New Features of *MutationView*:
A Module to Search for Disease-Causing Genes from Protein Functional Domain

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1 Introduction

In order to investigate the association between disease and genetic diversity, we have developed an integrated database system, *MutationView* ([http://mutview.dmb.med.keio.ac.jp/](http://mutview.dmb.med.keio.ac.jp/)). The *MutationView* principally focuses on mutations in the monogenic diseases and has various functions for real-time data analysis and display of the results with graphics. The characteristic features are as follows:

(i) Access to the gene or disease of interest is possible in several ways, such as through the chromosomal map of the gene or disease, anatomical chart of disease-associated organ or tissue and molecular diagram for the interaction of key gene products (proteins).

(ii) Various data are graphically displayed such as the genomic/cDNA structures of gene, functional domains of protein, nucleotide/amino acid sequences, histogram of mutation case number, changes of restriction sites, and various experimental conditions to detect mutations.

(iii) Various statistical analyses can be performed based on the mutation-associated patient’s information such as age, ethnic origin, disease-specific diagnostic results and symptoms.

2 Data Contents

To date, we have collected 18,293 entries of mutations from 2,585 literatures. These data deal with 324 genes and 590 diseases, which are sorted out according to the categories such as eye, brain, muscle, ear, heart, bone, autoimmunity and familial tumor (Table 1). The information includes not only molecular biological data but also patients data such as age, ethnic origin, disease-specific diagnostic results and symptoms.

3 A Module to Search from Protein Functional Domain

Information on the protein functional domain is invaluable when the effect of mutation is considered in relation to the protein function and disease symptom. The current version of *MutationView* is capable to store and display protein functional domain, however we developed a new module (Fig. 2 and 3) to carry out the following analysis: (a) to find the relationship between mutation hot spots of the gene and functional domains of protein; (b) to search for the genes that possess pathogenic mutation in the same functional domain of the protein product; (c) to retrieve all the disease-causing genes that produce proteins with the same functional domain. The domain information was obtained by *in silico* analysis using Pfam or by curator’s manual extraction from literatures. This new search function will be added to current *MutationView* system.

4 Data Presentation as html Table Format
**MutationView** has a sophisticated graphical data presentation system, however simple tables in the html format are often desired. Therefore, we developed a new function to create such tables from the built-in database (Fig. 1).

5 **Accessibility**

Users can access to MutationView without ID/password. However, this database is strictly for academic research purpose and not intended for commercial use. The softwares of MutationView are made available to LSDB curators in a spirit of cooperative database development.

6 **Acknowledgments**

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### Table 1: Contents of MutationView, 2006.

<table>
<thead>
<tr>
<th>KMDB</th>
<th>#Gene</th>
<th>Typical Diseases</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eye</td>
<td>130</td>
<td>Retinitis pigmentosa, Corneal dystrophy</td>
</tr>
<tr>
<td>Heart</td>
<td>37</td>
<td>Cardiomyopathy, Heart dysmorphism</td>
</tr>
<tr>
<td>Ear</td>
<td>61</td>
<td>Deafness</td>
</tr>
<tr>
<td>Brain/Neuron</td>
<td>74</td>
<td>Familial Parkinsonism, Alzheimer disease</td>
</tr>
<tr>
<td>Bone</td>
<td>45</td>
<td>Craniofacial Dysplasia</td>
</tr>
<tr>
<td>Cancer-related</td>
<td>43</td>
<td>Retinoblastoma, Neurofibromatosis</td>
</tr>
<tr>
<td>Muscle</td>
<td>35</td>
<td>DMD, MD Fukuyama type</td>
</tr>
<tr>
<td>Blood</td>
<td>46</td>
<td>CML, Citrullinemia</td>
</tr>
</tbody>
</table>

![Figure 1: Dynamically created html-format display](image1)

![Figure 2: Domain search function](image2)

![Figure 3: Gene structure window with predicted domain](image3)

**References**
