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BEADS

The Indian Specific Mutation Database

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***Abstract*— India is a country with incredible, biological and cultural diversity. Its natural resources attracted to the subcontinent many streams of people at different times, from different directions. This migration brought out a great diversity in human genes and cultures, resulting in a racial admixture of the Indian population. However, till date no database accounting for genetic mutations, abnormalities and variants of genetic diseases specific to the Indian population has been created. We have designed an Indian specific mutation database – BEADS based on SQL and JAVA. It's easy-to-use web interface will allow a remote user to retrieve (and submit) data on patient-specific mutation spectrum of genetic diseases through interactive web forms. The database will be helpful to the Physicians, Geneticists and other professionals in India as it provides information about the specific mutations pertaining to the Indian population. It will also provide links to other major public databases based on Human genetic disorders, type of mutation, clinical, biochemical data, geographical location and other common mutations.**

***Keywords*—Abnormalities, Diversity, Genetic diseases, Incredible, Locus heterogeneity, Mutations, Racial, Subcontinent, Variants.**

I. Introduction

Until now there is no exclusive bibliometric studies carried out on diseases specific to Indian population [1]. These studies have the potential to trace relationships amongst academic journal citations. The Indian population is one of the most diverse populations in the world, consisting of different races, castes and religion languages. The mixing of different cultural groups in India occurred between 4,200 and 1,900 years ago but started to decline as people began marrying only within their social castes, The modern-day Indians share connections to all the groups that intermarried in the distant past [2].

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However, till date no database accounting for genetic mutations, abnormalities and variants of genetic diseases specific to the Indian population has been created on one platform. **BEADS** – Is an effort in the form of database to meet this challenge. This database aims to investigate genetic distances among Indians to enable better understanding of diseases, SNPs, genetic mutations specific to Indians. The evolutionary history of primitive Indian ethnic groups and migration from Africa, middle-east and west Asia, southern China and south-east Asia has added to the genetic diversity of the country[4].

However, religion, language and geographical location of habitat serve as barriers for carrying out genetic studies in the Indian population. One of the our main objectives was to design a comprehensive and authoritative database of all known Indian disease specific literatures and research articles. By bringing this material together in one place and making it readily available our goal is to turn what is a largely untapped resource into usable material[3] . However, there is no common source of information to assess the load of specific genetic diseases reported in India, extent of locus and mutational heterogeneity, common mutations in the causal genes and the extent of molecular studies carried out vis-à-vis lack of it in the context of the disease load [4].

II. Materials and methodology

The primary source of data is peer-reviewed published reports specific to Indian population. With exception of a few reports all others are cited in PUBMED and NATURE. All the data sources are duly referred to and respective bibliographic pages are hyperlinked for further detailed information. As it is user friendly, the diseases enlisted in BEADS are divided into various categories such as 'Mutation specific', 'Geographic Location', 'based on caste, creed and religion' etc[6] .

We developed this database using MySql in the back end and JAVA in front end. MySql will help us to specialize in storage and easier to use. Data structured using MySql will easily adapt itself to the newer versions of our database. Using MySql for our database provides most effective communication to the end user. Using JAVA at the front end will make our database machine and platform

independent helping in reusability of code which will help in further easy updations of newer research material and data generated from Indian population.

TABLE 1: TIMELINE

Task	Duration	Jan	Feb	Mar	Apr	May	Jun	Jul	Aug	Sep	Oct	Nov	Dec
Literature Survey	12W	■	■	■									
Initial Schema	4W		■										
Data Curation	16W				■	■	■	■					
Back end design using SQL	8W								■	■			
Front end design using JAVA	12W								■	■	■		
Normalization-Trial run	4W											■	
Updation	4W												■

TimeLine : The workflow and time duration involved for BEADS database design

A. *Data submission and Updates*

We intend to take up this initiative in the future by providing a web-based interface for submission of new mutation data related to Indian population. Mutation data submission can be done by submitting the gene in the query box that will be provided in the web interface. However, the mutational data will be displayed based on either their publication in peer-reviewed journal or supportive documentary evidence leading to identification of the mutations or a plug in link will be created to integrate with other major public-domain databases. This will provide the users complete information about disease identification, Lead Identification, Diagnosis, Clinical research, Kit preparation etc.

III. Results

From our Literature survey it was evident that very little Research effort has gone into studying Indian specific mutation information. Our area of interest is to create 'The First Indian Specific Mutation Database – BEADS' that will provide mutation specific information and also it adds value to that information by including relevant publications, pathway maps, links to other databases etc. Updation of literature with respect to recent publications on Indian specific mutations has been dealt simultaneously, with other database creation tasks.

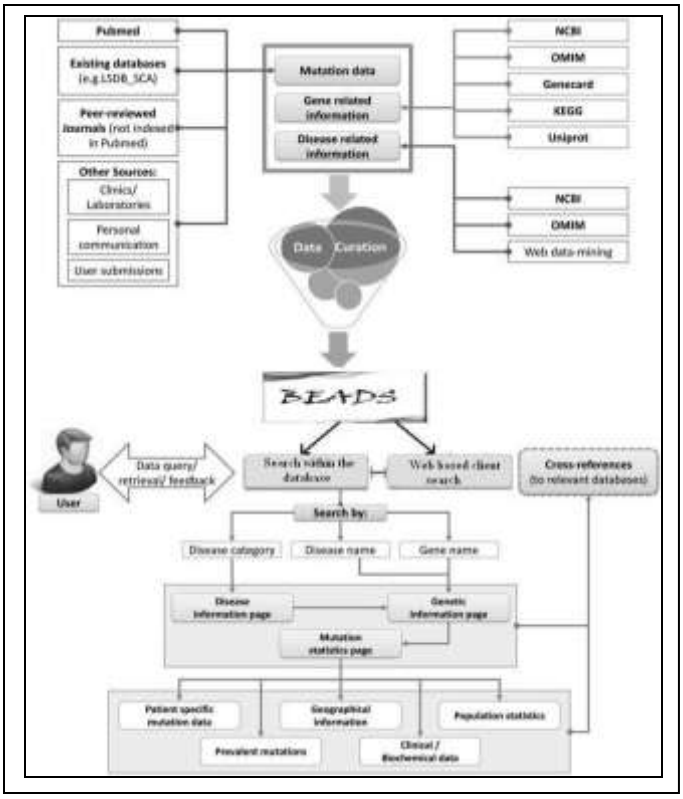

Figure 1: Initial basic Schema for the Database

IV. Discussion

Mutation variations specific to Indian population is studied to have personalized therapy and prevention of diseases. BEADS will help Physicians, Geneticists and other professionals by providing information about the specific mutations pertaining to the Indian population. It will also provide links to other major public-domain knowledge bases on Human genetic disorders, locus heterogeneity, type of mutation, clinical, biochemical data, geographical location and other common mutations. It will help physicians to design and develop new assays, diagnosing kits specific to Indian population. This will in turn help in diagnosing mutated genes, genetic diseases and identifying disease carriers among Indian population in future.

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