

KMDB : Keio Mutation Database for Human Disease Gene

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1 Introduction

The mutation data associated with human gene diseases are important not only for basic study of gene functions but also for clinical medicine including DNA diagnosis and gene therapy. We have been collecting mutation data of various diseases. Here, we report KMDB, a mutation database for human disease genes.

2 KMDBs Constructed on *MutationView*

KMDB has been constructed on a database software *MutationView* [1] which was designed to compile various mutation data as a distributed database and to provide graphical presentation of data analysis. To date, we constructed five separate KMDBs for eye, heart, ear, brain and cancer in which a total of 1593 entries of mutations are found for 38 different genes which are involved in 35 distinct diseases collected from 311 literatures. They were designated KMeyeDB [2], KMheartDB, KMearDB, KMbrainDB and KMcancerDB, respectively, and integrated as components of *MutationView*.

3 The Feature of KMDBs and *MutationView*

The KMDBs as well as the *MutationView* have the following features: (i) The KMDBs have a common entrance window (Fig. 1A). (ii) The KMDBs have two different versions, JAVA and HTML versions for viewing data (Fig. 1B). The JAVA version provides dynamic and interactive user interface which can be used through the recent browser software with JAVA 1.1. The JAVA version has an Anatomy menu (Fig. 1C-F) and Chromosome menu (not shown). (iii) For the gene (AIRE in Fig. 2 for example) selected through either menu, the gene structure is graphically shown and various mutations are located on exact positions in the Gene Structure Window (Fig. 2A, C and D). This window has various functions as follows. By clicking "About this gene" button, a window appears (Fig. 2B), showing further information from other databases such as OMIM, GDB and HGMD. By clicking "Zoom-in" button, the X-axis can be changed from the genomic structure to even the nucleotide sequence (Fig. 2D). In the default, the height of each bar represents the case number of mutation which appeared in the literature (Fig. 1A). A variety of information (including mutation frequency, mutation type, symptoms, ethnic origin, hereditary pattern, onset age) can be shown on the Y-axis using "Classify" menu. Fig. 2C shows a re-classified view by "Ethnic origin". Clicking the mutation symbol (Arrow in Fig. 2D for example) opens a new window for the details of mutation (Fig. 2E), including the change in nucleotide sequence and its consequences such as alterations of amino acid, splicing signal and restriction site. Probe information such as PCR primers can be displayed by switching "View type"

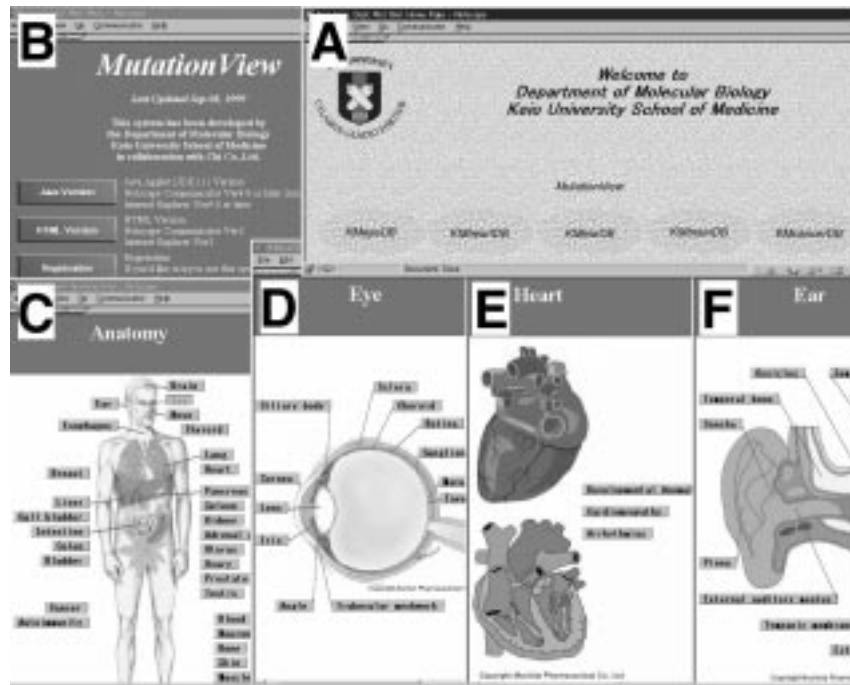


Figure 1: The Start-up menu and Anatomy windows of KMDBs. See text for details.

menu (Fig. 2C). Clicking the “PCR primer name” (indicated by an arrow) creates a new window, displaying the detailed information such as primer sequences and reaction conditions (Fig. 2F). The *MutationView* and KMDBs are accessible *via* <http://mutview.dmb.med.keio.ac.jp>. The user ID and password are issued after receiving applications through the same URL. The software *MutationView* is now available to all the interested research groups under the conditions that users participate actively in the establishment of a world-wide distributed database system for disease gene mutations.

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References

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