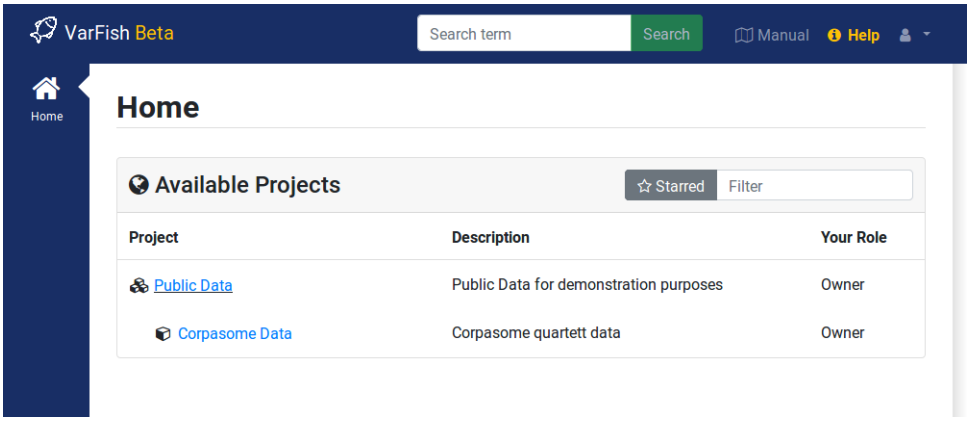
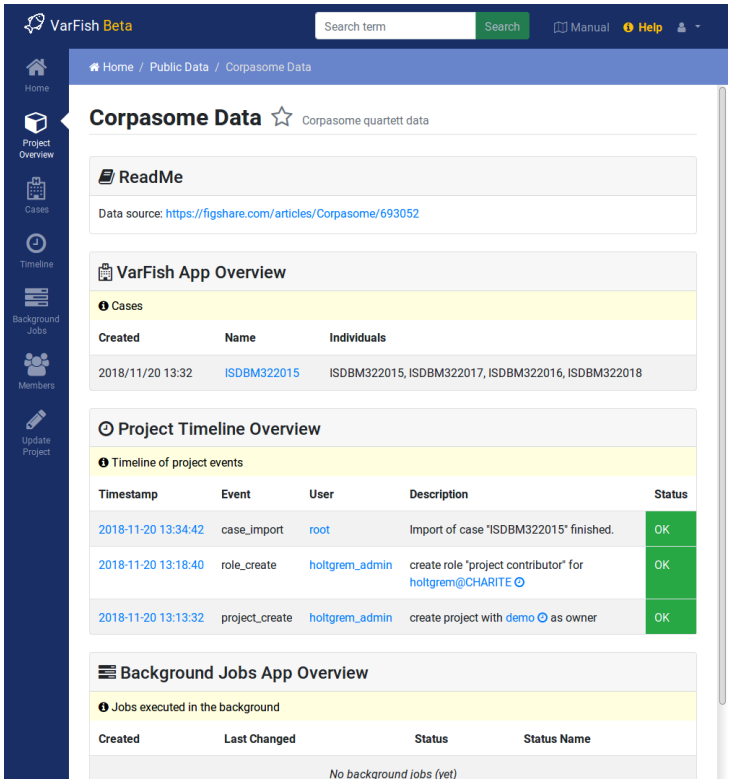
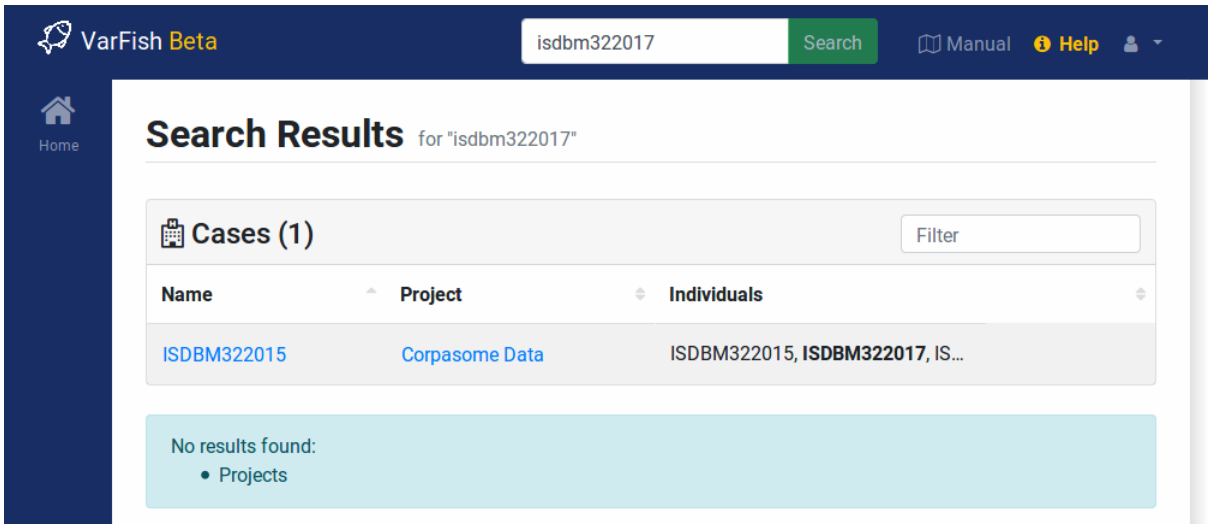
20210728

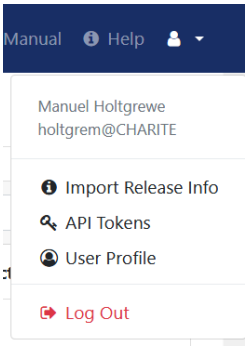
X-Beacon-User
Date Header

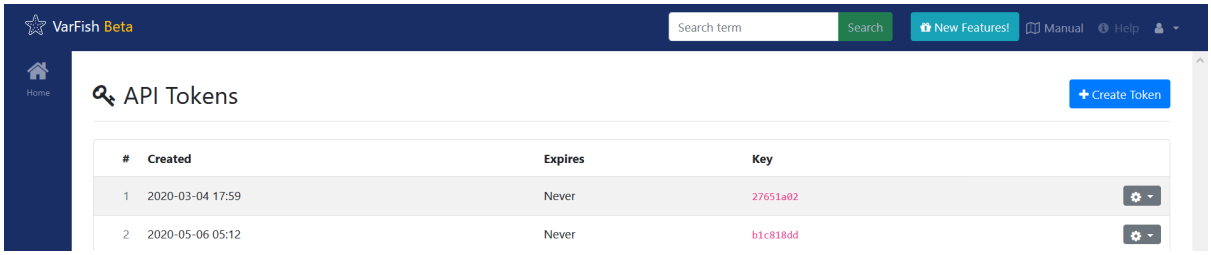
varfish-docker-compose

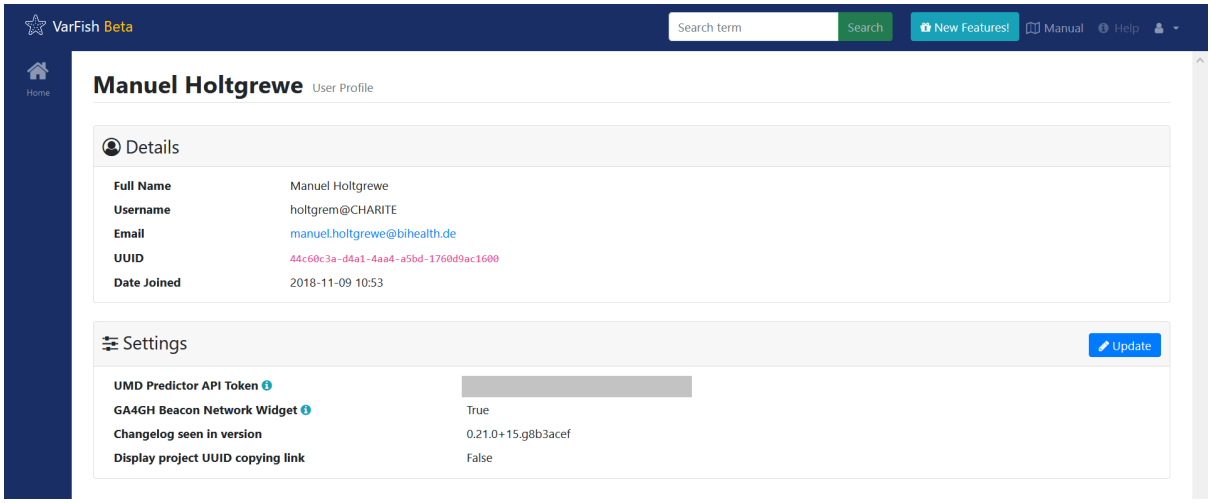

VarFish Beta
Search term
Search
Manual
Help
Home
Home
Available Projects
Starred
Filter
Project
Description
Your Role
Public Data
Public Data for demonstration purposes
Owner
Corpasome Data
Corpasome quartett data
Owner

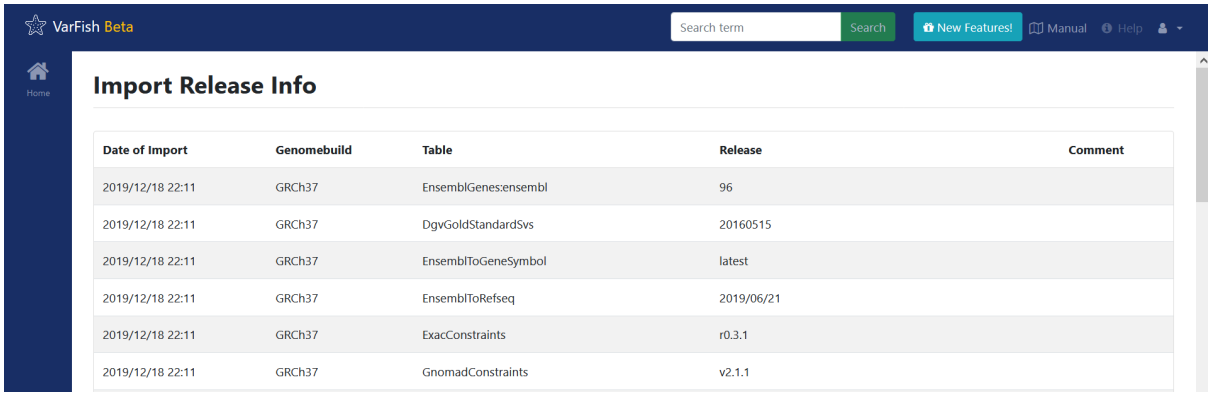

VarFish Beta
Search term
Search
Manual
Help
Home
Public Data
Corpasome Data
Home
Project Overview
Cases
Timeline
Background Jobs
Members
Update Project
Corpasome Data
Corpasome quartett data
ReadMe
Data source: https://figshare.com/articles/Corpasome/693052
VarFish App Overview
Cases
Created
Name
Individuals
2018/11/20 13:32
ISDBM322015
ISDBM322015, ISDBM322017, ISDBM322016, ISDBM322018
Project Timeline Overview
Timeline of project events
Timestamp
Event
User
Description
Status
2018-11-20 13:34:42
case_import
root
Import of case "ISDBM322015" finished.
OK
2018-11-20 13:18:40
role_create
holtgrem_admin
create role "project contributor" for holtgrem@CHARITE
OK
2018-11-20 13:13:32
project_create
holtgrem_admin
create project with demo as owner
OK
Background Jobs App Overview
Jobs executed in the background
Created
Last Changed
Status
Status Name
No background jobs (yet)


VarFish Beta
isdbm322017
Search
Manual
Help
Home
Search Results
for "isdbm322017"
Cases (1)
Filter
Name
Project
Individuals
ISDBM322015
Corpasome Data
ISDBM322015, ISDBM322017, IS...
No results found:
Projects

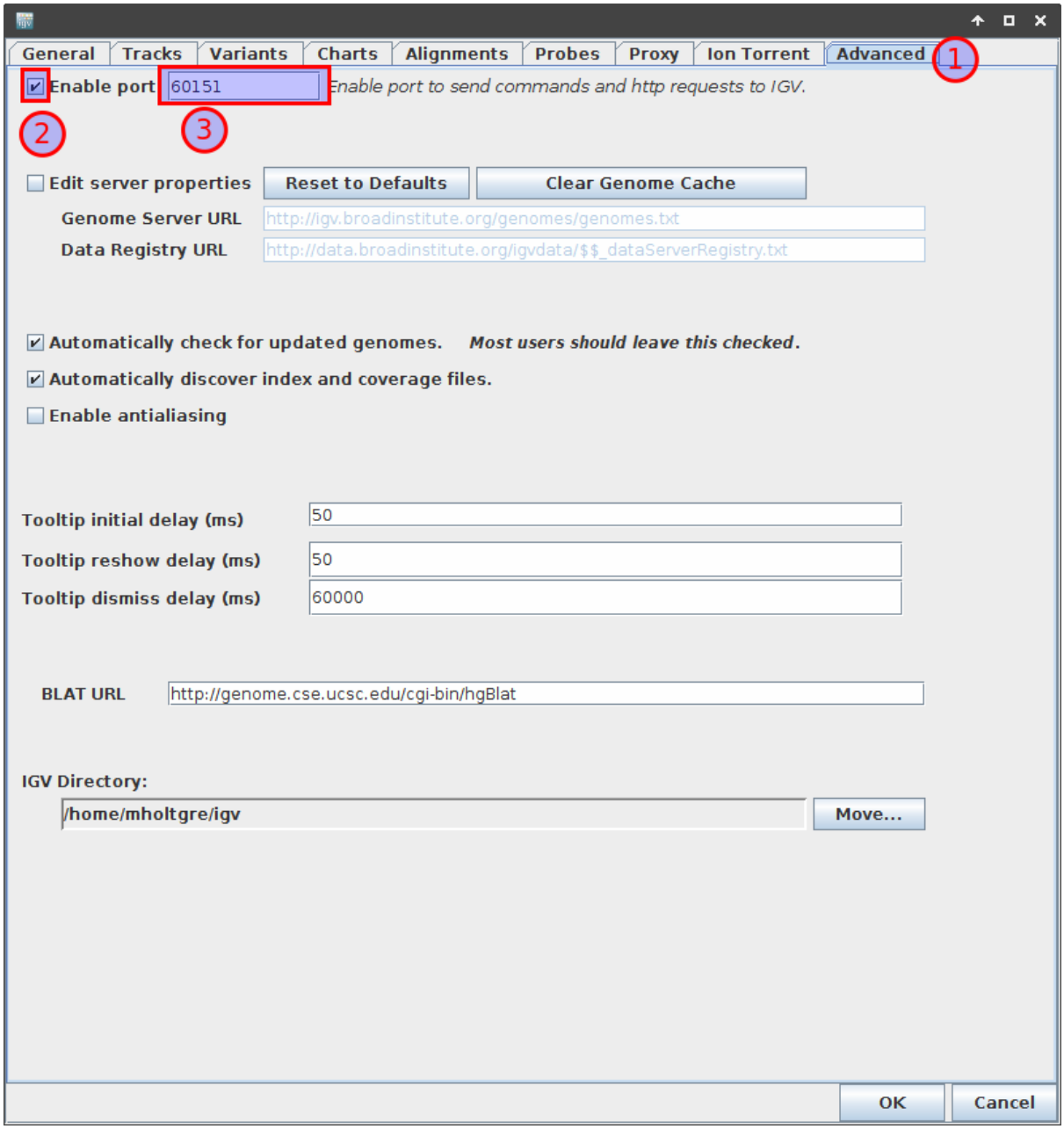

Manual
Help
Manuel Holtgrewe
holtgrem@CHARITE
Import Release Info
API Tokens
User Profile
Log Out


