

Curator Login

Curator Home  
(Dashboard)

Search Variant  
(Allele Registry)

Return Different  
Variant Expressions  
(Allele Registry)

Does this go  
here?  
Customize  
guidelines based  
on disease

Return Different  
Variant Expressions  
(Allele Registry)

Return grayed-out  
criteria description when  
non-computational or  
disease-dependent

Tools  
Search/Curate  
Variant

Search Variant  
Enter variant:

Return Variant

- unique ID
- known expressions
- HGNC, NCBI gene
- all associated diseases (OMIM, Orphanet)

[Curate this variant](#)

Dashboard will  
contain tools for  
curators, including  
ability to search on,  
and begin curating, a  
variant

First step in variant  
curation will be to search  
for a variant using one of  
many expressions. Allele  
Registry required at this  
step

Need Allele Registry

Allele Registry will  
return unique ID for  
variant and all known  
expressions of variant  
(HGVS, etc.)

Return criteria predictions for  
criteria where computational  
information for variant (Allele  
frequency, Computational &  
Predictive Data); populate  
correct cells

Need to define information  
resources, how information  
returned and versioned, how  
to determine whether criteria  
met

# Variant Expression(s)

	Benign			Pathogenic		
	Strong	Supporting	Supporting	Moderate	Strong	Very Strong
<b>Population Data</b>	High frequency in large population groups (e.g. 1000 Genomes, dbSNP, HapMap, etc.)	High frequency in large population groups (e.g. 1000 Genomes, dbSNP, HapMap, etc.)	Low frequency in large population groups (e.g. 1000 Genomes, dbSNP, HapMap, etc.)	Low frequency in large population groups (e.g. 1000 Genomes, dbSNP, HapMap, etc.)	Low frequency in large population groups (e.g. 1000 Genomes, dbSNP, HapMap, etc.)	Low frequency in large population groups (e.g. 1000 Genomes, dbSNP, HapMap, etc.)
<b>Computational Annotation Data</b>	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)
<b>Functional Data</b>	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)
<b>Segregation Data</b>	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)
<b>De novo data</b>	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)
<b>Allelic data</b>	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)
<b>Other databases</b>	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)	Indicates absence of deleteriousness (e.g. no predicted impact on protein structure, no predicted impact on gene expression, etc.)
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