Mutation View : A Network-Transparent Database System for Mutations in the Human Disease Genes

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More than 4,300 human diseases caused by gene mutation have been reported. The responsible genes for these diseases including hereditary disorders and cancers 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. We have developed a computer software Mutation View to systematically integrate genetic, molecular biological and clinical findings of each disease and to assist molecular diagnosis and gene therapy. 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) Primary structure of the gene or its cDNA is shown on X-axis schematically and various mutations are located on appropriate positions with symbols (Fig. 1). The detailed information of each mutation is displayed by clicking of the mutation symbol. Zooming-in is possible in wide ranges and the nucleotide sequence of the gene can be displayed in high magnification (Fig. 2). Relative frequency or case-number of each mutation can be shown as a histogram (Fig. 1) and classifications of mutation types or symptoms are available. One of these information is chosen with a switch and is displayed on Y-axis. (3) Input and editing of new mutations are easily manipulated and new data can be shown together with previously existing data (Fig. 2). (4) The exact mutation is shown with reference of normal nucleotide sequence. Change of restriction sites on the mutation position can be automatically analyzed even for newly added data (Fig. 2). (5) Since this Mutation View system has been designed as a distributed database, any laboratory can take a role as a server site for a particular disease and the users can access to any disease information via internet. A prototype of Mutation View using a cystic fibrosis database compiled by Dr. Lap-Chee Tsui will be demonstrated using a workstation. (This work was done in collaboration with Chi Co., Ltd. and NEC Corporation.)
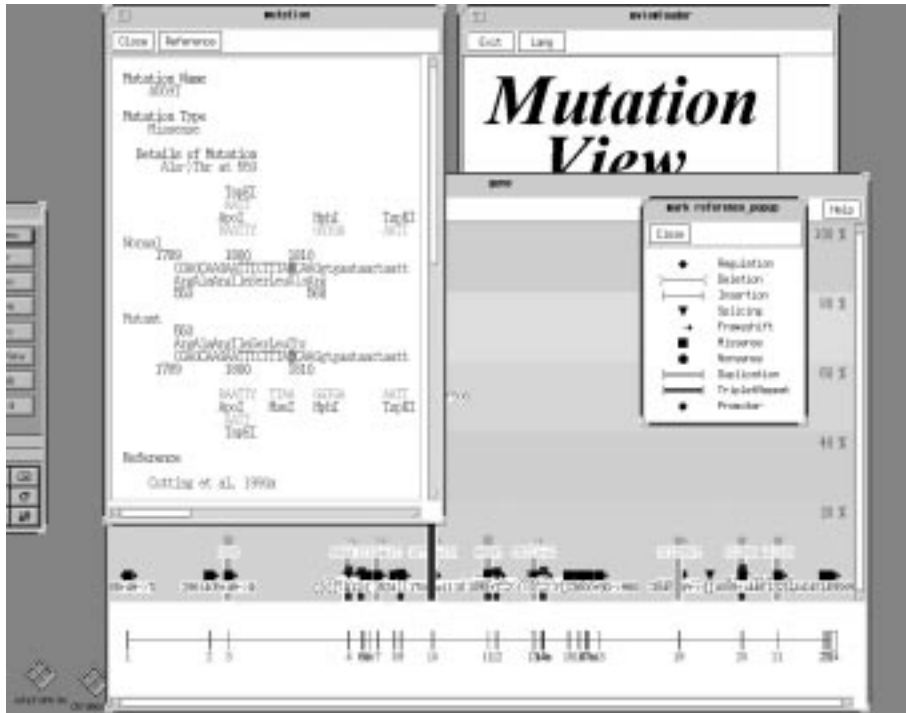
Figure 1:

Figure 2: