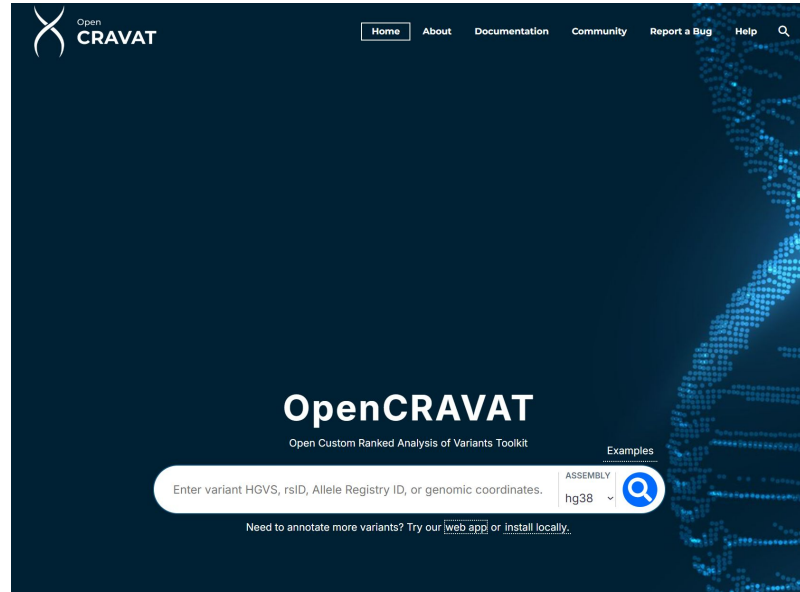


# **Tutorial to analyze and interactively visualize a VCF file using free web-based tools**

**Gurpreet Kaur, Ph.D.**

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# 1. OpenCRAVAT Web-app



<https://www.opencravat.org/>  
<https://github.com/KarchinLab/open-cravat>

JOBS

STORE

SINGLE VARIANT

User Survey

Open

CRAVAT

gurpreet

Varsiants

Genome:  
hg38/GRCh38

Add input files

OR

```
##fileformat=VCFv4.2
##FILTER=<ID=PASS,Description="All filters passed">
##contig=<ID=chr1,length=248956422>
##contig=<ID=chr2,length=242193529>
##contig=<ID=chr3,length=196295559>
##contig=<ID=chr4,length=190214555>
##contig=<ID=chr5,length=181538259>
##contig=<ID=chr6,length=170805979>
```

Example:  
VCF TSV HGVS ClinGen dbSNP CLEAR

Note:  
Enter a note for the analysis (optional)

Annotations

Packages

Categories

Variant Effect Prediction  
Allele Frequency  
Regulation  
Variants  
Evolution  
Visualization  
Cardiovascular  
Functional Studies  
Gwas

Show All None Selected

Cancer  
Genes  
Non Coding  
Proteins  
Mendelian Disease  
Networks  
Literature  
Drugs

☒ ☐

Additional Analysis

Case-Control cohorts ⓘ

No file selected

Job ID

Input File

Variants

Annotators

Genome

Note

Status

Download

Refresh Table

Import Job

1. Select example VCF

←

Link to download example VCF file:  
<https://github.com/KarchinLab/open-cravat/blob/f3e242e671d88486221a32d060e5bb9c487d9a1c/cravat/websubmit/input-examples/vcf.hg38.txt>

<https://run.opencravat.org/submit/nocache/index.html>

ANNOTATE

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**Annotations** **Packages**

Categories Show All None Selected

Variant Effect Prediction Cancer

Allele Frequency Genes

Regulation Non Coding

Variants Proteins

Evolution Mendelian Disease

Visualization Networks

Cardiovascular Literature

Functional Studies Drugs

Gwas

☒ ☒

+ 1000 Genomes

+ ALFA Allele Frequenc...

✓ ALFT

✓ Arrhythmia Channelo...

✓ BioGRID

✓ BioP

✓ Cancer Gene Census

✓ Cancer Genome Inter...

✓ Candidate os Regulat...

✓ CCR Constrained Co...

✓ CGD Clinical Genomi...

✓ CIVIC

✓ ClinGen Gene

✓ ClinVar

✓ COSMIC

✓ CScape

✓ DANN

✓ dbCID: Database of C...

✓ dbSNP

✓ Denovo-DB

✓ DIDA: Digenic Disease...

✓ ENCODE TFBS

✓ ESMb

✓ Essential Genes

✓ ExAC Gene and CNV

✓ FATHMM MSL

✓ FATHMM XF Coding

✓ Flanking Sequence

✓ Gene Ontology

✓ GenoCanyon

✓ Geuvadis eQTLs

✓ gMVP

✓ gnomAD

✓ GTEx eQTLs

✓ HaploReg

✓ HCP

✓ IntAct

✓ Likelihood Ratio Test

✓ LiVar

✓ LoFtool

✓ MetaLR

✓ MetaSVM

✓ MISTIC (Missense de...

✓ MuPT

✓ MutationTaster

✓ MultiPred

✓ NCBI Gene

✓ ncRNA

✓ ABRaOM

✓ All of Us

✓ AlphaMissense

✓ BayesDel

✓ BRCA1 Saturation Ge...

✓ CADD Exome

✓ Cancer Gene Landsc...

✓ Cancer Hotspots

✓ CardioBoost

✓ CEDAR

✓ CHASPlus

✓ CIVIC Gene

✓ ClinPred

✓ ClinVar ACMC

✓ COSMIC Gene

✓ CScape Coding

✓ DANN Coding

✓ dbSNV

✓ dbSNP Common

✓ DGkb: The Drug Inter...

✓ DITTO

✓ Ensembl Regulatory B...

✓ ESP6500

✓ EVE

✓ FATHMM

✓ FATHMM XF

✓ fitCons

✓ FunSeq2

✓ GeneLancer

✓ GERP+

✓ GHIS

✓ gnomAD Gene

✓ GRASP

✓ GWAS Catalog

✓ hg19 coordinates

✓ Human Phenotype On...

✓ InterPro

✓ LINSIGHT

✓ LiVar Full

✓ MaveDB

✓ MetaRNN

✓ miRBase

✓ MITOMAP

✓ Mutation Assessor

✓ Multipanning

✓ MultiPred

✓ MultiPred Indel

✓ ncER: non-coding ess...

✓ NDEx

← **2. Select Annotator (I selected all)**

JOBS STORE SINGLE VARIANT User Survey

✓ CIVIC

✓ ClinGen Gene

✓ ClinVar

✓ COSMIC

✓ CScape

✓ DANN

✓ dbCID: Database of C...

✓ dbSNP

✓ Denovo-DB

✓ DIDA: Digenic Disease...

✓ ENCODE TFBS

✓ ESMb

✓ Essential Genes

✓ ExAC Gene and CNV

✓ FATHMM MSL

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✓ Likelihood Ratio Test

✓ LiVar

✓ LoFtool

✓ MetaLR

✓ MetaSVM

✓ MISTIC (Missense de...

✓ MuPT

✓ MutationTaster

✓ MultiPred

✓ NCBI Gene

✓ ncRNA

✓ OMIM

✓ p(h)

✓ PangaloOB

✓ Phast Cons

✓ PhyloP

✓ PrimateAI

✓ PROVEAN: Protein Va...

✓ PubMed

✓ RegulomeDB

✓ REVEL

✓ Segway

✓ SIFT

✓ SIFTy

✓ Swiss-Prot Binding

✓ Swiss-Prot PTM

✓ Trinity CTRF

✓ UK10K Cohorts

✓ Uniprot Domain

✓ VEST4

✓ CIVIC Gene

✓ ClinPred

✓ ClinVar ACMC

✓ COSMIC Gene

✓ CScape Coding

✓ DANN Coding

✓ dbSNV

✓ dbSNP Common

✓ DGkb: The Drug Inter...

✓ DITTO

✓ Ensembl Regulatory B...

✓ ESP6500

✓ EVE

✓ FATHMM

✓ FATHMM XF

✓ fitCons

✓ FunSeq2

✓ GeneLancer

✓ GERP+

✓ GHIS

✓ gnomAD Gene

✓ GRASP

✓ GWAS Catalog

✓ hg19 coordinates

✓ Human Phenotype On...

✓ InterPro

✓ LINSIGHT

✓ LiVar Full

✓ MaveDB

✓ MetaRNN

✓ miRBase

✓ MITOMAP

✓ Mutation Assessor

✓ Multipanning

✓ MultiPred

✓ MultiPred Indel

✓ ncER: non-coding ess...

✓ NDEx

✓ OncoKB

✓ P(re)

✓ PharmGKB

✓ Phd SNPg

✓ PolyPhen-2

✓ Promoter IR

✓ Pseudogene

✓ Regeneron

✓ Repeat Sequences

✓ RVS

✓ SIFT

✓ SpliceAI

✓ Swiss-Prot Domains

✓ TARGET

✓ UCSC Genome Brows...

✓ Uniprot

✓ VARIETY\_LR

✓ VISTA Enhancer Brow...

**Additional Analysis**

Case-Control cohorts

No file selected

**ANNOTATE**

**3. Click ANNOTATE to submit job**

JOB

STORE

SINGLE VARIANT

User Survey

Genome

hg38/GRCh38

Add input files

OR

Enter variants in a supported input format such as VCF or TSV, or type a list of the paths to input files in the server

Example:

VCF

TSV

HGVs

ClinGen

dbSNP

CLEAR

Note:

Enter a note for the analysis (optional)

Annotations

Packages

Categories

Show

All

None

Selected

Variant Effect Prediction

Cancer

Allele Frequency

Genes

Regulation

Non Coding

Variants

Proteins

Evolution

Mendelian Disease

Visualization

Networks

Cardiovascular

Literature

Functional Studies

Drugs

Gwas

☒

☐

Additional Analysis

Case-Control cohorts

No file selected

ANNOTATE

Open CRAVAT

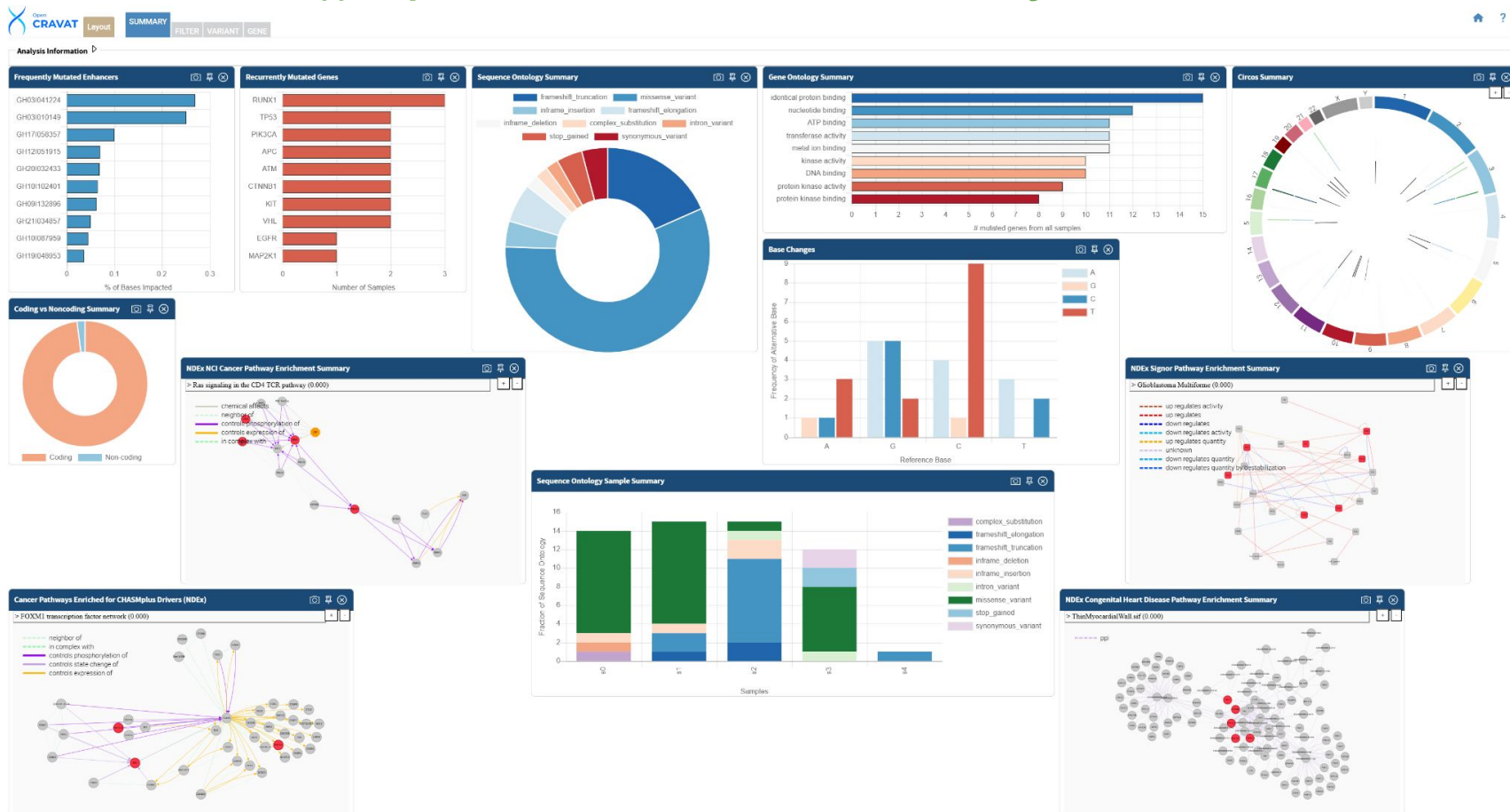
gurpreet

Job ID	Input File	Variants	Annotators	Genome	Note	Status	Download
250906-153841	input	49	181	hg38		Open Result Viewer	DB Log Errors

Refresh Table Import Job

4. Click Open Result Viewer under Status Tab

# 5. Result Viewer: (i) Explore **SUMMARY** tab to interactively visualize annotated variants



## 5. Result Viewer: (ii) Explore FILTER tab for various filtering options



LayoutSUMMARYFILTERVARIANTGENE

▶ Samples

▼ Genes

Type a list of gene names to include. One per line. Or, load a gene list from a file.  
Gene names are case-sensitive.

Choose FileNo file chosen

Clear

▶ Sample Properties

▶ Variant Properties

49/49 variants 

Apply filter  

## 5. Result Viewer: (iii) Explore **VARIANT** tab for detailed variant table and export results

**CRAVAT** **Layout** **SUMMARY** **FILTER** **VARIANT** **GENE** View table Home Help Download

Variant Annotation										1000 Genomes	1000 Genomes sAd Mixed American	1000 Genomes sAfrican	1000 Genomes sEast Asian	1000 Genomes sEuropean	1000 Genomes sSouth Asian	ABRADM	ALFA Allele Frequency Aggregator	ALFA Allele Frequency Aggregator African	ALFA Allele Frequency Aggregator Asian	ALFA Allele Frequency Aggregator European	ALFA Allele Frequency Aggregator Latin American	ALFA Allele Frequency Aggregator Others	All of Us										AL			
Chrom	Position	Ref Base	Alt Base	Variant Note	Coding	Gene	Transcript	Sequence Ontology	Exon Number	cDNA change	Protein Change	Samples	AF	CLM AF	ASW AF	CDX AF	GBR AF	BEB AF	AF	Global AF	African American AF	East Asian AF	European AF	Latin American AF	Others AF	All AF	Max AF	Max Sub-Population	African AF	Ad-Mixed American AF	East Asian AF	European AF	Middle Eastern AF	Other AF	South Asian AF	Dominant Probability
chr1	6197732	T	-		Yes	RPL22	ENST00000234075.9	frameshift, truncation	2	c.44del	p.Lys15_...s152del																									
chr2	147926124	A	-		Yes	ACVR2A	ENST00000241416.12	frameshift, truncation	10	c.1310del	p.Lys443_...																									
chr3	10149836	G	C		Yes	VHL	ENST00000256474.3	missense_variant	3	c.513G>C	p.Lys17_...																									
chr3	10149886	T	A		Yes	VHL	ENST00000256474.3	missense_variant	3	c.563T>A	p.Leu1_...																									
chr3	30650389	A	-		Yes	TCF8R2	ENST00000298754.10	frameshift, truncation	3	c.383del	p.Lys12_...																									
chr3	41224371	C	T		Yes	CTNAB1	ENST00000249496.11	missense_variant	3	c.395C>T	p.Ala20_...																									
chr3	41224610	C	A		Yes	CTNAB1	ENST00000249496.11	missense_variant	3	c.398C>A	p.Ser33_...																									
chr3	179191156	A	G		Yes	PIK3CA	ENST00000263967.4	missense_variant	2	c.331A>G	p.Lys11_...																									
chr3	179218794	G	A		Yes	PIK3CA	ENST00000263967.4	missense_variant	10	c.1674G>A	p.Glu54_...																									
chr3	179234296	C	T		Yes	PIK3CA	ENST00000263967.4	missense_variant	21	c.3139C>T	p.His10_...																									
chr4	54726020	-	GCCT...		Yes	KIT	ENST00000288135.6	inframe_insertion	9	c.1504_15...	p.Ala50_...																									
chr4	54733174	T	A		Yes	KIT	ENST00000288135.6	missense_variant	17	c.2465T>A	p.Asn6_...																									
chr5	112839543	G	C		Yes	APC	ENST00000257430.9	missense_variant	15	c.3949G>C	p.Glu13_...		0.003	0.0106	0	0	0.0165	0		0.0057	0.0076	0	0.006	0	0.0043	0.0065	0.0076	mid	0.0064	0.0044	9.300e-5	0.0075	0.0076	0.0055	0.0017	
chr5	112840261	-	A		Yes	APC	ENST00000257430.9	frameshift, inframe_deletion	16	c.4665dup	p.Thr1_...																									
chr5	171410544	-	TCTG		Yes	NFMI	ENST00000294930.10	frameshift, inframe_deletion	11	c.860_863...	p.Trp28_...																									
chr7	55174015	G	C		Yes	EGFR	ENST00000275493.7	missense_variant	18	c.2156G>C	p.Gly71_...																									
chr7	55174772	GGAA...	-		Yes	EGFR	ENST00000275493.7	inframe_deletion	19	c.2230_22...	p.Glu74_...																									
chr7	140753336	AC	TT		Yes	BRAF	ENST00000264499.2	complex_substitution	15	c.1918A>G	p.Val64_...																									
chr9	5079742	G	C		Yes	JAK2	ENST00000281652.4	missense_variant	14	c.1821G>C	p.Lys60_...																									
chr9	21979002	C	A		Yes	CDKN2A	ENST00000284494.10	missense_variant	2	c.457G>T	p.Asp1_...																									
chr9	132897140	-	ATA			TSC1	ENST00000298552.9	intron_variant		c.2975+4...	s2s3																									
chr9	136496197	AG	-		Yes	NOTCH1	ENST00000261671.1	frameshift, truncation	34	c.7541_75...	p.Pro2_...																									
chr10	87961047	ACTT	-		Yes	PTEN	ENST000002371933.8	frameshift, truncation	8	c.955_958...	p.Trp3_...																									
chr10	102402138	C	T		Yes	NFKB2	ENST00000261543.1	stop_gained	22	c.2557C>T	p.Arg8_...																									
chr11	534286	C	A		Yes	HRA5	ENST00000231189.8	missense_variant	2	c.37G>C	p.Gly13_...																									
chr11	106244076	C	G		Yes	ATM	ENST00000275843.1	missense_variant	5	c.820C>G	p.Ser20_...																									

