Biological Institute Medical Faculty
Charles University of Prague, Czech Republic

Department of Computer Science
Dresden University of Technology, Germany

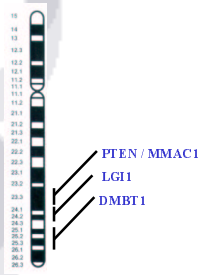
Mutation Database of Three Genes Located on Chromosome 10q (23-24)

Koufaki Olga-Niki, Hinze Thomas

Introduction:

In the last years the rapid expansion of human genetics particularly on the molecular level has enabled the effective and precise diagnosis of genetical disorders. The progress of the human genome project enables the identification and characterization of genes responsible for a plethora of deseases associated with alterations in the human genome. Particularly on chromosome 10 up to now 212 genes are identified. A large region of this chromosome is associated with brain malignant leasions. Genes like PTEN, LGI1, and DMBT1 exibits frequent alterations (mutations) in human glioblastomas.

Chromosome 10



Aims:

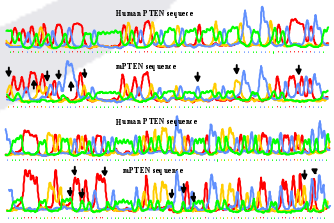
This project is focused on the easy and effective analysis of genomic alterations associated with brain tumour malignances. It is well known that three genes located on chromosome 10 are highly associated with this type of malignant leasions. It has been shown that the PTEN/MMAC1 gene is frequently mutated or partially deleted in glioblastoma multiforme. Since numerous mutations were up to now identified in this gene we have created a database that is able to identify mutations in the input sequence.

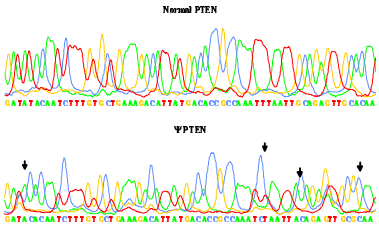
According to the last update (Sun Sep 19 23:22:39 EDT 199) of the genome database chromosome 10 contains 212 genes, 13 pseudogenes, and 3 putative genes in the total number of 7808 genes and 54 pseudogenes stored in the genetic database (GDB).

Functions

Automatical and manual identification of an input sequence

- **Automatical identification:** includes all possible genes available in the database
- **Manual identification:** uses a selected gene from the database
- **Handling Security:** only the reasonable features are available
- The input sequence accepts only characters encoding nucleotides





The **Oligo Navigator Kit (ONK)** is a gene analysis system that is flexible as far as conciders the language, the gene database, and the running platform. The database can be easily adapted to the language demands of the user. The usage of another running platform like OS/2, Linux, etc. makes the ONK system a flexible tool. Also the addition of any new gene and the ability of the system to identify and analyze the input sequence according to the new gene or according to any other gene in the database makes it a useful research tool.

System requirements

- **Hardware:**
  - Processor 80486 or higher
  - 2MB hard disk space
  - At least 16MB random access memory
  - Floating point unit NOT necessary
- **Operating system:**
  - Windows 95, Windows NT, Win32S

Technical Feedback

Program consists of three hierarchical components („Moduls“) with different tasks:

1. **Database**
   contains the „internal database“ by holding the gene sequences, the mutation information and the exon regions
2. **Application kernel**
   contains the search functions and the identification functions
   has access to the database by the database methods
3. **User surface**
   composed by control elements
   has access to the database and to the application kernel by prepared methods

Summary:

The Oligo Navigator Kit falls into the category of software information retrieval systems. Its functions and features lead to an application for medical diagnostics, biological research, bioinformatics, and DNA computing. Identifying, evaluating, and analysing sequences belongs to the daily tasks in these subjects. The database system offers a powerful extendable tool to transform the effort from the human to the computer. The software is conceived in a modul structure, divided into the main parts database, application kernel and user interface. This concept leads to a portability between different platforms, languages, and extensions of the database. The aim is to include all genes of chromosome 10 to have a complete collection with all derivable information about possible mutations and diseases in consequence.