

# ASSISTANCE: HOW TO SEARCH THE DERMAGENE DATABASE

## Home Page

The DermaGene Home-page exhibits a query form that allows users to perform selective querying, enabling them to apply multiple filters and subsequently obtain the desired records. The Query form enables users to search across multiple Dermatological disorders by searching either through Gene name, SNP, Ethnicity, PubMed ID or selecting from the Disease name and disease class drop down menu.

## Database Filters

DermaGene enables users to extract specific records through application of correct queries:

Query	Results
<b>Genes</b>	Filters records based on HGNC gene symbols. The database can suggest gene names as one continues to type the complete gene name.
<b>SNP (Reference SNP cluster ID)</b>	Filter records based on rsIDs
<b>Disease Name</b>	Select the dermatological disease from the provided drop-down menu.
<b>Disease Class</b>	Records can be restricted to only the subset that has disease-SNP associations from a particular disease class.
<b>Ethnicity</b>	Records can be filtered based on the population ethnicity. Select the ethnicity from the dynamic drop down menu provided which provides suggestions as you type in the search box.
<b>PubMed ID</b>	Relevant data can be extracted using the unique identifier provided by PubMed

## Functionality

Users can opt for AND & OR functionality provided by our database.

**1. OR Functionality:** User can obtain results using either of the filters provided in the database. Example: Upon querying, SNP (Reference SNP cluster ID) = rs121913105; Disease Name= Bannayan Riley Ruvalcaba syndrome; Disease class = Aging disease, we obtain the set of values that represents either of the specified criteria, the result shown below:



QUERY GENE

Select Operation:  AND  OR

Gene:

SNP (Reference SNP cluster ID):

Disease Name:

Disease class:

Ethnicity:

PubMed ID:

Note: selected operation will be applicable for all filters. And user can use 1 or more filter at a time.

Examples:

Gene:	PPARG	SNP (Reference SNP cluster ID):	rs121913105
Disease Name:	Acne Vulgaris	Disease class:	Angioedema
Ethnicity:	AMERICAN	PubMed ID:	24984871

**Operation behaviour**

**AND behaviour**  
Gene: PPARG, Disease Name: Acne Vulgaris  
Result: records which is having both gene as PPARG AND disease as Acne Vulgaris

**OR behaviour**  
Gene: PPARG, Disease Name: Acne Vulgaris  
Result: records which is having either gene as PPARG OR disease as Acne Vulgaris OR both



# DermaGene

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Disease Name	Gene	SNP	PubMed ID	Ethnicity	P-value	Class
<a href="#">Acanthosis Nigricans</a>	<a href="#">EGFR3</a>	<a href="#">rs121913105</a>	<a href="#">10053006</a>	AMERICAN	0.014	Skin and Connective Tissue Disease
<a href="#">Bannayan Riley Ruvyalcaba syndrome</a>	<a href="#">PTEN</a>	<a href="#">rs121909221</a>	<a href="#">27489861</a>	N.A	0.454	Neoplasia
<a href="#">Hutchinson Gilford progeria syndrome</a>	<a href="#">LMNA</a>	<a href="#">rs60652225</a>	<a href="#">12927431</a>	MULTI-ETHNICITY	0.627	Aging disease
<a href="#">Hutchinson Gilford progeria syndrome</a>	<a href="#">LMNA</a>	<a href="#">rs60310264</a>	<a href="#">12714972</a>	N.A	0.627	Aging disease
<a href="#">Hutchinson Gilford progeria syndrome</a>	<a href="#">LMNA</a>	<a href="#">rs60310264</a>	<a href="#">21941106</a>	N.A	0.627	Aging disease
<a href="#">Hutchinson Gilford progeria syndrome</a>	<a href="#">LMNA</a>	<a href="#">rs28928902</a>	<a href="#">12768443</a>	CAUCASIAN	0.627	Aging disease
<a href="#">Hutchinson Gilford progeria syndrome</a>	<a href="#">LMNA</a>	<a href="#">rs28928902</a>	<a href="#">16772334</a>	N.A	0.627	Aging disease
<a href="#">Hutchinson Gilford progeria syndrome</a>	<a href="#">LMNA</a>	<a href="#">rs58912633</a>	<a href="#">15622532</a>	GERMAN	0.627	Aging disease
<a href="#">Hutchinson Gilford progeria syndrome</a>	<a href="#">LMNA</a>	<a href="#">rs58912633</a>	<a href="#">17881656</a>	GERMAN	0.627	Aging disease
<a href="#">Hutchinson Gilford progeria syndrome</a>	<a href="#">LMNA</a>	<a href="#">rs58912633</a>	<a href="#">25738644</a>	GERMAN	0.627	Aging disease

**2. AND Functionality:** Users can obtain specific results by providing defined queries, thereby getting selective results. Example: Upon querying Gene=IKZF4 & Disease Name = Alopecia areata, we get selective results that satisfy both the criteria, but not any one individually. The results are as shown:



▼ QUERY GENE

**Select Operation:**     AND     OR

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Gene:

SNP (Reference SNP cluster ID):

Disease Name:

Disease class:

Ethnicity:

PubMed ID:

Note: selected operation will be applicable for all filters. And user can use 1 or more filter at a time.

