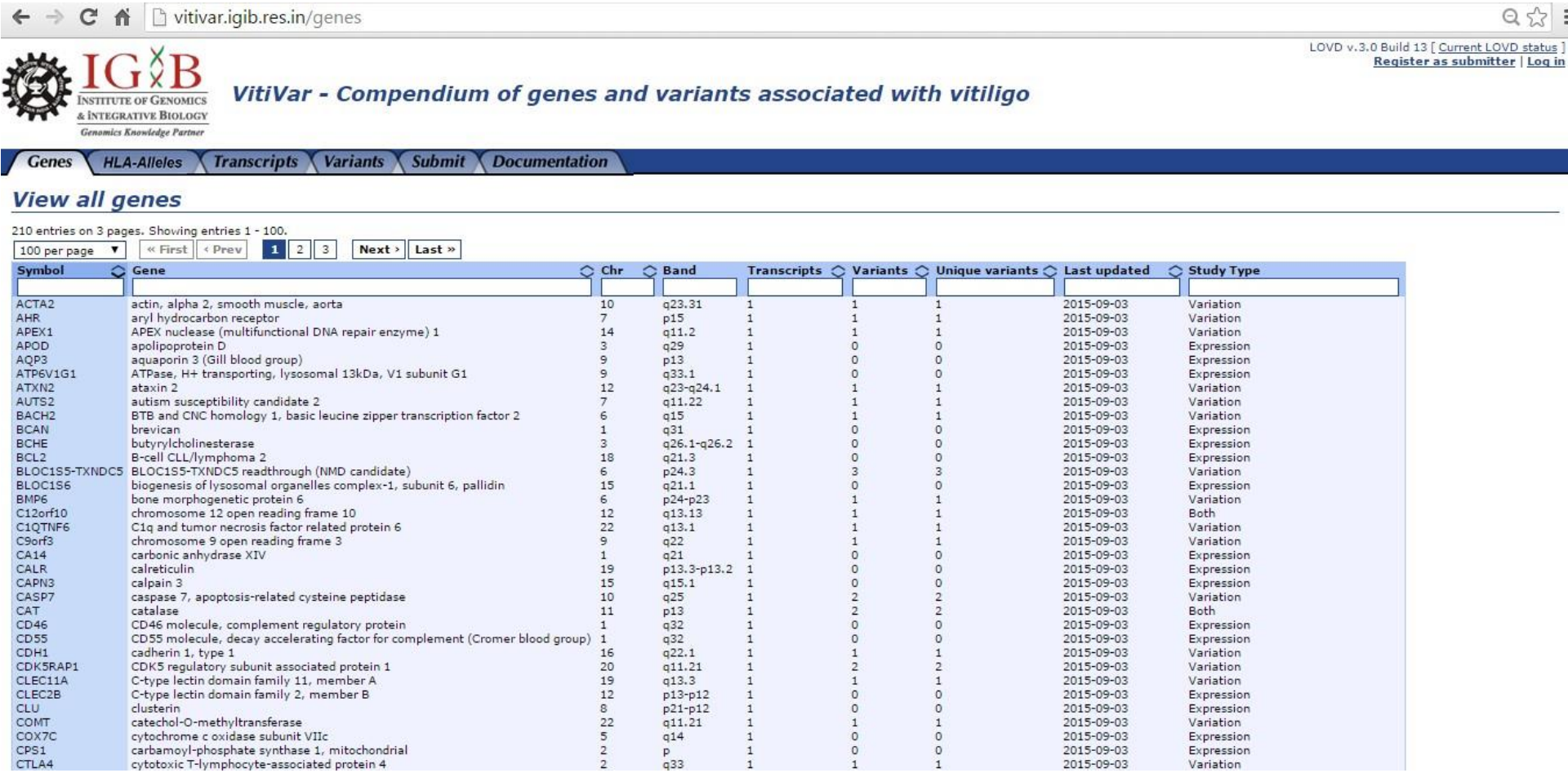
A quick guide to VitiVar

Home page of VitiVar
(http://vitivar.igib.res.in)



