Mutation View: A Distributed Database for Human Disease Gene Mutations

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More than 4,200 human diseases including hereditary disorders and cancers have been reported to be caused by gene mutation. The genes responsible for these diseases are being investigated as one of the most important aspects of the human genome project. To date, approximately 1,100 diseases have been mapped to particular chromosomal regions and their responsible genes have been identified for over 500 diseases. The mutation data provide invaluable informations on the function of genes, and are indispensable for clinical medicine such as developing DNA diagnosis and gene therapy. At present, human gene mutation data are being collected by individual researchers for their interested disease using various formats and softwares. Some of them are open to public via internet/WWW, but they are still character-based, not graphical and not so friendly to the general users. Thus, there have been a great demand to create a new database system. The ideal system should have a unified user interface, common data format and visual data display. It should be integrated with the other related databases such as GDB, OMIM and GenBank/EMBL/DDBJ. For this purpose, we have developed a multi-server/client database system Mutation View. Mutation View allows us to systematically integrate various informations including genetic, molecular biological and clinical findings of each disease. The features of Mutation View are as follows.

(1) Chromosome ideograms are drawn to list diseases in the mapped regions. OMIM documents can be cited for each disease. (2) Human body and organs/tissues are schematically shown, on which disease names and the symbols of responsible genes are listed. (3) For the gene selected by either (1) or (2), primary structure or its cDNA is shown on X-axis and various mutations are located on appropriate positions (Fig. 1). The detailed information of each mutation is displayed by clicking the mutation symbol. Zooming-in is possible and the nucleotide sequence of the gene can be displayed at high magnification (Fig. 2). Frequency or case-number of each mutation can be shown as a histogram on Y-axis. Other information such as classified mutation types, symptoms, onset age, and hereditary pattern can be obtained on the Y-axis display with a switch. (4) Input and editing of new mutations are easily manipulated and new data can be shown together with the previous data (Fig. 2). (5) The exact mutation is shown with reference of normal nucleotide sequence. Change of restriction sites on the mutation position can be automatically analyzed and displayed (Fig. 1 and 2). (6) Since this Mutation View was designed as a distributed database, any laboratory can act as a server site for a particular disease and the users can access to any sites via internet.

Current version of Mutation View offers data on eye diseases1 such as retinitis pigmentosa and glaucoma, cystic fibrosis2, tumor suppressor gene TP533 as model cases. Prototype Mutation View at present works only on a UNIX workstation, however a WWW server using JAVA is under development.

(This work has been carried out in collaboration with Chi Co., Ltd.)

References

[1] Data curated from literatures in our laboratory.

[2] L.-C. Tsui. Cystic Fibrosis Mutation Data Base, http://www.genet.sickkids.on.ca/cftr/

[3] D. Liao. P53/APC Database, http://www.mayo.edu/research/papers/P53%20Mutations/