

# Vitamin B12 Database User Manual

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## 1. Introduction

The Vitamin B12 Database is designed to provide comprehensive information on genetic variants associated with Vitamin B12 levels, including detailed annotations from scientific literature. This user manual will guide you through the features and functionalities of the database, ensuring you can easily navigate and utilize its full potential.

## 2. System Requirements

- A modern web browser (e.g., Google Chrome, Mozilla Firefox, Safari).
- Stable internet connection.

## 3. Getting Started

1. **Accessing the Database:** ○ Open your preferred web browser and navigate to the Vitamin B12 Database URL.
2. **Interface Overview:**
  - The homepage features a search bar, example searches, gene and phenotype charts, and various informational sections.



## About Us

The following Pubmed Search String [link](#) was used to identify 433 research publications which are being carefully reviewed to curate B12 associated variants. The search string utilizes a combination of MeSH Terms and plain text of relevant keywords. Of the 433 research publications, a review of 22 relevant publications has currently led to the creation of a Google spreadsheet database consisting of 153 associations with serum/plasma B12 levels or B12 deficiency status involving 91 variants belonging to 30 genetic loci. This is an ongoing work.

## Data Statistics :

Last Updated	22nd Aug, 2024.
No. of Publications	49
No. of Genes	94
No. of DBSNP IDs	264
No. of Phenotypes	6

DBSNP ID / Phenotype or Trait / Variant / Gene

Search

User Manual : Click to view

## Example Search :

Field	Example 1	Example 2
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## Phenotype Classification :

■ Total serum B12 
 ■ Total plasma B12 
 ■ HoloTC 
 ■ HoloHC 
 ■ MMA 
 ■ TotalTC

DBSNP ID / Phenotype or Trait / Variant / Gene

Search

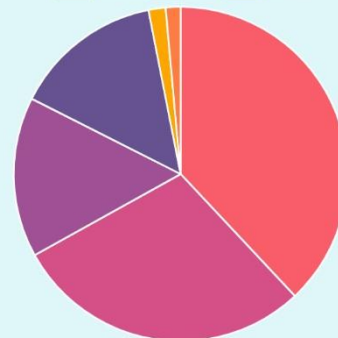
User Manual : Click to view

## Example Search :

Field	Example 1	Example 2
DBSNP ID	rs602662	rs1005887
Phenotype/Trait	Total serum B12	HoloTC
Variant	2:190319749	19:48703417
Gene	TcblR	BHMT

## Phenotype Classification :

■ Total serum B12 
 ■ Total plasma B12 
 ■ HoloTC 
 ■ HoloHC 
 ■ MMA 
 ■ TotalTC



## Contact Us

If you have any questions or need assistance, feel free to reach out to us:  
 Email: [contact@example.com](mailto:contact@example.com)  
 Address: 123 Example Street, City, Country

## 4. Using the Database

### Search Functionality

- **Search Bar:** You can search for entries by typing a query into the search bar.
- **Searchable Fields:**
  - DBSNP ID: Search by specific SNP IDs.
  - Phenotype/Trait: Look for associated traits or phenotypes.  
 Note: here it is important to note that the searchable Phenotype/Trait include: HoloTC, HoloHC, TotalTC, MMA, Total B12, Total serum B12, Total plasma B12. It is also important to note that Total B12 is a consolidation of both Total serum B12 and Total plasma B12.
  - Variant: Identify specific genetic variants. (Syntax for variants is ChrNo:Position)

(iv) Gene: Find information linked to specific genes.

- The database will search across all relevant columns (DBSNP id, Variant, Phenotype/Trait, Gene) to find matches.

### Understanding the Results

- Results Page:** Each search result is displayed in individual boxes, showing details like Gene, Variant, Phenotype, and more. Results will be displayed, paginated, if necessary, with each result showing aggregated data related to the search query, with a 'View Details' button under Actions.

Total serum B12

Search

User Manual : Click to view

Please find the results for your query below:

Number of results: 109

Results per page: 5

DBSNP ID	Phenotype/Trait	Variant	Gene	Actions
rs34324219	Total serum B12	11:59855905	TCN1	<a href="#">View Details</a>
rs601338	Total serum B12	19:48703417	FUT2	<a href="#">View Details</a>
rs41281112	Total serum B12	13:99866380	CLYBL, LOC101927437	<a href="#">View Details</a>
rs602662	Total serum B12	19:48703728	FUT2	<a href="#">View Details</a>
rs526934	Total serum B12	11:59866020	TCN1	<a href="#">View Details</a>

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### Viewing Details

- Details Page:** Clicking on 'View Details' will take you to the details page, where specific information about that entry is displayed.
- Column Headers:** The column names appear on the left, with corresponding values on the right.
- Top box(blue):** the first box that appears on the details page consists of all the columns whose value remains the same in all the results and is hence, consolidated into one box at the top to keep it organised.

