Chapter 5

Results and Discussion

The dbPedigree database (Database of Pedigree Analysis), a relational database, having a collection of heritable genetic defect; its causable gene; associated mutations and polymorphisms. The counts of disease, gene, pedigree, mutation, SNP, citations are shown in Table 5.1.

DISEASE	360
GENE	450
PEDIGREE	2850
MUTATION	1170
SNPs	408
CITATION	1050

Table 5.1 Shows the counts of disease, gene, pedigree, mutation, SNP, citations.

5.1 The dbPedigree

DiseaseName

The disease name is a broad category of disease. The database has around 360 diseases, therefore 360 Disease-ID.

SubDiseaseName

Represented as SDName in the maintable of the database contains the different categories of the same disease. The Figure 5.1 illustrations Subclasses of Androgen Insensitivity Syndrome namely Androgen Insensitivity Complete and Androgen Insensitivity Partial.

AI15	Androgen Insensitivity Complete	k-Linked	AR	1-BP INS; 179A	Xq12		http://jcem.endojournals.org/content/8
AI15	Androgen Insensitivity Complete	k-Linked	AR	2-BP DEL; 180GC	Xq12		http://jcem.endojournals.org/content/8
AI15	Androgen Insensitivity Complete	(-Linked	AR	INS/DEL; EX5	Xq12	Danish	http://www.nature.com/jhg/journal/v48
AI15	Androgen Insensitivity Complete	K-Linked	AR	PARTIAL DEL	Xq12		http://www.ncbi.nlm.nih.gov/pmc/artic
AI15	Androgen Insensitivity Complete	(-Linked	AR	TRP717TER	Xq12		http://www.ncbi.nlm.nih.gov/pmc/artic
AI15	Androgen Insensitivity Complete	C-Linked	AR	5-KB DEL; EX E	Xq12		http://www.ncbi.nlm.nih.gov/pmc/artic
AJ15	Androgen Insensitivity Partial	C-Linked	AR	TYR761CYS	Xq12		http://www.ncbi.nlm.nih.gov/pmc/artic
AI15	Androgen Insensitivity Partial	k-Linked	AR	TYR761CYS	Xq12		http://www.ncbi.nlm.nih.gov/pmc/artic
AJ15	Androgen Insensitivity Partial	I-Linked	AR	ARG839HIS	Xq12		http://www.ncbi.nlm.nih.gov/pmc/artic
AI15	Androgen Insensitivity Partial	C-Linked	AR	ARG839CYS	Xq12		http://www.ncbi.nlm.nih.gov/pmc/artic
AI15	Androgen Insensitivity Partial	C-Linked	AR	GLU2LYS	Xq12		http://www.ncbi.nlm.nih.gov/pmc/artic
AI15	Androgen	X-Linked	AR	ARG846HIS	Xq12		http://jcem.endojournals.org/content/8

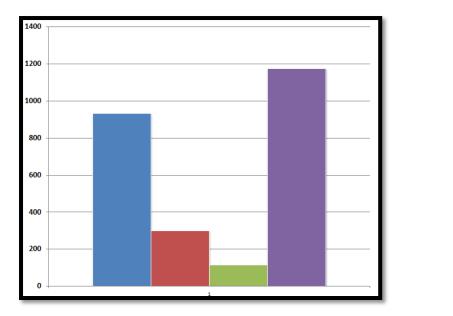
Figure 5.1 Illustrations Subclasses of Androgen Insensitivity Syndrome namely Androgen Insensitivity Complete and Androgen Insensitivity Partial.

DiseaseType

The number of records mentioned under each category autosomal dominant, autosomal recessive, X-linked dominant, X-linked recessive, Y- linked or mitochondrial mutation as reported from literature have been shown in Table 4.2.

DISEASE TYPE	COUNT
AUTOSOMAL DOMINANT :	1176
AUTOSOMAL RECESSIVE :	934
X-LINKED DOMINANT :	64
X-LINKED RECESSIVE :	11
X-LINKED (UNCLASSIFIED) :	227
TOTAL X-LINKED :	302
MITOCHONDRIAL :	116

Table 5.2 Shows the number of records mentioned under each category of disease type.



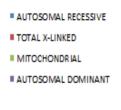


Figure 5.2 Shows the bar chart of data recorded under different modes of inheritance.

