Query another mutation

Documentation

OpenCRAVAT Variant Report

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- Non-Coding Annotations
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Variant Information

	Variant Type	Genomic location	Variant consequence	dbSNP ID	UniProt Accession Number
POMT1	single nucleotide variant (C>T)	chr9:131510048 (genome build GRCh38)	nonsynonymous (intron variant)	No dbSNP ID is available	Q9Y6A1

Allele Frequency

Variant Allele Frequencies in global populations

gnomADv3 Allele Frequency: No annota

1000 Genomes Allele Frequency

Clinical Relevance

Clinical significance and conditions associated with this variant in ClinVar

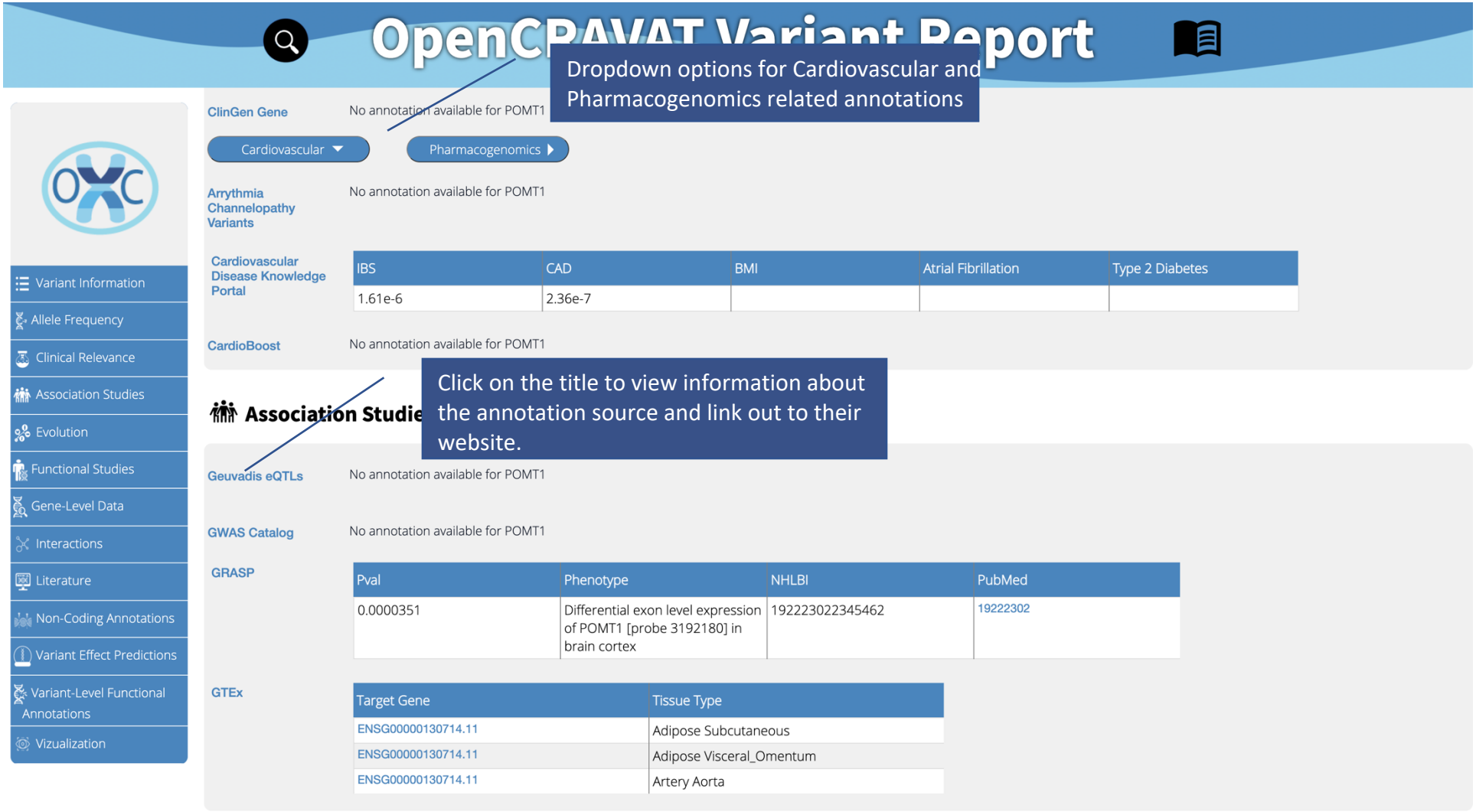
ClinVar Significance: Benign/Likely benign (ID: 95466)