Symbol	Gene	Chr	Band	Transcripts	Variants	Unique variants	Last updated	Study Type
ACTA2	actin, alpha 2, smooth muscle, aorta	10	q23.31	1	1	1	2015-09-03	Variation
AHR	aryl hydrocarbon receptor	7	p15	1	1	1	2015-09-03	Variation
APEX1	APEX nuclease (multifunctional DNA repair enzyme) 1	14	q11.2	1	1	1	2015-09-03	Variation
APOD	apolipoprotein D	3	q29	1	0	0	2015-09-03	Expression
AQP3	aquaporin 3 (Gill blood group)	9	p13	1	0	0	2015-09-03	Expression
ATP6V1G1	ATPase, H+ transporting, lysosomal 13kDa, V1 subunit G1	9	q33.1	1	0	0	2015-09-03	Expression
ATXN2	ataxin 2	12	q23-q24.1	1	1	1	2015-09-03	Variation
AUTS2	autism susceptibility candidate 2	7	q11.22	1	1	1	2015-09-03	Variation
BACH2	BTB and CNC homology 1, basic leucine zipper transcription factor 2	6	q15	1	1	1	2015-09-03	Variation
BCAN	brevican	1	q31	1	0	0	2015-09-03	Expression
BCHE	butyrylcholinesterase	3	q26.1-q26.2	1	0	0	2015-09-03	Expression
BCL2	B-cell CLL/lymphoma 2	18	q21.3	1	0	0	2015-09-03	Expression
BLOC1S5-TXNDC5	BLOC1S5-TXNDC5 readthrough (NMD candidate)	6	p24.3	1	3	3	2015-09-03	Variation
BLOC1S6	biogenesis of lysosomal organelles complex-1, subunit 6, pallidin	15	q21.1	1	0	0	2015-09-03	Expression
BMP6	bone morphogenetic protein 6	6	p24-p23	1	1	1	2015-09-03	Variation
C12orf10	chromosome 12 open reading frame 10	12	q13.13	1	1	1	2015-09-03	Both
C1QTNF6	C1q and tumor necrosis factor related protein 6	22	q13.1	1	1	1	2015-09-03	Variation
C9orf3	chromosome 9 open reading frame 3	9	q22	1	1	1	2015-09-03	Variation
CA14	carbonic anhydrase XIV	1	q21	1	0	0	2015-09-03	Expression
CALR	calreticulin	19	p13.3-p13.2	1	0	0	2015-09-03	Expression
CAPN3	calpain 3	15	q15.1	1	0	0	2015-09-03	Expression
CASP7	caspase 7, apoptosis-related cysteine peptidase	10	q25	1	2	2	2015-09-03	Variation
CAT	catalase	11	p13	1	2	2	2015-09-03	Both
CD46	CD46 molecule, complement regulatory protein	1	q32	1	0	0	2015-09-03	Expression
CD55	CD55 molecule, decay accelerating factor for complement (Cromer blood group)	1	q32	1	0	0	2015-09-03	Expression
CDH1	cadherin 1, type 1	16	q22.1	1	1	1	2015-09-03	Variation
CDK5RAP1	CDK5 regulatory subunit associated protein 1	20	q11.21	1	2	2	2015-09-03	Variation
CLEC11A	C-type lectin domain family 11, member A	19	q13.3	1	1	1	2015-09-03	Variation
CLEC2B	C-type lectin domain family 2, member B	12	p13-p12	1	0	0	2015-09-03	Expression
CLU	clusterin	8	p21-p12	1	0	0	2015-09-03	Expression
COMT	catechol-O-methyltransferase	22	q11.21	1	1	1	2015-09-03	Variation
COX7C	cytochrome c oxidase subunit VIIc	5	q14	1	0	0	2015-09-03	Expression
CPS1	carbamoyl-phosphate synthase 1, mitochondrial	2	p	1	0	0	2015-09-03	Expression
CTLA4	cytotoxic T-lymphocyte-associated protein 4	2	q33	1	1	1	2015-09-03	Variation

