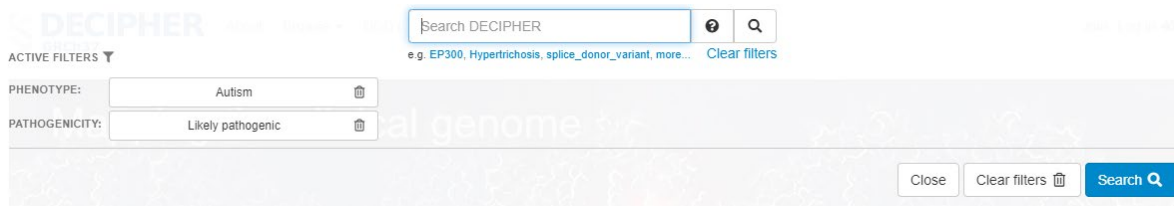
PHENOTYPE: ✕ CONSEQUENCE: + PATHOGENICITY: +

Search results for "phenotype:Autism, gene:EP300" ([Refine Search](#))

PATHOGENICITY: +

Close

The search term can be selected with your mouse.



Additional search terms can be added in the same way, as required. Search terms added to the active filters list can be removed by clicking on the rubbish bin icon next to the search term. Once all search terms have been added, click on “Search” to perform your search.




Search results from the DECIPHER database that match the query terms are shown as below.

Search results for "phenotype:Autism, pathogenicity:"Likely pathogenic"" ([Refine Search](#))

Patient variants 197 | CNV syndrome variants 0 | DDD research variants 0 | Genes 0

Results | [Browser](#)

Variants: 1 to 10 of 214 | Show: All variant types | Filter...

DECIPHER Patient	Sex	Location	Size	Inheritance / Genotype	Pathogenicity / Contribution	Phenotype(s)	Contact
257998	46XY	1 145432842 146068297 Duplication	635.46 kb	Paternally inherited Heterozygous	Likely pathogenic Partial	Abnormality of the dentition, Autism, Intellectual disability, Restrictive behavior	
258332	46XY	2 166848945 166848945 A > T	SNV	De novo Heterozygous	Likely pathogenic Full	Autism, Developmental stagnation at onset of seizures, Focal clonic seizures, Global developmental delay, Hyperactivity, Seizures, Status epilepticus	
258350	46XY	12 116421998 116421998 G > T	SNV	De novo Heterozygous	Likely pathogenic Full	Adducted thumb, Autism, Brachycephaly, Bruxism, Global developmental delay, Protruding ear, Talipes equinovarus, Uplifted earlobe	

Searching Patient IDs and Internal References

Any search that only contains a set of numbers is automatically interpreted by the search system as a patient identifier. So typing in “259614” for example will return results for DECIPHER patient 259614.

For any searches for patient internal references (accessible to logged in users for their projects only), please prefix your search with “ref:”. For example, if you want to find your patient with your internal identifier “12345” please search with “ref:12345”.

Viewing search results




Search results can be viewed in a table as shown below. There is a dropdown filter at the top of the table, which can be used to filter the results by variant type. By default, “All variant types” are displayed. A free text filter is also available.

If you are a logged-in user, an additional dropdown filter will be shown which allows you to filter by your access to the patient, for example “Patients in my projects” or “Open access patients”. By default, “All patients” are displayed.

In addition, the results table can be sorted by clicking on the arrows on the relevant column heading.