VarFish Beta
Search term
Search
New Features!
Manual
Help
Home
API Tokens
+ Create Token
# Created Expires Key
1 2020-03-04 17:59 Never 27651a02
2 2020-05-06 05:12 Never b1c818dd

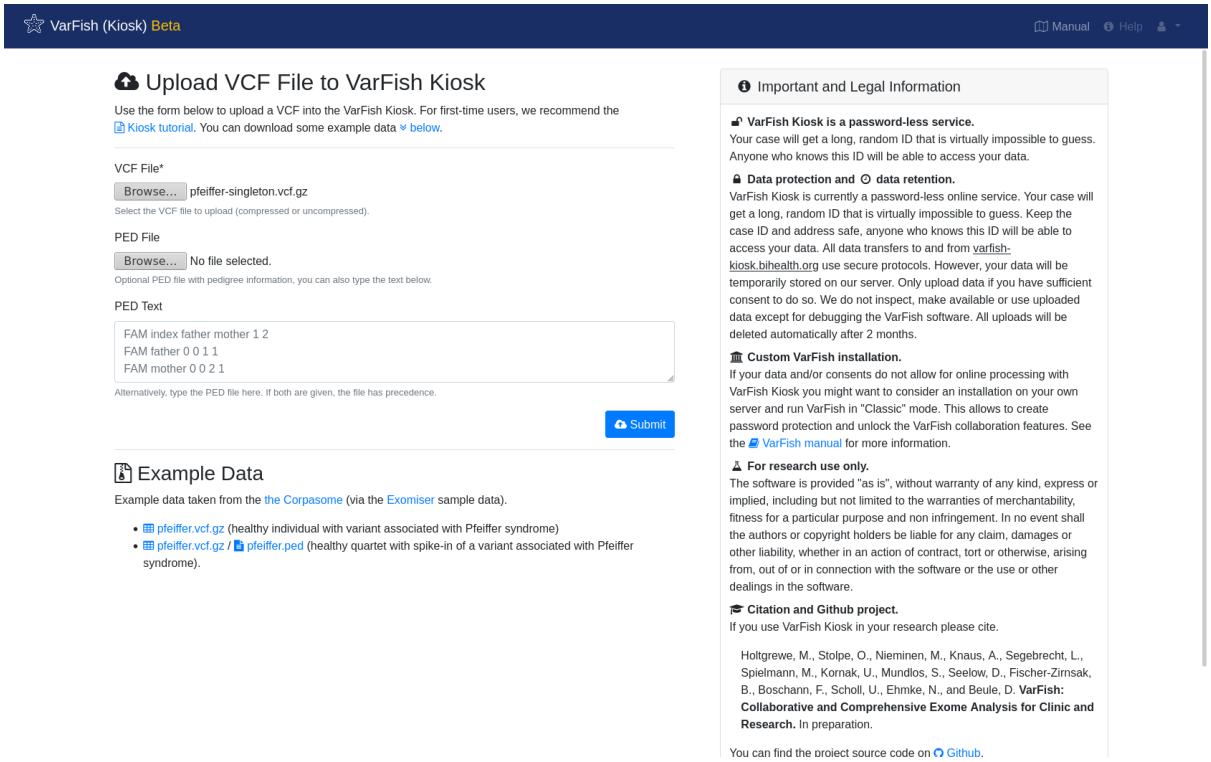

VarFish Beta
Search term
Search
New Features!
Manual
Help
Home
Manuel Holtgrewe
User Profile
Details
Full Name
Manuel Holtgrewe
Username
holtgrem@CHARITE
Email
manuel.holtgrewe@bihealth.de
UUID
44c60c3a-d4a1-4aa4-a5bd-1760d9ac1600
Date Joined
2018-11-09 10:53
Settings
Update
UMD Predictor API Token
GA4GH Beacon Network Widget
True
Changelog seen in version
0.21.0+15.g8b3acef
Display project UUID copying link
False


VarFish Beta
Search term
Search
New Features!
Manual
Help
Home
Import Release Info
Date of Import
Genomebuild
Table
Release
Comment
2019/12/18 22:11
GRCh37
EnsemblGenes:ensembl
96
2019/12/18 22:11
GRCh37
DgvGoldStandardSvs
20160515
2019/12/18 22:11
GRCh37
EnsemblToGeneSymbol
latest
2019/12/18 22:11
GRCh37
EnsemblToRefseq
2019/06/21
2019/12/18 22:11
GRCh37
ExacConstraints
r0.3.1
2019/12/18 22:11
GRCh37
GnomadConstraints
v2.1.1

60151


General
Tracks
Variants
Charts
Alignments
Probes
Proxy
Ion Torrent
Advanced
1
Enable port
60151
Enable port to send commands and http requests to IGV.
2
3
Edit server properties
Reset to Defaults
Clear Genome Cache
Genome Server URL
http://igv.broadinstitute.org/genomes/genomes.txt
Data Registry URL
http://data.broadinstitute.org/igvdata/$$_dataServerRegistry.txt
Automatically check for updated genomes.
Most users should leave this checked.
Automatically discover index and coverage files.
Enable antialiasing
Tooltip initial delay (ms)
50
Tooltip reshow delay (ms)
50
Tooltip dismiss delay (ms)
60000
BLAT URL
http://genome.cse.ucsc.edu/cgi-bin/hgBlat
IGV Directory:
/home/mholtgre/igv
Move...
OK
Cancel

FGFR2:p.Glu566Gly

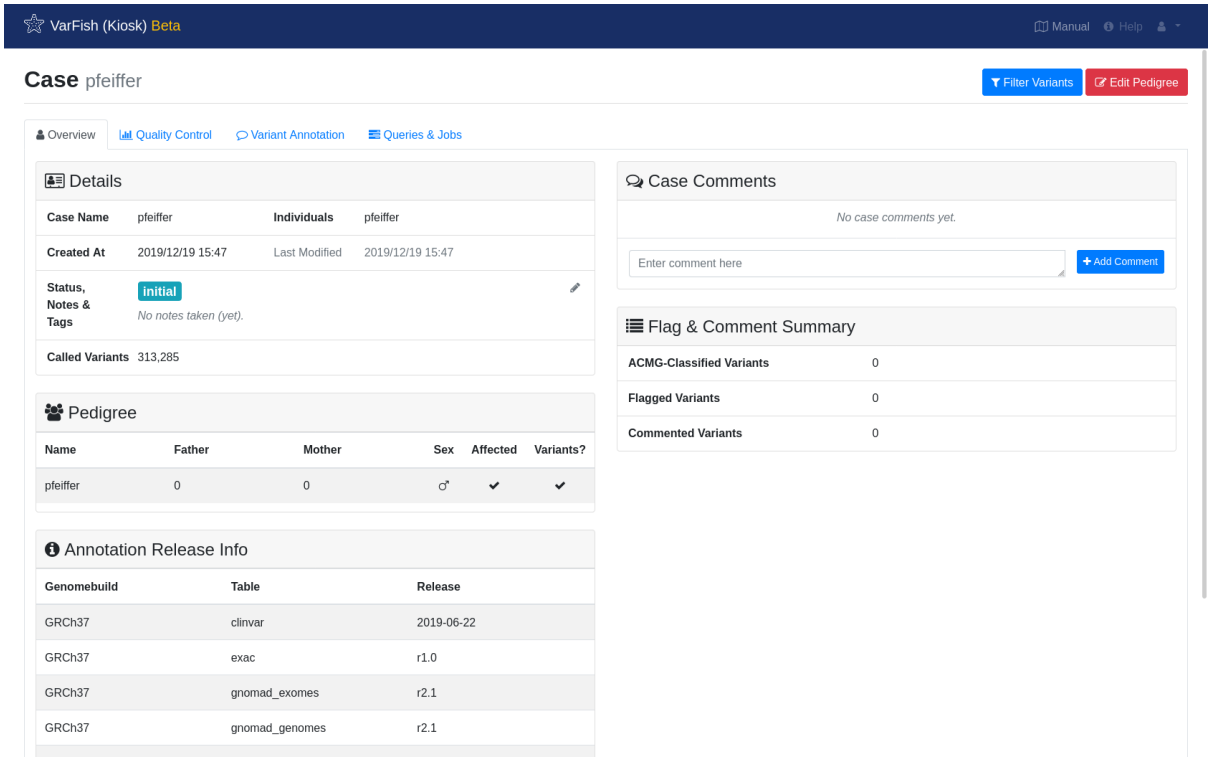


pfeiffer-singleton.vcf.gz

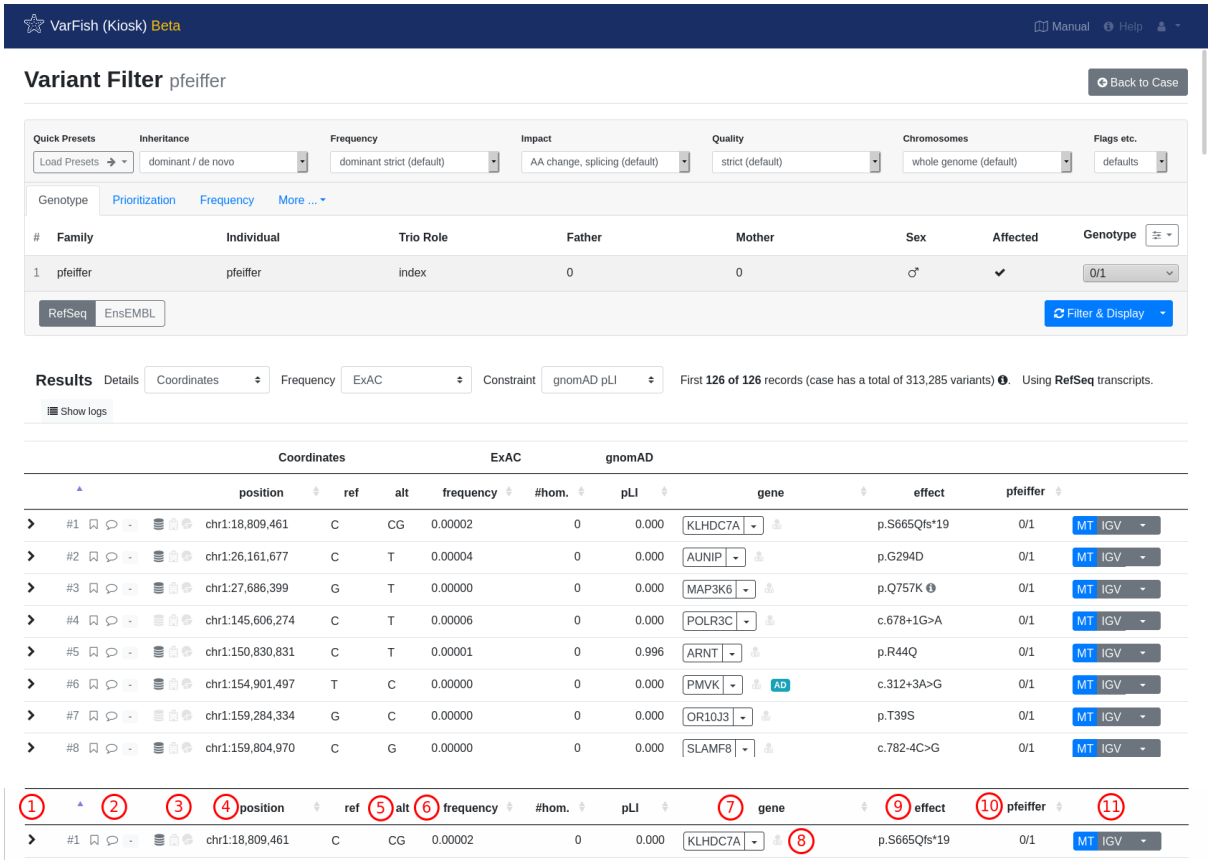
FAM00120120

```
FAM index  father mother 1 2
FAM father 0      0      1 1
FAM mother 0      0      2 1
```

varfish-kiosk.bihealth.org

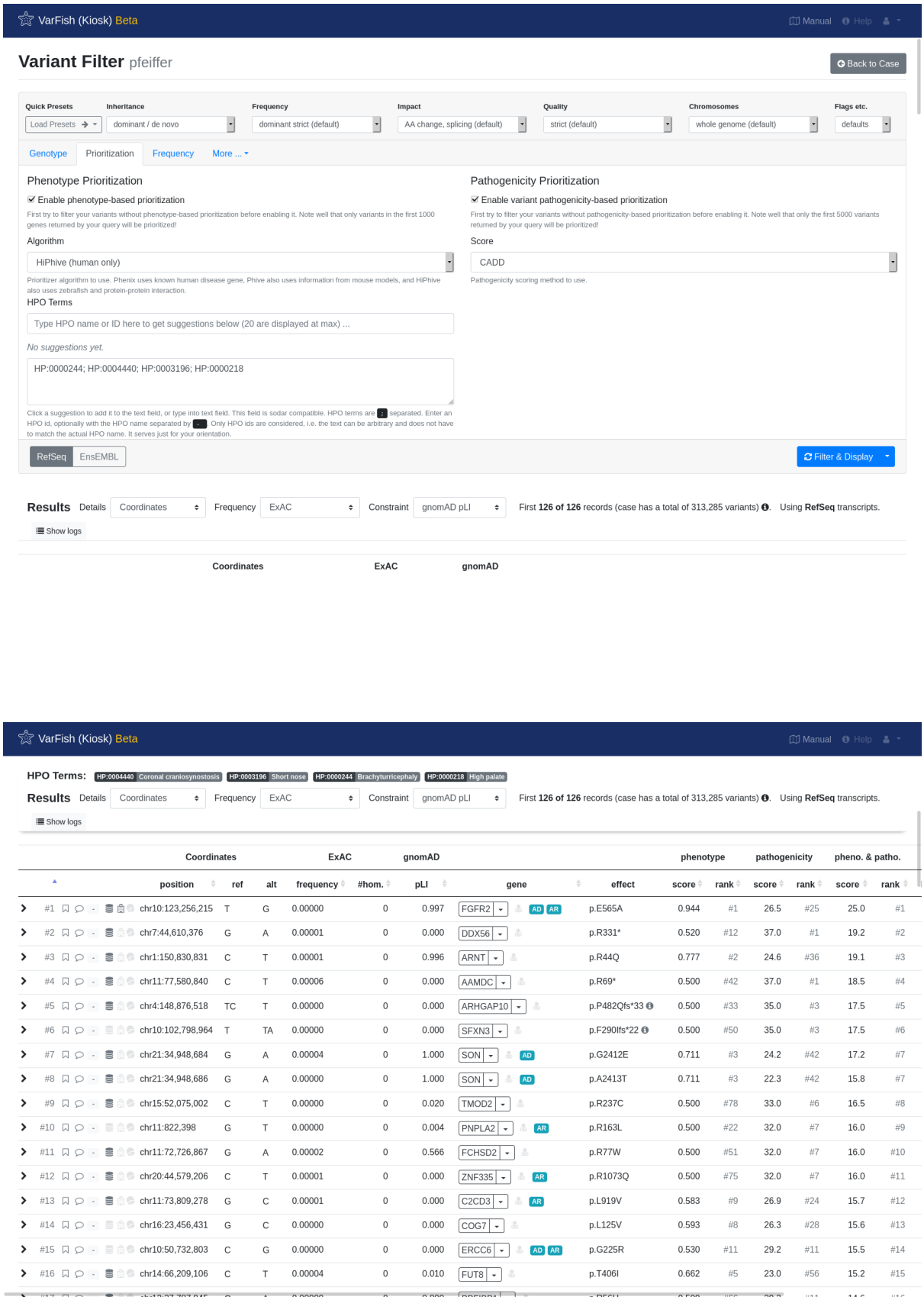

VarFish (Kiosk) Beta
Manual
Help
Case pfeiffer
Filter Variants
Edit Pedigree
Overview
Quality Control
Variant Annotation
Queries & Jobs
Details
Case Name pfeiffer
Individuals pfeiffer
Created At 2019/12/19 15:47
Last Modified 2019/12/19 15:47
Status, Notes & Tags
initial
No notes taken (yet).
Called Variants 313,285
Pedigree
Name Father Mother Sex Affected Variants?
pfeiffer 0 0 ♂ ✔ ✔
Annotation Release Info
Genomebuild Table Release
GRCh37 clinvar 2019-06-22
GRCh37 exac r1.0
GRCh37 gnomad_exomes r2.1
GRCh37 gnomad_genomes r2.1
Case Comments
No case comments yet.
Enter comment here
Add Comment
Flag & Comment Summary
ACMG-Classified Variants 0
Flagged Variants 0
Commented Variants 0