ClinVar Conditions: Congenital muscular dystrophy-dystroglycanopathy with mental retardation, type B1 | Limb-girdle muscular dystrophy-dystroglycanopathy, type C1 | Limb-girdle muscular dystrophy-dystroglycanopathy, type C1 | Walker-Warburg congenital muscular dystrophy

Denovo-DB: No annotation available for POMT1

OMIM: 609308 613155

Sidebar for navigation to all sections in the variant report


OpenCRAVAT Variant Report
Dropdown options for Cardiovascular and Pharmacogenomics related annotations
Click on the title to view information about the annotation source and link out to their website.
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ClinGen Gene
No annotation available for POMT1
Cardiovascular
Pharmacogenomics
Arrythmia Channelopathy Variants
No annotation available for POMT1
Cardiovascular Disease Knowledge Portal
IBS
CAD
BMI
Atrial Fibrillation
Type 2 Diabetes
1.61e-6
2.36e-7
CardioBoost
No annotation available for POMT1
Association Studie
Geuvadis eQTLs
No annotation available for POMT1
GWAS Catalog
No annotation available for POMT1
GRASP
Pval
Phenotype
NHLBI
PubMed
0.0000351
Differential exon level expression of POMT1 [probe 3192180] in brain cortex
192223022345462
19222302
GTEx
Target Gene
Tissue Type
ENSG00000130714.11
Adipose Subcutaneous
ENSG00000130714.11
Adipose Visceral_Omentum
ENSG00000130714.11
Artery Aorta

Evolution

GERP++

GERP++
0.0932 rankscore
-1.10 score

GERP++ Neutral Rate
2.20

GHIS

GHIS Score
0.563

RVIS

Score	Percentile Rank	ExAC-based RVIS	ExAC-based Percentile	FDR p-value
-0.300	-0.460	32.3	23.4	0.297

ALoFT

Transcript	Transcripts Affected	Tolerated Probability	Recessive Probability	Dominant Probability	Classification	Confidence
ENST00000372228	some	0.44185	0.5133	0.04485	Recessive	High Confidence

LINSIGHT

No annotation available for POMT1

Phast Cons

Vertebrate
0.0639 rankscore
0 score

Mammalian
0.0192 rankscore
0 score

Conservation scores and rank scores, found in Phast Cons and PhyloP.

PhyloP

Vertebrate
0.119 rankscore

Mammalian
0.0139 rankscore

Primate
0.134 rankscore

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Functional Studies

MaveDB No annotation available for POMT1

Gene-Level Data

This section explores gene level annotations including gene description from NCBI, scores from gnomAD gene, and more.

NCBI Gene

The protein encoded by this gene is an O-mannosyltransferase that requires interaction with the product of the POMT2 gene for enzymatic function. The encoded protein is found in the membrane of the endoplasmic reticulum. Defects in this gene are a cause of Walker-Warburg syndrome (WWS) and limb-girdle muscular dystrophy type 2K (LGMD2K). Several transcript variants encoding different isoforms have been found for this gene.

Essential Genes

Essential	CRISPR	CRISPR2	Gene Trap	Indespensibility Score	Indespensibility Prediction
Essential	Non-essential phenotype-changing	Non-essential phenotype-changing	Non-essential phenotype-changing	0.473	Loss-of-function tolerant

gnomAD Gene

Transcript	Obv/Exp LoF	Obv/Exp Mis	Obv/Exp Syn	LoF Z-Score	Mis Z-Score	Syn Z-Score	pLI	pRec	pNull
ENST00000341012	0.71237	0.90461	1.0704	1.7396	0.67678	-0.70886	7.6698e-16	0.58851	0.41149
ENST00000354713	0.73499	0.89564	1.0941	1.655	0.7548	-0.96317	1.0178e-17	0.38789	0.61211
ENST00000372220	0.56598	0.88619	1.1278	1.8625	0.58517	-0.95165	0.0000038787	0.95208	0.047914
ENST00000372228	0.71345	0.89738	1.0697	1.8163	0.7701	-0.73855	2.4725e-17	0.56752	0.43248

Gene ontology

Biological Process	Cellular Component	Molecular Function
protein O-linked mannosylation	endoplasmic reticulum membrane	dolichyl-phosphate-mannose-protein mannosyltransferase activity

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P(rec)

Score 0.989

Known Status recessive

Interpro

No annotation available for EGFR M1?

