Resource Summary Report

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Database of Human Hemoglobin Variants and Thalassemias

RRID:SCR_007084
Type: Tool

Proper Citation

Database of Human Hemoglobin Variants and Thalassemias (RRID:SCR_007084)

Resource Information

URL: http://globin.cse.psu.edu/globin/hbvar

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Description: HbVar is a relational database of information about hemoglobin variants and mutations that cause thalassemia. The initial data came from Syllabi authored by Prof. Titus H.J. Huisman, Mrs. Marianne F.H. Carver, Dr. Erol Baysal, and Prof. Georgi D. Efremov. This information was converted to a database, and now new entries are added and old entries are corrected by curators. HbVar results from a collaboration among several investigators at Penn State University (USA), INSERM Creteil (France), and Boston University Medical Center (USA). Visit our query page or summary page to see the types of information available.

Synonyms: HbVar

Resource Type: database, data or information resource

Keywords: hemoglobin, hemoglobin mutation, hemoglobin variant, thalassemia

Resource Name: Database of Human Hemoglobin Variants and Thalassemias

Resource ID: SCR_007084

Alternate IDs: nif-0000-02942

Record Creation Time: 20220129T080239+0000

Record Last Update: 20260803T040501+0000

Ratings and Alerts

No rating or validation information has been found for Database of Human Hemoglobin Variants and Thalassemias.

No alerts have been found for Database of Human Hemoglobin Variants and Thalassemias.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 21 mentions in open access literature.

Listed below are recent publications. The full list is available at T1D .

Zhang H, et al. (2025) Analysis of genotypic distribution and rare variants of patients with ?/?-thalassemia screened in one hospital in Beijing, China. Human genomics, 19(1), 13.

Sadoon TA, et al. (2025) Complete molecular spectrum of ?-globin gene mutations via direct sequencing identifies seven novel variants in ?-thalassemia major. PloS one, 20(11), e0336610.

Shi Q, et al. (2025) A novel targeted long-read sequencing-based preimplantation genetic testing method for ?-thalassemia (tlrPGT-?-thal). Journal of assisted reproduction and genetics, 42(8), 2565.

Huang J, et al. (2025) Catalog of 73 novel variants in thalassemia: discoveries and insights. Human molecular genetics, 34(24), 2067.

Xia Y, et al. (2024) A particular focus on the prevalence of ?-thalassemia and ?-thalassemia among pregnant women in Changsha County, Hunan Province. Frontiers in genetics, 15, 1422462.

Zhuang J, et al. (2024) Molecular characterization of similar Hb Lepore Boston-Washington in four Chinese families using third generation sequencing. Scientific reports, 14(1), 9966.

Selvatici R, et al. (2024) Relevance of Next-Generation Sequencing in the Diagnosis of Thalassemia and Hemoglobinopathies: The Experience of Four Italian Diagnostic Hubs. Genes, 16(1).

Bao X, et al. (2023) Identification of four novel large deletions and complex variants in the ?-globin locus in Chinese population. Human genomics, 17(1), 38.

Long J, et al. (2022) Third-generation sequencing: A novel tool detects complex variants in the ?-thalassemia gene. Gene, 822, 146332.

Chauhan W, et al. (2021) A Novel Frameshift Mutation, Deletion of HBB:c.199_202delAAAG [Codon 66/67 (-AAAG)] in ?-Thalassemia Major Patients from

the Western Region of Uttar Pradesh, India. The application of clinical genetics, 14, 77.

Mahmud N, et al. (2021) Hemoglobin A2 and Heterogeneous Diagnostic Relevance Observed in Eight New Variants of the Delta Globin Gene. Genes, 12(11).

Zhao J, et al. (2020) Combined use of gap-PCR and next-generation sequencing improves thalassaemia carrier screening among premarital adults in China. Journal of clinical pathology, 73(8), 488.

Zhao S, et al. (2019) Pilot study of expanded carrier screening for 11 recessive diseases in China: results from 10,476 ethnically diverse couples. European journal of human genetics : EJHG, 27(2), 254.

Zhang H, et al. (2019) Next-generation sequencing improves molecular epidemiological characterization of thalassemia in Chenzhou Region, P.R. China. Journal of clinical laboratory analysis, 33(4), e22845.

Qadah T, et al. (2019) Computational Analysis of Protein Structure Changes as a Result of Nondeletion Insertion Mutations in Human ?-Globin Gene Suggests Possible Cause of ?-Thalassemia. BioMed research international, 2019, 9210841.

Zhang L, et al. (2019) LOVD-DASH: A comprehensive LOVD database coupled with diagnosis and an at-risk assessment system for hemoglobinopathies. Human mutation, 40(12), 2221.

Sürün D, et al. (2018) High Efficiency Gene Correction in Hematopoietic Cells by Donor-Template-Free CRISPR/Cas9 Genome Editing. Molecular therapy. Nucleic acids, 10, 1.

Warghade S, et al. (2018) Prevalence of hemoglobin variants and hemoglobinopathies using cation-exchange high-performance liquid chromatography in central reference laboratory of India: A report of 65779 cases. Journal of laboratory physicians, 10(1), 73.

Turner A, et al. (2016) Rapid detection of pathological mutations and deletions of the haemoglobin beta gene (HBB) by High Resolution Melting (HRM) analysis and Gene Ratio Analysis Copy Enumeration PCR (GRACE-PCR). BMC medical genetics, 17(1), 75.

Domingues-Hamdi E, et al. (2014) Role of ?-globin H helix in the building of tetrameric human hemoglobin: interaction with ?-hemoglobin stabilizing protein (AHSP) and heme molecule. PloS one, 9(11), e111395.