49 total rows | [Export](#)

**Variant Annotation**

Gene: RPL22  
 Chrom: chr1  
 Position: 6197732  
 Ref base(s): T  
 Alt base(s): -  
 Gene: UniProt  
 Prot Chng: -  
 cDNA Chng: c.44del  
 Seq Ont: NMD\_transcript\_variant, 5\_prime\_UTR\_variant  
 Transcript: ENST00000234075.9

**PanglossDB**

No data

**HaploReg American**

No data

**BayesDel**

No data

**1000 Genomes Ad Mixed A**

No data

**COSMIC Gene**

Occurrences: 400

Tissue	Count
stomach	93
large intestine	72
liver	40
skin	36
haematopoietic and lymphoid tissue	36
prostate	17
pancreas	14
lung	13
bladder	12
breast	11
endometrium	10
ovary	6

**Candidate cis Regulatory Elements by ENCODE (SCREEN)**

No data

**FunSeq2**

No data

**REVEL**

No data

**InSight**

BP-22 collect  
 Interactors:  
 ACTB  
 AP2M1  
 ARRB1  
 ARRB2

**Swiss-Prot PTM**

No data

VARIETY\_R



# 5. Result Viewer: (iv) Explore GENE tab for gene-based table and export results

Open CRAVAT

Layout

SUMMARY

FILTER

VARIANT

GENE

View table

Home

Help

Download

Variant Annotation				BiOGRID	CCD Clinical Database	CHASM plus	CHASM plus ACC	CHASM plus BLCA	CHASM plus BRCA	CHASM plus CESC	CHASM plus CHOL	CHASM plus COAD	CHASM plus DLBC	CHASM plus ESCA	CHASM plus GBMLGG	CHASM plus HNSC	CHASM plus KICH	CHASM plus KIRC	CHASM plus KIRP	CHASM plus LAML	CHASM plus LGG	CHASM plus LIHC	CHASM plus LUAD	CHASM plus LUSC	CHASM plus MESO	CHASM plus OV	CHASM plus PAAD	CHASM plus PCPG	CHASM plus PRAD	CHASM plus READ	CHASM plus SARC	CHASM plus SKCM	CHASM plus STAD	CHASM plus TCGT	CHASM plus THCA	CHASM plus UCEC	CHASM plus UCS	CHASM plus UVM	CI G			
Gene	Gene Note	Number of Coding Variants	Number of Noncoding Variants	Sequence Ontology	All Sequence Ontologies	Interactors	Condition	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value	Gene p-value		
RPL22		1	0	frameshift.truncation	frameshift.tru...	AP2M1,ARF...																																				
ACVR2A		1	0	frameshift.truncation	frameshift.tru...	ACVR1,ACV...																																				
VHL		2	0	missense_variant	missense_vari...	AAP2,ACTR...		0	0.12	0.049	0.173	0.162	0.12	0.032	0.129	0.138	0.019	0.027	0.027	0	0.007	0.103	0.031	0.143	0.024	0.013	0.126	0.155	0.157	0.008	0.029	0.141	0.132	0.002	0.006	0.126	0.167	0.041	0.056	0.147	0.153	38
TGFB2		1	0	frameshift.truncation	frameshift.tru...	AAAS,ABCA...																																				
CTNMB1		2	0	missense_variant	missense_vari...	AASAS,ABCA...		0	0	0	0.042	0.017	0.24	0	0.241	0.005	0.239	0.001	0.248	0.069	0.044	0.229	0.002	0	0	0.039	0.03	0.054	0.025	0.259	0	0.02	0.24	0	0	0.256	0.056	0.054	0	0.259	0.284	125
PIK3CA		3	0	missense_variant	missense_vari...	ADAP1,AKT...		0	0.007	0	0	0	0.001	0	0.195	0	0	0	0.122	0	0	0.104	0	0	0	0	0	0.003	0.026	0	0	0.019	0.001	0	0	0.002	0.134	0	0	0.016	0.32	25
KIT		2	0	inframe_insertion	inframe_inser...	ABL1,ABL2...	Gastrointest...	0	0.263	0.13	0.19	0.13	0.086	0.169	0.285	0.269	0.212	0.126	0.293	0.334	0.234	0.002	0.185	0.069	0.152	0.043	0.196	0.151	0.337	0.249	0.048	0.062	0.106	0.003	0.316	0	0.178	0.167	0.16	0.304	0.32	25
APC		2	0	frameshift_elongation	frameshift_el...	ACTN1,ADF...		0	0.028	0	0.031	0.04	0.165	0	0.032	0.003	0.024	0.007	0.171	0.014	0.028	0.017	0.047	0	0.001	0.008	0.177	0.022	0.056	0.031	0	0	0.032	0.001	0	0.02	0.026	0.197	0.001	0.19	0.028	66
MTM1		1	0	frameshift_elongation	frameshift_el...	AATF,ABCD...																																				
EDFR		2	0	inframe_deletion	inframe_dele...	AAAS,AAAD...		0	0.03	0.02	0.01	0.011	0.006	0.031	0.143	0.017	0	0.007	0.144	0.029	0.02	0.12	0	0.011	0	0.006	0.144	0.025	0.173	0.15	0.027	0.028	0.013	0.019	0.02	0.012	0.178	0.167	0.028	0.161	0.166	19
BRAF		1	0	complex_substitution	complex_subs...	AKT1,ALDO...																																				
JAK2		1	0	missense_variant	missense_vari...	AMYT,CAH1...	Thrombocyto...	0.167	0.24	0.299	0.246	0.162	0.448	0.232	0.46	0.218	0.206	0.23	0.438	0.226	0.186	0.442	0.198	0.243	0.149	0.036	0.462	0.108	0.241	0.249	0.475	0.222	0.454	0.299	0.147	0.202	0.178	0.495	0.293	0.481	0.493	22