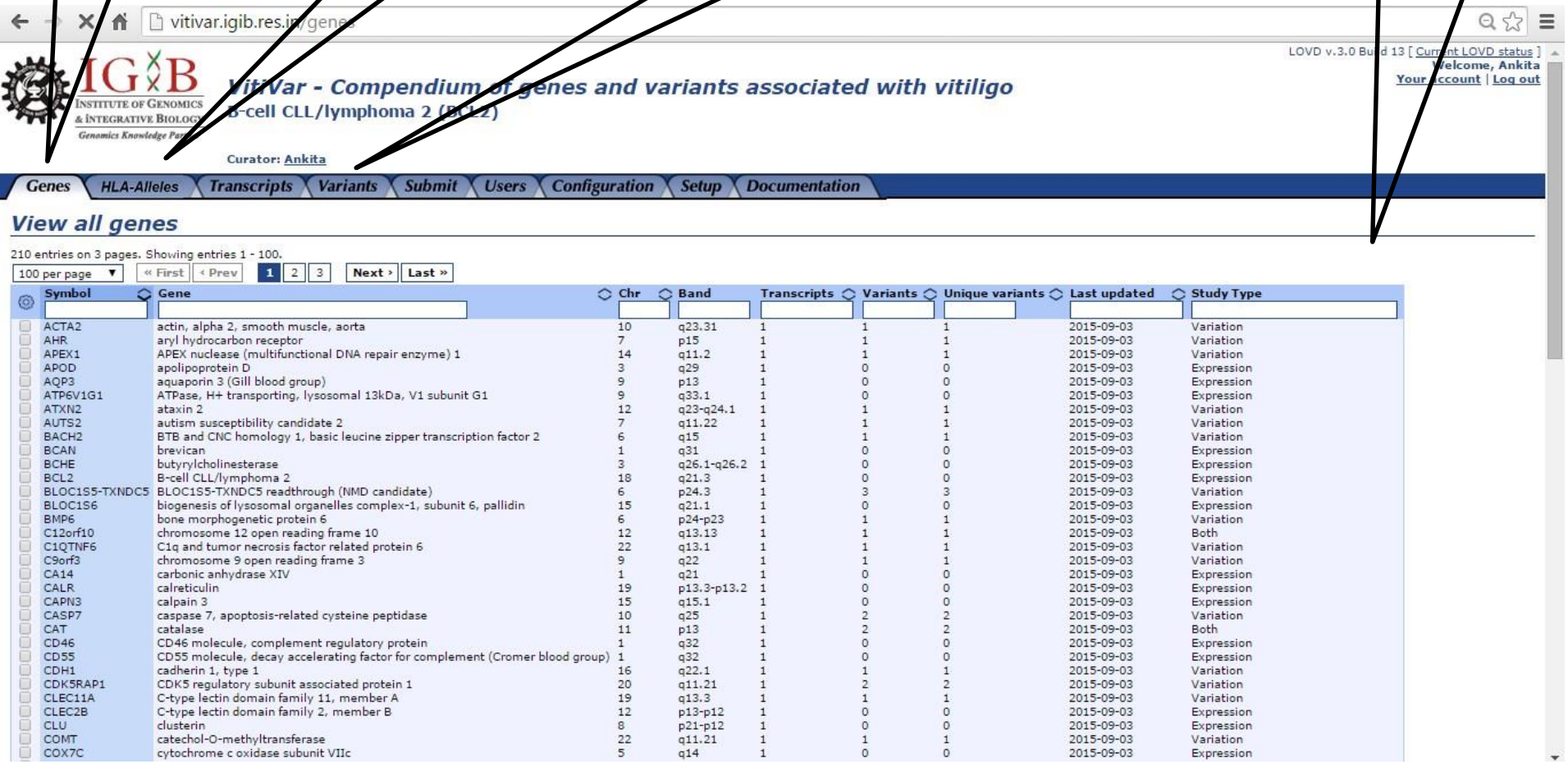
Description of fields on Home Page

List of manually curated genes found to be associated with vitiligo

HLA-alleles that are reported to be associated with vitiligo (Downloadable excel file).

