

# Searching DECIPHER

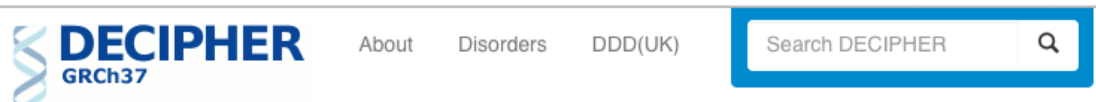
DECIPHER Release 8.5 introduces advanced search facility that allows permits querying the database using a combination of terms including phenotypes, gene symbols, chromosomal locations/bands and other fields. We have also introduced an autocomplete feature that can be used to quickly find the search term of interest as you begin typing your search term. Please follow this short guide to help you to 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 without any term filled in the box will open up a page that has some search examples.



Please enter a search term

[Search examples](#)

Please enter a search term

Search examples

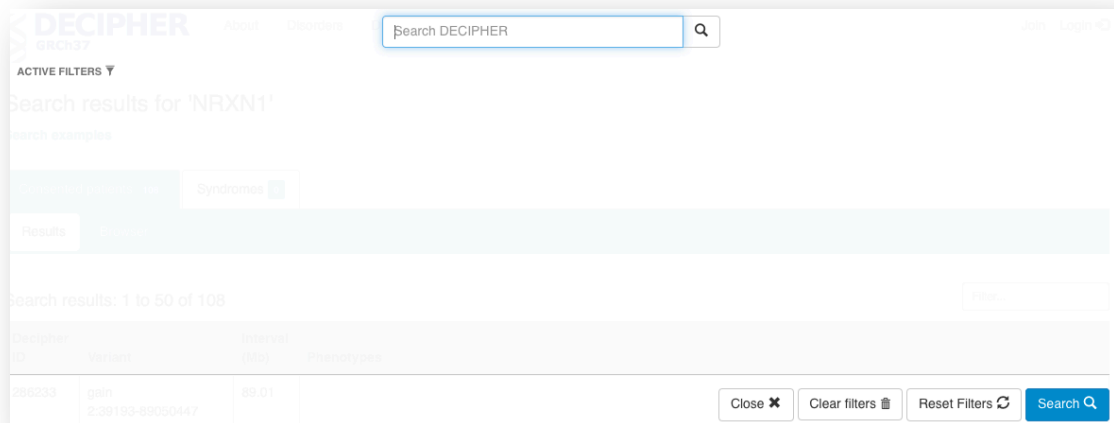
- [Short stature](#) - by phenotype
- [6:157099063-157531913](#) - by overlapping position
- [17p11.2](#) - by band (band will be converted to position)
- [BRCA2](#) - by gene
- [Benign](#) - by *pathogenicity*
- [Biparental](#) - by *inheritance*
- [255882](#) - by DECIPHER patient ID
- [dendritic spines](#) - plain text search in syndrome names, descriptions, etc.

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

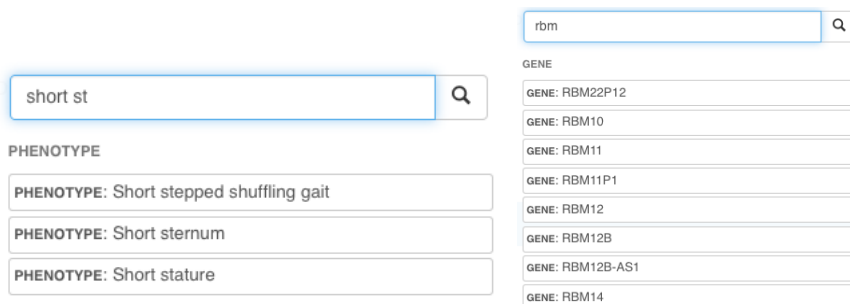
- [17p11.2, Dyscalculia](#) - Combine band and phenotype
- [Arachnodactyly, High palate](#) - Search for patients with more than one phenotype