CDKN2A		1	0	missense_variant	missense_vari...	ACY1,ACTB...		0.022	0.232	0	0.283	0.13	0.156	0.162	0.242	0.002	0.016	0.001	0.237	0.284	0.048	0.224	0.054	0.003	0.003	0	0.243	0.165	0.005	0.207	0.04	0.245	0.034	0.001	0.002	0.248	0.05	0.266	0.232	0.257	0.266	14
TSC1		0	1	intron_variant	intron_varian...	ABT1,ACTA1...	Tuberculo...																																			
NOTCH1		1	0	frameshift.truncation	frameshift.tru...	ADAM10,AN...	Aortic valve d...																																			
PTEN		1	0	frameshift.truncation	frameshift.tru...	ABCF2,ACA...	PTEN hamar...																																			
NFKB2		1	0	stop_gained	stop_gained(1)	ATAD3B,BC...																																				
HNR4S		1	0	missense_variant	missense_vari...	AAAS,ABCC...		0	0.155	0	0.007	0.008	0.152	0.109	0.163	0.164	0.17	0	0.159	0.194	0.199	0.137	0.198	0.175	0.125	0	0.161	0.186	0.189	0	0	0.172	0.168	0	0.171	0.162	0	0	0.086	0.18	0.183	275
ATM		2	0	stop_gained	missense_vari...	ABL1,ABCT...		0.134	0.159	0.105	0.11	0.261	0.416	0.121	0.265	0.234	0.162	0.474	0.259	0.064	0.291	0.269	0.22	0.314	0.051	0.191	0.393	6.407	0.322	0.34	0.192	0.129	0.169	0.27	0.077	0.438	0.255	0.642	0.131	0.632	0.641	69
KRAS		1	0	synonymous_variant	intron_varian...	A2M,AAAS...																																				
ACVRL1		1	0	missense_variant	missense_vari...	AMH,HR23A...		0.032	0.215	0.179	0.157	0.118	0.132	0.184	0.143	0.468	0.266	0.111	0.14	0.35	0.177	0.12	0.28	0.091	0.093	0.076	0.14	0.256	0.159	0.146	0.223	0.153	0.148	0.107	0.042	0.14	0.174	0.164	0.562	0.161	0.212	
BRCA2		1	0	missense_variant	missense_vari...	AB1,ACV1...		0.023	0.216	0.025	0.001	0.003	0.007	0.049	0.064	0.093	0.034	0.036	0.218	0.112	0.151	0.117	0.146	0.006	0.084	0.027	0.224	0	0.247	0.11	0.007	0.008	0.029	0.097	0.007	0.229	0.125	0.146	0.075	0.029	0.246	7
RS1		1	0	missense_variant	intron_varian...	AATF,ABT1...		0	0.001	0	0	0	0.149	0.018	0.158	0.001	0	0.159	0.034	0.194	0.137	0.002	0	0	0	0.161	0	0.185	0.166	0.012	0.168	0	0	0.009	0.157	0.026	0.183	0	0.002	0.183	425	
B2M		1	0	frameshift.truncation	frameshift.tru...	A2M,B2M2...	Immunodef...																																			
MAP2K1		2	0	missense_variant	intron_varian...	ABCF1,AKA...		0	0.15	0.104	0.138	0.004	0.047	0.088	0.156	0.006	0.157	0.127	0.155	0.201	0.212	0.134	0.192	0.063	0.033	0.118	0.157	0.188	0.194	0.163	0.176	0.065	0.161	0	0.017	0.161	0.198	0.006	0.284	0.175	0.188	21
PALB2		1	0	missense_variant	intron_varian...	ATR,ATXN7...		0.232	0.246	0.285	0.259	0.316	0.243	0.269	0.075	0.248	0.253	0.368	0.244	0.169	0.19	0.236	0.306	0.148	0.235	0.008	0.256	0.031	0.272	0.262	0.329	0.258	0.264	0.381	0.114	0.308	0.291	0.279	0.275	0.263	0.281	125
TP53		4	0	missense_variant	missense_vari...	AAAGAB,AA...		0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0	0.017	85
NF1		1	0	missense_variant	missense_vari...	AGAP1,AKT...		0.002	0.003	0.002	0	0.012	0.236	0.017	0.061	0.055	0.001	0.045	0.244	0.002	0.027	0.042	0	0.011	0.001	0	0.249	0	0.084	0	0.106	0.067	0.005	0.002	0.002	0.038	0.062	0.149	0.024	0.051	0.273	386

35 total rows

Export

Variant Annotation

CDSMC Gene

IntAct

NCBI Gene

ChnGen Gene

gnomAD Gene

Cancer Gene Census

Gene: RPL22

# Coding Variants: 1

Most Severe Seq On: frameshift.truncation

Seq On: #transcript

frameshift.truncation: 3

3\_prime\_UTR\_variant: 3

CDSMC Gene

Discovered: 400

Tissue

Count

large intestine: 93

stomach: 72

liver: 40

skin: 40

hematopoietic and lymphoid tissue: 36

prostate: 17

pancreas: 14

lung: 13

biliary tract: 12

breast: 11

endometrium: 10

esophagus: 9

IntAct

Biological Process

Interactors

ACTB

AP2M1

ARHGAP1

ARHGAP2

BAG4

BRD9

CBX8

CDC42

CDC5L

CSE1L

CTNNA1

NCBI Gene

Ribosomes, the organelles that catalyze protein synthesis, consist of a small 40S subunit and a large 60S subunit. Together these subunits are...