Vitamin B12 Database

St. John's  
Research Institute



Details for Total serum B12

No. of results: 4

DBSNP id: [rs34324219](#)

Chromosome number: 11

End Position: 59855905

Reference : C

Gene: [TCN1](#)

Other Function:

Amino Acid Change:

G Build: hg38

Start Position: 59855905

Variant: 11:59855905

Alternate: A

Gene Function: Distribution

Genomic Location: Intronic

- **Record box:** Each record box provides comprehensive information about the result. Here, it is important to note that:
  - (i) Each record box is showing you aggregated results for a particular PMID, i.e., each record box shows all the results coming from the same paper in one box.

<b>1. From Publication: Genome-wide association study of vitamin B6, vitamin B12, folate, and homocysteine blood concentrations.</b> <a href="#">PMID: 19303062</a>  <b>Phenotype/Trait: Total serum B12</b>	
<b>Study Parameters</b>	
<b>Ethnicity:</b> Caucasian (InCHIANTI+SardinIA+BLSA)	<b>Geographical Origin:</b> Mixed
<b>Country:</b> Mixed	<b>Study Type:</b> GWAS
<b>Effect Allele:</b> A	<b>Effect Allele Frequency:</b> 0.47
<b>Effect/Beta (SE/CI):</b> 44.2	<b>Odds Ratio[CI]:</b>
<b>Pvalue:</b> 2.43E-12	<b>Pvalue_Dup:</b> 2.43E-12
<b>Net Effect:</b> Increase	<b>Effect description:</b> pg/ml
<b>Description:</b> Tanaka et al	

- (ii) In-case there are multiple values for any column, it is due to the presence of multiple cohorts in the paper, and therefore, multiple values are shown, each separated by a '|' or ','.

<b>Study Parameters</b>	
<b>Ethnicity:</b> Danish - Inter99   Danish - Health2006	<b>Geographical Origin:</b> Western Europe
<b>Country:</b> Denmark	<b>Study Type:</b> GWAS
<b>Effect Allele:</b>	<b>Effect Allele Frequency:</b>
<b>Effect/Beta (SE/CI):</b> -15 (-19;-10)   -7 (-12;2)	<b>Odds Ratio[CI]:</b>
<b>Pvalue:</b> 8.00E-11   7.00E-03	<b>Pvalue_Dup:</b> 8.00E-11   7.00E-03
<b>Net Effect:</b> Decrease	<b>Effect description:</b>
<b>Description:</b> Allin et al	

- (iii) If multiple phenotypes are observed in one paper, then it will be shown in the record box with multiple 'Phenotype/Variant' sub-headings and the relevant columns and their values below.

**1. From Publication: The FUT2 secretor variant p.Trp154Ter influences serum vitamin B12 concentration via holo-haptocorrin glycosylation.**

[PMID: 29040465](#)

**Phenotype/Trait: HoloHC**

**Study Parameters**

<b>Ethnicity:</b> Irish	<b>Geographical Origin:</b> North America and Caribbean
<b>Country:</b> Ireland {Republic}	<b>Study Type:</b> GWAS
<b>Effect Allele:</b>	<b>Effect Allele Frequency:</b>
<b>Effect/Beta (SE/CI):</b> -0.05961 (0.006145)	<b>Odds Ratio[CI]:</b>
<b>Pvalue:</b> 8.03E-22	<b>Pvalue_Dup:</b> 8.03E-22
<b>Net Effect:</b> Decrease	<b>Effect description:</b>
<b>Description:</b> Velkova et al	

