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The KMDB/*MutationView*: a mutation database for human disease genes

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ABSTRACT

The KMDB/*MutationView* is a graphical database of mutations in human disease-causing genes and its current version consists of nine category-based sub-databases including diseases of eye, heart, ear, brain, cancer, syndrome, autoimmunity, muscle and blood. The KMDB/*MutationView* stores mutation data of 97 genes involved in 87 different disease and is accessible through http://mutview.dmb.med.keio.ac.jp.

INTRODUCTION

We previously developed the KMDB for mutation data in human disease-causing genes using a database software called *MutationView*, which was designed to serve as a distributed database system (1). The previous KMDB contained six category-based sub-databases such as KMeyeDB, KMheartDB, KMbrainDB, KMearDB, KMaiDB and KMcancerDB. Here, we report a more advanced version of KMDB (v. 1.2), which now includes three additional sub-databases such as KMsyndromeDB, KMmuscleDB and KMbloodDB with a substantial increase in genes and mutations.

DATA CONTENT

The KMDB/*MutationView* has collected 3092 mutation entries from 606 literature sources, dealing with 97 genes involved in 87 distinct diseases (Fig. 1, top left; Table 1). The KMsyndromeDB deals with the syndromes such as Waardenburg syndrome, which is caused by single gene mutations. The KMaiDB collects mutations in the hereditary autoimmune diseases such as APECED (autoimmune polyendocrinopathy-candidiasis-ectodermal dystrophy). The KMmuscleDB deals with various neuromuscular diseases such as Duchenne muscular dystrophy and the KMbloodDB is designed for the genetic diseases of blood cells such as chronic myeloid leukemia and the genetic deficiencies in serum components such as albumin.

Figure 1 shows the entrance window of KMDB/*MutationView*. Clicking a part of the body will show up a list of the associated diseases and genes (not shown). Choosing a genetic disease will bring up its mutation data displayed in the 'gene structure window'. Figure 1 (top right) shows the gene structure of PARKIN which is a pathogenic gene for ARJP (autosomal recessive juvenile parkinsonism) (2). The gene structure can easily be switched to the nucleotide sequence and/or amino acid sequence. The frequency of each mutation type is shown as a histogram along with the genomic exon/intron strucutre. Details for each mutation can be seen by clicking the appropriate symbol listed in the 'Help' menu. Figure 1 (bottom right) shows the mutation detail for a large deletion mutation DelEx3-4 in the PARKIN gene.

DISTRIBUTION OF THE DATABASE

The software *MutationView* was designed to manage and coordinate multiple category-based sub-databases located at different web sites. Currently, the coordinating server for KMDB/*MutationView* is located at Keio University, Tokyo, while a category-based KMcancerDB dealing with E-cadherin and β-catenin mutations in cancer is located at Setsunan University, Osaka. Users can access all the data in KMDB/*MutationView* through Keio University.

ACCESSIBILITY AND AVAILABILITY

The KMDB/*MutationView* employs Java1.1 interpreter for entire function and hence most Internet browsing softwares can be used except Netscape on a Macintosh (Apple computer, Inc.). The coordinating server of KMDB/*MutationView* is located at Keio University School of Medicine (URL: http://mutview.dmb.med.keio.ac.jp). The user ID and password are issued upon formal application through the above URL. The software *MutationView* is made available to any research groups that are interested in establishing a world-wide distributed database for disease gene mutations. For inquiries, contact the first author (mino@dmb.med.keio.ac.jp).