Cancer Gene Landscape

ChnGen Gene

Note

Save

gnomAD Gene

Cancer Gene Census

Class: TSC, Fusion

Inheritance: Somatic

Somatic Types: AML, CML, Genotoxic Types

Rank Score Comparison

## 6. For more details: [https://docs.opencravat.org/en/latest/getting\\_started\\_web.html](https://docs.opencravat.org/en/latest/getting_started_web.html)

### Getting Started with the Website and GUI

#### Learning Objectives

Understanding the Context for our Task

Creating an OpenCRAVAT Account (Web)

#### Installing OpenCRAVAT via Installer (Local)

Start OpenCRAVAT GUI

Browse Annotators (Web)

Install Annotators (Local)

Convert to Input File Format

Submit Annotation Job

Filter Results

Visualize Results

What you learned

#### Getting Started with Open-Cravat on the Command Line

Installation Instructions

Command line usage

Viewing Results

GUI usage

Uninstallation

Update instructions

#### ADDITIONAL TOOLS

Single Variant Report

Large VCF Annotation

#### ADVANCED

Filter tutorial

Case-Control Analysis

🏠 / Getting Started with the Website and GUI

[View page source](#)

## Getting Started with the Website and GUI

### Contents

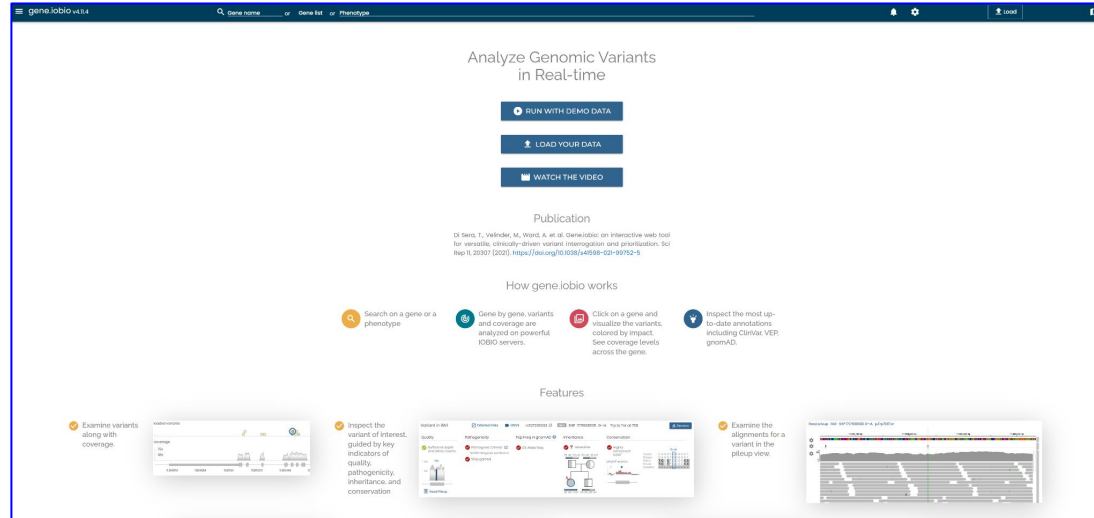
- [Getting Started with the Website and GUI](#)
  - [Learning Objectives](#)
  - [Understanding the Context for our Task](#)
  - [Creating an OpenCRAVAT Account \(Web\)](#)
  - [Installing OpenCRAVAT via Installer \(Local\)](#)
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  - [Convert to Input File Format](#)
  - [Submit Annotation Job](#)
  - [Filter Results](#)
  - [Visualize Results](#)
  - [What you learned](#)

### Learning Objectives

By the end of this tutorial, you should be able to:

- **Create** as user account on the OpenCRAVAT site
- **Install** OpenCravat locally using an installer
- **Search** for available annotators on the open cravat site
- **Upload** and **annotate** variant files on the site
- **Filter** annotated results for visualization
- **Visualize** and **Summarize** Results in OpenCRAVAT
- **Export** and **Share** annotation results with others


## 2. Gene.iobio Web-app



<https://gene.iobio.io/>  
<https://github.com/iobio/gene.iobio>

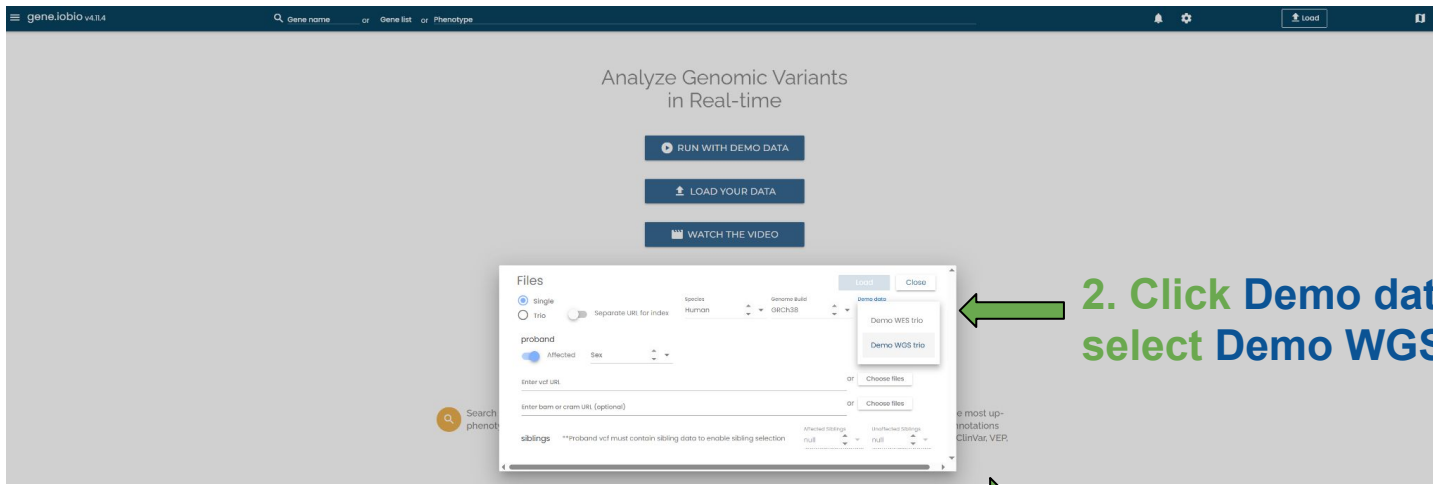
 WATCH THE VIDEO

Di Sera, T., Velinder, M., Ward, A. et al. GeneJobio: an interactive web tool for versatile, clinically-driven variant interrogation and prioritization. *Sci Rep* 11, 20307 (2021). <https://doi.org/10.1038/s41598-021-99752-5>

 Inspect the most up-to-date annotations including ClinVar, VEP, gnomAD.