OtherDisease

Represented as ODisease in the maintable of the database shows the name of any other disease which could have been present in the proband's family. The Figure 5.3 shows that while studying the pedigree for Li-Fraumeni-Like Syndromes, Sarcomas; premenopausal breast cancers were also considered.

DID	SDName	DType	ODisease	Gene	Locus	Mutation	SNP	Ethnicity	PNo	Link	PName
LI 198	Li- Fraumeni- Like Syndromes	Autosomal Dominant	Sarcomas; premenopausal breast cancers	TP53	17p12	ARG248TRP exon 7			PID1	http://www.nejm.org/action/showImage?doi=10.1056%2	family 6
L) 198	Li- Fraumeni- Like Syndromes	Autosomal Dominant	Sarcomas; premenopausal breast cancers	TP53	17p13	ARG248TRP exon 7			PID1947	http://www.nejm.org/action/showImage?doi=10.1056%2	family 6
NH249			Colon cancer	TP53	17p13	GLY325VAL exon 9		10,00	PID10	http://www.nejm.org/action/showImage?doi=10.1056%2.	family 49
OL253				LEP	7q32.1	homozygosity for the delta- 133G		PAKISTANI	PID100	http://jcem.endojournals.org/content/89/10/4821/F1	
EB118		Autosomal		COL7A1	3p21.31	GLY2563ARG			PID1000	http://www.ncbi.nlm.nih.gov/pmc/articles/PMC191467	family 5

Figure 5.2 Shows the data included in the OtherDisease column.

Gene

Genes hold the information to build and maintain an organism's cells and pass genetic traits to offspring. Any change in the sequence of the gene may give rise to a defective protein or may silent the gene itself. In both the cases the individual is affected. This has been considered as an important feature for searching. The search can be carried out using Gene HGNC Symbol. The HGNC symbol is unique. It is necessary to provide a unify symbol for each gene so that it can be used globally and this also facilitates electronic data retrieval from publications and databases. The DbPedigree database has around 450 Gene HGNC symbols.

GeneLocus

The specific place on a chromosome where a gene is located is referred as locus. The Figure 5.4 shows the number of gene locus located on each chromosome analysed during making of dbPedigree. The maximum number of diseases lies on chromosome 2 and X while the chromosome 22 is least related.

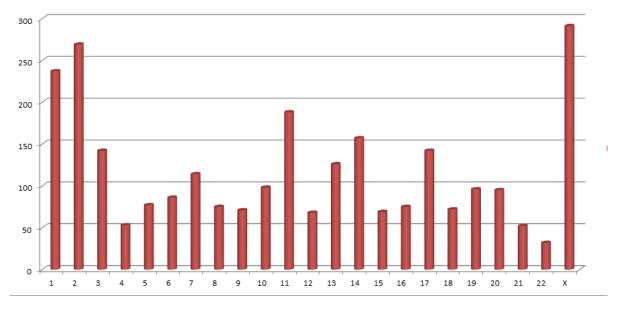


Figure 5.4 Shows the number of gene locus located on each chromosome.

Associated Mutation

The mutation can be insertion, deletion, duplication, single nucleotide change, Translocation etc. The single nucleotide change around 1200 entries of mutational data, rest 1000 entries are all others. The Fig. depicts contribution of each type of mutation.

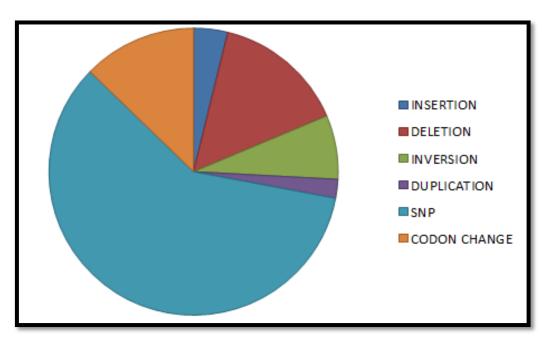


Figure 5.5 Depicts contribution of each type of mutation in building of dbPedigree.

Single Nucleotide Polymorphism

This field contains only those SNP which have been reported in dbSNP. The database contains 408 rs_id's.

Ethnicity

The field contains the region to which the proband belongs. Most of the proband's belonging to African, European, French, United States, and Chinese ethnicity respectively.

PedigreeNumber

This field was introduced so that the problem of a single pedigree involving more than one mutation can be sorted. The Figure 5.6 shows a single pedigree PID1004 showing 2 mutations.