Examples:

<b>Gene:</b>	PPARG	<b>SNP (Reference SNP cluster ID):</b>	rs121913105
<b>Disease Name:</b>	Acne Vulgaris	<b>Disease class:</b>	Angioedema
<b>Ethnicity:</b>	AMERICAN	<b>PubMed ID:</b>	24984871

**Operation behaviour**

**AND behaviour**  
Gene: PPARG, Disease Name: Acne Vulgaris  
Result: records which is having both gene as PPARG AND disease as Acne Vulgaris

**OR behaviour**  
Gene: PPARG, Disease Name: Acne Vulgaris  
Result: records which is having either gene as PPARG OR disease as Acne Vulgaris OR both



Disease Name	Gene	SNP	PubMed ID	Ethnicity	P-value	Class
<a href="#">Alopecia Areata</a>	<a href="#">IKZF4</a>	<a href="#">rs1701704</a>	<a href="#">20596022</a>	MULTI-ETHNICITY	0.242	Auto-immune disease
<a href="#">Alopecia Areata</a>	<a href="#">IKZF4</a>	<a href="#">rs1701704</a>	<a href="#">20596022</a>	MULTI-ETHNICITY	0.002	Auto-immune disease
<a href="#">Alopecia Areata</a>	<a href="#">IKZF4</a>	<a href="#">rs1701704</a>	<a href="#">20596022</a>	EUROPEAN	0.243	Skin and Connective Tissue Disease

### Hyperlinks to websites

Every entry in the database provides links to associated WebPages for Disease, Genes, SNPs & PubMed IDs.

1. Clicking under 'Disease Name', e.g. [Alopecia Areata](#) opens OMIM webpage, where users can browse through data available about this disease.

The screenshot shows the OMIM database search interface. At the top, there is a search bar containing 'Alopecia Areata' and a search button. Below the search bar, there are options for 'View Results as:' (Gene Map Table, Clinical Synopsis) and 'Display:' (Highlights). A section titled 'Would you also like:' includes checkboxes for 'hair loss', 'hypotrichosis', 'Add All', 'sparse hair', and 'thinning hair'. The search results are listed below, showing 274 entries. The first three results are:

- 1: \* 104000. ALOPECIA AREATA 1; AA1  
ALOPECIA UNIVERSALIS, INCLUDED  
Cytogenetic location: 18p11.3-p11.2, Genomic coordinates (GRCh38): 180-15,400,000  
Matching terms: areata, alopecia  
▶ Gene-Phenotype Relationships ▶ Phenotypic Series ▶ ICD+ ▶ Links
- 2: \* 147150. MYXOVIRUS RESISTANCE 1, MOUSE, HOMOLOG OF; MX1  
Cytogenetic location: 21q22.3, Genomic coordinates (GRCh38): 21-41,420,328-41,459,213  
Matching terms: areata, alopecia  
▶ Links
- 3: \* 191160. TUMOR NECROSIS FACTOR; TNF  
Cytogenetic location: 6p21.33, Genomic coordinates (GRCh38): 6,31,575,966-31,578,335  
Matching terms: areata, alopecia  
▶ Gene-Phenotype Relationships ▶ Links

The fourth result is partially visible:

- 4: \* 610753. ALOPECIA AREATA 2; AA2  
Cytogenetic location: 16q11-q22, Genomic coordinates (GRCh38): 16,36,800,000-74,100,000  
Matching terms: areata, alopecia  
▶ Gene-Phenotype Relationships ▶ Phenotypic Series ▶ ICD+ ▶ Links

2. Clicking under 'Gene', e.g. **IKZF4** opens NCBI webpage, where users can browse through data available about this Gene.

The screenshot shows the NCBI Gene database interface. The search term 'IKZF4' is entered in the search bar. The results are displayed in a table with the following columns: Name/Gene ID, Description, Location, Aliases, and MIM. The table lists several entries for IKZF4 in different species and tissues. On the right, there are filters for 'Results by taxon' and 'Find related data'.