## Simple Searching

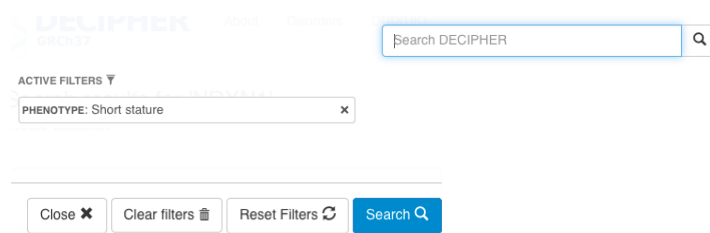
The moment you place your cursor inside the search box, a new panel opens up as below:



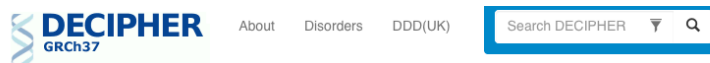
Begin typing the query text: gene name or phenotype term. As you type the term, the system will begin to suggest terms from our database.



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



Search results from the DECIPHER database that match the query term are shown as below. The search box now shows a “funnel” icon to let you know that a filter is currently active. You can remove the filter by clicking inside the search box and then choosing the filter to be removed by clicking on the “x” next to the filter. Or you can add additional search terms to further refine your search.



Search results for 'Short stature'

[Search examples](#)

Search results: 1 to 50 of 886

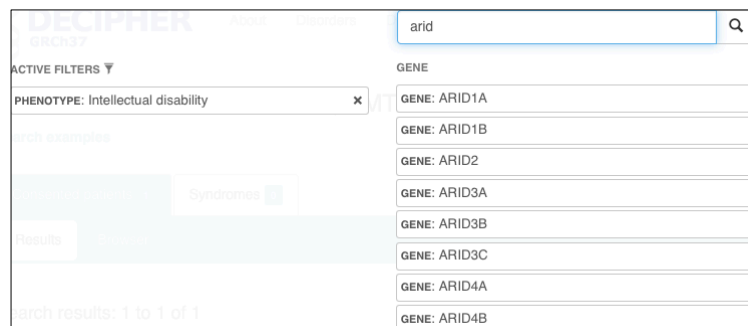
Filter...

| Decipher ID | Phenotype(s)   |
|-------------|--|
| 1019        | Arachnodactyly, Delayed speech and language development, Intellectual disability, Kyphosis, Microcephaly, Open mouth, Pes cavus, Proportionate short stature   |
| 1024        | Abnormality of the hand, Ataxia, Delayed speech and language development, Hypoplasia of the corpus callosum, Intellectual disability, Microcephaly, Pes planus, Scoliosis, Seizures, Short stature, Strabismus |

## Combination Queries

DECIPHER search now permits combining multiple queries with the “and” qualifier. For example you could search for the co-occurrence of two phenotypes, or a specific phenotype – gene symbol combination etc.

The example below shows a combination search for patients in DECIPHER with phenotype: intellectual disability and variant affecting the gene: ARID1B.



DECIPHER  
GRCh37

Search DECIPHER

ACTIVE FILTERS

PHENOTYPE: Intellectual disability

GENE: ARID1B

Search results for 'Intellectual disability, ARID1B'

[Search examples](#)

Search results: 1 to 13 of 13

| Decipher ID | Variant                       | Interval (Mb) | Phenotypes  |
|-------------|-------------------------------|---------------|---|
| 248472      | loss<br>6:155893417-158639123 | 2.75          | Cystic lung disease, Deeply set eye, Deep palmar creases, Defect in the atrial septum, Hyperconvex nail, Intellectual disability, Microtia, Plagiocephaly, Upslanted palpebral fissure, Wide intermamillary distance  |
| 248770      | loss<br>6:150977264-159129307 | 8.15          | Abnormality of the nervous system, Deeply set eye, Delayed speech and language development, Intellectual disability, Localized hirsutism, Low anterior hairline, Seizures, Thick lower lip vermillion, Vertical nystagmus, Visual impairment  |
| 250126      | gain<br>6:153063292-159666476 | 6.60          | Abnormality of the mouth, Cleft palate, Cryptorchidism, Focal seizures with impairment of consciousness or awareness, Intellectual disability, Kyphosis, Non-midline cleft lip  |
| 250455      | loss<br>6:157126309-157761085 | 0.63          | Abnormality of the nose, Autism, Broad hallux, Choanal atresia, Coarse facial features, Facial asymmetry, Intellectual disability, Low posterior hairline, Macroglossia, Myopia, Proportionate short stature, Self-mutilation, Thick lower lip vermillion, Thick upper lip vermillion, Ureteral duplication |

