

Abstract

How to curate 100 LSDBs?

Mauno Vihinen, Institute of Medical Technology, FIN-33014 University of Tampere, Finland

We have been working for almost ten years on information systems related to immunodeficiencies. We started by establishing mutation database for X-linked agammaglobulinemia named after the affected gene as BTKbase. The first public release was made on 1995. Since that a large number of immunodeficiency related genes and mutations have been identified. To make this information available for the physicians and research community we have established this far 60+ mutation databases.

Our registries are patient based i.e. there is one entry per patient. This choice was made in the beginning to facilitate also retrospective and prospective analyses or patient related, clinical and other information.

For the maintenance and distribution of the databases we have developed MUTbase program. A key issue for parallel curation of a large number of databases is automation. We start new databases by establishing an electronic submission form. After that many functionalities are automated.

Information to the databases comes either from literature or as direct submissions. We are in grateful for the immunodeficiency community for active submission. Active researchers in the field have greatly contributed to the management and contents of the databases.

Public web distribution versions are automatically generated. The databases are available also for mobile devices with WAP protocol.

Mutation databases as such are very useful for many people. We have expanded our scope to providing also other information for all parties interested in these disorders. Therefore, we have established also ImmunoDeficiency Resource (IDR) knowledgebase and IDdiagnostics, registry of laboratories performing both clinical and genetic tests.

We are looking forward to establishing new mutation databases for each immunodeficiency as genes and mutations are identified. There are more than 100 immunodeficiencies...