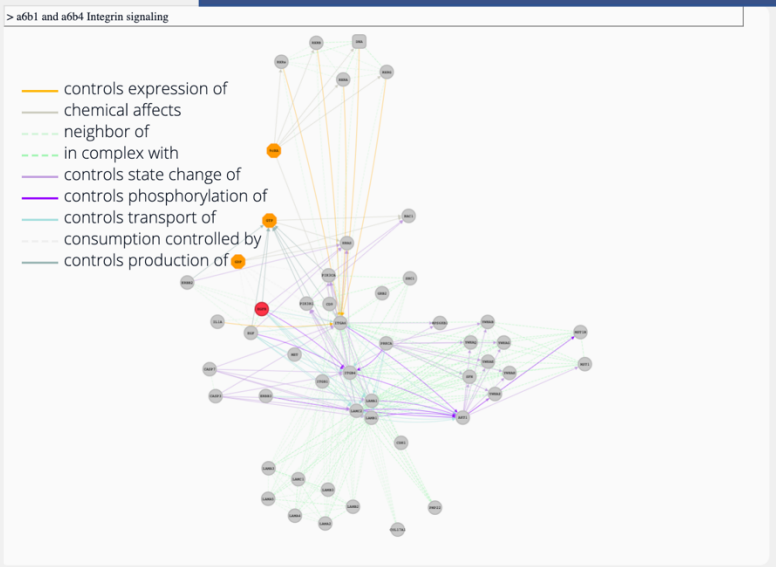
Interactions

Network visualization from NDEx. Click the buttons of the left side to switch the type of NDEx that is viewed.

NDEx NCI Cancer Pathways

NDEx Congenital Heart Disease

NDEx SIGNOR



List of genes that interact with partner gene, from BioGRID and IntAct.

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BioGRID ID 116446

BioGRID

AAR2 ACTN1 ALYREF ANXA7 AR ARF1 ATP5PF BABAM1 BAG1 BAX BBS1 BCL2L1 BCLAF1 BOLA2 BOLA2B CASP8 CCT8 CDH1 CFTR CHD4
CHTOP CLCF1 COPE CPSF6 CSN1S1 CTNNB1 DAXX DBN1 DKC1 DLD DNM1L DUSP1 DUSP12 DUSP19 DUSP2 DUSP28 DUSP4 DUSP6 DUSP9
EEF1G EEF2 EFCAB6 EFTUD2 EGLN3 EIF3B ENO1 EPB41L5 FADD GATAD2B GEMIN4 GOPC H2AC4 H2AC8 HADHA HDAC1 HDAC2 HDGFRP2
HINT1 HIPK1 HNRNPA1L2 HNRNPDL HRNR HSD17B10 HSPA4 HSPA5 ILF2 IMPA2 IMPDH2 ITPA LDHA LDHB LIMA1 LYPLA1 MAP1B MAP3K5
MAPKAPK5 MBD3 MCM2 MDH2 MIF MRPS18B MRPS9 MT-ND1 MTA1 MTA2 MTHFD1 MTRF1 MYCBP MYH11 MYL12A NDUFA4 NDUFS3 NIT2
NONO NPM1 NXF1 OAT OTUD7B PABPC1 PARK7 PDHA1 PDHB PDIA6 PHOSPHO1 PIAS2 PIH1D1 PINK1 PIR PITX3 PML PNN PPARGC1A PPIA
PPM1B PPM1D PPP2R1A PPP2R1B PPTC7 PRDX2 PRDX3 PRDX5 PRDX6 PRKN PRPF4 PRPF8 PTEN RAB1A RBBP4 RBBP7 RNPS1 SAP18 SERBP1
SFPQ SLC18A2 SNCA SOD1 SPINDOC SQSTM1 SREBF2 SSBP1 STUB1 SUMO1 TAB1 TALDO1 TDP2 THRAP3 TMOD2 TMOD3 TP53 TPI1 TPM1
TPM3 TRAF6 TRAP1 TRIM28 TXN U2AF2 UBA2 UBE2I UBE2M UNK USP19 VHL WBP11 YWHAB YWHAQ YWHAZ

