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Finding mutations in your favorite gene

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Abstract

The Universal Mutation Database (UMD) was developed to allow researchers interested in particular human disease genes to query gene-specific databases for mutational information in a dynamic and interactive fashion.

Content

The Universal Mutation Database (UMD) was developed to allow researchers interested in particular human disease genes to query gene-specific databases for mutational information in a dynamic and interactive fashion. The value of this site lies in allowing the user to query specific mutations and to obtain a wealth of data about mutational hotspots, comparisons between subsets of data, mutation distributions, CpG islands, and base and amino-acid modifications. Rather than providing a basic locus-specific repository, the site is generated with generic software that allows the construction of locus-specific databases with an optimized search structureand the input of secure clinical data. Eight genes (for example, *p53*, *VHL* and *MEN1*) have currently been adapted for use with this software and are represented in the core of the website; a ninth gene has been adapted but is found in a different section of the website. The number of mutations for each gene depends on the particular gene-specific database queried, and varies between 42 and 11,106. The site also contains links to numerous other databases.

Navigation

The website is well laid out, with one entry point directing the user to the UMD software necessary to access and query each database. Once you have accessed the software, you are presented with entry points to query each gene-specific database. After selecting the gene database of interest, there are direct access points for specific issues including 'CpG, amino-acid modification, deletions analysis,' and so on and it even has an 'I found a mutation' entry point, with which to determine the frequency of a given mutation in the database and display a table of the various mutational events for this position. Each database also includes an exhaustive search engine that allows the user to select up to 20 different search criteria, including clinical data. The search engine allows Boolean search parameters.

Some users may encounter access difficulties with firewalls. This can usually be resolved by contacting both UMD and your own administrator to check the necessary settings. (The UMD main

administrator, Christophe Beroud, once contacted did respond in a reasonably prompt fashion and was as helpful as possible. As it turned out, the problems I encountered were caused by my server and not UMD's.) Another problem with the software is that the 'back' button of your browser is incompatible with the software and should not be used once a search has been initiated.

Reporter's comments

Best feature

The site makes a concerted effort to provide the user with a clear and easy way to query a gene-specific mutation database in an exhaustive fashion. The capacity for extensive mutation searches should allow researchers to focus on the particular question they wish to address. The database also acts as a depository that researchers can query to see if another researcher has already screened or found a particular mutation in a particular cell line or tissue.

Worst feature

The free-search engine is extremely powerful if you know how to use it. It took me some time to figure out how to get from the basic initial query to a final result that was accessible. Such difficulties could easily be resolved if the website maintained a help file or a FAQ section, to help the general user through a basic analysis. Also, the user is not told how often the database is updated, only how many times it has been accessed. While that is nice to know, most users would prefer to be told whether or not the site is up to date.

Table of links

References

1. Universal Mutation Database.