

# HGV&TB: A comprehensive online resource on Human genes and genetic variants associated with Tuberculosis

## Home Page:

The home page provides a brief overview to the content of the database. The users can query the database with multiple parameters that include GeneID, rsID, PMID, Technique, Template, Geographic Location, Phenotype and Concluded Pathogenicity. Three examples to access the database with each parameter have also been provided. Important characteristics of the data are shown in the figurative form under the highlights section.

**HGV&TB** Database for Human Genes and Genetic Variants associated with Tuberculosis

HOME MANUAL CONTACT

### About HGV&TB

Tuberculosis is a major cause of mortality in the developing world. Since the establishment of genetic factors as a key player in TB, large volumes of work has been done elucidating the role of individual genes in TB susceptibility. Despite this we are far from reaching a consensus regarding the genes involved in TB susceptibility. To bring an end to this prevailing disagreement, a compilation of data on genetic variants involved in Tuberculosis susceptibility is utmost required. To date, no such systematic effort has been made in this regard.

We have, thus, comprehensively curated all genetic variations reported to be associated with Tuberculosis susceptibility in human genome and established a novel Locus Specific Database that stores all Human genes and genetic variants associated with Tuberculosis (HGV&TB). The database currently house 102 genes associated with various forms of tuberculosis with 307 variants supported with pertinent patient data, obtained after an exhaustive literature study. 101 of these variations have been found to be exonic and 78 in intronic region. In addition, the database concludes the pathogenicity of the genetic variations investigated their phenotypic consequences and ethnic origin, concluding 299 genetic variants in 71 genes to be pathogenic.

Search: GeneID, RSID, PMID, Technique, Template, Geographic Location, Phenotype or Concl.

Database can be accessed by:

Browsing Genes Example: AKT1   CCL1   CCL5	Browsing RSID Example: rs4586   rs159290   rs210837
Browsing PMID Example: 16352737   19057661   20176143	Browsing Technique Example: PCR   MALDI-TOF   Genotyping
Browsing Template Example: DNA   RNA   mRNA	Browsing Geographic Location Example: China   India   Vietnam
Browsing Phenotype Example: Tuberculosis   Pulmonary Tuberculosis   Extrapulmonary Tuberculosis	Browsing Pathogenicity Example: Pathogenic   Probably Pathogenic

### Highlights:

Citation:  
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## Different types of queries available:

- ❖ **Gene:** The searching can be carried out based on gene HGNC symbol.
- ❖ **rsID:** Searching can also be performed by using rsid. In case you don't remember the entire rsid, you can use a partial rsID as a search query as well.
- ❖ **PMID:** The searching can also be performed by using the PMID of the corresponding literature.
- ❖ **Phenotype:** The searching can be carried out based on the tuberculosis phenotype for example, Pulmonary Tuberculosis, Extrapulmonary Tuberculosis etc
- ❖ **Template:** Searching can also be performed by using the detection template like DNA, RNA etc
- ❖ **Technique:** Searching can also be performed by using the detection technique like PCR, MALDI-TOF etc.

- ❖ **Geographic Location:** Searching can also be performed by using the geographic location of the population studied.
- ❖ **Pathogenicity:** Searching can also be performed by the pathogenicity status of the variant.

Database can be accessed by:

Browsing Genes

Example: [AKT1](#) | [CCL1](#) | [CCL5](#)

Browsing PMID

Example: [16352737](#) | [19057661](#) | [20176143](#)

Browsing Template

Example: [DNA](#) | [RNA](#) | [mRNA](#)

Browsing Phenotype

Example: [Tuberculosis](#) | [Pulmonary Tuberculosis](#) | [Extrapulmonary Tuberculosis](#)

Browsing RSID

Example: [rs4586](#) | [rs159290](#) | [rs210837](#)

Browsing Technique

Example: [PCR](#) | [MALDI-TOF](#) | [Genotyping](#)

Browsing Geographic Location

Example: [China](#) | [India](#) | [Vietnam](#)

Browsing Pathogenicity

Example: [Pathogenic](#) | [Probably Pathogenic](#)

## Query Result:

The result is divided in four different sections namely Gene, Variant, Details and External Links.

Unique **HGVID** has been provided to all the entries within the database.

The **Gene** Section gives information regarding the gene that includes its name, associated haplotypes, location of the gene (Exonic, Intronic, 5' UTR, 3' UTR, Upstream or Downstream), the exon number on which the variant is present and the variation.

The **Variant** section details the variant information, quoting the type of variation reported in literature, the associated phenotype, the reported p-value and odds ratio corresponding to the variation, the reported and concluded pathogenicity and the HGVS notation for the query.

The **Details** section quotes the experimental details regarding the variant queried. It provides information on the type of detection technique and template used and the origin, ethnicity and geographic location of the sample in which the variation has been found.

The **External Links** section takes the user to dbSNP, PUBMED, UCSC and Gene Card with the variation into consideration.

Gene, Phenotype, Concluded Pathogenicity, Detection Technique, Detection Template and Geographic location in the query result have been linked to query the database as well.

In case of multiple results for a single query, all the sections for each associated result are reported one by one under their unique HGVID.

**HGVID** **HG17:rs4586**

**Gene Information**

Gene	CCL2
Haplotypes	-
Location	Exonic
Exon	-
Values	c.900C>T

**Variant Information**

Type	SNP
Phenotype	Pediatric Tuberculosis disease
P-Value	0.029
Odds Ratio	1.34
Reported Pathogenicity	Probably Pathogenic
Concluded Pathogenicity	Pathogenic
HGVS	-

**Experimental Details**

Detection Template	ONA
Detection Technique	PCR
Origin	-
Ethnicity	Han Chinese
Geographic Location	North China

**External Links**

dbSNP	rs4586
PUBMED	Feng WX et. al.,21556333
UCSC	-
Gene Card	CCL2

**Links to submit as a query**

Result for rs4586 query