200

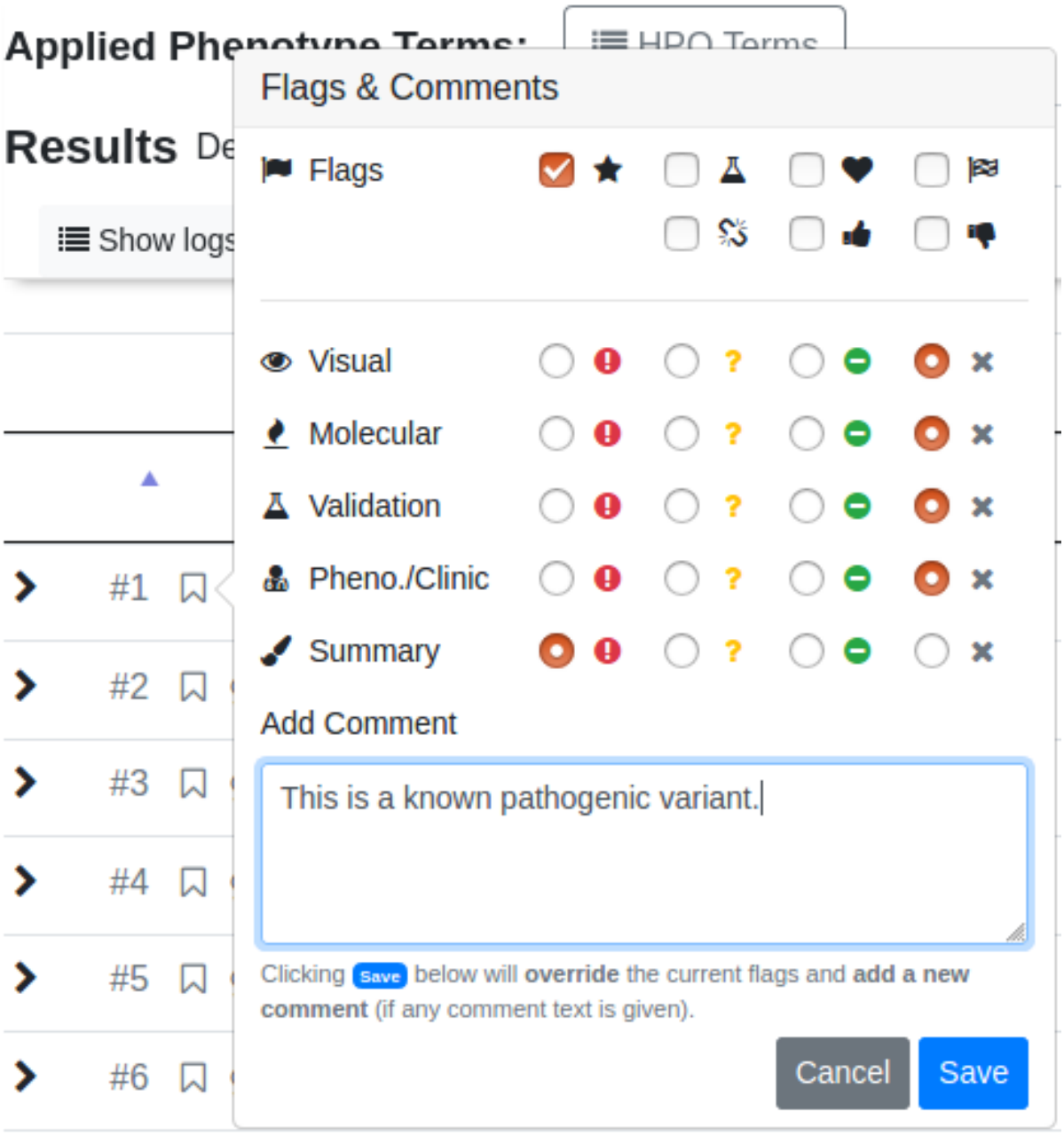


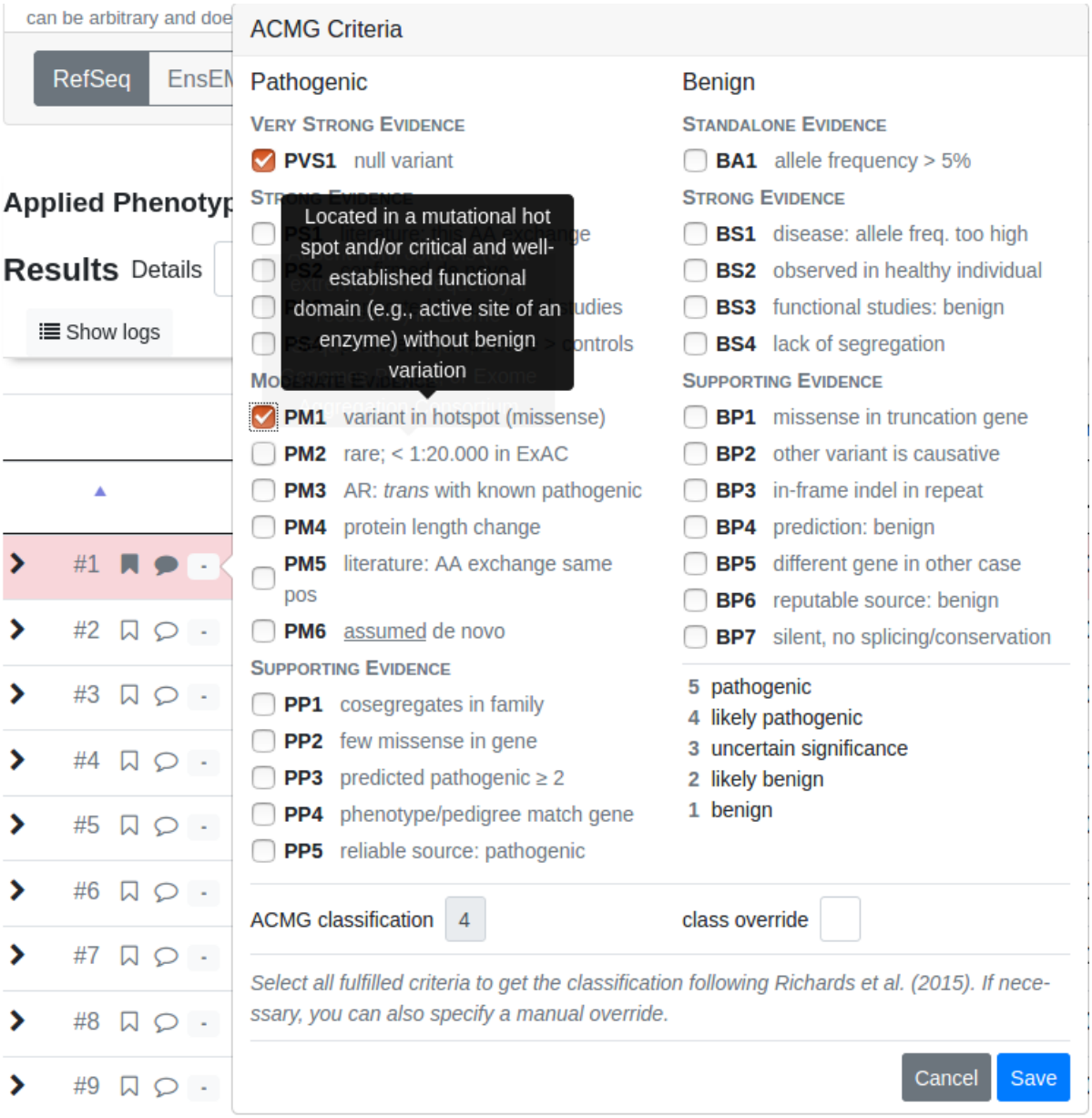
126

HP:0004440; HP:0003196; HP:0000244; HP:0000218



		Coordinates			ExAC	gnomAD				phenotype		pathogenicity		pheno. & patho.	
		position	ref	alt	frequency	#hom.	pLI	gene	effect	score	rank	score	rank	score	rank
>	#1	chr10:123,256,215	T	G	0.00000	0	0.997	FGFR2 AD AR	p.E565A	0.944	#1	26.5	#25	25.0	#1
>	#2	chr7:44,610,376	G	A	0.00001	0	0.000	DDX56	p.R331*	0.520	#12	37.0	#1	19.2	#2
>	#3	chr1:150,830,831	C	T	0.00001	0	0.996	ARNT	p.R44Q	0.777	#2	24.6	#36	19.1	#3
>	#4	chr11:77,580,840	C	T	0.00006	0	0.000	AAMDC	p.R69*	0.500	#42	37.0	#1	18.5	#4
>	#5	chr4:148,876,518	TC	T	0.00000	0	0.000	ARHGAP10	p.P482Qfs*33	0.500	#33	35.0	#3	17.5	#5
>	#6	chr10:102,798,964	T	TA	0.00000	0	0.000	SFXN3	p.F290Ifs*22	0.500	#50	35.0	#3	17.5	#6
>	#7	chr21:34,948,684	G	A	0.00004	0	1.000	SON AD	p.G2412E	0.711	#3	24.2	#42	17.2	#7
>	#8	chr21:34,948,686	G	A	0.00000	0	1.000	SON AD	p.A2413T	0.711	#3	22.3	#42	15.8	#7
>	#9	chr15:52,075,002	C	T	0.00000	0	0.020	TMOD2	p.R237C	0.500	#78	33.0	#6	16.5	#8
>	#10	chr11:822,398	G	T	0.00000	0	0.004	PNPLA2 AR	p.R163L	0.500	#22	32.0	#7	16.0	#9
>	#11	chr11:72,726,867	G	A	0.00002	0	0.566	FCHSD2	p.R77W	0.500	#51	32.0	#7	16.0	#10
>	#12	chr20:44,579,206	C	T	0.00001	0	0.000	ZNF335 AR	p.R1073Q	0.500	#75	32.0	#7	16.0	#11
>	#13	chr11:73,809,278	G	C	0.00001	0	0.000	C2CD3 AR	p.L919V	0.583	#9	26.9	#24	15.7	#12
>	#14	chr16:23,456,431	G	C	0.00000	0	0.000	COG7	p.L125V	0.593	#8	26.3	#28	15.6	#13
>	#15	chr10:50,732,803	C	G	0.00000	0	0.000	ERCC6 AD AR	p.G225R	0.530	#11	29.2	#11	15.5	#14
>	#16	chr14:66,209,106	C	T	0.00004	0	0.010	FUT8	p.T406I	0.662	#5	23.0	#56	15.2	#15


Applied Phenotype Terms:
HPO Terms
Results De
Show logs
Flags & Comments
Flags
Visual
Molecular
Validation
Pheno./Clinic
Summary
Add Comment
This is a known pathogenic variant.
Clicking Save below will override the current flags and add a new comment (if any comment text is given).
Cancel
Save
#1
#2
#3
#4
#5
#6