If you wish to search for any term in DECIPHER using a broader text-search, prefix your search term with “text:”. The text-search will generally be effective in finding your own patients if you are a logged-in user as it will query fields not available to public users such as Notes and Internal Reference etc.

*Example:* Searching for patients with specific phenotype within a chromosomal region/band.

Type “17p11” in the search box followed by a comma, the input is parsed as a band and shown in the filters. Now type in a phenotype like “obesity” to find all patients with a variant in the 17p11 region with obesity as a recorded phenotype.

DECIPHER  
GRCh37

Search DECIPHER

ACTIVE FILTERS

BAND: 17p11

PHENOTYPE: Obesity

Search results for '17p11, Obesity'

Search examples

Consented patients 3   Syndromes 2

Results   Browser

Search results: 1 to 3 of 3 Filter...

| Decipher ID | Variant                      | Interval (Mb) | Phenotypes   |
|-------------|------------------------------|---------------|--|
| 1643        | loss<br>17:16892401-20193169 | 3.30          | Intellectual disability, Macroglossia, Mandibular prognathia, Obesity, Protruding tongue, Round face, Small feet, Strabismus, Synophrys, Thick eyebrow, Wide mouth |
| 249307      | loss<br>17:16802930-20221836 | 3.62          | Abnormality of the nervous system, Intellectual disability, Obesity, Strabismus, Synophrys   |
| 251420      | loss<br>17:16728248-20407149 | 3.68          | Intellectual disability, Obesity, Sleep disturbance  |

### Search Constraints:

- All queries are of the type “AND” i.e., results must match ALL of the criteria
- Only one positional query is allowed in a combination search. For example gene symbol searches cannot be combined with bands or two genes cannot be searched together, because you would be looking for variants that are in multiple positions at once.

ACTIVE FILTERS ▼

GENE: ARID1B x

GENE: RBM8A x

You should only search for one band, gene, or position at a time.

ACTIVE FILTERS ▼

GENE: RBM8A x

BAND: 17p11 x

You should only search for one band, gene, or position at a time.

## Searching Patient IDs and Internal References

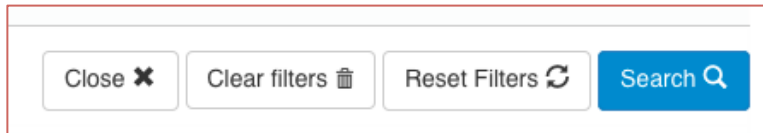
Any search that only contains a set of numbers is automatically interpreted by the search system as a patient identifier. So typing in “275854” for example will get you results for DECIPHER patient with identifier 257854. To search for any occurrence of this number in DECIPHER, please prefix with “text:” e.g. text: 257854.

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 “CG258456” please search with “ref:CG258456”.

## Reset Searches

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The funnel on the search box indicates that a search filter is active. To add/edit your filters or to restart from scratch, click inside the search box to open up the search dialog. Click on the "Clear filters" to remove all filters, and "Close" to close the search dialogue box. "Reset filters" appears after you have done a search and can be used to return the filters to the terms you searched for if you have changed them since.



### Finally...

We welcome your feedback on this new development. Please send us your comments and suggestions for improvement to [decipher@sanger.ac.uk](mailto:decipher@sanger.ac.uk) and we will endeavour to incorporate these in future developments.