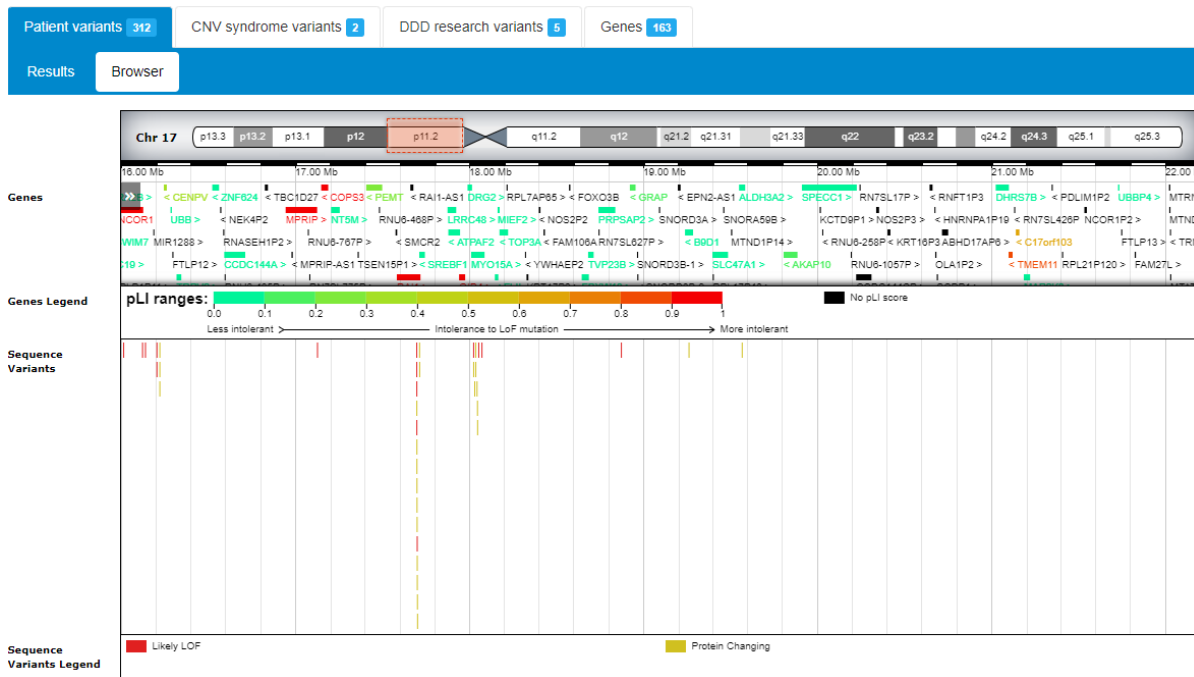
Variants: 1 to 10 of 214

Show: All variant types

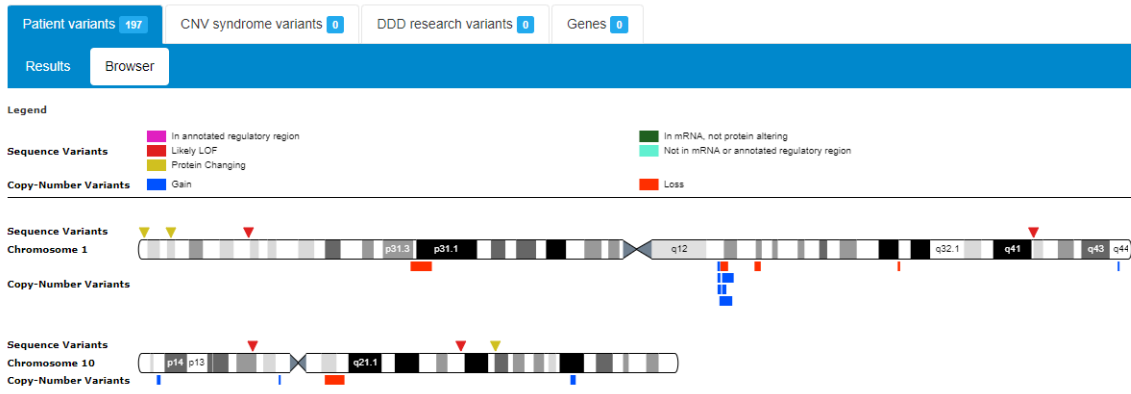
DECIPHER Patient	Sex	Location	Size	Inheritance / Genotype	Pathogenicity / Contribution	Phenotype(s)	Contact
257998	46XY	1 145432842 146068297 Duplication	635.46 kb	Paternally inherited Heterozygous	Likely pathogenic Partial	Abnormality of the dentition, Autism, Intellectual disability, Restrictive behavior	
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258350	46XY	12 116421988 116421988 G > T	SNV	De novo Heterozygous	Likely pathogenic Full	Adducted thumb, Autism, Brachycephaly, Bruxism, Global developmental delay, Protruding ear, Talipes equinovarus, Uplifted earlobe	

It is also possible to view the results in the Genome Browser, by clicking on the “Browser” tab.

If the search results refer to a single genomic location (e.g. a gene or band) the results will be displayed on the genome browser as below.



If the search results are across the genome the results will be displayed as a karyotype view as shown below (not available in IE11).



Any search results for CNV syndromes, DDD research variants or genes will be displayed on the relevant tabs.