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Full Length Research Paper

ATID: Analytical tool for inherited diseases

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The body of information surrounding molecular and genomic experiments and clinical investigations is rapidly growing. Bioinformatics tools are software programs that are designed for extracting the meaningful information from the mass of molecular biology/biological databases and carry out sequence or structural analysis. Analytical tool for inherited disease (ATID) is a classification database that can hold the complete available information about the inherited diseases. Our designed tool is a set of search programs for the windows platform and is used to perform fast searches for comparison of nucleotide sequences, mutation identification and single nucleotide polymorphism (SNP). Also protein sequences can be searched to find a match against the queried protein sequence. The information comprises their clinical phenotypes, causes and type of disease. Designed software has the ability to classify the disease based on the comparison of given affected individual information (phenotypic and genotypic) with existing information. We analyzed fake data as well as individual's original data on our tool by comparing that with the existing information. That was easy to use and also analyzed results within few seconds. The ATID windows application is designated to solve the problem of extracting the information about the inherited diseases in a fast and simple way. The designed tool can be modified for other diseases as well in future or a single database on the same pattern can be developed comprising of multiple diseases for phenotypic as well as molecular profiling.

Key words: Inherited disease, analytical tool for inherited diseases, bioinformatics, tools.

INTRODUCTION

Inherited genetic variation has a critical but as yet largely characterized role in human disease. Despite the ever-accelerating pace of biomedical research, the root causes of common human disease remain largely unknown, preventative measures are generally inadequate and available treatments are rarely remedial (The international HapMap consortium, 2005). According to the international HapMap consortium, more than a thousand genes for rare, highly heritable "mendalian" disorders have been identified, in which variation in a single gene is both necessary and sufficient to cause disease. Need of the day is to store and to access this huge data, for whom, bioinformatics or computational biology, interdisciplinary fields comprises.

The field of computational biology has seen dramatic growth over the past few years, both in terms of available data, scientific questions and challenges for learning and inference. The rapid pace of discovery in the genomic and proteomic arena requires parallel growth in the field of bioinformatics. Bioinformatics should built databases in a way that facilitates not just the storage of data, but efficient handling and retrieval of information from these databases. This is evident from the fact that modern biology and related sciences are increasingly becoming dependent on this new technology. It is expected that bioinformatics will especially contribute in the future as the leading edge in biomedicine to pharmaceutical companies by expediently yielding a greater quantity of lead drugs for therapy.

The enormous growth of biological data led to the development of several things. First, all these data need to be stored. The second requirement is the need for radical new methods for analyzing these huge databases. Thirdly, powerful hardware is required to carry out the task of analyzing these databases. Development of existing databases as well as the conceptualization and

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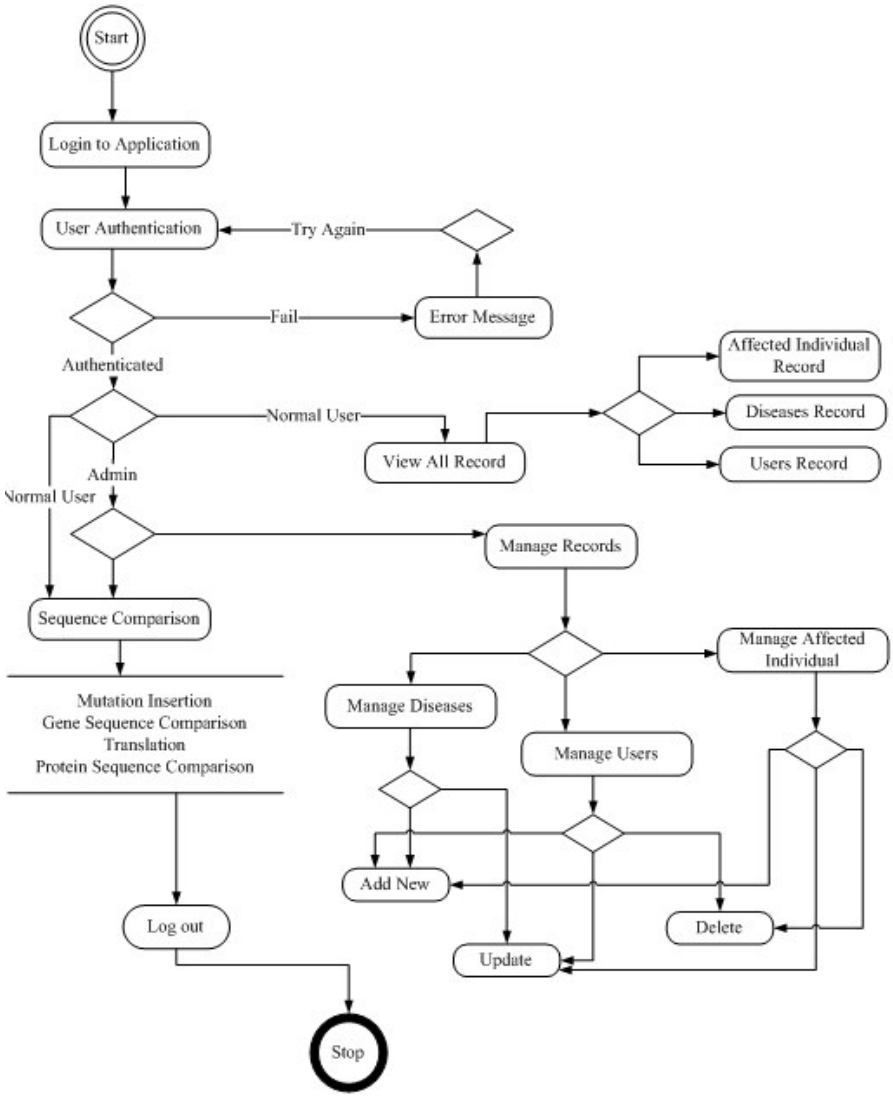