←T→	DID	PNo 🖕	Mutation
📄 🥔 Edit 🖉 Inline Edit 👫 Copy 🥥 Delete	LI198	PID1	ARG248TRP exon 7
🖂 🥔 Edit 📝 Inline Edit 👫 Copy 🥥 Delete	NH249	PID10	GLY325VAL exon 9
🔲 🥒 Edit 📝 Inline Edit 👫 Copy 🤤 Delete	OL253	PID100	homozygosity for the delta-133G
📄 🥜 Edit 📝 Inline Edit 👫 Copy 🥥 Delete	EB118	PID1000	GLY2569ARG
📄 🥔 Edit 📝 Inline Edit 👫 Copy 🤤 Delete	EB118	PID1001	GLY2749ARG
📄 🥔 Edit 📝 Inline Edit 👫 Copy 🥥 Delete	MD238	PID1002	VAL154ILE
🔲 🥜 Edit 📝 Inline Edit 👫 Copy 🤤 Delete	OD01	PID 1003	PHE540BER
📄 🥜 Edit 📝 Inline Edit 👫 Copy 🤤 Delete	CD81	PID1004	ALA546ASP
📄 🥔 Edit 📝 Inline Edit 👫 Copy 🥥 Delete	CD81	PID1004	PRO551GLN
📄 🥔 Edit 📝 Inline Edit 👫 Copy 🥥 Delete	CD81	BID1005	APC124LELL
📄 🥜 Edit 📝 Inline Edit 👫 Copy 🤤 Delete	CD81	PID1006	PRO501THR
🔲 🥔 Edit 📝 Inline Edit 👫 Copy 🥥 Delete	CD81	PID1007	PRO501THR
📄 🥔 Edit 📝 Inline Edit 👫 Copy 🥥 Delete	CD81	PID1008	PRO501THR
📄 🥜 Edit 📝 Inline Edit 👫 Copy 🥥 Delete	BR50	PID1009	PRO267LEU
📄 🥜 Edit 📝 Inline Edit 👫 Copy 🤤 Delete	BR50	PID1009	IVS12AS; A-G; -2
📄 🥜 Edit 📝 Inline Edit 👫 Copy 🤤 Delete	RS304	PID101	1-BP DEL; 806G
🔲 🥜 Edit 📝 Inline Edit 👫 Copy 🤤 Delete	BR50	PID1010	GLY294SER

Figure 5.6 Shows a single pedigree PID1004 showing 2 mutations.

Link

The pedigree has been reported in the form of hyperlinks to the images in the research article.

Pedigree Name

If the pedigree link contains more than 1 pedigree, then this field will report the family name or pedigree reference name.

p-value

A measure of how much evidence there is against the null hypothesis. The smaller the p-value, the more evidence exists against. Traditionally, researchers will reject the null hypothesis if the p-value is less than 0.05. A small p-value is evidence against the null hypothesis while a large p-value means little or no evidence against the null hypothesis.

LOD

It is the LOD score, logarithm (base 10) of odds, is a statistical test often used for linkage analysis. It compares the likelihood of obtaining the test data if the two loci are indeed linked,

to the likelihood of observing the same data purely by chance. Positive LOD scores favour the presence of linkage, whereas negative LOD scores indicate that linkage is less likely.

Citation

The PubMed ID or the PubMed Central ID of every literature associated with the pedigree has been reported.

It is a manually curated database expected to reporting 2850 pedigrees. It is an open access database, and user will be able to access it from DTU server.

5.2 The Webpages

A web-based graphical user interface has been designed using HTML. The software used for designing the interface is Macromedia Dreamweaver 8. The dbPedigree has 5 main pages.

The Homepage dbPedigree About Database Search Page Glossary Contacts

5.2.1 The "HOMEPAGE" dbPedigree

When the user visits the dbPedigree website, the following homepage will be displayed in the browser. It will also have links to types of disease.

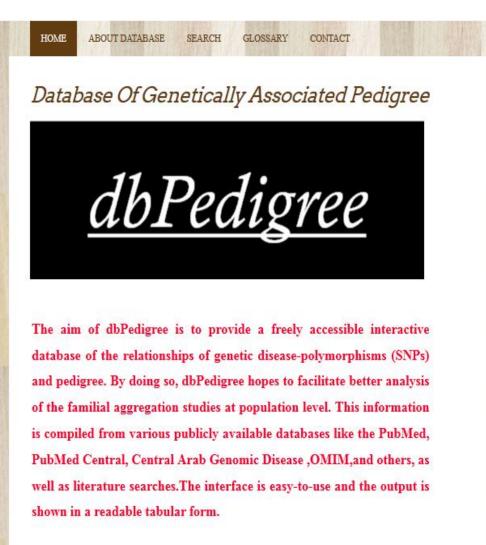


Figure 5.3 The dbPedigree homepage.