Name/Gene ID	Description	Location	Aliases	MIM
IKZF4 ID: 64375	IKAROS family zinc finger 4 [Homo sapiens (human)]	Chromosome 12, NC_000012.12 (56007647-56038436)	EOS, ZNFN1A4	608239
Ikzf4 ID: 22787	IKAROS family zinc finger 4 [Mus musculus (house mouse)]	Chromosome 10, NC_000076.6 (129638839-129669609, complement)	A639029-I08Rik, Eos, Zfn1a4, Znf1a4	
Ikzf4 ID: 100361823	IKAROS family zinc finger 4 [Rattus norvegicus (Norway rat)]	Chromosome 7, NC_005106.4 (13061422-3098142, complement)		
IKZF4 ID: 549071	IKAROS family zinc finger 4 [Bos taurus (cattle)]	Chromosome 5, AC_000162.1 (57613472-57638544, complement)	ZNFN1A4	
IKZF4 ID: 101747801	IKAROS family zinc finger 4 [Gallus gallus (chicken)]	Chromosome 33, NC_008465.3 (630411-642769)		
Ikzf4 ID: 100496195	IKAROS family zinc finger 4 [Xenopus tropicalis (tropical clawed)]	Chromosome 2, NC_030678.1 (134504409-134526294, complement)	eos, znf1a4	

3. Clicking under 'SNP', e.g. **rs1701704** opens NCBI-dbSNP webpage, where users can browse through data available about this SNP.

The screenshot shows the NCBI dbSNP database interface. The search term 'rs1701704' is entered in the search bar. The results are displayed in a table with the following columns: RefSNP, Allele, and HGVS Names. The table lists various alleles and their corresponding HGVS names. The page also includes a 'GENERAL' sidebar and a 'RELATED SITES' section.

RefSNP	Allele	HGVS Names
Organism: human ( <i>Homo sapiens</i> )	Variation Class: SNV, single nucleotide variation	NC_000012.11.g.56412487T>G
Molecule Type: Genomic	RefSNP Allele: A/C (REV)	NC_000012.12.g.56018703T>G
Created/Updated in build: 89/150	Allele Origin: A/C (REV)	NR_135623.1.n.63+6326A>C
Map to Genome Build: <a href="#">HG19/Weight 1</a>	Ancestral Allele: A	XM_005269088.1.c.21+526T>G
Validation Status: <a href="#">P</a> <a href="#">V</a> <a href="#">H</a>	Variation Viewer: <a href="#">View</a>	XM_005269089.1.c.40+526T>G
Citation: <a href="#">PubMed</a>	Clinical Significance: NA	XM_005269089.2.c.40+526T>G
Association: <a href="#">MIGRBLQWAS</a>	G=0.2019/1011 (1000 Genomes)	XM_005269090.1.c.40+526T>G
	MAF: <a href="#">MinorAlleleCount</a>	XM_005269090.3.c.40+526T>G
	G=0.2344/6826 (TOPMED)	XM_005269091.1.c.40+526T>G
		XM_005269092.1.c.40+526T>G

4. Clicking under 'PubMed ID', e.g. **20596022** opens NCBI-Pubmed webpage, where users can browse through literature source discussing the disease.



The screenshot shows a PubMed search result for the article ID 20596022. The page layout includes a header with the NCBI logo and navigation options, a search bar with the ID entered, and a main content area. The article title is 'Genome-wide association study in alopecia areata implicates both innate and adaptive immunity.' The abstract text describes the genetic basis of alopecia areata (AA) through a genome-wide association study (GWAS) in a sample of 1,054 cases and 3,278 controls. It identifies 159 single nucleotide polymorphisms (SNPs) significantly associated with AA, including genes like CTLA4, IL21, IL2, and Eos. The study also highlights the upregulation of ULBP ligands in the hair follicle dermal sheath during active disease, suggesting a novel disease mechanism involving both innate and acquired immunity. The right sidebar contains links to full-text versions on Nature and PMC, options to save items, and a list of similar articles.

Format: Abstract +

Send to =

Full text links

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Similar articles

Treatment of alopecia areata: "What is new on the horizon?" [Dermatol Ther. 2011]

Follow-up study of the first genome-wide association scan in alopecia areata [J Invest Dermatol. 2012]

Abstract The role of hair follicle immune privilege collapse in alopecia areata [J Invest Dermatol Symp Proc...]

ULBP3: a marker for alopecia areata incognita [Arch Dermatol Res. 2015]

Review Alopecia areata: autoimmunity—the evidence is in [J Invest Dermatol Symp Proc...]

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