Variants known to be associated with vitiligo

Genes categorized on the basis of the literature based information. Reported association either due to variation and/or expression



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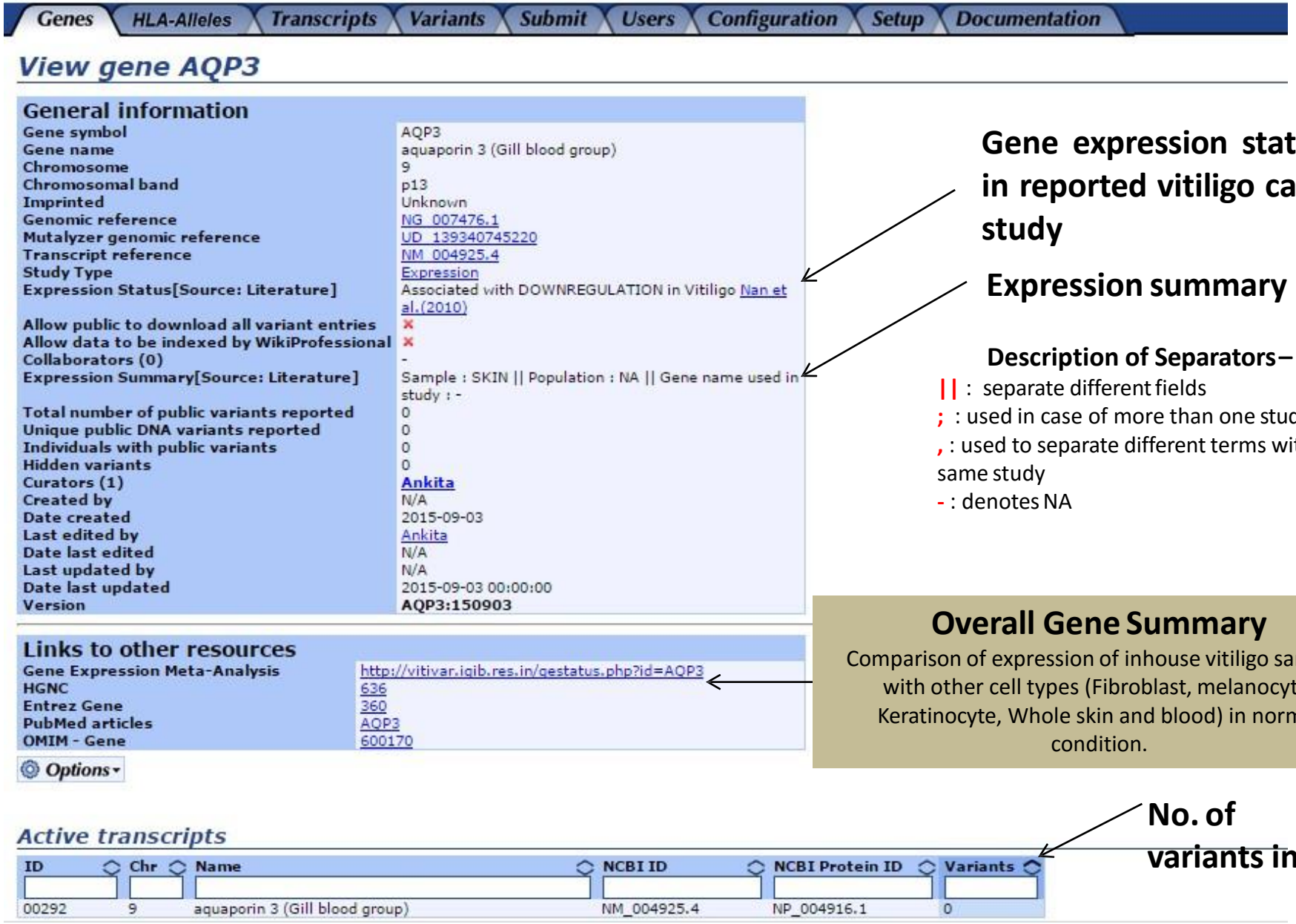
Description of Gene page (appears on clicking gene)