5.2.2 The "ABOUT DATABASE" Page

The "About Database" gives the brief introduction about the different modes of inheritance of genetic disorder.

ABOUT DATABASE SEARCH

GLOSSARY CONTACT

About Database Of Genetically Associated Pedigree :

The dbPedigree (Database of Genetically Associated Pedigree); contains extensive information about genetic disorders studied by building pedigree. The inheritance patterns trace the genetically encoded traits, condition and disease to an offspring. The pedigree gives the aggregation of the occurrence of more cases of a given disorder in close relatives of the proband than in control families. The process includes building up of a questioner to know about the genetic history of a family or a "genetic family tree".

Types of Genetic Disorder

<u>Autosomal Dominant Disorder</u>: The disease condition in which only one abnormal allele of a gene is present .The affected male or female have equal probability of passing the disease to offspring. A heterozygous affected parent and an unaffected parent have, on average, an equal number of affected and unaffected children; i.e., risk of occurrence for each child of an affected parent is 50%, because the affected individual can be heterozygous for the trait that is having one normal copy of gene and one affected copy, thus the offspring have 50% chances of inheriting

Figure 5.4 The About Database page.

5.2.3 The "SEARCH" Page

The search page allows user to search the database using disease name or the gene name. The page also contains links to list of genetic disorders and list of associated gene.

	s of genetic variability, and hence
of evolution. By T. Dobzhansky	
5	
The Database can be search	hed by either the name of the disease or the
	be displayed giving the details about the associate y.To pedigree has been reported in the form of
matation and the cumficity	rio peargree mas ocen reporteu in the form of
Hyperlinks.	
Hyperlinks.	
Hyperlinks. KNOW ABOUT	
KNOW ABOUT	
KNOW ABOUT List Of Genetic Diseases	

Figure 5.5 The search page.

Search page has links to list of disease and links of genes pages.

5.2.3.1 The "List of Disease" Page

The list of disease contains 360 disorders whose pedigree has been recorded in dbPedigree.

List Of Genetic Disease

17beta-Hydroxysteroid Dehydrogenase 10 Deficiency Acheiropody Acrodermatitis Enteropathica Adenylosuccinate Lyase Deficiency Adolescent Nephronophthisis Adrenocortical Carcinoma Pediatric Albinism Alkaptonuria Alopecia Universalis Congenita Alstrom Syndrome Alzheimer Disease Amyloidosis **Amyotrophic Lateral Sclerosis** Andersen Cardiodysrhythmic Periodic Paralysis Androgen Insensitivity Syndrome Anemia Angelman Syndrome Angioedema Aniridia Aortic Valve Disease Apolipoprotein C-II Arthritis Ascaris Ataxia Ataxia-Telangiectasia Atrial Fibrillation Atrial Septal Defect Atrichia Atypical Thiel-Behnke CD Avascular Necrosis of Femoral Head Axenfeld-Rieger Syndrome Axial Myopia Bannayan-Riley-Ruvalcaba Bardet-Biedl Syndrome Barth Syndrome **Basal Cell Nevus Syndrome**



Figure 5.6 The list of genetic disease.

5.2.3.2 The "List of genes" Page

The list of genes contains 450 HGNC symbols of genes associated with genetic disorders that have been recorded in dbPedigree. The list contains 4 column and 113 rows.

		1	List Of Gene		
	MTATP6	DSRAD	CO1	ATXN8OS	
	MTMR13	DYSF	CO3	CRYBA1	
	MTND1	DYT7	COCH	CRYGD	
	MTND6	DYT7	COHI	GLRAI	
	MTRNRI	EFNB1	COL10A1	PRNP	
	MTTI	EFNB1	COLIAI	SQSTM1	
	MTTL1	EGR2	COL1A2	TBC1D24	195
	MTTS1	EIF2AK3	COL2A1	TCF2	
	MYBPC1	ELN	COL5A1	AARS	THE REAL
	MYH7	EMD	COL7A1	ABCA3	
Manit	МҮН9	EPHX2	COL9A2	ABCA4	
1.1.1	MYO15A	ERBB3	COL9A3	ABCC6	
	муос	ESR1	CRB1	ACADM	
	MYP1	EVC2	CRX	ACADM	
		1000			1.141、1112-1

Figure 5.7 The list of genes.