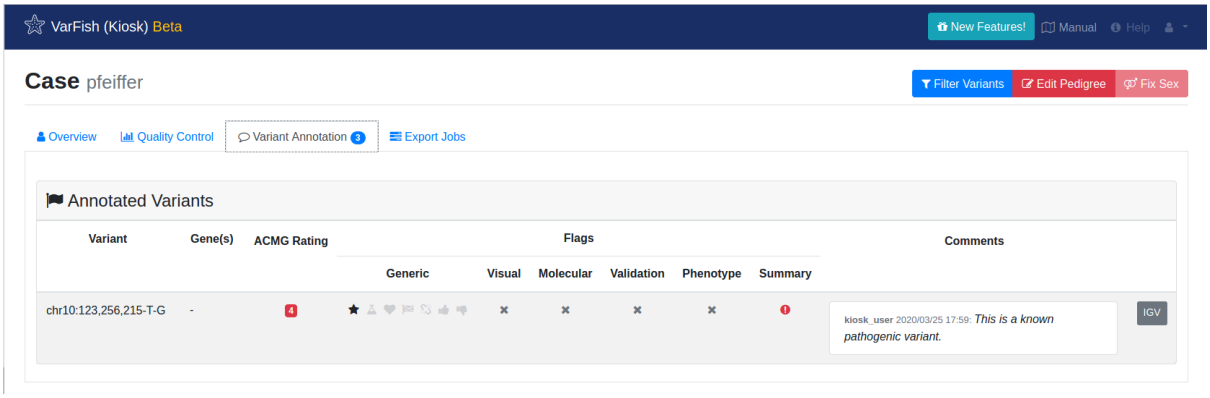


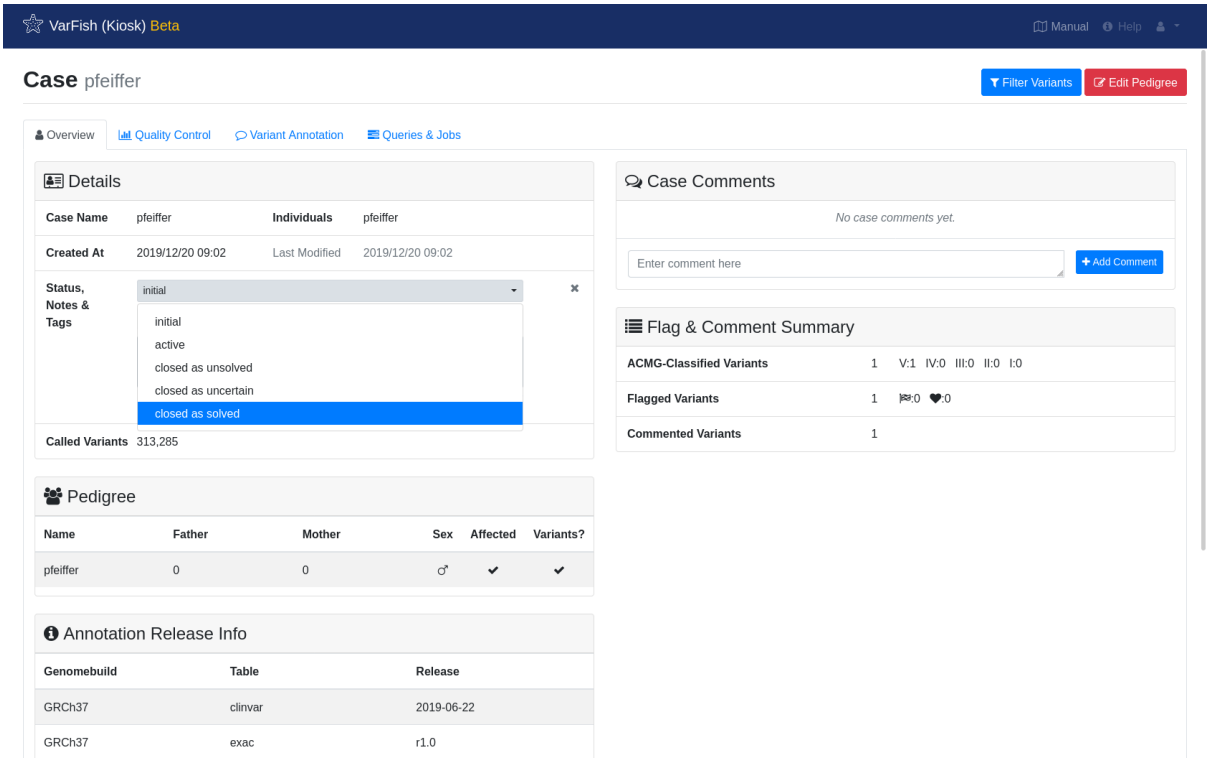
Applied Phenotype Terms: HPO Terms

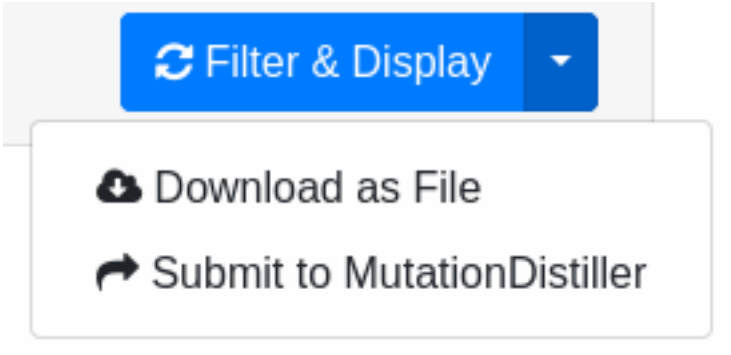
Results Details Coordinates Frequency ExAC Constraint gnomAD pLI First **126 of 126** records (case has a total of 313,285 variants). Using **RefSeq** transcripts.

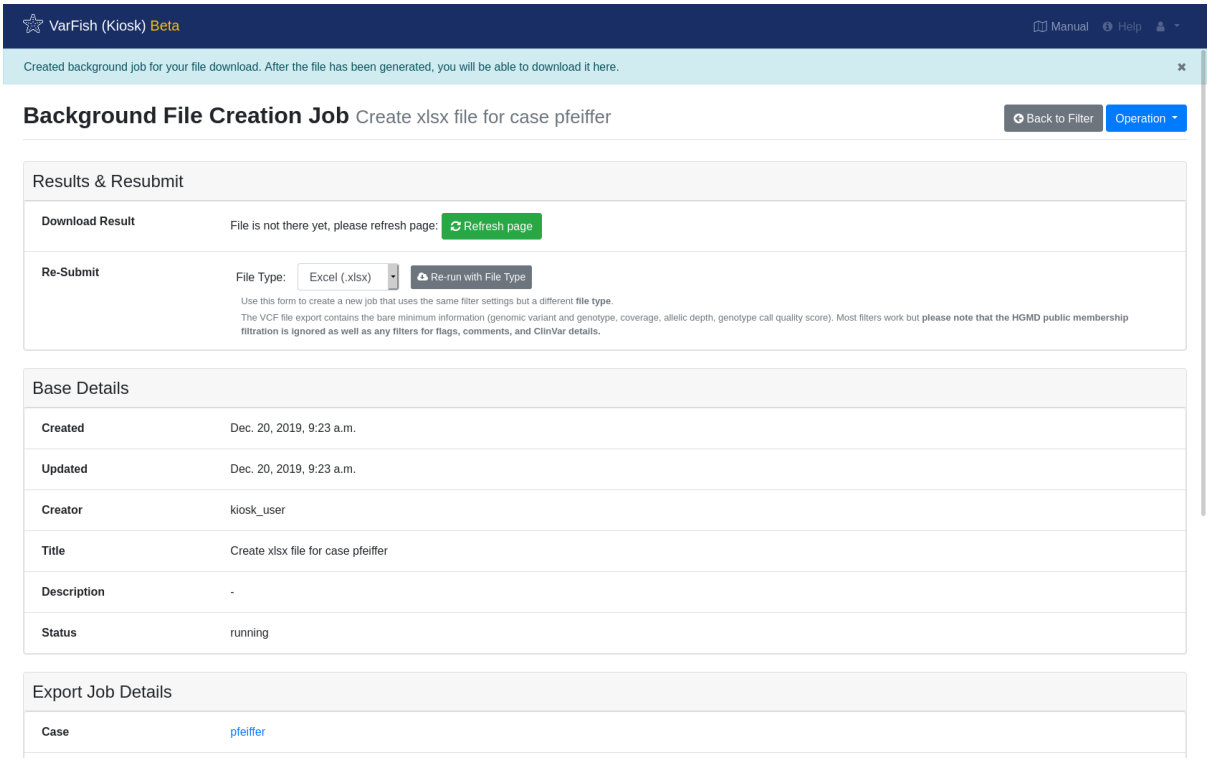
Show logs

		Coordinates			ExAC	gnomAD					phenotype		pathogenicity		pheno. & patho.	
		position	ref	alt	frequency	#hom.	pLI	gene		effect	score	rank	score	rank	score	rank
▸	#1	chr10:123,256,215	T	G	0.00000	0	0.997	FGFR2	AD AR	p.E565A	0.903	#1	26.5	#25	23.9	#1
▸	#2	chr7:44,610,376	G	A	0.00001	0	0.000	DDX56		p.R331*	0.514	#11	37.0	#1	19.0	#2
▸	#3	chr11:77,580,840	C	T	0.00006	0	0.000	AAMDC		p.R69*	0.500	#41	37.0	#1	18.5	#3

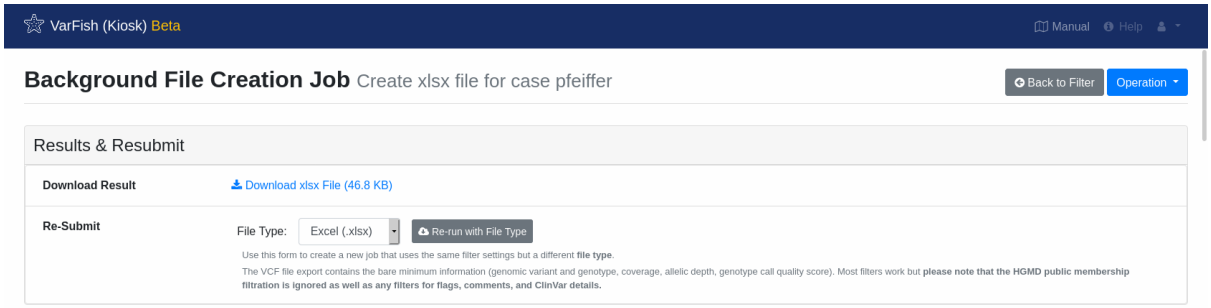

VarFish (Kiosk) Beta
New Features!
Manual
Help
Case pfeiffer
Filter Variants
Edit Pedigree
Fix Sex
Overview
Quality Control
Variant Annotation 3
Export Jobs
Annotated Variants
Variant
Gene(s)
ACMG Rating
Flags
Generic
Visual
Molecular
Validation
Phenotype
Summary
Comments
chr10:123,256,215-T-G
-
4
kiosk_user 2020/03/25 17:59: This is a known pathogenic variant.
IGV

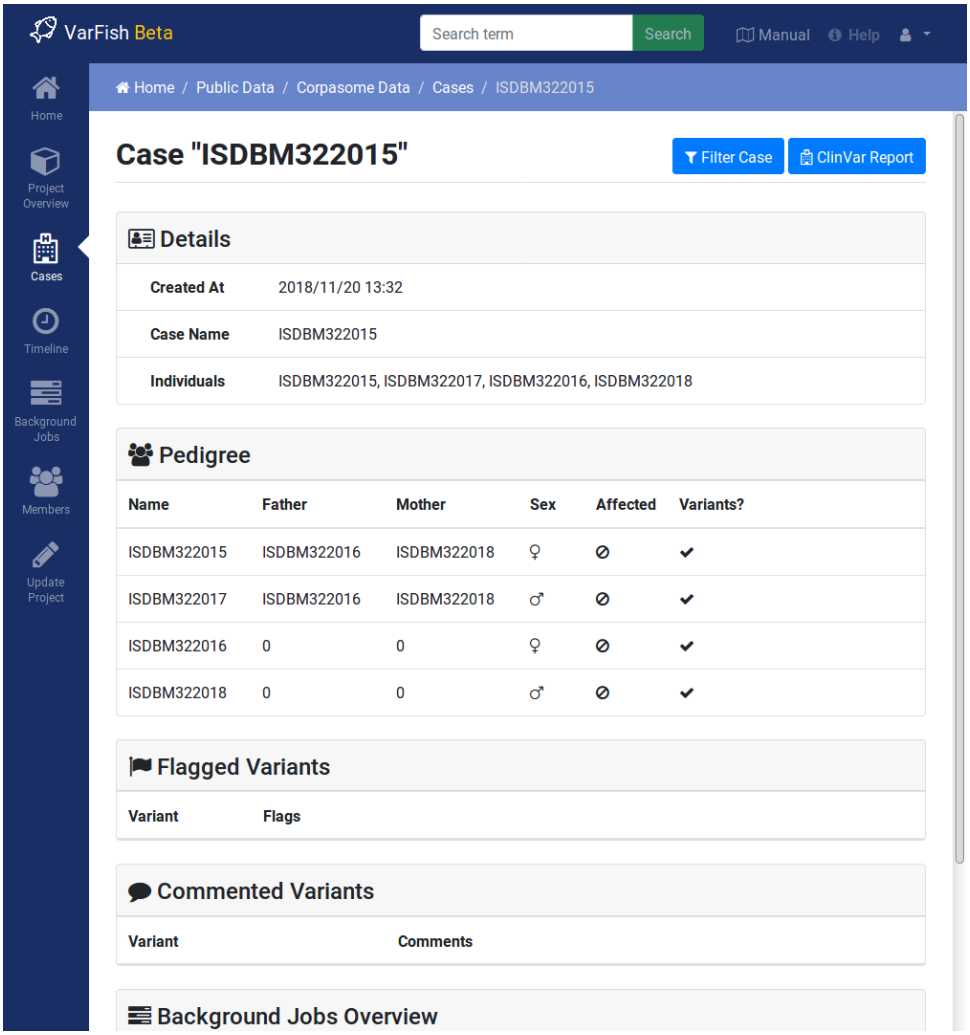

VarFish (Kiosk) Beta
Manual
Help
Case pfeiffer
Filter Variants
Edit Pedigree
Overview
Quality Control
Variant Annotation
Queries & Jobs
Details
Case Name
pfeiffer
Individuals
pfeiffer
Created At
2019/12/20 09:02
Last Modified
2019/12/20 09:02
Status, Notes & Tags
initial
initial
active
closed as unsolved
closed as uncertain
closed as solved
Called Variants
313,285
Case Comments
No case comments yet.
Enter comment here
Add Comment
Flag & Comment Summary
ACMG-Classified Variants
1
V:1 IV:0 III:0 II:0 I:0
Flagged Variants
1
:0 :0
Commented Variants
1
Pedigree
Name
Father
Mother
Sex
Affected
Variants?
pfeiffer
0
0
Annotation Release Info
Genomebuild
Table
Release
GRCh37
clinvar
2019-06-22
GRCh37
exac
r1.0






```
FAM index   father mother 1 2
FAM sibling father mother 1 1
FAM father  0      0      1 1
FAM mother  0      0      2 1
```


VarFish (Kiosk) Beta
Manual
Help
Background File Creation Job Create xlsx file for case pfeiffer
Back to Filter
Operation
Results & Resubmit
Download Result
Download xlsx File (46.8 KB)
Re-Submit
File Type:
Excel (.xlsx)
Re-run with File Type
Use this form to create a new job that uses the same filter settings but a different file type.
The VCF file export contains the bare minimum information (genomic variant and genotype, coverage, allelic depth, genotype call quality score). Most filters work but please note that the HGMD public membership filtration is ignored as well as any filters for flags, comments, and ClinVar details.