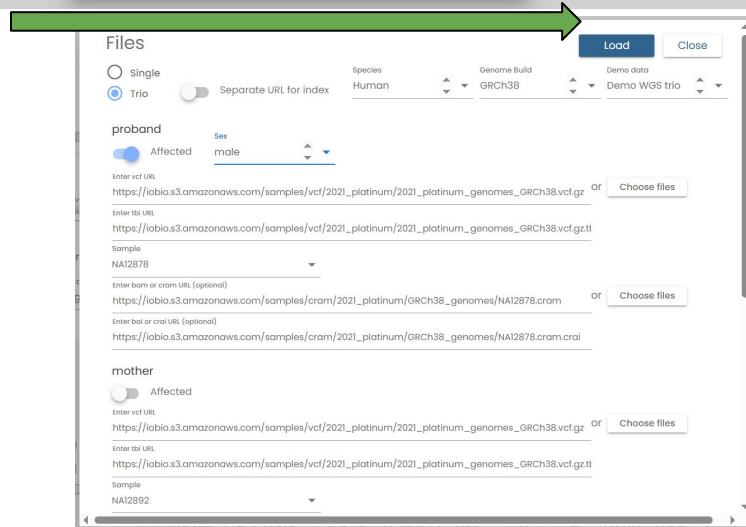
[illegible]

1. Click **Load** and select **Specify files**

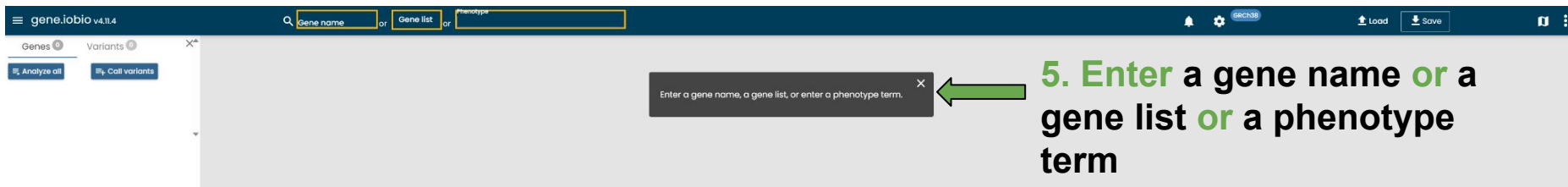


2. Click Demo data and select Demo WGS trio

4. Click Load to load Demo data



3. It will open WGS trio samples information: proband, mother and father for exploration



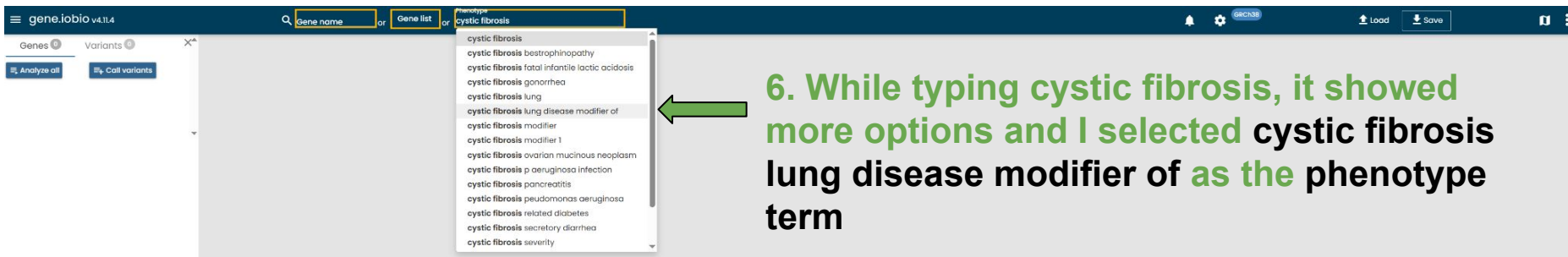
gene.iobio v4.11.4

Search bar: Gene name or Gene list or Phenotype

Buttons: Genes, Variants, Analyze all, Call variants

Message box: Enter a gene name, a gene list, or enter a phenotype term.

5. Enter a gene name **or** a gene list **or** a phenotype term



gene.iobio v4.11.4

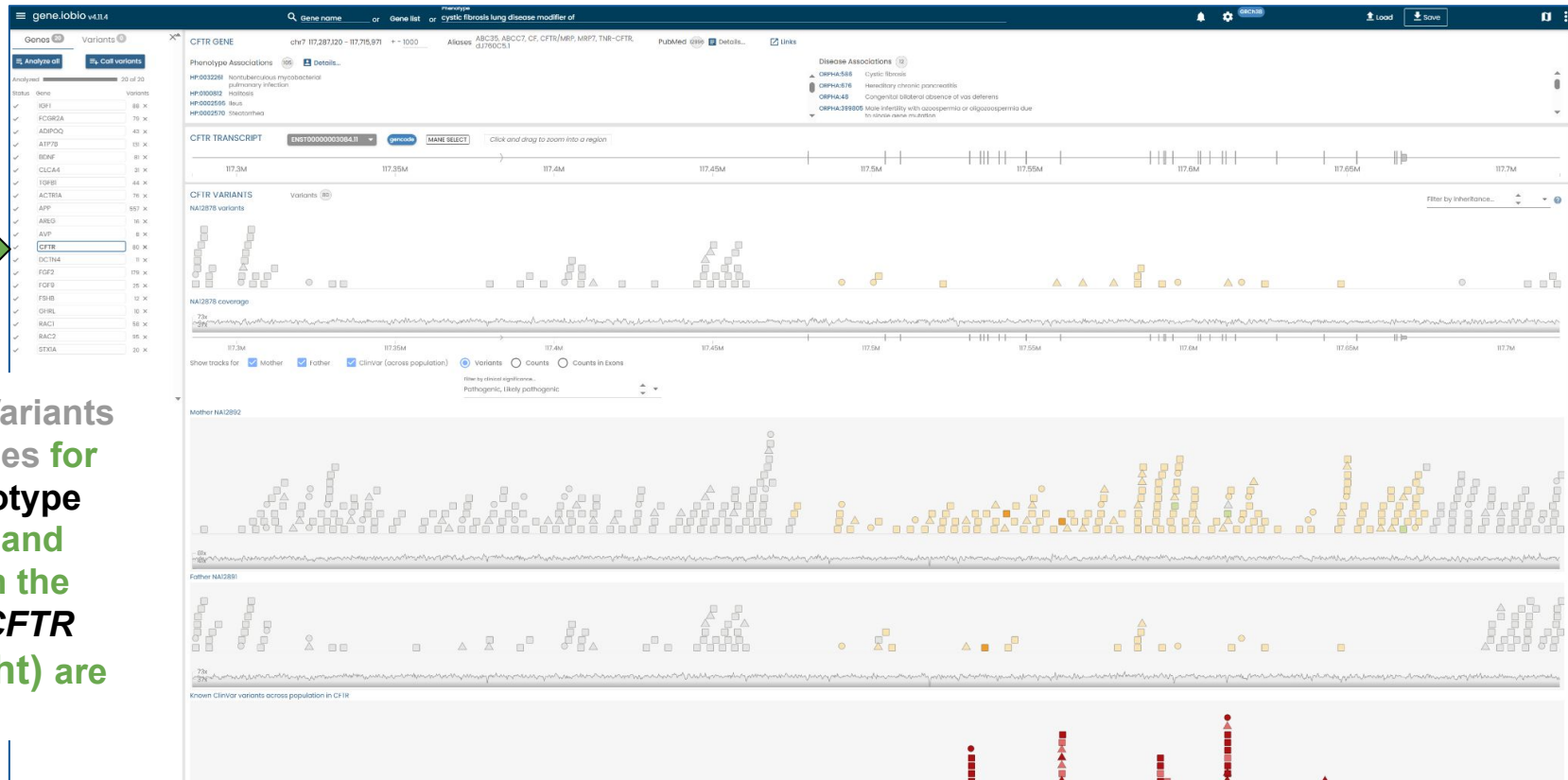
Search bar: Gene name or Gene list or Phenotype