Gene expression status in reported vitiligo case study

Expression summary

Description of Separators–

|| : separate different fields

; : used in case of more than one study.

, : used to separate different terms within same study

- : denotes NA

Overall Gene Summary

Comparison of expression of inhouse vitiligo samples with other cell types (Fibroblast, melanocyte, Keratinocyte, Whole skin and blood) in normal condition.

No. of variants in gene

Description of Gene summary page

Gene Summary		
Gene Name	**Study Type**	**No. of Variants**
CAT	Both	1

Expression of gene from cataloged data (from literature based information)

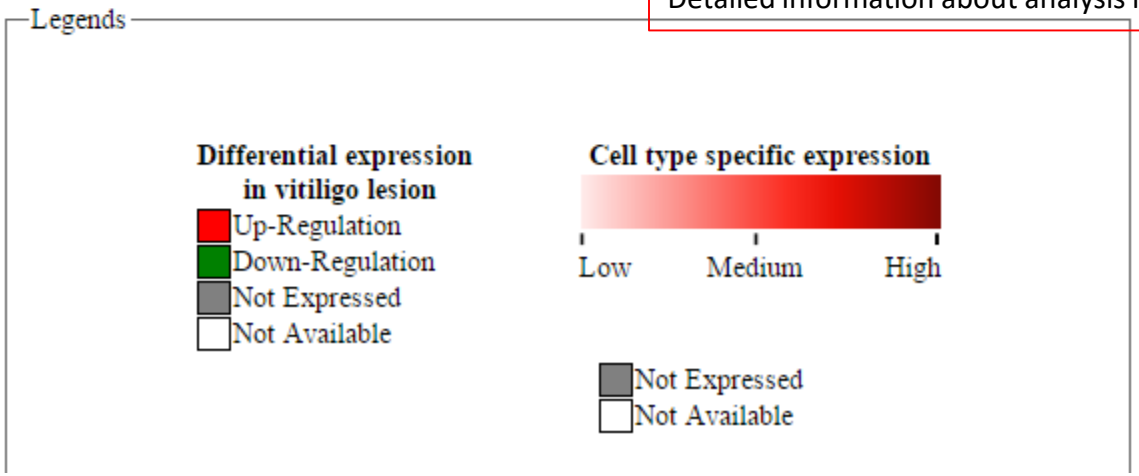
Expression Summary from Catalog Data				
Gene Name	**Expression Status**	**Sample**	**Population**	**PMID**
CAT	DOWNREGULATION	BLOOD;SKIN	NA;NA	{PMID:Ozel et al.(2013):24333267};{PMID:Sravani et al.(2009):19439879}

Meta Analysis of Expression Data

Differential expression in vitiligo lesion	Fibroblast	Keratinocyte	Melanocyte	Whole Skin	Blood

Meta-analysed gene expression data

Detailed information about analysis is provided in manuscript



Variants Tab

User can find description of fields by mouse hover on respective fields. For example:

Link of variants to 1000 genomes browser

Gene or Locus mapped in study	dbSNP ID	IGV	1000 Genomes	Population
DNAH5	rs2277046	NA		
DDX6 and CXCR5	rs638893	NA		
IL2RA	rs3134883	NA	1000G	HAN CHINESE
CD80	rs59374417	NA	1000G	EUROPEAN
CLNK	rs11940117	NA	1000G	EUROPEAN
CASP7	rs4353229	NA	1000G	EUROPEAN
TYR	rs11021232	NA	1000G	EUROPEAN
IKZF4	rs2456973	NA	1000G	EUROPEAN
SH2B3	rs4766578	NA	1000G	EUROPEAN
MC1R	rs4785587	NA	1000G	EUROPEAN
MC1R	rs9926296	NA	1000G	EUROPEAN
TOB2	rs4822024	NA	1000G	EUROPEAN
TOB2	rs79008	NA	1000G	EUROPEAN
HLA-C-HLA-B	rs9468925	NA	1000G	HAN CHINESE;HAN CHINESE;HAN CHINESE
HLA-A-HCG9	rs12206499	NA	1000G	EUROPEAN;EUROPEAN;EUROPEAN
PTPN22	rs2476601	NA	1000G	EUROPEAN;EUROPEAN;MULTIPLE POPULATIONS(META-ANALYSIS);MULTIPLE POPULATIONS(META-ANALYSIS);ROMANIAN;CAUCASIAN/EUROPEAN;MEXICAN
LPP	rs1464510	NA	1000G	EUROPEAN
IL2RA	rs706779	NA	1000G	EUROPEAN
UBASH3A	rs2839511	NA	1000G	EUROPEAN
RERE	rs301819	NA	1000G	EUROPEAN
TYR	rs1847134	NA	1000G	EUROPEAN
RERE	rs11121194	NA	1000G	EUROPEAN
HLA-G	rs2975033	NA	1000G	EUROPEAN
HLA-A-HCG9	rs3903160	NA	1000G	EUROPEAN
HLA-A-HCG9	rs6457110	NA	1000G	EUROPEAN
HCG9-HLA-J	rs4947244	NA	1000G	EUROPEAN

Frequency of variant in different Indian populations .Source : Indian Genome Variation Consortium.

Click over IGV link, User can visualize the frequency spectrum of variations in indian populations
Source : Indian Genome Variation Consortium (IGVC) data

SNP	Chromosome	Allele1	Allele2
rs4908760	1	A	G

Population	Frequency A1
AA-C-IP1	0.78
AA-E-IP2	0.80
DR-C-IP1	0.72
DR-S-IP1	0.50
DR-S-LP5	0.48
DR-S-LP7	0.40
DR-S-LP8	0.63
IE-E-IP1	0.47
IE-E-LP2	0.50
IE-E-LP4	0.50
IE-NE-IP1	0.70
IE-NE-LP1	0.50
IE-N-IP2	0.69
IE-N-LP18	0.50
IE-N-LP1	0.46
IE-N-LP5	0.36
IE-N-LP9	0.67

Description about Population names is provided in link (**Population Descriptions**) at the page end.