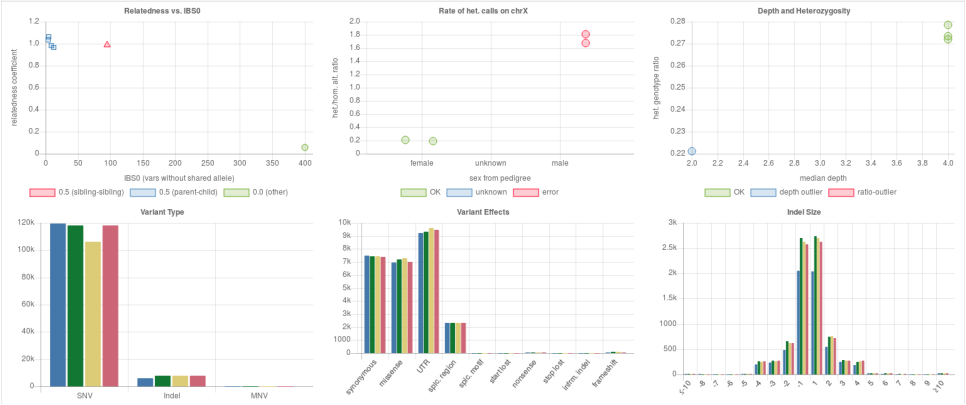

VarFish Beta
Search term
Search
Manual
Help
Home
Project Overview
Cases
Timeline
Background Jobs
Members
Update Project
Home / Public Data / Corpasome Data / Cases / ISDBM322015
Case "ISDBM322015"
Filter Case
ClinVar Report
Details
Created At 2018/11/20 13:32
Case Name ISDBM322015
Individuals ISDBM322015, ISDBM322017, ISDBM322016, ISDBM322018
Pedigree
Name Father Mother Sex Affected Variants?
ISDBM322015 ISDBM322016 ISDBM322018 ♀ ⊘ ✔
ISDBM322017 ISDBM322016 ISDBM322018 ♂ ⊘ ✔
ISDBM322016 0 0 ♀ ⊘ ✔
ISDBM322018 0 0 ♂ ⊘ ✔
Flagged Variants
Variant Flags
Commented Variants
Variant Comments
Background Jobs Overview

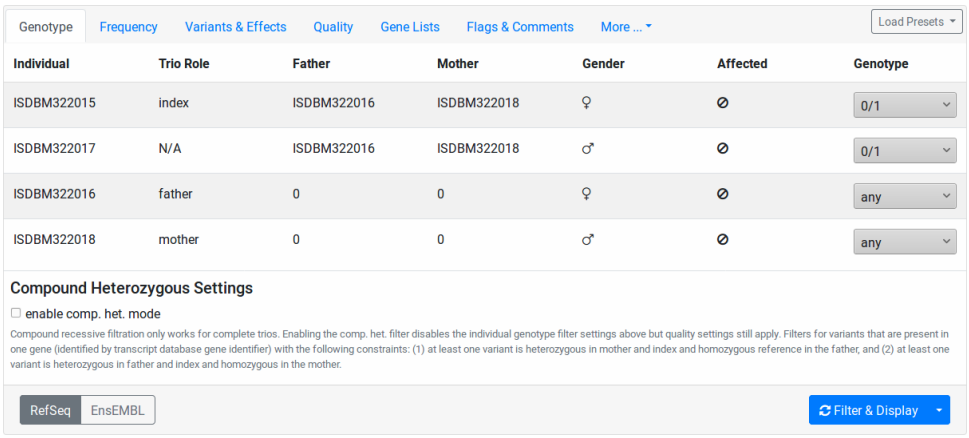
VarFish App Overview

Cases

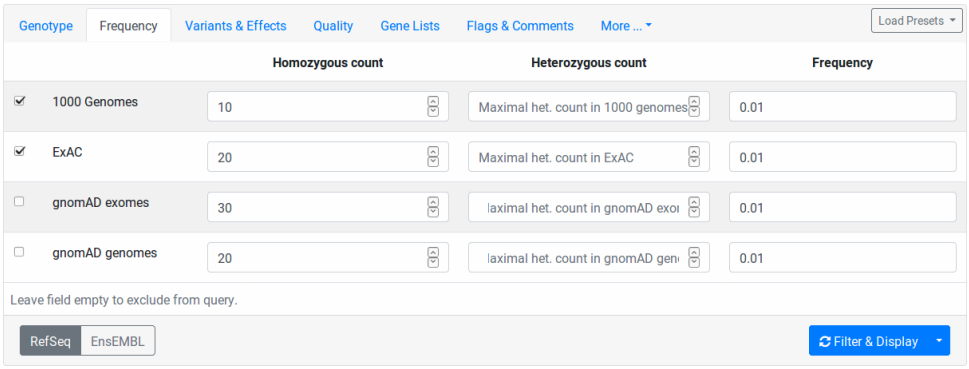
Created	Name	Individuals
2018/11/26 13:38	ISDBM322015	ISDBM322015, ISDBM322017, ISDBM322016, ISDBM322018

QC Plots

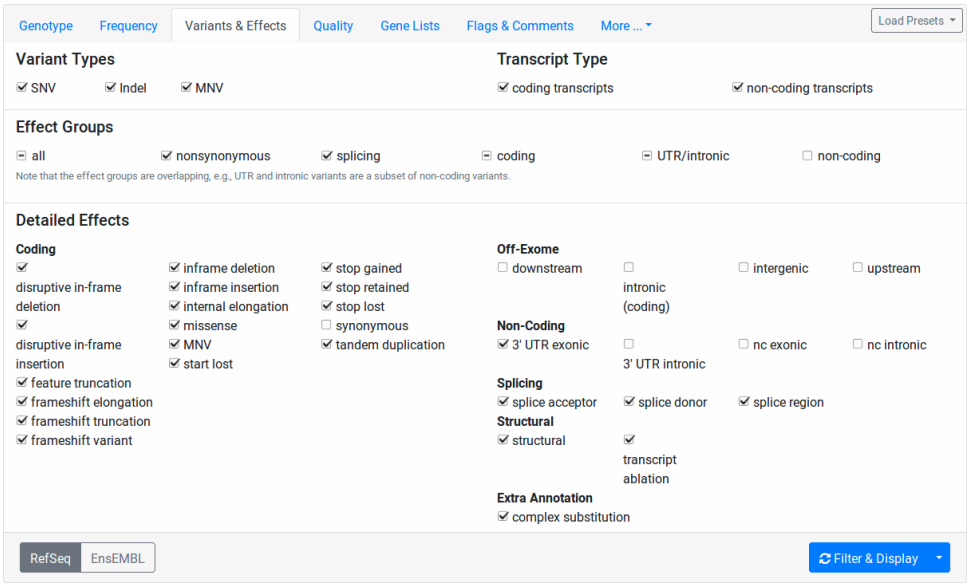


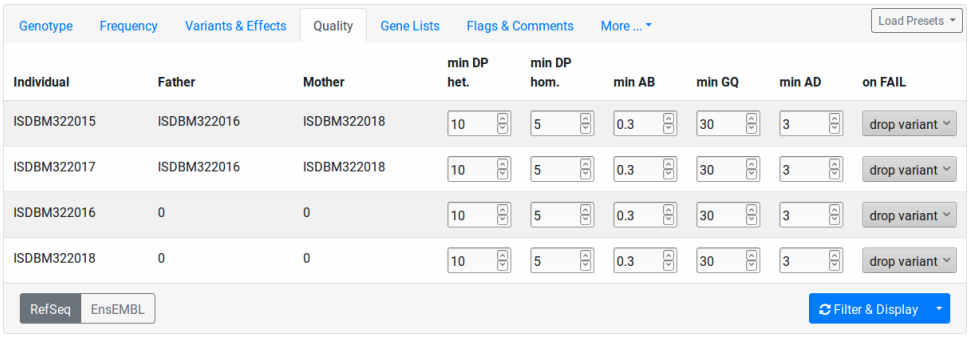

Genotype
Frequency
Variants & Effects
Quality
Gene Lists
Flags & Comments
More ...
Load Presets
Individual
Trio Role
Father
Mother
Gender
Affected
Genotype
ISDBM322015
index
ISDBM322016
ISDBM322018
0/1
ISDBM322017
N/A
ISDBM322016
ISDBM322018
0/1
ISDBM322016
father
0
0
any
ISDBM322018
mother
0
0
any
Compound Heterozygous Settings
enable comp. het. mode
Compound recessive filtration only works for complete trios. Enabling the comp. het. filter disables the individual genotype filter settings above but quality settings still apply. Filters for variants that are present in one gene (identified by transcript database gene identifier) with the following constraints: (1) at least one variant is heterozygous in mother and index and homozygous reference in the father, and (2) at least one variant is heterozygous in father and index and homozygous in the mother.
RefSeq
EnsEMBL
Filter & Display

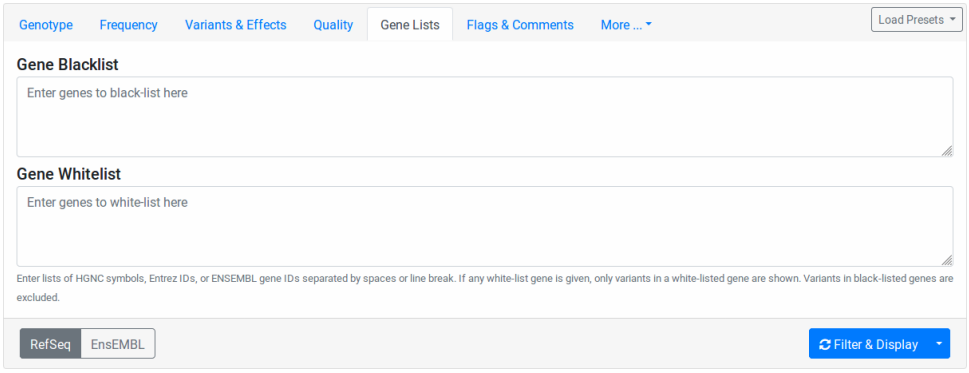
```
./.
./.
```

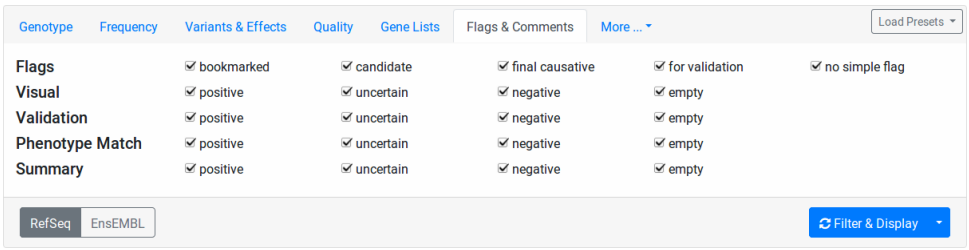

Genotype
Frequency
Variants & Effects
Quality
Gene Lists
Flags & Comments
More ...
Load Presets
Homozygous count
Heterozygous count
Frequency
1000 Genomes
10
Maximal het. count in 1000 genomes
0.01
ExAC
20
Maximal het. count in ExAC
0.01
gnomAD exomes
30
0.01
gnomAD genomes
20
0.01
Leave field empty to exclude from query.
RefSeq
EnsEMBL
Filter & Display

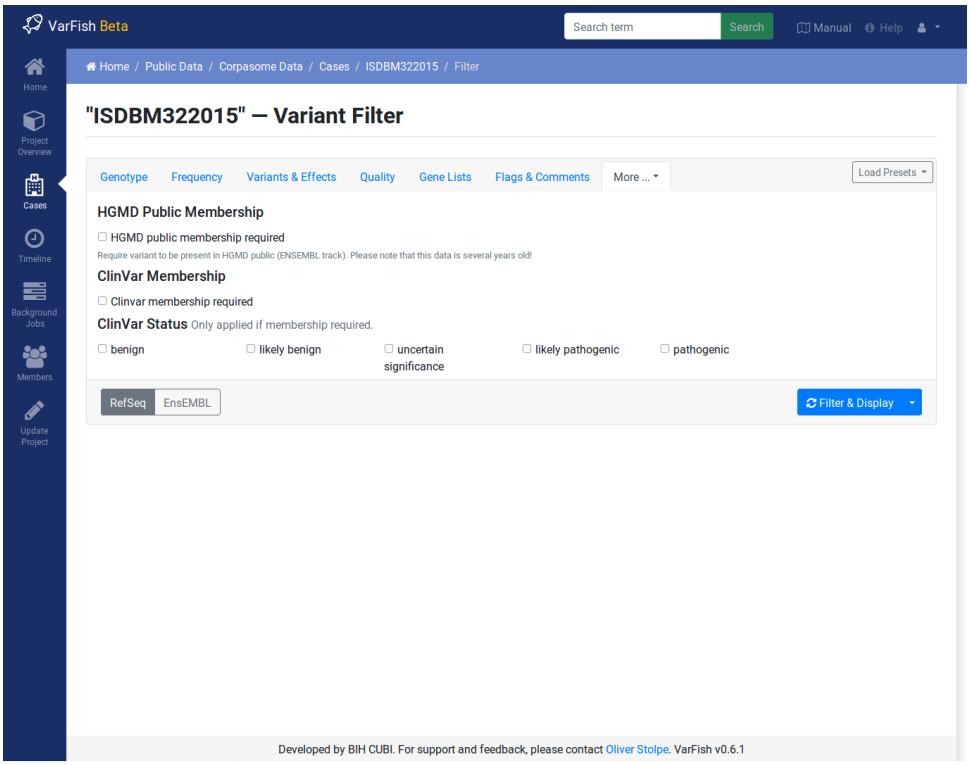
```
10
0.01
```


Genotype
Frequency
Variants & Effects
Quality
Gene Lists
Flags & Comments
More ...
Load Presets
Variant Types
SNV
Indel
MNV
Transcript Type
coding transcripts
non-coding transcripts
Effect Groups
all
nonsynonymous
splicing
coding
UTR/intronic
non-coding
Note that the effect groups are overlapping, e.g., UTR and intronic variants are a subset of non-coding variants.
Detailed Effects
Coding
disruptive in-frame deletion
disruptive in-frame insertion
feature truncation
frameshift elongation
frameshift truncation
frameshift variant
inframe deletion
inframe insertion
internal elongation
missense
MNV
start lost
stop gained
stop retained
stop lost
synonymous
tandem duplication
Off-Exome
downstream
intronic (coding)
intergenic
upstream
Non-Coding
3' UTR exonic
3' UTR intronic
nc exonic
nc intronic
Splicing
splice acceptor
splice donor
splice region
Structural
structural
transcript ablation
Extra Annotation
complex substitution
RefSeq
EnsEMBL
Filter & Display

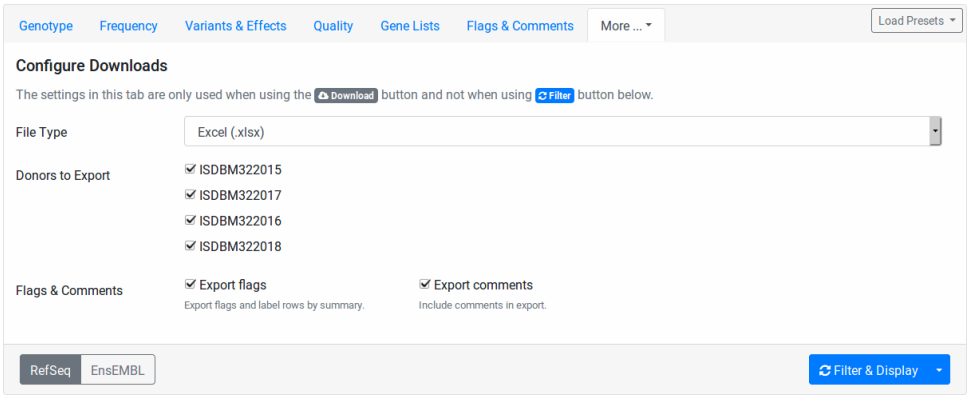

Genotype
Frequency
Variants & Effects
Quality
Gene Lists
Flags & Comments
More ...
Load Presets
Individual
Father
Mother
min DP het.
min DP hom.
min AB
min GQ
min AD
on FAIL
ISDBM322015
ISDBM322016
ISDBM322018
ISDBM322017
ISDBM322016
0
0
ISDBM322018
0
0
10
5
0.3
30
3
drop variant
RefSeq
EnsEMBL
Filter & Display