Figure 1. Activity diagram of analytical tool for inherited diseases (ATID).

creation of new types of databases will be critical focal point for the advancement of biological discovery. Analytical tool for inherited diseases (ATID) is a classification database that holds the complete available information about the inherited diseases.

The information comprises their clinical phenotypes, causes, underlying known genes, type of disease, mutation identification and protein comparison. Phenotypic as well as molecular profile of an affected individual can be compared to the existing information and disease can be classified.

MATERIALS AND METHODS

Programming language used for the software was JAVA using the API's of BioJava. And the database used was Microsoft access 2007 that uses the ODBC, JDBC drivers for connectivity.

RESULTS

Design is a meaningful representation of any product/software that is to be built and is the most artistic part of the software development process (Pressman, 2005). ATID design consists of mutation identification, gene sequence comparison and protein sequence comparison. Activity diagram of ATID is given in Figure 1, which describes the work flow behaviour of system and represents the execution state of a mechanism as a sequence of steps grouped sequentially as parallel control flow branches. Hence activity diagram make it easier to understand the design and internal behaviour of ATID. Major modules are gene information, sequence comparisons either gene or protein and affected individual records (Figures 2, 3 and 4). Diseases information can also be added or updated with respect to

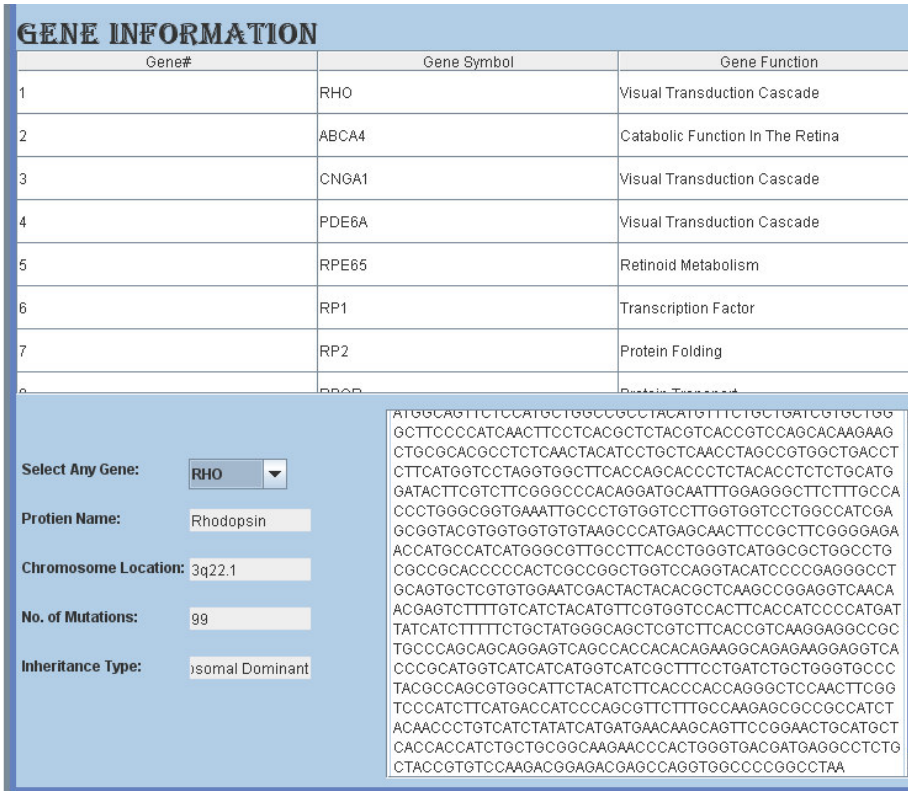

Figure 2. Gene selection for detail Information.

