

KMDB / MutationView : An Integrated Knowledge Base for Mutations and Polymorphisms in Human Disease Genes – Data Expansion and Further Functional Development –

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1 Introduction

More than 1000 disease-causing genes have been reported. Databases for mutations in these disease-causing genes are indispensable for the diagnostics, therapeutics and basic research of the diseases. At present, mutation data are maintained as individual Locus-specific Databases (LSDBs) for some 200 diseases and therefore more comprehensive database system with common graphical user-interface is required. We previously developed the *KMDBs*, an integrated graphical knowledge base for mutations in human disease genes using a database system *MutationView* which was designed to serve as a distributed database system [2, 3]. Current *KMDBs* consist of 9 sub-databases including *KMeyeDB*, *KMheartDB*, *KMearDB*, *KMbrainDB*, *KMcancerDB*, *KMsyndromeDB*, *KMautoimmuneDB*, *KMmuscleDB* and *KMbloodDB*. Here, we report an extensive data addition to *KMDBs* and various new improvement of *MutationView* system.

2 Data Contents

KMDB/MutationView has now collected 6383 (3092) entries of mutations from 1405 (606) literatures, dealing with 192 (97) genes involved in 183 (87) distinct diseases (the numbers in parentheses are of September 2000). Numbers of genes and typical diseases are shown in the following table.

3 Characteristics of *MutationView*

(i) Several ways are available to access to the gene of interest such as through the chromosomal map of the gene or disease, anatomical chart of disease-associated organ or tissue and diagram of

KMDB	# Gene	Typical Diseases
eye	44	Glaucoma, Cataract, Corneal dystrophy
heart	16	Arrhythmia, Cardiomyopathy, Dysmorphology
ear	32	Deafness
brain	30	Parkinson Disease, Alzheimer disease
cancer	15	Breast, Retinoblastoma
syndrome	37	Waardenburg, Long QT, Cardiomyopathy
autoimmune	2	APECED
muscle	27	Duchenne, Limb-Girdle, Charcot-Marie-Tooth
blood	31	CML, Citorulinemia

causative gene product. (ii) Various functions for data analysis and display are available such as genomic/cDNA structure of gene, functional domain of protein, zooming in/out of the nucleotide/amino acid sequences, histogram of mutations, case number, changes in the nucleotide sequence and restriction sites, classification based on mode of inheritance and clinical symptom, practical information such as PCR primers and reaction conditions. (iii) *MutationView* system can be linked to global LSDBs while maintaining their independency.

4 New Features of *MutationView*

(i) The Chromosome Overview window (Fig. 1A) shows all the genes in *KMDB/MutationView* along chromosome idiograms. (ii) Protein functional domain can be shown with the cDNA/Coding Region display mode. Fig. 1B shows the case of PARKIN gene [1]. (iii) Molecular action diagram can be used as an entrance to select a gene of interest. Fig. 1C shows the muscular dystrophy-related genes.

5 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 Macintosh. The coordinating server of *KMDB/MutationView* is located at Keio University School of Medicine [4]. The user ID and password are issued upon formal applications 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 Shinsei Minoshima (mino@dmb.med.keio.ac.jp).

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References

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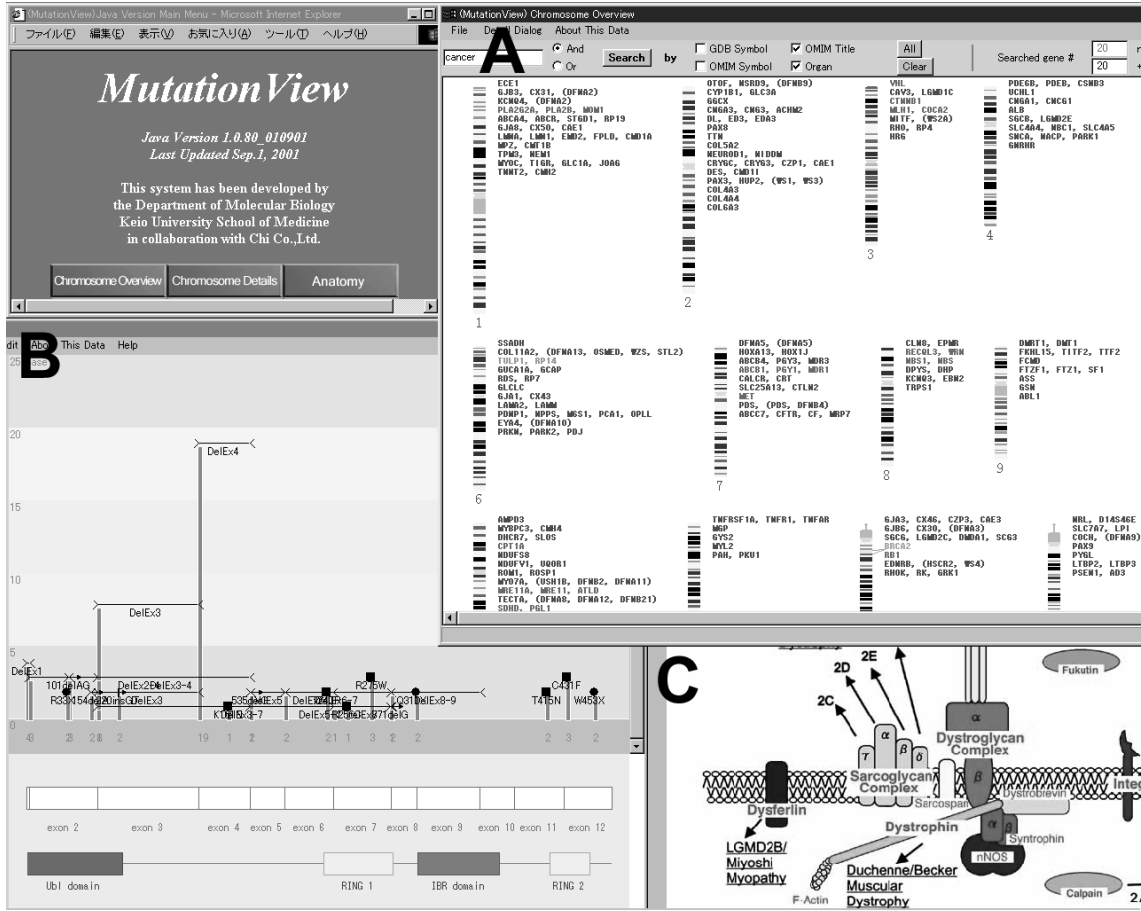


Figure 1: New features of *KMDB/MutationView*. See text for details.

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[4] <http://mutview.dmb.med.keio.ac.jp>