5.2.4 The Glossary page

The Glossary page has also been designed to make users familiar to the terms that they can come across while going through the literature of human genetic disorders. The page contains 14 such terms.

	GLOSSARY						
Allele	One of two or more alternative nucleotide sequences at a single gene locus on a chromosome.						
Allele Frequency	The frequency of a population used to characterize the genetic diversity of a species population or equivalently the richness of its gene pool.						
Association Studies	The primary means of establishing an association between a given phenotype and the other covariates, such as other phenotype data or genotype data.						
Genetic Model	The overall specification of how the disease allele(s) act to the influence the disease. A genetic model consists of three main components: -a model for disease susceptibility, connecting disease phenotypes to genotypes at disease susceptibility (DS) loci for the sibs; -a population genetics model, describing the population joint distribution of genotypes at the DS loci of the parents; and -a segregation model, describing the segregation of alleles at the DS loci during meiosis.						
Haplotype	Set of closely linked genetic markers present on one chromosome which tend to be inherited together (not easily separable by recombination).						
Heritability	A measure of the degree to which the variance of the distribution of a phenotype is due to genetic causes. Specifically, heritability is defined as the proportion of phenotypic variance explained by the analyzed marker.						
Linkage	Two genes or markers that are so close together on a chromosome that they are rarely separated by recombination are said to be linked.						
	A statistical method for detecting linkage between a disease allele and markers of known location by following their inheritance in families. linkage disequilibrium						

Figure 5.8 The Glossary Page.

5.2.5 The Contacts Page

For any further queries the users can mail at the specified E-mail ID given on the contact page.

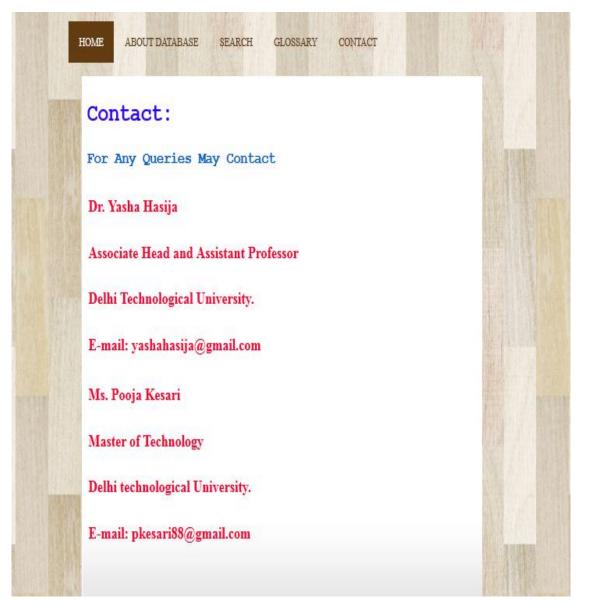


Figure 5.9 The Contact Page.

5.3 The steps for querying dbPedigree

- Disease-Wise Search
 - 1. Select the **disease** radio button.

Search Options:							
Do You Want To Search By :							
Disease Name Gene Name Search							

Figure 5.10 Disease radio button.

2. Specify the name of the disease and click on the **search** button.

Search Options:	Search Options:								
Do You Want To Search By :									
Oisease Name	© Gene Name								
Albinism	Search								

Figure 5.11 Searching "Albinism" in the database.

3. The result would be shown in a tabular format containing different column like sub-disease category (if any),type of disorder, other disease included during the pedigree study, causative gene along with its locus, associated mutation ,SNP, ethnicity of the family ,pedigree number and name, p-value, LOD score and the reference of the literature .The result contains 23 records against "Albinism".