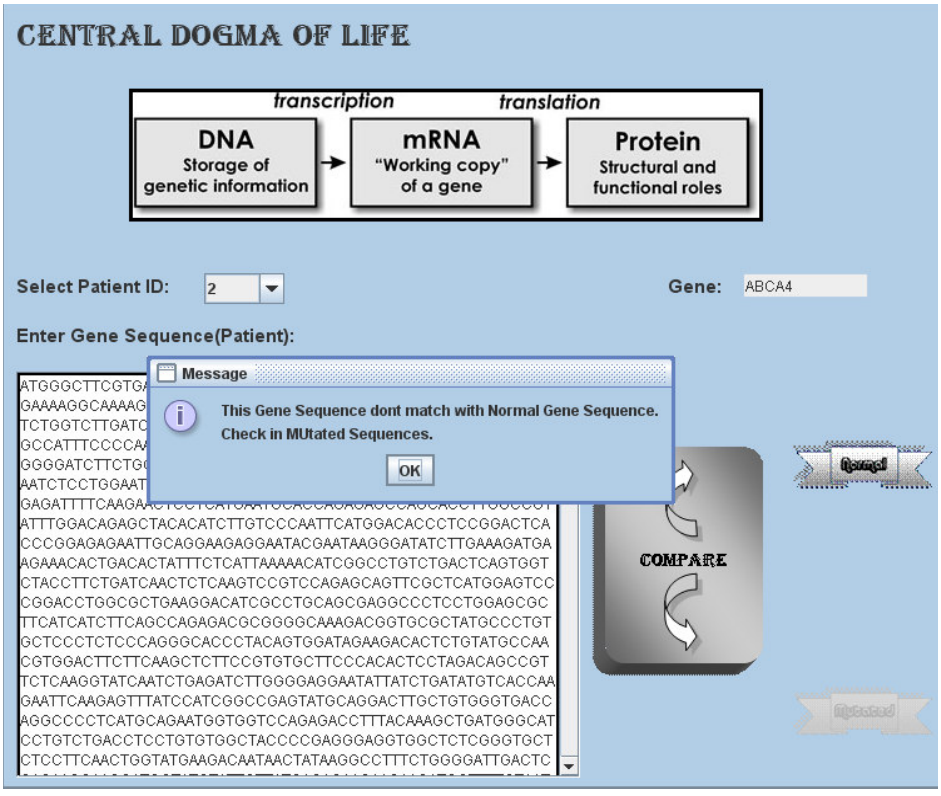

Figure 3. Comparison with normal gene sequence.

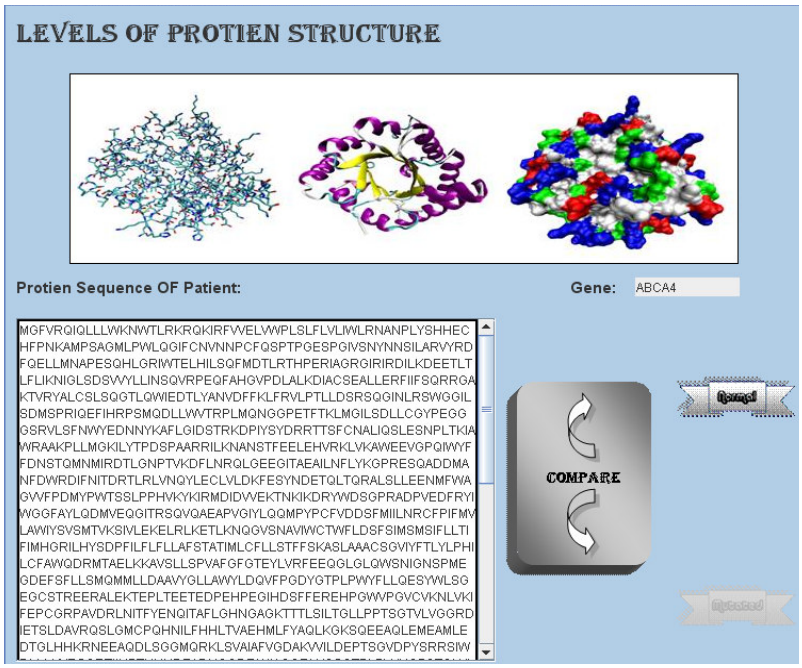

Figure 4. Comparison with mutated gene sequence and conversion into protein sequence.

new discovery or report. It was kept in consideration that testing is to be done at every stage because testing plays a critical role in the success of system.

Analysis to the implementation phase, different testing strategies like black box testing, white box testing, integrated and unit testing are applied to fix the defects and to modify the tool accordingly.

DISCUSSION

Many scientists today complain that it gets increasingly difficult to find useful information in the resulting “data labyrinth”. This may largely be due to the fact that the information gets more and more scattered over an increasingly number of heterogeneous resources (Altschul et al, 1990) and as the things become complicated, system become a dilemma. The ATID windows application is designated to solve the problem of extracting the information about the inherited diseases in a fast and simple way. An application with proper “graphical user interface” provides no switching from one program to another is required as it is sequential software. No as such training is required for its practise. All the data is united at a single place.

The designed tool can be modified for other diseases as well in future or a single database on the same pattern can be developed comprising of multiple diseases for phenotypic as well as molecular profiling.

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BioJava:http://www.biojava.org/download/bj16/bin/biojava.jar.