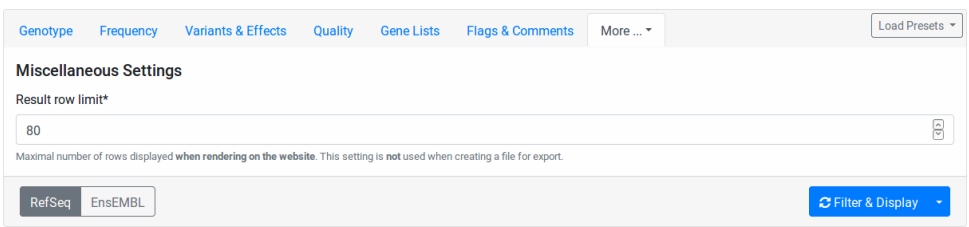

Genotype
Frequency
Variants & Effects
Quality
Gene Lists
Flags & Comments
More ...
Load Presets
Gene Blacklist
Enter genes to black-list here
Gene Whitelist
Enter genes to white-list here
Enter lists of HGNC symbols, Entrez IDs, or ENSEMBL gene IDs separated by spaces or line break. If any white-list gene is given, only variants in a white-listed gene are shown. Variants in black-listed genes are excluded.
RefSeq
EnsEMBL
Filter & Display

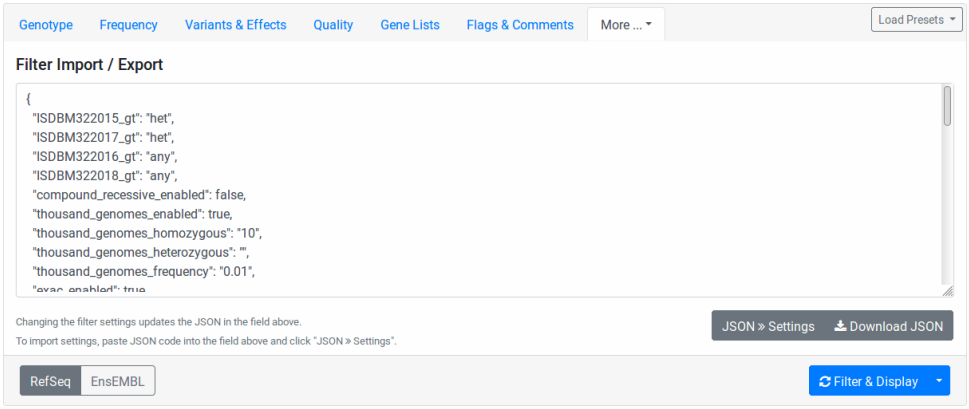

Genotype
Frequency
Variants & Effects
Quality
Gene Lists
Flags & Comments
More ...
Load Presets
Flags
bookmarked
candidate
final causative
for validation
no simple flag
Visual
positive
uncertain
negative
empty
Validation
positive
uncertain
negative
empty
Phenotype Match
positive
uncertain
negative
empty
Summary
positive
uncertain
negative
empty
RefSeq
EnsEMBL
Filter & Display

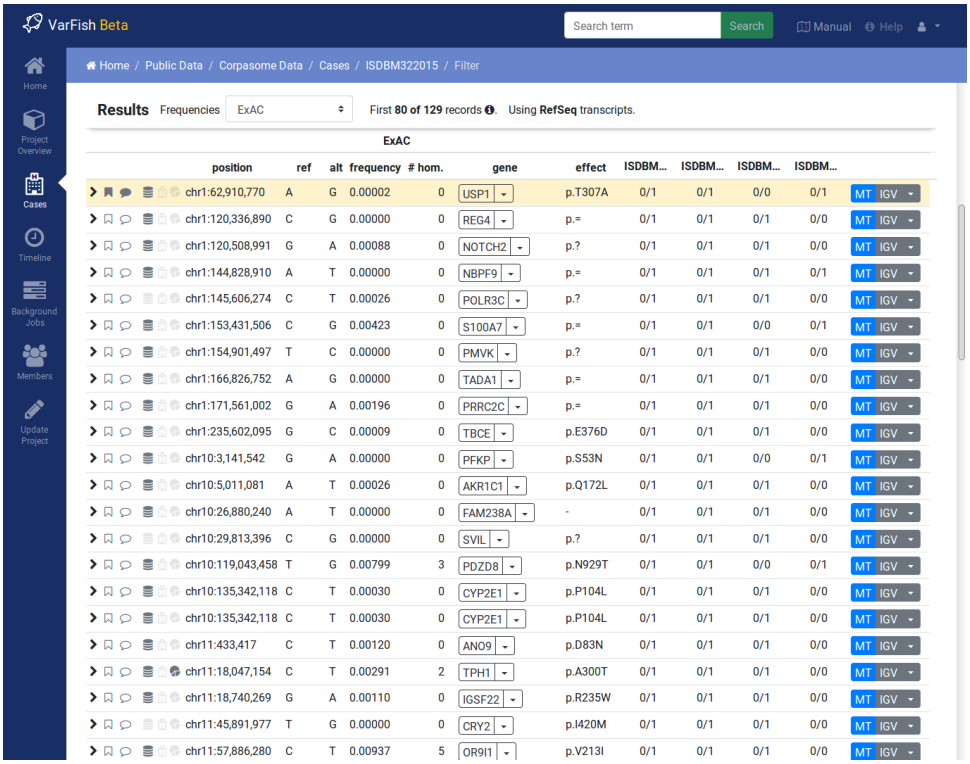


{# Bookmark for the variant bookmark popup #}

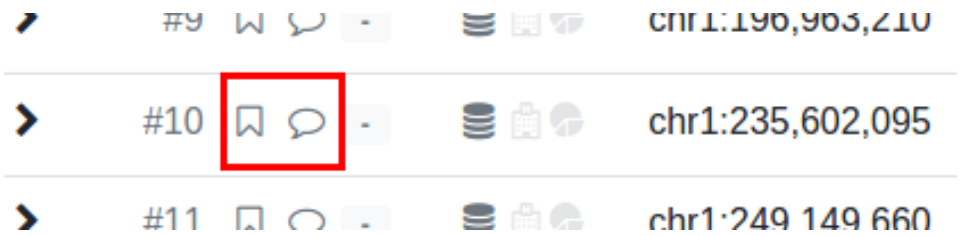


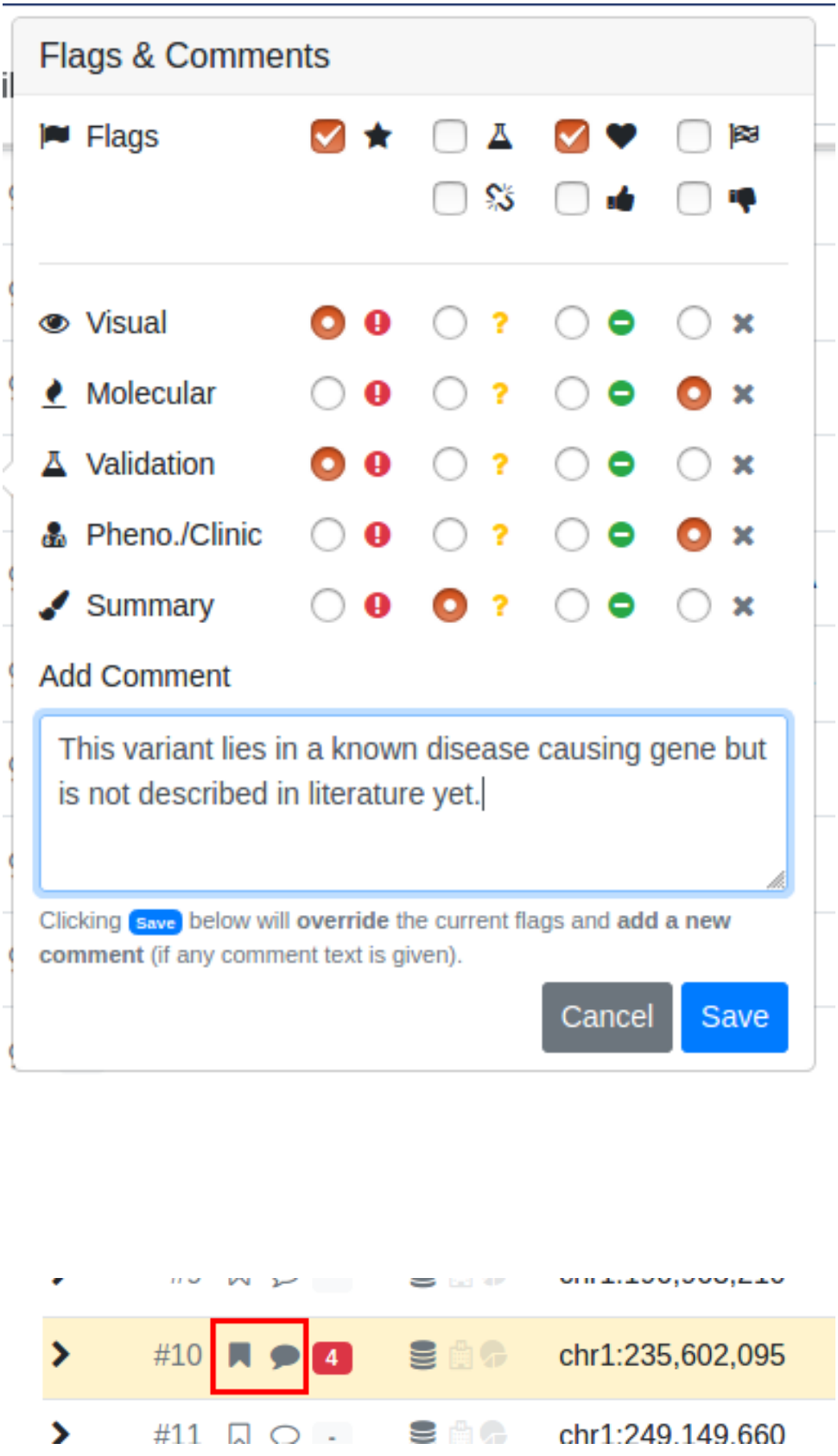


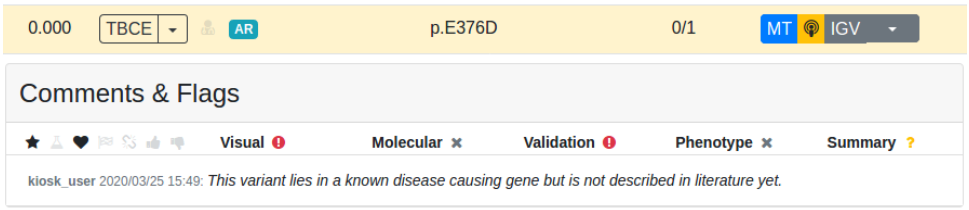

Genotype
Frequency
Variants & Effects
Quality
Gene Lists
Flags & Comments
More ...
Load Presets
Filter Import / Export
{
"ISDBM322015_gt": "het",
"ISDBM322017_gt": "het",
"ISDBM322016_gt": "any",
"ISDBM322018_gt": "any",
"compound_recessive_enabled": false,
"thousand_genomes_enabled": true,
"thousand_genomes_homozygous": "10",
"thousand_genomes_heterozygous": "",
"thousand_genomes_frequency": "0.01",
Changing the filter settings updates the JSON in the field above.
To import settings, paste JSON code into the field above and click "JSON » Settings".
JSON » Settings
Download JSON
RefSeq
EnsEMBL
Filter & Display

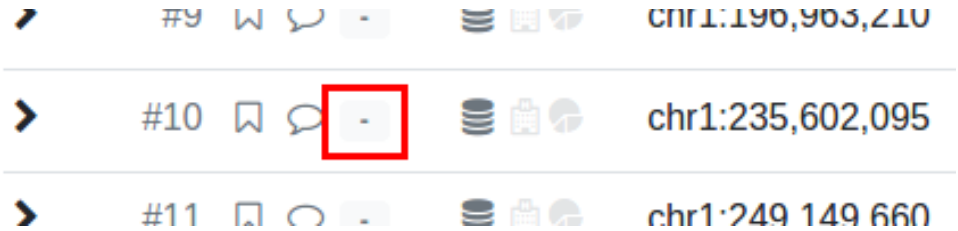

VarFish Beta
Search term
Search
Manual
Help
Home / Public Data / Corpasome Data / Cases / ISDBM322015 / Filter
Home
Project Overview
Cases
Timeline
Background Jobs
Members
Update Project
Results
Frequencies
ExAC
First 80 of 129 records.
Using RefSeq transcripts.
ExAC
position ref alt frequency # hom. gene effect ISDBM... ISDBM... ISDBM... ISDBM...
chr1:62,910,770 A G 0.00002 0 USP1 p.T307A 0/1 0/1 0/0 0/1 MT IGV
chr1:120,336,890 C G 0.00000 0 REG4 p.= 0/1 0/1 0/1 0/0 MT IGV
chr1:120,508,991 G A 0.00088 0 NOTCH2 p.? 0/1 0/1 0/1 0/0 MT IGV
chr1:144,828,910 A T 0.00000 0 NBPF9 p.= 0/1 0/1 0/1 0/1 MT IGV
chr1:145,606,274 C T 0.00026 0 POLR3C p.? 0/1 0/1 0/1 0/0 MT IGV
chr1:153,431,506 C G 0.00423 0 S100A7 p.= 0/1 0/1 0/0 0/1 MT IGV
chr1:154,901,497 T C 0.00000 0 PMVK p.? 0/1 0/1 0/1 0/0 MT IGV
chr1:166,826,752 A G 0.00000 0 TADA1 p.= 0/1 0/1 0/1 0/0 MT IGV
chr1:171,561,002 G A 0.00196 0 PRRC2C p.= 0/1 0/1 0/1 0/0 MT IGV
chr1:235,602,095 G C 0.00009 0 TBCE p.E376D 0/1 0/1 0/1 0/0 MT IGV
chr10:3,141,542 G A 0.00000 0 PFKP p.S53N 0/1 0/1 0/0 0/1 MT IGV
chr10:5,011,081 A T 0.00026 0 AKR1C1 p.Q172L 0/1 0/1 0/1 0/0 MT IGV
chr10:26,880,240 A T 0.00000 0 FAM238A - 0/1 0/1 0/1 0/0 MT IGV
chr10:29,813,396 C G 0.00000 0 SVIL p.? 0/1 0/1 0/1 0/0 MT IGV
chr10:119,043,458 T G 0.00799 3 PDZD8 p.N929T 0/1 0/1 0/0 0/1 MT IGV
chr10:135,342,118 C T 0.00030 0 CYP2E1 p.P104L 0/1 0/1 0/1 0/0 MT IGV
chr10:135,342,118 C T 0.00030 0 CYP2E1 p.P104L 0/1 0/1 0/1 0/0 MT IGV
chr11:433,417 C T 0.00120 0 ANO9 p.D83N 0/1 0/1 0/1 0/0 MT IGV
chr11:18,047,154 C T 0.00291 2 TPH1 p.A300T 0/1 0/1 0/1 0/0 MT IGV
chr11:18,740,269 G A 0.00110 0 IGSF22 p.R235W 0/1 0/1 0/1 0/0 MT IGV
chr11:45,891,977 T G 0.00000 0 CRY2 p.I420M 0/1 0/1 0/1 0/0 MT IGV
chr11:57,886,280 C T 0.00937 5 OR9I1 p.V213I 0/1 0/1 0/1 0/0 MT IGV

