

Abstract

findis.org – Finnish Disease Database

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Limited number of founders and the subsequent isolation of the population, has resulted in the enrichments of some rare disease alleles in Finland. Some 35, mostly recessive diseases are more common in this population, although are reported also from other global populations. This group of “Finnish diseases” often referred as the Finnish disease heritage, was first introduced by Norio et al. in 1973.

findis.org is a newly established database of 35 monogenic diseases belonging to the Finnish disease heritage. For each disease the database provides a short description of clinical symptoms, prevalence and incidence of the disease as well as genetic locus and molecular description for identified mutations. The regional distribution within Finland is also provided. The character and consequences of all known mutations, Finnish and foreign, are described at the DNA and polypeptide level. Disease allele frequencies are typically reported for Finnish mutations. Precise mutations are presently known for 29 of these disorders and the well defined locus assignment exists for 5, making the Finnish disease heritage one among the best characterized in the world.

The database provides links to OMIM, EDDNAL, and Ensembl. If the disease belongs to a group of diseases with already existing specific database, findis.org refers to this database for the non-Finnish mutations. References to the original publications are included, and when available, links to original html- and pdf-format publication files are provided.

The technical structure of findis.org has been developed using a web application oriented programming language PHP for the application logic and PostgreSQL as the relational database backend. This allows for a very portable and scalable environment. A custom, windows GUI application, the Disease Editor, has been implemented in Borland Delphi, which allows updates of the database to be reflected online instantly.

The information for the database has been gathered from researchers, original and review publications and reliable websites. The database follows the Quality Criteria for Health Related Websites recommended by the European Commission.

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URLs: www.findis.org