Resource Summary Report

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FH HUS Mutation Database

RRID:SCR_008512
Type: Tool

Proper Citation

FH HUS Mutation Database (RRID:SCR_008512)

Resource Information

URL: http://www.FH-HUS.org

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Description: The database has now been updated to include ALL mutations found in HUS patients, including those in Factor I(FI) and Membrane (MCP). Homology models are available for the domains of FI and MCP and all analysis previously available for Factor H (FH) are now also available for FI and MCP. All SNP records for FH, FI and MCP are also now included in the database on the SNP pages. Only those SNPs within coding regions will be included in the full list of mutations and within the advanced search. For more information on the different versions of the database click here. We have also redesigned the site in order to display information more clearly. Please let us know what you think of the new design. Home Information Mutations Models References Links Submit Contact Us Help Collaborators NEWS !! SEP 2009 The database has now been recovered. Please report any bugs that you notice. NEWS !! MAY 2009 We have suffered from a complete server failure this month but these issues have been sorted out and work is being carried out to restore all the data within our FH-HUS database. Sorry for any inconvenience this may have caused. NEWS !! JAN 2007 Mutations within complement Factor B have also been associated with aHUS. (Goicoechea de Jorge et al., 2007) NEW !! Nov 2006 FH-HUS Database Version 2.1 The database has now been updated to include ALL mutations found in HUS patients, including those in Factor I(FI) and Membrane (MCP). Homology models are available for the domains of FI and MCP and all analysis previously available for Factor H (FH) are now also available for FI and MCP. All SNP records for FH, FI and MCP are also now included in the database on the SNP pages. Only those SNPs within coding regions will be included in the full list of mutations and within the advanced search. For more information on the different versions of the database click here. We have also redesigned the site in order to display information more clearly. Please let us know what you think of the new design. Quick Search Enter Codon No : Choose Protein : Advanced Search Have you or someone you know been diagnosed with aHUS? The information contained on this web site is provided for scientific

research purposes only. We do not give medical advice or recommend any particular treatment for specific individuals. Here are several links for patient information on aHUS: http://renux.dmed.ed.ac.uk/ http://en.wikipedia.org/ http://kidney.niddk.nih.gov http://www.webmd.com HUS HUS (Haemolytic Uraemic Syndrome) is a disease associated with microangiopathic haemolytic anemia, thrombocytopenia and acute renal failure. A subgroup of the syndrome is strongly associated with abnormalities within the complement regulator factor H gene. To read information on HUS click here. To read information on Factor H (FH) click here. FH Mutations There are currently 74 Factor H mutations, 10 Factor I mutations and 25 MCP mutations linked with HUS patients within this database. There are also 5 mutations within FH that are associated with MPGN patients. . Following HGVS guidelines, mutations are numbered starting from the ATG initiation codon and include the 18-residue signal peptide. The number of the codon with respect to the mature FH protein and consistent with the RSCB PDB entry for secreted FH (1haq.pdb) is shown alongside in parenthesis. Type I and Type II Phenotype Type I indicates that the mutant protein is either absent from the plasma or present in lower amounts. This indicates the mutation has a structural effect on the mutant protein - ie reducing the stability Type II indicates that the mutant protein is present in normal amounts in plasma. This indicates that the mutation has a functional effect on the protein ie affecting substrate binding References There are three references you can use to reference this database Saunders et al, 2007. The interactive Factor H-atypical hemolytic uremic syndrome mutation database and website: update and integration of membrane cofactor protein and Factor I mutations with structural models. Hum Mutat. 2007 28:222-234. Saunders et al, 2006. An interactive web database of factor H-associated hemolytic uremic syndrome mutations: insights into the structural consequences of disease-associated mutations. Hum Mutat. 2006 27:21-30. Saunders & Perkins, 2006. A user''s guide to the interactive Web database of factor H-associated hemolytic uremic syndrome. Semin Thromb Hemost. 2006 32:160-8. Abstract. BACKGROUND: cblC disease is a cause of hemolytic uremic syndrome (HUS), which has been primarily described in neonates and infants with severe renal and neurological lesions. PATIENTS: Two sisters aged 6 and 8.5 years presented with a latent hemolytic process characterized by undetectable or low plasma haptoglobin, respectively, associated with renal failure and gross proteinuria. Renal biopsies performed in both patients found typical findings of thrombotic microangiopathy suggesting the diagnosis of HUS. Both patients were free of neurologic signs. RESULTS: Biochemical investigations found a cobalamin processing deficiency of the cblC type. Search for additional factors susceptible to worsen endothelial damage revealed homozygosity 677C--> T mutation in the methylenetetrahydrofolate reductase gene as well as heterozygosity for a 3254T--> C mutation in factor H in the patient with the most severe clinical presentation. Long-term subcutaneous administration of hydroxocobalamin in combination with oral betaine and folic acid resulted in clinical and biological improvement in both patients. CONCLUSION: cblC disease may be a cause of chronic HUS with delayed onset in childhood. Superimposed mutation of factor H gene might influence clinical severity.

