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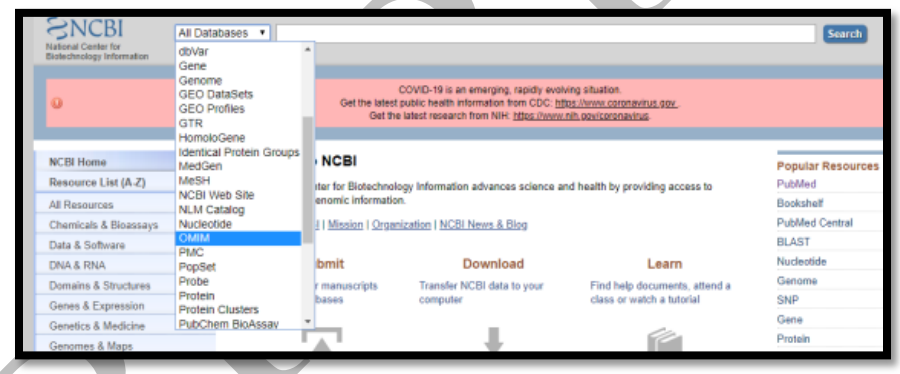
OMIM Overview

It is the short form of ‘Online Mendelian Inheritance in Man’ and allows us access to information related to genetic disorders.

On the NCBI front page, we can search for the OMIM from the list of the databases present in the drop down menu.

In the search bar, we search for the name of the genetic disorder or its ID or the name of the gene.

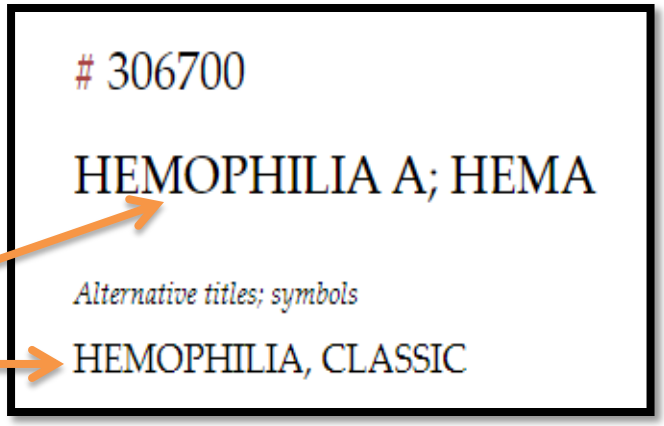


When we search for our term, we will get our results in the following format.

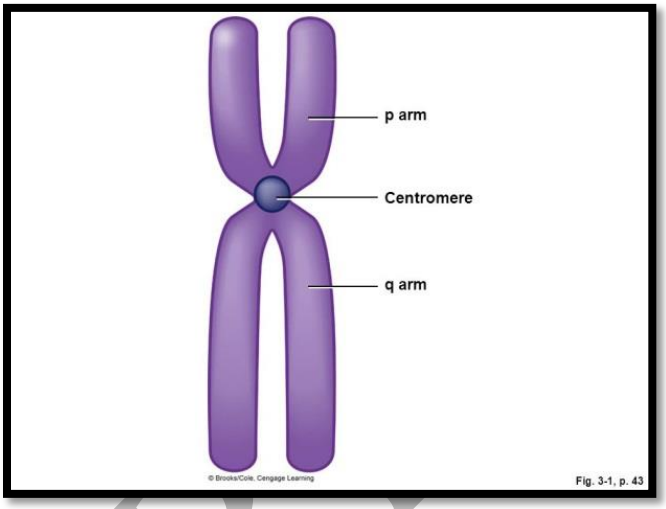
After clicking on our desired result we get the following near the top of the screen.

The number is the MIM number (detailed later) and the ‘Hemophilia A; HEMA’ refer to the ‘Genetic disorder; Abbreviation’

Underneath them we see the title.



Location



The site where the gene can be found within the genome.

In this example, the gene is found on the 'X' chromosome on the 'q' arm of chromosome number 28.

Phenotype

Indicates that this gene gives us the phenotype specified in this field.