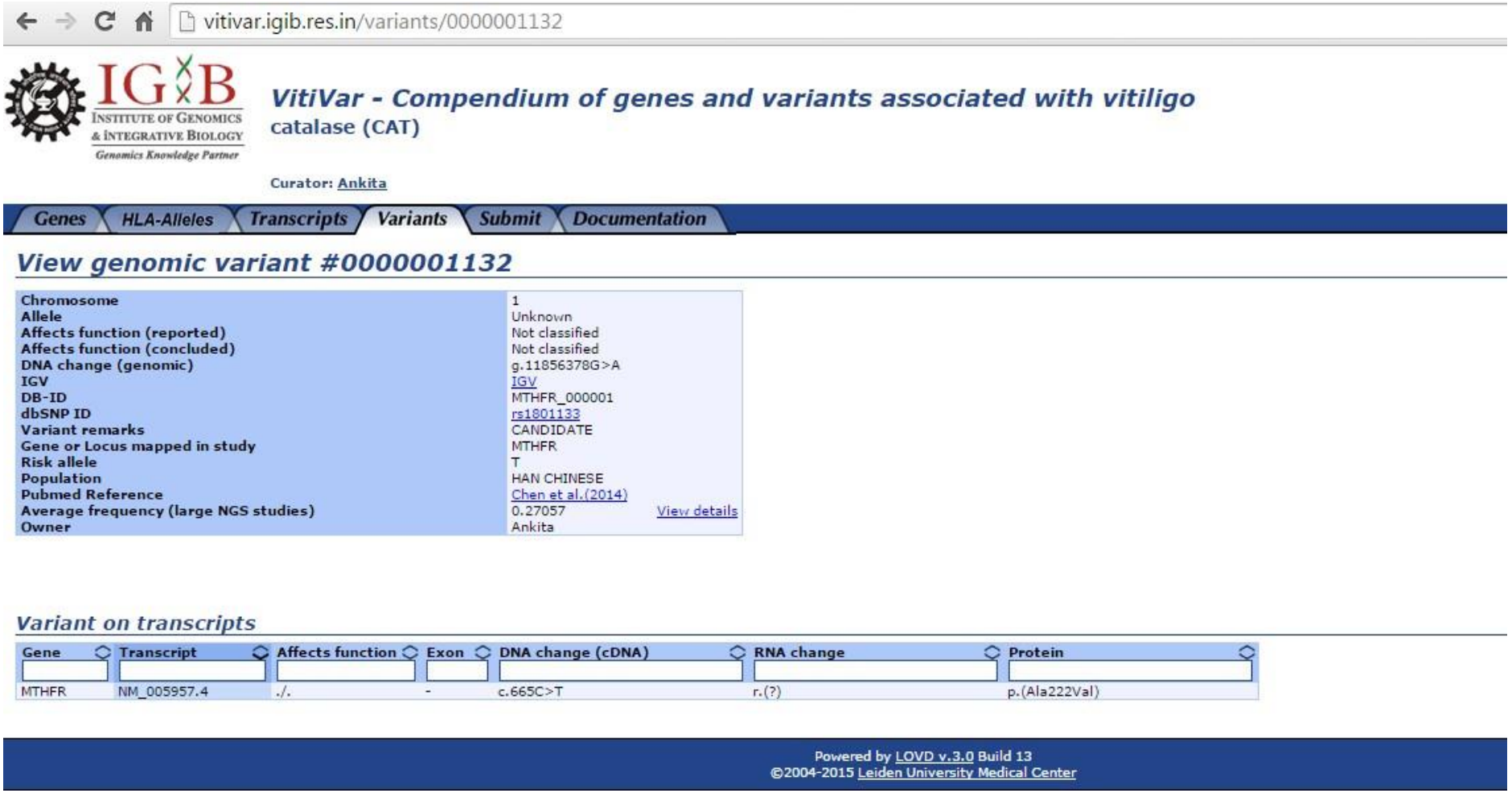
Scroll Right for more annotation fields

Risk allele	Population	Pubmed Reference	Owner
A	EUROPEAN	Jin et al.(2010)	Ankita
G	EUROPEAN	Jin et al.(2010)	Ankita
C	EUROPEAN	Jin et al.(2010)	Ankita
T	HAN CHINESE	Chen et al.(2014)	Ankita
A;T;T;T;T;T;T	EUROPEAN;EUROPEAN;MULTIPLE POPULATIONS(META-ANALYSIS);MULTIPLE POPULATIONS(META-ANALYSIS);ROMANIAN;CAUCASIAN/EUROPEAN;MEXICAN	Jin et al.(2010);CantÃ³n et al.(2005);Song et al.(2013);Zheng et al.(2012);Laberge et al.(2008);LaBerge et al.(2008);Garcia et al.(2014)	Ankita
G	HAN CHINESE	Li et al.(2009)	Ankita
-;G	SAUDI;NA	Abanmi et al.(2005);AydÄ±ngÃ¶z et al.(2015)	Ankita
-	INDIAN	Laddha et al.(2014)	Ankita
-	EUROPEAN	Jin et al.(2011)	Ankita
-	EUROPEAN	Jin et al.(2011)	Ankita
-	EUROPEAN	Birlea et al.(2010)	Ankita
-;-;G	INDIAN;EUROPEAN;ROMANIAN,CAUCASIAN	Dwivedi et al.(2011);Birlea et al.(2010);Birlea et al.(2009)	Ankita
G;G	EUROPEAN;INDIAN	Song et al.(2013);Dwivedi et al.(2011)	Ankita
-	KOREAN	Yun et al.(2010)	Ankita
-	KOREAN	Yun et al.(2010)	Ankita
-	KOREAN	Yun et al.(2010)	Ankita
G	EUROPEAN	Jin et al.(2010)	Ankita
-	EUROPEAN	Jin et al.(2011)	Ankita
-	EUROPEAN	Jin et al.(2011)	Ankita
T	HAN CHINESE	Jin et al.(2010)	Ankita
T	EUROPEAN	Quan et al.(2010)	Ankita
T	EUROPEAN	Jin et al.(2010)	Ankita
T	EUROPEAN	Jin et al.(2010)	Ankita
T	EUROPEAN	Jin et al.(2010)	Ankita
-	EUROPEAN	Jin et al.(2010)	Ankita

Description of all annotation tabs on Variants page

Column Header	Annotation
Variant_id	The id given to the variant by LOVD database
Chr	Chromosome # on which the variant is present
DNA change (genomic)	The nucleotide change for that variant
IGV	Link to frequency of variant in different Indian populations
1000 Genomes	Link of variants to 1000 genomes browser
DB-ID	Database id for that variant
dbSNP ID	dbSNP ID or rs_id for that variant
Variant remarks	Variant obtained from which study type: GWAS, Expression, Candidate etc.
Gene or Locus mapped in the study	Gene or locus for that particular variant as mentioned in thel iterature