**Phenotype/Trait: Total serum B12**

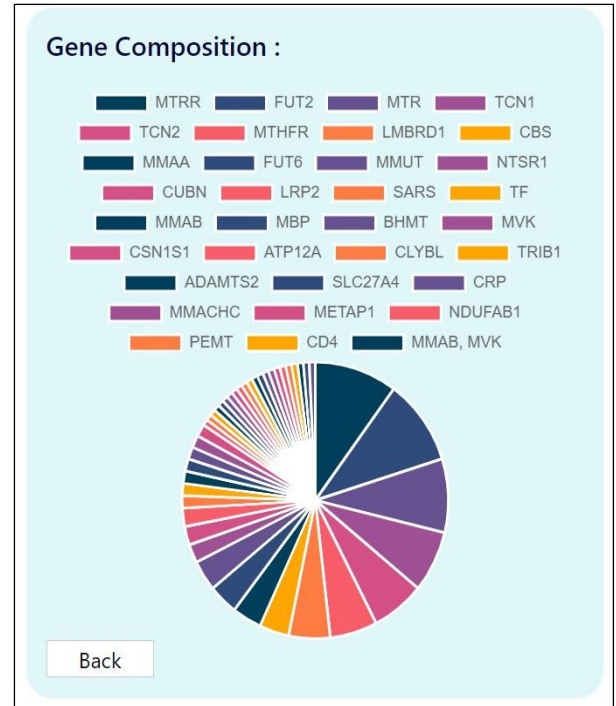
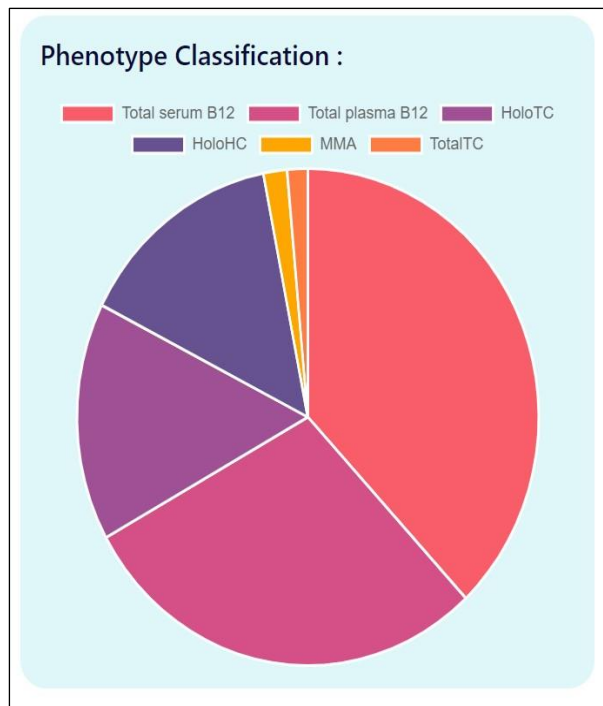
**Study Parameters**

<b>Ethnicity:</b> Irish	<b>Geographical Origin:</b> North America and Caribbean
<b>Country:</b> Ireland {Republic}	<b>Study Type:</b> GWAS
<b>Effect Allele:</b>	<b>Effect Allele Frequency:</b>
<b>Effect/Beta (SE/CI):</b> -0.05113 (0.00544)	<b>Odds Ratio[CI]:</b>
<b>Pvalue:</b> 1.32E-20	<b>Pvalue_Dup:</b> 1.32E-20

- **Hyperlinks:** Clicking on a DBSNP ID will take you to its NIH entry, clicking on the gene name will take you to its GeneCards website and clicking on PMID will take you to the NCBI entry for further reading
- **Search by Column:** The details page allows for targeted searches within specific columns, providing focused results for in-depth analysis.
- **JSON Response:** The data is returned in JSON format, with all associated documents retrieved from the database based on the selected column and query.

## Gene and Phenotype Charts

- **Phenotype Chart:** Displays a pie chart representing the distribution of phenotypes in the dataset. Each slice is clickable, leading to more detailed information about the associated genes. If you want to view the charts without a particular phenotype, just click on its name and its corresponding slice will be removed from the pie chart.
- **Gene Chart:** This chart shows the gene composition for a selected phenotype. Use the "Back" button to return to the Phenotype Pie Chart. If you want to view the charts without a particular gene, just click on its name and its corresponding slice will be removed from the pie chart



## 5. Additional Features Example Searches

- **Predefined Searches:** Use the example searches provided on the homepage to quickly explore the database.

**Example Search :**

Field	Example 1	Example 2
DBSNP ID	rs602662	rs1005887
Phenotype/Trait	Total serum B12	HoloTC
Variant	2:190319749	19:48703417
Gene	TcbIR	BHMT

## User Manual

- **Accessing the User Manual:** A "User Manual" box on the homepage provides a link to this document in PDF format for easy reference.

[User Manual : Click to view](#)

## Contact Us

- **Support:** The "Contact Us" section on the homepage provides contact information for further assistance.

## 6. Troubleshooting

- **Search Issues:** Ensure that the search term is entered correctly and that your internet connection is stable.
- **Display Issues:** If elements are not aligning correctly, try refreshing the page or using a different web browser.

## 7. Contact Information

- **Email:** [contact@example.com](mailto:contact@example.com)
- **Address:** 123 Example Street, City, Country