IntAct

ABL1 ACTN1 ALYREF AR ARF1 BABAM1 BBS1 BCLAF1 BOLA2 CCT8 CHD2 CHTOP CPSF6 DAXX DBN1 DKC1 DLD DNM1L EEF1G EEF2 EIF3B
EPB41L5 FADD GATAD2B GEMIN4 GOPC HADHA HDAC1 HDAC2 HDGFL2 HIST1H2AE HNRNPA1L2 HNRNPDL HRNR HSD17B10 HSP90AA5P ILF2
IMPDH2 IQGAP1 KIF1B LIMA1 LIN37 LIN54 MBD3 MDH2 MRPS18B MRPS9 MTA1 MTA2 MTHFD1 MYCBP MYH11 MYL12A OAT OTUB1 OTUD7B
PABPC1 PARK7 PDHA1 PDHB PDIA6 PHOSPHO1 PNN POTEKP PPIA PPM1B PPM1D PPP1R12A PPP2R1A PPP2R1B PPTC7 PRDX3 PRDX6 PRPF4
PTBP1 PYCR1 RAB1A RACK1 RBBP4 RBBP7 RNPS1 SAP18 SERBP1 SLX4 SMARCB1 SNCA SOD1 SPINDOC SSBP1 TAB1 THRAP3 TMOD2 TMOD3
TNFRSF10A TPI1 TPM1 TPM3 TRAP1 TRIM28 TXN UBA2 UBE2I USP19 VHL WBP11 YWHAB YWHAQ YWHAZ

Literature

LitVar No annotation available for PARK7 A39S

Non-Coding Annotations

This section includes information about non coding variants, including scores from Segway, GeneHancer target genes, and more.

Non-Coding Annotations

Candidate cis_Regulatory Elements by ENCODE (SCREEN)

Classification	CTCF Bound	cCRE Accession ID
pELS	Yes	EH38E1324288

ENCODE TFBS

No annotation available for UBR4

GeneHancer Type

Enhancer

GeneHancer

Target Gene	Link Strength
ENSG00000272084	561.28
EMC1	30.99
UBR4	4.41
LOC105376815	0.34