Synonyms: FH HUS Mutation Database

Resource Type: database, data or information resource

Funding:

Resource Name: FH HUS Mutation Database

Resource ID: SCR_008512

Alternate IDs: nif-0000-30558

Record Creation Time: 20220129T080247+0000

Record Last Update: 20250517T055900+0000

Ratings and Alerts

No rating or validation information has been found for FH HUS Mutation Database.

No alerts have been found for FH HUS Mutation Database.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We found 24 mentions in open access literature.

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

Santoro D, et al. (2020) Identification of a New Complement Factor H Mutation in a Patient With Pregnancy-Related Acute Kidney Injury. Kidney international reports, 5(9), 1603.

Urban A, et al. (2020) Gain-of-function mutation in complement C2 protein identified in a patient with aHUS. The Journal of allergy and clinical immunology, 146(4), 916.

Geerlings MJ, et al. (2018) Genotype-phenotype correlations of low-frequency variants in the complement system in renal disease and age-related macular degeneration. Clinical genetics, 94(3-4), 330.

Gaut JP, et al. (2017) Routine use of clinical exome-based next-generation sequencing for evaluation of patients with thrombotic microangiopathies. Modern pathology : an official journal of the United States and Canadian Academy of Pathology, Inc, 30(12), 1739.

Besbas N, et al. (2017) Turkish pediatric atypical hemolytic uremic syndrome registry: initial analysis of 146 patients. BMC nephrology, 18(1), 6.

Ding Y, et al. (2017) A haplotype in CFH family genes confers high risk of rare glomerular nephropathies. Scientific reports, 7(1), 6004.

Noris M, et al. (2015) Glomerular Diseases Dependent on Complement Activation, Including Atypical Hemolytic Uremic Syndrome, Membranoproliferative Glomerulonephritis, and C3 Glomerulopathy: Core Curriculum 2015. American journal of kidney diseases : the official journal of the National Kidney Foundation, 66(2), 359.

Rodriguez E, et al. (2014) New functional and structural insights from updated mutational databases for complement factor H, Factor I, membrane cofactor protein and C3. Bioscience reports, 34(5).

Posnik B, et al. (2013) Acute Progression of Adult-Onset Atypical Hemolytic-Uremic Syndrome due to CFH Mutation: A Case Report. Case reports in nephrology, 2013, 739820.

Kopp A, et al. (2012) Factor h: a complement regulator in health and disease, and a mediator of cellular interactions. Biomolecules, 2(1), 46.

Roumenina LT, et al. (2011) Alternative complement pathway assessment in patients with atypical HUS. Journal of immunological methods, 365(1-2), 8.

Johnson SA, et al. (2010) Impact of compound heterozygous complement factor H mutations on development of atypical hemolytic uremic syndrome-A pedigree revisited. Molecular immunology, 47(7-8), 1585.

Küntzer J, et al. (2010) Human variation databases. Database : the journal of biological databases and curation, 2010, baq015.

Okemefuna AI, et al. (2009) Multimeric interactions between complement factor H and its C3d ligand provide new insight on complement regulation. Journal of molecular biology, 391(1), 119.

Okemefuna AI, et al. (2009) Electrostatic interactions contribute to the folded-back conformation of wild type human factor H. Journal of molecular biology, 391(1), 98.

Boon CJ, et al. (2009) The spectrum of phenotypes caused by variants in the CFH gene. Molecular immunology, 46(8-9), 1573.

Botto M, et al. (2009) Complement in human diseases: Lessons from complement deficiencies. Molecular immunology, 46(14), 2774.

Boon CJ, et al. (2008) Basal laminar drusen caused by compound heterozygous variants in the CFH gene. American journal of human genetics, 82(2), 516.

Ponce-Castro IM, et al. (2008) Molecular characterization of Complement Factor I deficiency in two Spanish families. Molecular immunology, 45(10), 2764.

Jokiranta TS, et al. (2007) Where next with atypical hemolytic uremic syndrome? Molecular immunology, 44(16), 3889.