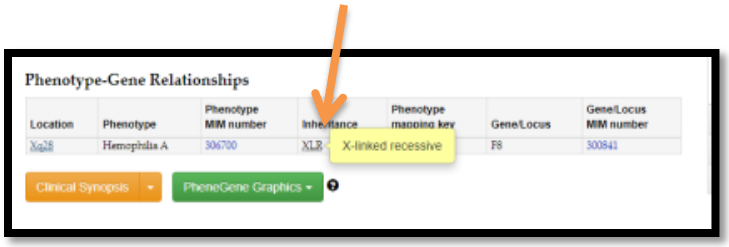
In this example, Hemophilia A.

Phenotype MIM number

It serves as a personal identification number for this phenotype. This is specific for the OMIM database.

In this example, it is 306700.

Inheritance



Allows us to know what is the type of inheritance of this genetic disorder. They could be autosomal dominant or recessive or x-linked dominant or recessive.

In this case it is referred to as XLR (X-linked recessive). In order to get the full form of the abbreviation, hover your indicator over the abbreviation.

Phenotype mapping key

Every phenotype on the map is assigned a phenotype mapping **key** numbered 1–4:

1. The disorder was positioned by mapping of the wild-type gene
2. The disorder itself was mapped
3. The molecular basis of the disorder is known
4. The disorder is a chromosome deletion or duplication syndrome.

In this example, the molecular basis of the disorder is known, therefore it was assigned the phenotype mapping key 3.

Gene/locus

It is the protein (end result) that we get as a result of the expression of this gene.

In this example, it is F8 (Coagulation factor VIII).

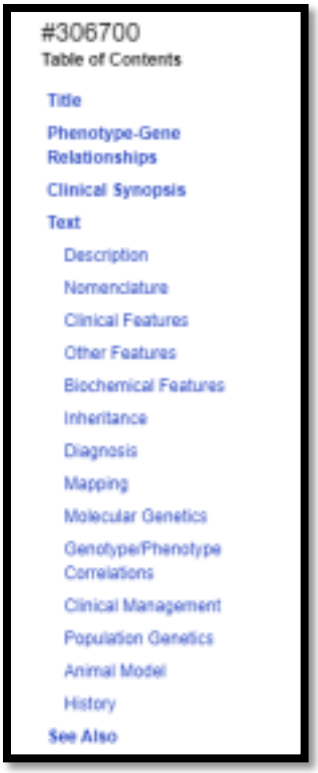
Gene/locus MIM number

It is the number that the protein (end result) gets assigned in the OMIM database. Clicking on it will direct you to the database of the gene at hand

In this example, it is 300841.

Additional information

There is a bar present on the left hand side of the screen, where we can skip to the desired section regarding the disorder we're studying.



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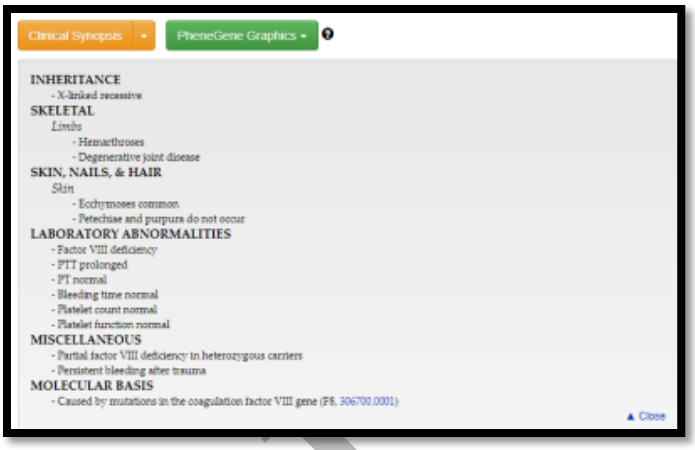
Clinical synopsis

A general summary regarding the clinical aspect of this disorder can be obtained by clicking on the clinical synopsis button of the arrow just next to it.



Gene details

Upon clicking on the MIM number of the gene we are directed to its page.

Details regarding it can be present and these include its name and alternative names, the cytogenic location (where and in which chromosome it is present) as well as its genomic coordinates (the gene extends from which nucleotide to which nucleotide).