Resource Summary Report

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Turkish Human Mutation Database

RRID:SCR_008246
Type: Tool

Proper Citation

Turkish Human Mutation Database (RRID:SCR_008246)

Resource Information

URL: http://hmut-tr.sourceforge.net/

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Description: The Molecular Biology and Genetics Department at Bogazii University is one of the major reference laboratories in Turkey, specialized in molecular analysis of common genetic disorders. Over the years, the rapid accumulation of mutation data in connection with detailed clinical and laboratory information, has led to the idea of establishing a national database for storing, analysing and presenting it in a more efficient and systematic way. For this purpose, an interdisciplinary project was initiated in 1995. b-Thalassemia and Hemophilia-B Databases were selected as preliminary models, for they offer alternative design and implementation strategies due to different clinical and genetic characteristics. b-Thalassemia is an autosomal recessive disorder, characterized by microcytosis and hemolytic anemia, which is the result of reduced b-Globin chain synthesis. In Turkey, the disease is represented with a gene frequency of 2 and reflected by a wide spectrum of clinical manifestations with the presence of more than 40 different mutation. Currently, there is no database available for thalassemia mutations. Hemophilia B is an X-linked recessive disorder caused by heterogenous mutations, resulting in a marked deficit of coagulation factor IX (FIX); an essential component of the clotting mechanism. A hemophilia B database was first published in 1990 as a list of point mutations and short additions and deletions with 115 mutations comprising 216 entries Gene-, System-, or Disease- Specific Databases

Synonyms: Human Mutation Database

Resource Type: data or information resource, database

Keywords: gene-, genetic, anemia, autosomal, b-globin, biology, b-thalassemia, clinical, clotting, coagulation, disorder, hemolytic, hemophilia-b, heterogenous, laboratory, mechanism, microcytosis, model, molecular, mutation, or disease- specific databases, synthesis, system-

Resource Name: Turkish Human Mutation Database

Resource ID: SCR_008246

Alternate IDs: nif-0000-21404

Old URLs: http://www.medinfo.hacettepe.edu.tr/hmuttr/

Record Creation Time: 20220129T080246+0000

Record Last Update: 20260912T200159+0000

Ratings and Alerts

No rating or validation information has been found for Turkish Human Mutation Database.

No alerts have been found for Turkish Human Mutation Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We have not found any literature mentions for this resource.