Sub Disease Name	Disease Type		100000	1000002	Associated Mutation	rs_id	1	Ethnicity	pedigree number	Link to pedigree	Pedigree Name	LOD Score	Citation
Ocular Albinism Type I	X-Linked		GPR143	Xp22.2	14-BP DEL; NT816				PID1578	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC1724103 /figure/F1/			11520764
Albinism Oculocutaneous Type IB	Autosomal Recessive		TYR	11q14.3	PRO406LEU	rs1048	394313	FRENCH ;USA;Swiss	PID1642	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC1706616 /?page=2			5516239
Albinism Oculocutaneous Type IB	Autosomal Recessive	Waardenburg Syndrome 2	TYR	11q14.3	ARG402GLN	rs1126	809		PID1643	http://hmg.oxfordjournals.org /content/6/5/659.long			9158138
Albinism Oculocutaneous Type IA	Autosomal Recessive		TYR	11q14.3	ARG59GLN	rs6175	53185		PID1644	http://www.jbc.org/content /265/29/17792.long			2120217
Temperature- Sensitive Oculocutaneous Albinism	Autosomal Recessive		TYR	11q14.3	ARG402GLN	rs1126	5809	Caucasian		http://www.ncbi.nlm.nih.gov /pmc/articles/PMC329899 /?page=2			1900307
Ocular Albinism Type 1	X-Linked		GPR143	Xp22.2	CYS116TRP	Contract of the second s		Canadian		http://www.ncbi.nlm.nih.gov /pmc/articles/PMC1468396 /figure/F1/	pedigree 11		PMC146839
Ocular Albinism Type 1	X-Linked		GPR143	Xp22.2	THR166ASN			FRENCH		http://www.ncbi.nlm.nih.gov /pmc/articles/PMC1468396 /figure/F1/	pedigree 1L		PMC146839
Ocular Albinism Type 1	X-Linked		GPR143	Xp22.2	163_170dup			FRENCH		http://www.ncbi.nlm.nih.gov /pmc/articles/PMC1468396 /figure/F2/	pedigree 2B		PMC146839

Figure 5.12 Results of search using keyword "Albinism".

4. The pedigree result is sorted by the pedigree number. The pedigree image can be opened by right clicking the link and opening it in a new window.

Sub Disease Name	Disease Type	Disease Include	and the second	1000	Associated Mutation	rs_id	Ethnicity	pedigree number	Link to pedigree		Pedigree Name	Probus	LOD Score
Ocular Albinism Type I	X-Linked		GPR143	Xn22.2	14-BP DEL; NT816				http://www.ncbi. /pmc/articles/PM /figure/F1/				
Albinism Oculocutaneous Type IB	Autosomal Recessive		TYR	11q14.3	PRO406LEU	rs104894313	FRENCH ;USA;Swiss	PID1642	http://www.ncbi. /pmc/articles/PM /?page=2	IC1706616	A MAN		and the second se
Albinism Oculocutaneous Type IB	Autosomal Recessive	Waardenburg Syndrome 2	TYR	11q14.3	ARG402GLN	rs1126809		PID1643	http://hm Op	en Link en Link in New <u>T</u> i en Link in New <u>W</u>	Sec		
Albinism Oculocutaneous Type IA	Autosomal Recessive		TYR	11q14.3	ARG59GLN	rs61753185		PID1644	/265/29/1	py ect <u>A</u> ll erch Google for "h	http://www.no	:bi"	
Temperature- Sensitive Oculocutaneous Albinism	Autosomal Recessive		TYR	11q14.3	ARG402GLN	rs1126809	Caucasian	and the second second	http://ww	w Selection Source			
Ocular Albinism Type 1	X-Linked		GPR143	Xp22.2	CYS116TRP	79109	Canadian	1.5 C 200 4 5 4 5 6	http://www.ncbi. /pmc/articles/PM /figure/F1/	CARL STREET	pedigree 11		

Figure 5.13 Right clicking the link to open the "Albinism" pedigree.

5. **Pedigree Image**

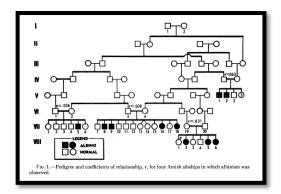


Figure 5.14 The pedigree image of Albinism.

• Gene-Wise Search

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1 Select the **Gene** radio button.

Search Options:									
Do You Want To Search By :									
© Disease Name Search									

Figure 5.15 Gene radio button.

2 Specify the name of the disease and click on the **search** button.

Search Options:										
Do You Want To Search By :										
Disease Name	• Gene Name									
alk	Search									

Figure 5.16 Searching "ALK" gene in the database.

3 The result would be shown in a tabular format containing different column like . disease category, type of disorder, other disease included during the pedigree study, causative gene along with its locus, associated mutation, SNP, ethnicity of the family, pedigree number and name, p-value, LOD score and the reference of the literature .The result contains 8 records against "ALK" gene.