Promoter IR Regions

No annotation available for UBR4

VISTA Enhancer Browser

No annotation available for UBR4

Ensembl Regulatory Build

promoter_flanking_region

ENSR00000920648

Trinity CTAT RNA Editing Database

No annotation available for UBR4

Segway

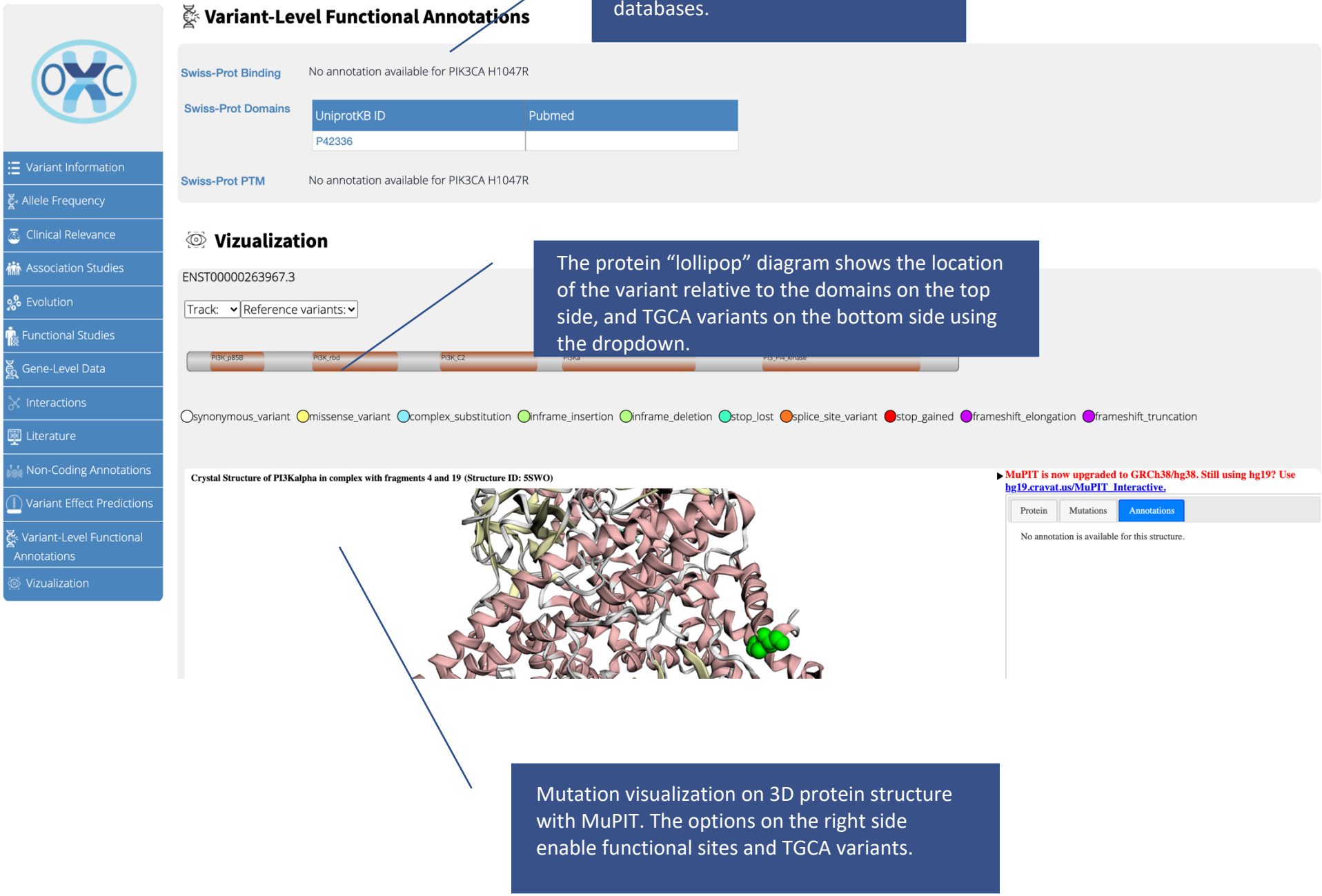
Sum Score	Mean Score
56.4	0.0176

Variant Effect Prediction

Compares damaging vs. tolerated predictions of the variant, from multiple annotations.

DANN		coding score	0.936	coding rankscore	0.235
FATHMM	No annotation available for EGFR M1?				
FATHMM MKL	prediction: Damaging	coding score	0.708	coding rankscore	0.348
FATHMM XF	prediction: Neutral	coding score	0.0141	coding rankscore	2.09e-3
LRT	prediction: Neutral	coding score	0.129	coding rankscore	0.187
MetaLR	prediction: Tolerated	coding score	0.272	rankscore	0.644
MetaSVM	prediction: Tolerated	score	-0.779	rankscore	0.564

Table that illustrates variant effect prediction, including a score, rank score, and prediction from many sources.


Information from multiple Swiss-Prot databases.
Variant-Level Functional Annotations
Swiss-Prot Binding
No annotation available for PIK3CA H1047R
Swiss-Prot Domains
UniprotKB ID
Pubmed
P42336
Swiss-Prot PTM
No annotation available for PIK3CA H1047R
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Vizualization
ENST00000263967.3
Track:
Reference variants:
The protein “lollipop” diagram shows the location of the variant relative to the domains on the top side, and TGCA variants on the bottom side using the dropdown.
PI3K_p85B
PI3K_rbd
PI3K_C2
synonymous_variant
missense_variant
complex_substitution
inframe_insertion
inframe_deletion
stop_lost
splice_site_variant
stop_gained
frameshift_elongation
frameshift_truncation
Crystal Structure of PI3Kalpha in complex with fragments 4 and 19 (Structure ID: 5SWO)
MuPIT is now upgraded to GRCh38/hg38. Still using hg19? Use hg19.cravat.us/MuPIT_Interactive.
Protein
Mutations
Annotations
No annotation is available for this structure.
Mutation visualization on 3D protein structure with MuPIT. The options on the right side enable functional sites and TGCA variants.