KMeyeDB: Keio Mutation Database for Eye Disease Genes Constructed on a Graphical Distributed Database System *MutationView*

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More than 4,300 human diseases have been identified to be inheritable. To date, ∼1,400 disease loci have been mapped to particular chromosomal regions and almost 800 disease-causing genes and the associated mutations have been identified. These mutation data are indispensable 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 KMeyeDB, a mutation database of human eye disease genes [1]. KMeyeDB has been constructed on a database software *MutationView* [2] which provides graphical data presentation and analysis as a smooth user-interface. At present, KMeyeDB covers mutation data of 16 different genes (RHO, RDS, PDE6A, PDE6B, ROM1, CNCG1, RP3, ABCR, RHOK, RB1, RS1, GSN, TGFBI, MYOC, CYP1B1, and CHM) for 18 eye diseases including retinitis pigmentosa, glaucoma, corneal dystrophy, choroideremia and others. The database software *MutationView* has the following features:

(i) Chromosome ideograms are displayed to list diseases in the mapped regions.

(ii) Human body is schematically shown to list diseases on the particular organ and/or tissues.

(iii) OMIM, GDB and HGMD information can be retrieved for each disease with hyperlink.

(iv) For the gene selected, the genomic and cDNA structures are graphically shown and various mutations are located on appropriate positions (Fig. 1D). Frequency and case number for each mutation are shown as a histogram. Other information such as mutation types, clinical symptoms, age of onset, or inheritance pattern is accompanied with individual mutation and can be used to classify the cases (Fig. 1A). PCR primer information to amplify various regions of the gene is also shown (Fig. 1B and D).

(v) Zoom-in allows to display the nucleotide sequence on which exact mutation is indicated and change of the restriction sites due to mutation is analyzed (Fig. 1C).

(vi) Entry of new mutation data is readily performed and analyzed with existing data, but modification of the data is permitted only to the qualified curator of each disease gene.

(vii) Since any data on WWW site with the same defined format can be made accessible, *MutationView* will be able to coordinate many existing locus-specific mutation databases as a distributed database.

The KMeyeDB is accessible *via* http://www.dmb.med.keio.ac.jp with user ID and password, which are issued after application through the same URL. The software *MutationView* is made available to the founders and qualified curators of locus-specific mutation databases on a collaborative basis to establish a world-wide distributed database system for disease gene mutations. The KMeyeDB and *MutationView* will be demonstrated at the meeting.

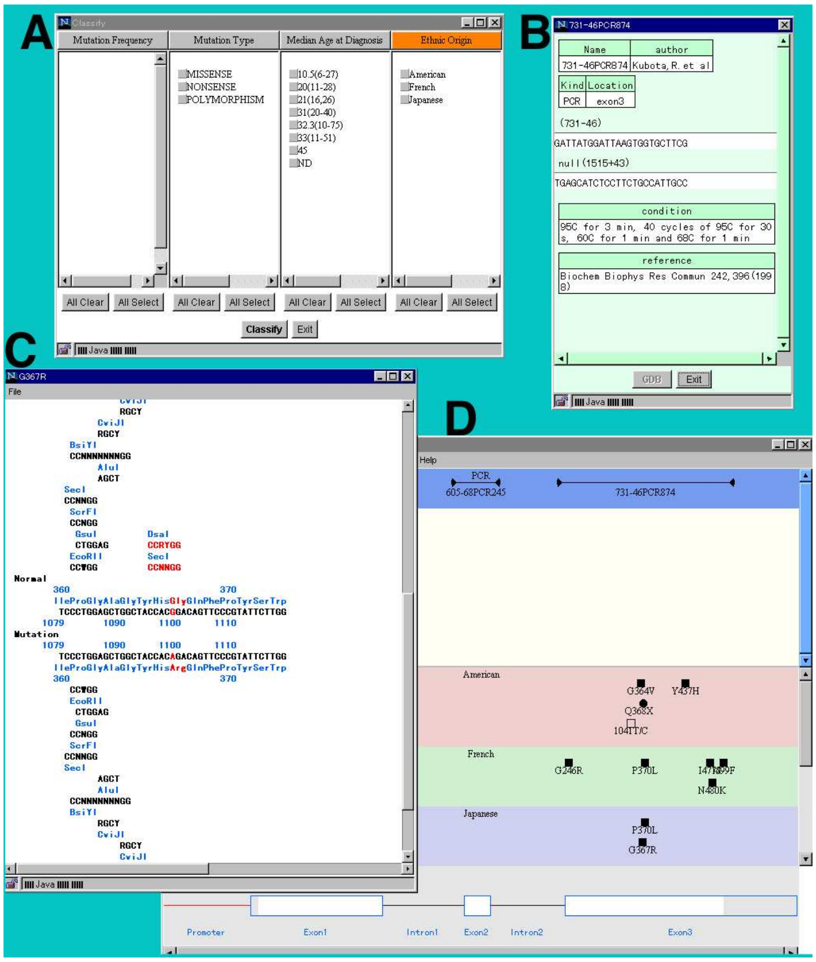

Figure 1: Various windows of KMeyeDB constructed on *MutationView*. Here, mutation data of the myocilin gene (MYOC) causing a type of glaucoma are shown. A. Classify window: One of 4 items, Mutation Frequency, Mutation Type, Median Age at Diagnosis and Ethnic Origin, can be used to classify data by clicking corresponding buttons. B. PCR detail window: Primer sequences and reaction condition are shown. C: Mutation detail window: Sequences of normal and mutation alleles are shown with restriction sites. D: Gene structure window: Classified mutations by Ethnic Origin are shown with PCR primers.

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References

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