Sub Disease Name		Disease Include		Gene	locus	Associated Mutation	rs_id	Ethnicity	pedigree number	Link to pedigree	Pedigree Name	P-value	LOD Score	Citation
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.	2-р23.1	GLY1128ALA c.3383G>C	rs113994088		PID422	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 /figure/F1/	FBN52			PMC2672043
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.	2-р23.1	ARG1192PRO c.3575G>C	rs113994089		PID423	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 /figure/F1/	FNB2			PMC2672043
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.	2-р23.1	ARG1275GLN c.3824G>A	rs113994087		PID424	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 /figure/F1/	FNB6			PMC2672043
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.	2-р23.1	ARG1275GLN c.3824G>A	rs113994087		PID425	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 /figure/F1/	FBN11			PMC2672043
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.	2-р23.1	ARG1275GLN c.3824G>A	rs113994087		PID426	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 /figure/F1/	FBN12			PMC2672043
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.	2-р23.1	ARG1275GLN c.3824G>A	rs113994087		PID427	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 /figure/F1/	FBN13			PMC2672043
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.	2-р23.1	ARG1275GLN c.3824G>A	rs113994087		PID428	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 /figure/F1/	FBN56			PMC2672043
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.	2-р23.1	ARG1192PRO c.3575G>C	rs113994089		PID429	http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 /figure/F1/	FBN32			PMC2672043

Figure 5.17 Results of search using keyword "ALK".

- 4 The pedigree result is sorted by the pedigree number. The pedigree image can be
- . opened by right clicking the link and opening it in a new window.

Second enderstein die	and the second second	Disease Include	254-19	Gene locus	Associated Mutation	rs_id	Ethnicity	pedigree number	Link to pedigree Re	P-value	LOD Score	Citation
Neuroblastoma Type 3	Autosomal Dominant		ALK	2р23.2-р23.1	GLY1128ALA c.3383G>C	rs113994088			http://www.ncbi.nlm.nih.gov /pmc/articles/PMC2672043 FF /figure/F1/	BN52		PMC267204
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.2-p23.1	ARG1192PRO c.3575G>C	rs113994089		PID423	http://www.ncbi.nlm.nih.gov /pmc/ar /figure/F Open Link in New Tab	<u>O</u> pen Link		
Neuroblastoma Type 3	Autosomal Dominant		ALK	2р23.2-р23.1	ARG1275GLN c.3824G>A	rs113994087		PID424	http://w Open Link in New Wind /pmc/art /figure/F Copy	Open Link in New <u>W</u> indow		PMC267204
Neuroblastoma Type 3	Autosomal Dominant		ALK	2р23.2-р23.1	ARG1275GLN c.3824G>A	rs113994087		PID425	http://w /pmc/art Search Google for "http			PMC267204
Neuroblastoma Type 3	Autosomal Dominant		ALK	2p23.2-p23.1	ARG1275GLN c.3824G>A	rs113994087		PID426	http://w ⁻ Inspect Element (Q) /pmc/articles/PMC2072043 FF /figure/F1/	5.N12		PMC267204

Figure 5. 18 Right clicking the link to open the "ALK" pedigree.

5 Pedigree Image

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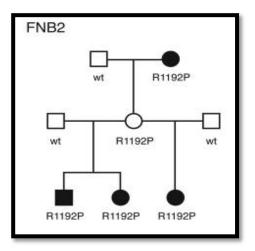


Figure 5.19 The pedigree image of "ALK" gene.

In case the disease or the gene name does not exist in the database. An error message would be shown.



Figure 5.20 The Error message.

5.4 The Working of PHP Script

The PHP script receives the query form from the browser and retrieves the records from the database after filtering them as required by the user. The selections made by the user are then obtained from the filtered records and sent to the server which then sends them to the browser. The detailed procedure is as follows:

- Connection is made with the database using command mysql_connect. This requires the database name, the hostname and port as well as the username and password that allows one to access the localhost.
- 2. Connection is made with the database using command **mysql_select_db**. This requires the database name, the hostname and port as well as the username and password that allow one to access the database.
- 3. A variable will point to the selection made among the two radio button.
- 4. The SQL command is run, where the query will be executed for whichever keyword specified in the text box.

In case query is created using disease name, first it would be searched in DiseaseTable and the diseaseID would be recorded. Then records would be selected from maintable against that diseaseID.

Similar is the process for Gene-wise search.

5. The selected records would be displayed in the HTML table.