Buttons: Genes, Variants, Analyze all, Call variants

Dropdown menu for 'cystic fibrosis':

- cystic fibrosis
- cystic fibrosis bestrophinopathy
- cystic fibrosis fatal infantile lactic acidosis
- cystic fibrosis gonorrhea
- cystic fibrosis lung
- cystic fibrosis lung disease modifier of
- cystic fibrosis modifier
- cystic fibrosis modifier 1
- cystic fibrosis ovarian mucinous neoplasm
- cystic fibrosis p aeruginosa infection
- cystic fibrosis pancreatitis
- cystic fibrosis pseudomonas aeruginosa
- cystic fibrosis related diabetes
- cystic fibrosis secretory diarrhea
- cystic fibrosis severity

6. While typing cystic fibrosis, it showed more options and I selected cystic fibrosis lung disease modifier of as the phenotype term



7. No. of Variants in the Genes for that phenotype term (left) and Variants in the selected **CFTR** Gene (right) are shown





gene.iobio v4.11.4

Gene name or Gene list or Phenotype cystic fibrosis lung disease modifier of

Genes 2 Variants 3

Analyze all Call variants

Phenotype Associations 20 of 20

HP:0002281 Non-tuberculous mycobacterial pulmonary infection

HP:0000882 Histoplasia

HP:0002595 Rash

HP:0002570 Stomatitis

CFTR GENE chr7 117,287,120 - 117,715,971 +- 1000

Aliases ABC35, ABCC7, CF, CFTR/MPR, MRP7, TNR-CFTR, c.1790C>T

PubMed Details Links

Disease Associations 2

ORPHA:596 Cystic fibrosis

ORPHA:578 Hereditary chronic pancreatitis

ORPHA:48 Congenital bilateral absence of vas deferens

ORPHA:395805 Male infertility with azoospermia or oligozoospermia due to single gene mutation

CFTR TRANSCRIPT1 ENST00000203864.11

CFTR VARIANTS NA12878 variants

NA12878 coverage

Show tracks for Mother Father ClinVar (across population) Variants Counts Counts in Exons

Filter by clinical significance Pathogenic, Likely pathogenic

Mother NA12892

Father NA12891

Known ClinVar variants across population in CFTR

CFTR VARIANT SNP chr7:117501772 G>A

Not reviewed Multiallelic rs1095689242 HGVS: ENST00000203864.1:c.54-248G>A

Quality Sufficient depth and allele counts

Read Pileup

Genotype/Phenotype Associations

Phenotype based search hits

Phenotype Cystic fibrosis lung disease modifier of

Phenotype Cystic fibrosis

Pathogenicity Intron variant

Population Frequency

gnomAD genomes v4

0.124 Allele frequency

0.180 Population max allele frequency

8536 alt of 88864 total

0 homozygotes

Population frequencies

gnomAD exomes

0.0 Allele frequency

Inheritance

De novo

Conservation

Not conserved -1.621

Annotations Select...

9. Click on a De novo Variant (new and not inherited) for details

## 10. For more details and options: <https://gene.iobio.io/tutorial>

### gene.iobio Tutorials

#### Datasets

Platinum trio - whole genome sequencing  
1000 Genomes project trio - exome sequencing

#### Use cases

Recessive variant - RAI1	description	screencast	gene.iobio
Realtime variant calling - MYLK2	description	screencast	gene.iobio
Low coverage variants - AIRE	description	screencast	gene.iobio
Alternate transcripts - PDGFB	description	screencast	gene.iobio
Phenotype driven analysis - PDHAT1	description	screencast	gene.iobio

#### Screencasts



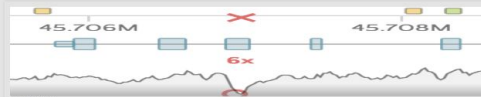
30 JULY 2015  
Introduction to gene.iobio  
Overview of basic usage and features



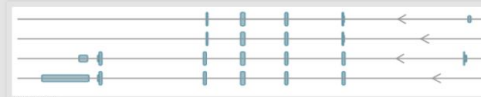
30 JULY 2015  
Use Case: recessive variants  
Simple view of a recessive variant, pathogenic in ClinVar



30 JULY 2015  
Use Case: real-time variant calling  
Recall variants in realtime from the underlying alignment data



30 JULY 2015  
Use Case: low coverage alignments  
Variants can be missed as a result of low coverage in the alignments



30 JULY 2015  
Use Case: alternative transcripts  
Annotate variants based on different transcripts