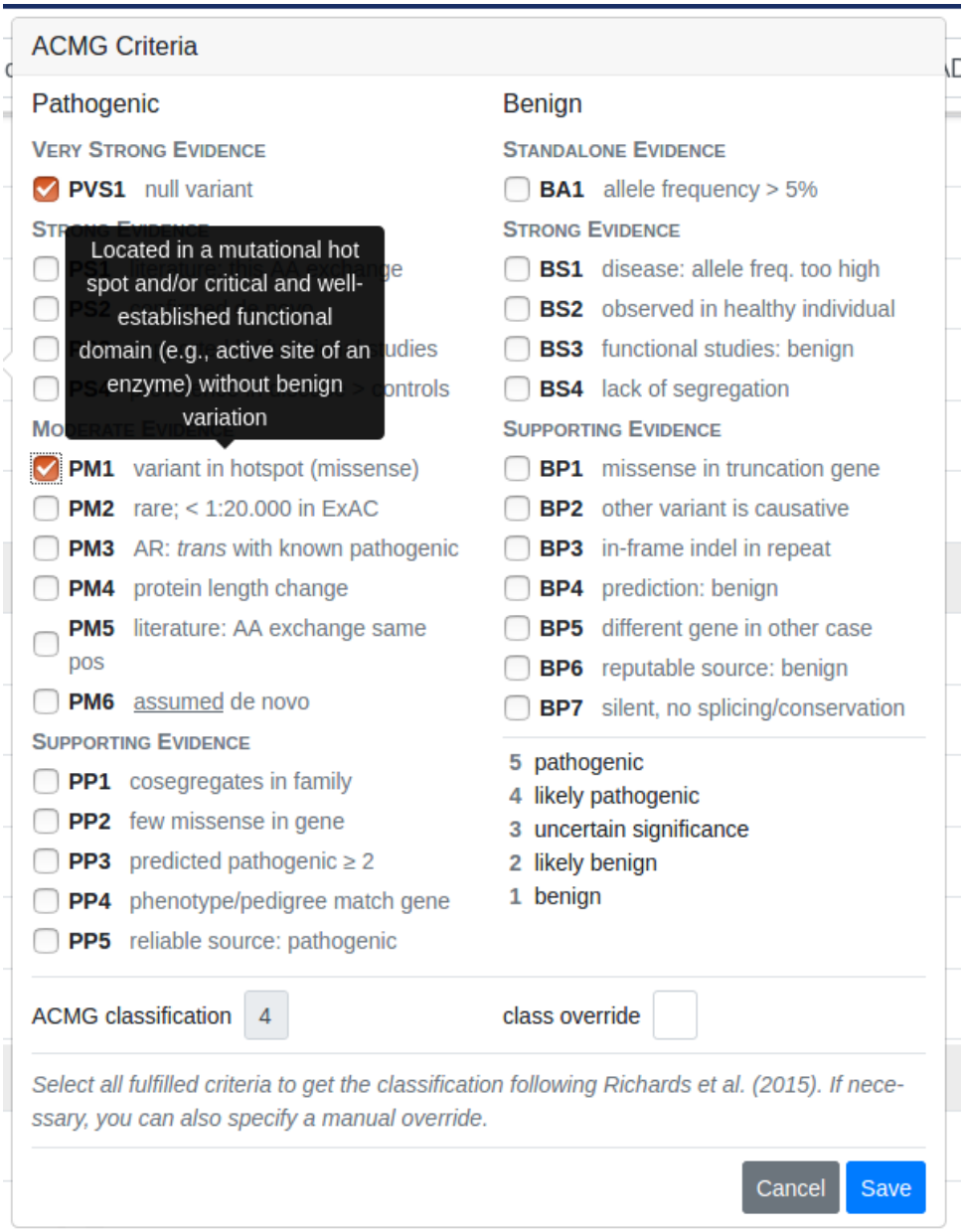
60151

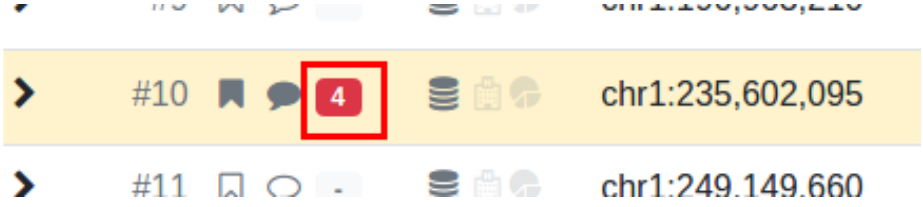

#10
chr1:235,602,095

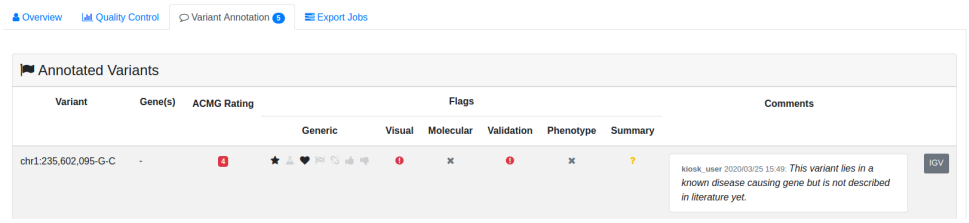

Flags & Comments
Flags
Visual
Molecular
Validation
Pheno./Clinic
Summary
Add Comment
This variant lies in a known disease causing gene but is not described in literature yet.
Clicking Save below will override the current flags and add a new comment (if any comment text is given).
Cancel
Save
#10
4
chr1:235,602,095
#11
chr1:249,149,660

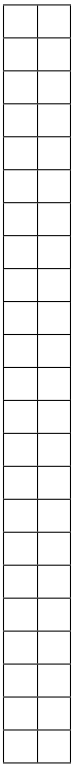

0.000
TBCE
AR
p.E376D
0/1
MT
IGV
Comments & Flags
Visual
Molecular
Validation
Phenotype
Summary
kiosk_user 2020/03/25 15:49: This variant lies in a known disease causing gene but is not described in literature yet.

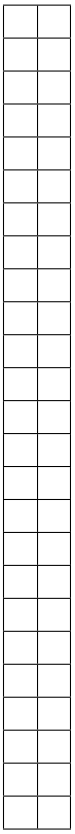

#10
chr1:235,602,095


ACMG Criteria
Pathogenic
Very Strong Evidence
PVS1 null variant
Strong Evidence
PS1 literature: this AA exchange
PS2
PS3 udies
PS4 ontrols
Located in a mutational hot spot and/or critical and well-established functional domain (e.g., active site of an enzyme) without benign variation
Moderate Evidence
PM1 variant in hotspot (missense)
PM2 rare; < 1:20.000 in ExAC
PM3 AR: trans with known pathogenic
PM4 protein length change
PM5 literature: AA exchange same pos
PM6 assumed de novo
Supporting Evidence
PP1 cosegregates in family
PP2 few missense in gene
PP3 predicted pathogenic ≥ 2
PP4 phenotype/pedigree match gene
PP5 reliable source: pathogenic
Benign
Standalone Evidence
BA1 allele frequency > 5%
Strong Evidence
BS1 disease: allele freq. too high
BS2 observed in healthy individual
BS3 functional studies: benign
BS4 lack of segregation
Supporting Evidence
BP1 missense in truncation gene
BP2 other variant is causative
BP3 in-frame indel in repeat
BP4 prediction: benign
BP5 different gene in other case
BP6 reputable source: benign
BP7 silent, no splicing/conservation
5 pathogenic
4 likely pathogenic
3 uncertain significance
2 likely benign
1 benign
ACMG classification 4
class override
Select all fulfilled criteria to get the classification following Richards et al. (2015). If necessary, you can also specify a manual override.
Cancel
Save


#10
4
chr1:235,602,095
#11
chr1:249,149,660


Overview
Quality Control
Variant Annotation 5
Export Jobs
Annotated Variants
Variant
Gene(s)
ACMG Rating
Flags
Generic
Visual
Molecular
Validation
Phenotype
Summary
Comments
chr1:235,602,095-G-C
-
4
kiosk_user 2020/03/25 15:49: This variant lies in a known disease causing gene but is not described in literature yet.
IGV





https://genome-euro.ucsc.edu/cgi-bin/hgTracks?db=hg19&position=21:11038733-11038733

```
$ git version
git version 1.8.3.1
$ docker-compose -version
docker-compose version 1.28.2, build 67630359
$ docker version
Client: Docker Engine - Community
 Version:           20.10.3
[...]
```

varfish-docker-composedocker-compose.yml

```
$ git clone https://github.com/bihealth/varfish-docker-compose.git
$ cd varfish-docker-compose
```

volumesgrch37grch38

```
$ wget --no-check-certificate https://file-public.cubi.bihealth.org/transient/
↪varfish/anthenea/varfish-site-data-v1-20210728-grch37.tar.gz{,.sha256}
$ sha256sum --check varfish-site-data-v1-20210728-grch37.tar.gz.sha256
$ tar xf varfish-site-data-v1-20210728-grch37.tar.gz
$ ls volumes
exomiser  jannovar  minio  postgres  redis  traefik
```

.envenv.exampleDJANGO_SECRET_KEY

```
$ cp env.example .env
$ $EDITOR .env
```

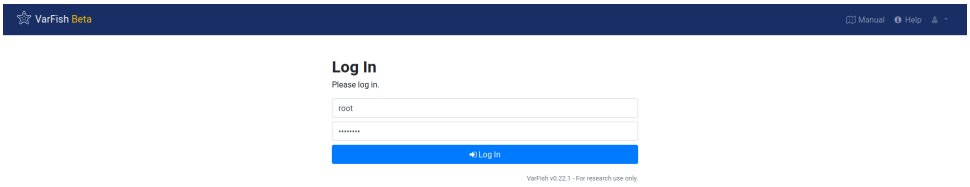
https://<your-host>/Ctrl-C

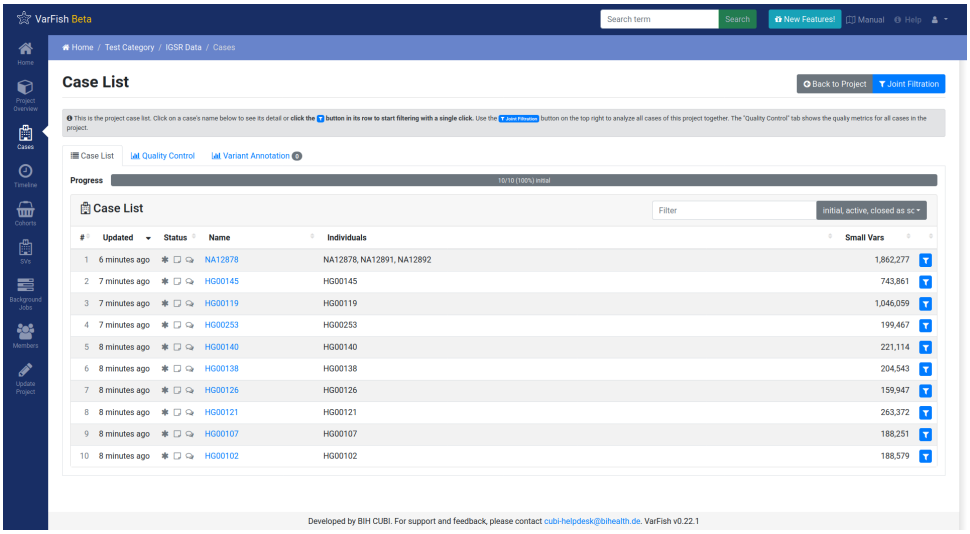
```
$ docker-compose up
```

```
$ docker-compose up -d
Starting compose_exomiser-rest-prioritiser_1 ... done
Starting compose_jannovar_1                  ... done
Starting compose_traefik_1                   ... done
Starting compose_varfish-web_1               ... done
Starting compose_postgres_1                  ... done
Starting compose_redis_1                     ... done
Starting compose_minio_1                     ... done
Starting compose_varfish-celeryd-query_1     ... done
Starting compose_varfish-celeryd-default_1   ... done
Starting compose_varfish-celeryd-import_1    ... done
Starting compose_varfish-celerybeat_1        ... done
```

```
$ docker ps
3ec78fb9f12c   bihealth/varfish-server:0.22.1-0                              "docker-
↪entrypoint.s…"   17 hours ago   Up 31 seconds   8080/tcp                  ␣
↪          compose_varfish-celeryd-import_1
313afb611ab1   bihealth/varfish-server:0.22.1-0                              "docker-
↪entrypoint.s…"   17 hours ago   Up 30 seconds   8080/tcp                  ␣
↪          compose_varfish-celerybeat_1
4d865726e83b   bihealth/varfish-server:0.22.1-0                              "docker-
↪entrypoint.s…"   17 hours ago   Up 31 seconds   8080/tcp                  ␣
↪          compose_varfish-celeryd-query_1
```

```
a5f90232c4da   bihealth/varfish-server:0.22.1-0                                  "docker-
↪entrypoint.s…"   17 hours ago   Up 31 seconds   8080/tcp                                 ␣
↪          compose_varfish-celeryd-default_1
96cec7caebe4   bihealth/varfish-server:0.22.1-0                                  "docker-
↪entrypoint.s…"   17 hours ago   Up 33 seconds   8080/tcp                                 ␣
↪          compose_varfish-web_1
8d1f310c9b48   postgres:12                                                       "docker-
↪entrypoint.s…"   17 hours ago   Up 32 seconds   5432/tcp                                 ␣
↪          compose_postgres_1
8f12e16e20cd   minio/minio                                                       "/usr/
↪bin/docker-ent…"   17 hours ago   Up 32 seconds   9000/tcp                             ␣
↪            compose_minio_1
03e877ac11db   quay.io/biocontainers/jannovar-cli:0.33--0
↪"jannovar -Xmx6G -Xm…"   17 hours ago   Up 33 seconds                                    ␣
↪                  compose_jannovar_1
6af09b819e59   traefik:v2.3.1                                                    "/
↪entrypoint.sh --pr…"   17 hours ago   Up 33 seconds   0.0.0.0:80->80/tcp, 0.0.0.
↪0:443->443/tcp   compose_traefik_1
514cb4386224   redis:6                                                           "docker-
↪entrypoint.s…"   19 hours ago   Up 32 seconds   6379/tcp                                 ␣
↪          compose_redis_1
5678b9e6797b   quay.io/biocontainers/exomiser-rest-prioritiser:12.1.0--1
↪"exomiser-rest-prior…"   19 hours ago   Up 34 seconds                                    ␣
↪                  compose_exomiser-rest-prioritiser_1
```






```
varfish-db-downloader/
    GRCh37/
        <table_group>/
            <version>/
                <table>.tsv
                <table>.release_info
    GRCh37/
        [...]
    noref/
        [...]
    import_versions.tsv
    [...]
```

```
varfish-db-downloader/
    noref/
        hpo/
            20220126/
                Hpo.release_info
                Hpo.tsv
                HpoName.release_info
                HpoName.tsv
        mim2gene/
            20220126/
                Mim2geneMedgen.release_info
                Mim2geneMedgen.tsv
    import_versions.tsv
    [...]
```

```
$ cd varfish-docker-compose  # make sure to be in the docker compose folder
$ docker-compose down
```