Risk allele	Risk allele for that variant as reported in the literature (these are separated by ";" according to references mentioned in pubmed reference column and "NA" if risk allele is not clearly mentioned in study)
Population	Population in which study was conducted (these are also separated by ";" as per their pubmed reference and "NA" if it was not available)
Pubmed reference	The reference of the article in which the variant was reported to be associated with vitiligo

Variant information in Vitivar Database



More information about interpretations of variants in Vitivar

Variants in Vitivar are stored according to standard format defined by HGVS [Human Genome Variation Society)

For example,
g.8501786A>G it means A to G substitution at genomic position 8501786 [Genomic chage]
cDNA changes are also described in similar format only g is replaced by c

Effect of variant at RNA and Protein level are described in following formats –
RNA level :
Example -
unknown effect

r.? - *RNA has not been analysed, an effect is expected but difficult to predict (e.g. when the transcription start site, the polyA-addition signal or the polyA-addition site is changed)*

r.(?) - *RNA has not been analysed but an effect other then that caused by the change at DNA level is not expected*

r.(=) - *RNA has not been analysed, but no change is expected*

probable effect

r.(76a>u) - *RNA has not been analysed, the change expected is r.76a>u (can also be described as r.(?))*

Protein level:
unknown effect

p.? - protein has not been analysed, an effect is expected but difficult to predict

p.(=) - protein has not been analysed, but no change is expected

p.= - protein has not been analysed, RNA was, but no change is expected (***silent change***)

Probable effect (without any experimental evidence)
p.(Arg22Ser)

Detail description about interpretation of variant nomenclature at DNA, RNA and Protein levels, please visit -
http://www.hgvs.org/

Content Courtesy: from http://www.hgvs.org/

For any query related to vitivar, please contact :
ddash@igib.in, ankita@igib.in