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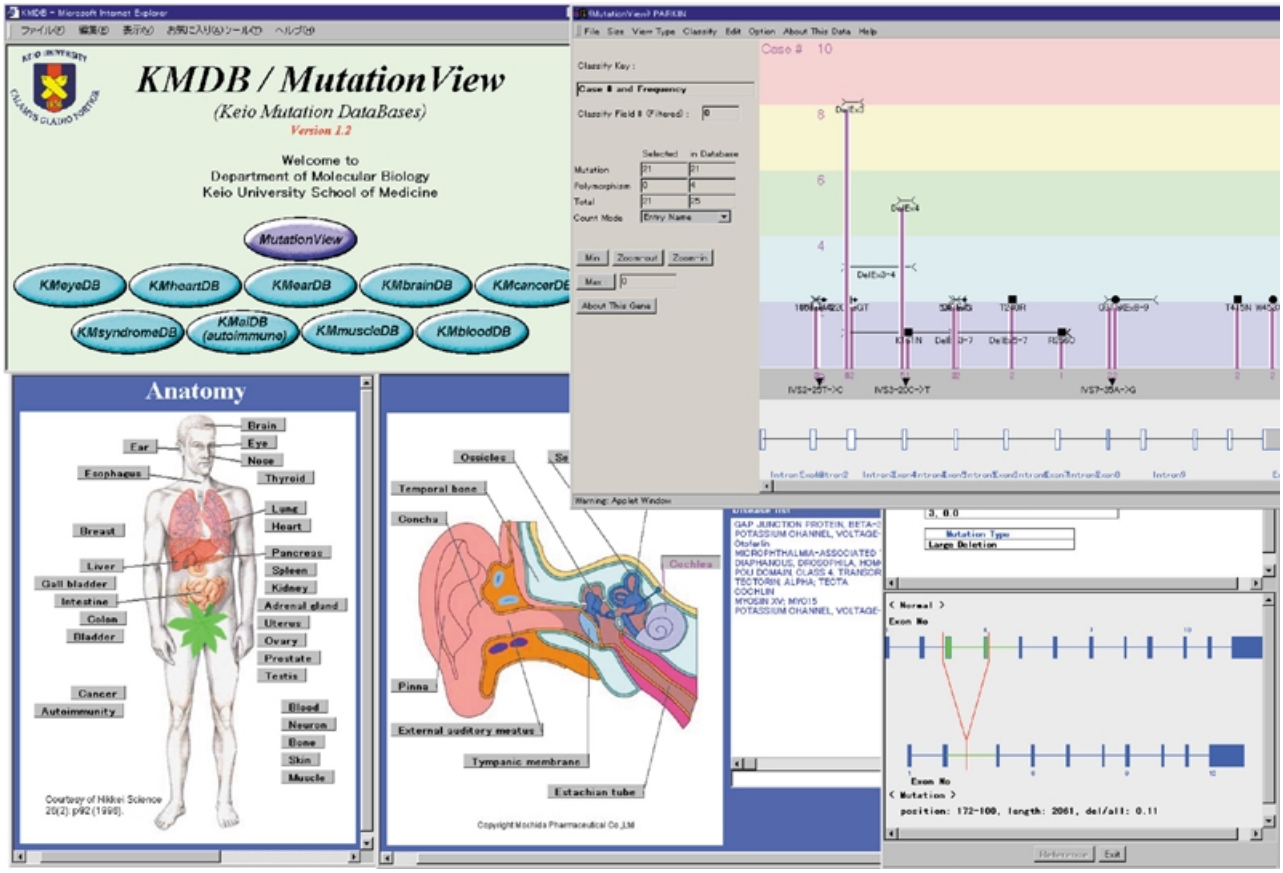

Figure 1. Features of the KMDB/*MutationView*. (**Top left**) Entrance window. (**Bottom left**) Anatomy window of *MutationView*. (**Bottom middle**) Anatomy window of KMearDB. (**Top right**) Gene structure window of KMbrainDB showing PARKIN gene with a histogram of mutation frequencies. (**Bottom right**) Mutation details window showing DelEx3-4 mutation in the PARKIN gene. See text for details.

Table 1. Mutation data in the KMDB/*MutationView*

Category	Number of diseases	Number of genes	Number of mutation entries	Number of literature references compiled
Eye	24	23[a]	385	112
Heart	7	7	79	19
Ear	3	11[a]	49	21
Brain/nerve	10	11[a]	227	77
Cancer-related	8	8	1303	164
Syndrome	15	17[a]	855	159
Autoimmunity	2	2	96	13
Muscle	3	3	42	15
Blood	4	4	13	7
Miscellaneous	11	11	43	19
Total	87	97	3092	606

[a]The number of diseases does not necessarily match with the number of genes because it has been known that some diseases with different clinical symptoms are caused by different mutation types in the same gene and other diseases with a common clinical phenotype are caused by multiple different genes involved in a certain physiological pathway.

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