`docker-compose.yml`

```
varfish-web:
  image: ghcr.io/bihealth/varfish-server:VERSION
  env_file:
    - .env
  networks:
    - varfish
  restart: unless-stopped
  volumes:
    - "/root/varfish-server-background-db-20210728:/data:ro"
  [...]
```

```
volumes:
  - "/root/varfish-server-background-db-20210728:/data:ro"
  - type: bind
    source: varfish-db-downloader/
    target: /data-db-downloader
    read_only: true
```

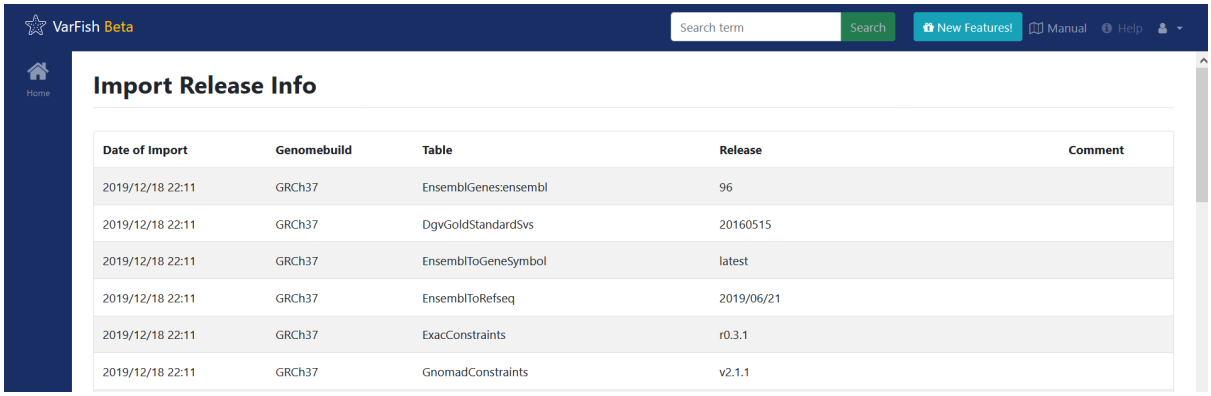
```
$ docker-compose up
```

```
$ docker exec -it varfish-docker-compose_varfish-web_1 bash -i
```

```
varfish-web-container$ cd /usr/src/app
varfish-web-container$ python manage.py import_tables --tables-path /data-db-
↪downloader
```

```
Disabling autovacuum on all tables...
Hpo -- Importing Hpo 2022/01/26 (, source: /data-db-downloader/noref/hpo/20220126/
↪Hpo.tsv) ...
Mim2geneMedgen -- Importing Mim2geneMedgen 2022/01/26 (, source: /data-db-
↪downloader/noref/mim2gene/20220126/Mim2geneMedgen.tsv) ...
Hpo -- Removing old Hpo results.
Mim2geneMedgen -- Removing old Mim2geneMedgen results.
Mim2geneMedgen -- Importing new Mim2geneMedgen data
Hpo -- Importing new Hpo data
Mim2geneMedgen -- Finished importing Mim2geneMedgen 2022/01/26 (Mim2geneMedgen.tsv)
Hpo -- Finished importing Hpo 2022/01/26 (Hpo.tsv)
HpoName -- Importing HpoName 2022/01/26 (, source: /data-db-downloader/noref/hpo/
↪20220126/HpoName.tsv) ...
HpoName -- Removing old HpoName results.
HpoName -- Importing new HpoName data
HpoName -- Finished importing HpoName 2022/01/26 (HpoName.tsv)
Enabling autovacuum on all tables...
```

Import Release Info


VarFish Beta
Search term
Search
New Features!
Manual
Help
Home
Import Release Info
Date of Import
Genomebuild
Table
Release
Comment
2019/12/18 22:11
GRCh37
EnsemblGenes:ensembl
96
2019/12/18 22:11
GRCh37
DgvGoldStandardSvs
20160515
2019/12/18 22:11
GRCh37
EnsemblToGeneSymbol
latest
2019/12/18 22:11
GRCh37
EnsemblToRefseq
2019/06/21
2019/12/18 22:11
GRCh37
ExacConstraints
r0.3.1
2019/12/18 22:11
GRCh37
GnomadConstraints
v2.1.1

volumes/cadd-rest-apivarfish-docker-compose

```
$ cd varfish-docker-compose
$ mkdir -p volumes/cadd-rest-api/db
$ curl https://raw.githubusercontent.com/kircherlab/CADD-scripts/7502f47/install.
↪sh \
    > volumes/cadd-rest-api/install.sh
```

install.sh

```
$ docker run -it -e CADD=/opt/miniconda3/share/cadd-scripts-1.6-0 \
    -v $PWD/volumes/cadd-rest-api:/data bihealth/cadd-rest-api:0.3.1-0 \
    bash /data/install.sh -b
Using kircherlab.bihealth.org as download server
CADD-v1.6 (c) University of Washington, Hudson-Alpha Institute for Biotechnology␣
↪and Berlin Institute of Health 2013-
2020. All rights reserved.

The following questions will quide you through selecting the files and␣
↪dependencies needed for CADD.
After this, you will see an overview of the selected files before the download and␣
↪installation starts.
Please note, that for successfully running CADD locally, you will need the conda␣
↪environment and at least one set of
annotations.

Do you want to install the virtual environments with all CADD dependencies via␣
↪conda? (y)/n n
Do you want to install CADD v1.6 for GRCh37/hg19? (y)/n y
Do you want to install CADD v1.6 for GRCh38/hg38? (y)/n n
Do you want to load annotations (Annotations can also be downloaded manually from␣
↪the website)? (y)/n y
Do you want to load prescored variants (Makes SNV calling faster. Can also be␣
↪loaded/installed later.)? y/(n) y
Do you want to load prescored variants for scoring with annotations (Warning:␣
↪These files are very big)? y/(n) y
Do you want to load prescored variants for scoring without annotations? y/(n) y
Do you also want to load prescored InDels? We provide scores for well known InDels␣
↪from sources like ClinVar, gnomAD/TOPMed etc. y/(n) y

The following will be loaded: (disk space occupied)
```

```
 - Download CADD annotations for GRCh37-v1.6 (121 GB)
 - Download prescored SNV inclusive annotations for GRCh37-v1.6 (248 GB)
 - Download prescored InDels inclusive annotations for GRCh37-v1.6 (3.4 GB)
 - Download prescored SNV (without annotations) for GRCh37-v1.6 (78 GB)
 - Download prescored InDels (without annotations) for GRCh37-v1.6 (0.6 GB)
Please make sure you have enough disk space available.
Ready to continue? (y)/n y
Starting installation. This will take some time.
[...]
Connecting to kircherlab.bihealth.org (kircherlab.bihealth.org)|141.80.169.4|:443..
↪. connected.
HTTP request sent, awaiting response... 200 OK
Length: 61 [application/x-gzip]
Saving to: ‘InDels_inclAnno.tsv.gz.tbi.md5’

InDels_inclAnno.tsv.gz.tbi.md5             100
↪%[=======================================================================================>
↪]      61  --.-KB/s    in 0s
2021-03-08 18:55:10 (19.9 MB/s) - ‘InDels_inclAnno.tsv.gz.tbi.md5’ saved [61/61]

InDels_inclAnno.tsv.gz: OK
InDels_inclAnno.tsv.gz.tbi: OK
```

.env.env

```
# Extra: CADD REST API ************************************************

# Uncomment the following lines to enable variant prioritization using the
# CADD score.  See the VarFish Server manual for installation instructions,
# in particular how to download the required data.
VARFISH_ENABLE_CADD=1
VARFISH_CADD_REST_API_URL=http://cadd-rest-api:8080
VARFISH_CADD_MAX_VARS=5000
```

docker-compose.ymlcadd-rest-api-servercadd-rest-api-celeryddocker-compose.yml

```
# Uncomment the following lines to enable the CADD REST API server that
# is used for variant prioritization using the CADD score.  We need both
# the server and the CADD-based worker.
cadd-rest-api-server:
  image: bihealth/cadd-rest-api:0.3.1-0
  env_file: cadd-rest-api.env
  command: ["wsgi"]
  # [...]

# You have to provide multiple cadd-rest-api-celeryd-worker container if
# you want to handle more than one query at a time.
cadd-rest-api-celeryd-worker-1:
[...]
cadd-rest-api-celeryd-worker-3:
  image: bihealth/cadd-rest-api:0.3.2-0
  env_file: cadd-rest-api.env
  command: ["celeryd"]
  networks: [varfish]
  restart: unless-stopped
  volumes:
    - "./volumes/cadd-rest-api/data/annotations:/opt/miniconda3/share/cadd-scripts-
↪1.6-0/data/annotations:ro"
    - "./volumes/cadd-rest-api/data/prescored:/opt/miniconda3/share/cadd-scripts-1.
↪6-0/data/prescored:ro"
    - "./volumes/cadd-rest-api/db:/data/db:rw"
```

```
docker-compose down && docker-compose up -d
```

varfish-docker-composevarfish-docker-compose

docker-compose.yml

docker-compose.override.ymldocker-compose upvarfish-docker-composedocker-compose.yml

docker-compose.override.yml

- docker-compose.override.yml-cert
- docker-compose.override.yml-letsencrypt
- docker-compose.override.yml-cadd

*.override.yml-*docker-compose.yml

varfish-docker-compose

docker-compose.override.yml-certconfig/traefik/tls/server.crtserver.key traefik

docker-compose.override.yml-letsencrypt--certificatesresolvers.le.acme.email=LICENSE

docker-compose down && docker-compose up -d

.envvarfish-docker-compose

ENABLE_LDAP=0 01

AUTH_LDAP_SERVER_URI= ldap://ldap.example.com:portldaps://...

AUTH_LDAP_BIND_DN=

AUTH_LDAP_BIND_PASSWORD=

AUTH_LDAP_USER_SEARCH_BASE= DC=com,DC=example,DC=ldap

AUTH_LDAP_USERNAME_DOMAIN= EXAMPLEuser@EXAMPLE

AUTH_LDAP_DOMAIN_PRINTABLE=${AUTH_LDAP_USERNAME_DOMAIN}

ENABLE_LDAP_SECONDARY=0 01

AUTH_LDAP2AUTH_LDAP

https://varfish.example.com/saml2_auth/acs/

docker-compose.ymldocker-compose-overrrided.yml

```
varfish-web:
  ...
  volumes:
    - "/path/to/my/secrets:/secrets:ro"
```

.env

ENABLE_SAML

SAML_CLIENT_ENTITY_ID

SAML_CLIENT_ENTITY_URL

SAML_CLIENT_METADATA_FILE

SAML_CLLIENT_IDP

SAML_CLIENT_KEY_FILE

SAML_CLIENT_CERT_FILE

SAML_CLIENT_XMLSEC1

SAML_ATTRIBUTES_MAP

SAML_ATTRIBUTES_MAP emailusernamefirst_namelast_nameSAML_ATTRIBUTES_MAP="email=email,
    username=uid,first_name=firstName,last_name=name"

SAML_NEW_USER_GROUPS

SAML_NEW_USER_ACTIVE_STATUS

SAML_NEW_USER_STAFF_STATUS

SAML_NEW_USER_SUPERUSER_STATUS

PROJECTROLES_SEND_EMAIL=0

EMAIL_SENDER= noreply@varfish.example.com

EMAIL_SUBJECT_PREFIX= [VarFish]

EMAIL_URL= smtp://user:password@mail.example.com:1234

DATABASE_URL=postgresql://postgres:password@postgres/varfish

docker-compose

VARFISH_LOGIN_PAGE_TEXT

FIELD_ENCRYPTION_KEY python -c 'import os, base64; print(base64.urlsafe_b64encode(os.urandom(32)))'

VARFISH_QUERY_MAX_UNION 20

ENABLE_SENTRY=0

SENTRY_DSN=

VARFISH_ENABLE_HGMD_PRO_LINKOUT=0

VARFISH_HGMD_PRO_LINKOUT_URL_PREFIX=https://my.qiagendigitalinsights.com/bbp/view/hgmd/

traefik

```yaml
services:
  traefik:
    ports:
      - "80:80"
      - "443:443"
```

80808443

webwebsecure

```yaml
services:
  traefik:
    command:
      # ...
      - "--entrypoints.web.address=:80"
      - "--entrypoints.websecure.address=:443"
```

```yaml
services:
  traefik:
    command:
      # ...
      - "--entrypoints.web.address=:8080"
      - "--entrypoints.websecure.address=:8443"
```

docker-compose.ymldocker-compose.override.ymldocker-compose.override.yml-*

```yaml
services:
  traefik:
    ports:
      - "8080:80"
      - "8443:443"
    command:
      - "--providers.docker=true"
      - "--providers.docker.exposedbydefault=false"
      - "--entrypoints.web.address=:80"
      - "--entrypoints.web.http.redirections.entryPoint.to=websecure"
      - "--entrypoints.web.http.redirections.entryPoint.scheme=https"
      - "--entrypoints.web.http.redirections.entrypoint.permanent=true"
      - "--entrypoints.web.address=:80"
      - "--entrypoints.websecure.address=:443"
```

docker-compose up -ddocker-compose.yml

traefik

ports

```yaml
services:
  traefik:
    ports:
      - "80:80"
      - "443:443"
```

```yaml
services:
  traefik:
    ports:
      - "10.0.0.1:80:80"
      - "10.0.0.1:443:443"
```

10.0.0.1:8080:80

firewalldufw

volumesvarfish-docker-compose

cadd-rest-api

exomiser

jannovar

minio

postgres

redis

traefik

redisexomiserpostgrescadd-rest-apiminio

minio

cadd-rest-api

postgres

VARFISH_ENABLE_BEACON_SITE=

conda-forgebiocondadefaultsvarfish-annotator

```
# EITHER
$ conda install -y varfish-annotator-cli==0.14.0
# OR
$ conda create -y -n varfish-annotator varfish-annotator-cli==0.14.0
$ conda activate varfish-annotator
```

mambamamba installcreateconda installcreate

```
$ GENOME=grch37      # alternatively use grch38
$ RELEASE=20210728
$ mkdir varfish-annotator-20210728-$GENOME
$ cd varfish-annotator-20210728-$GENOME
$ wget --no-check-certificate \
    https://file-public.cubi.bihealth.org/transient/varfish/anthenea/varfish-
↪annotator-db-$RELEASE-$GENOME.h2.db.gz{,.sha256} \
    https://file-public.cubi.bihealth.org/transient/varfish/anthenea/jannovar-db-
↪$RELEASE-$GENOME.tar.gz{,.sha256}
$ sha256sum --check varfish-annotator-db-$RELEASE-$GENOME.h2.db.gz.sha256
varfish-annotator-db-20210728-grch37.h2.db.gz: OK
$ sha256sum --check jannovar-db-$RELEASE-$GENOME.tar.gz.sha256
jannovar-db-20210728-grch37.tar.gz: OK
$ gzip -d varfish-annotator-db-$RELEASE-$GENOME.h2.db.gz
$ tar xf jannovar-db-$RELEASE-$GENOME.tar.gz
$ rm jannovar-db-20210728-$RELEASE.tar.gz{,.sha256} \
```

```
    varfish-annotator-db-$RELEASE-$GENOME.h2.db.gz.sha256
$ mv jannovar-db-$RELEASE-$GENOME/* .
$ rmdir jannovar-db-$RELEASE-$GENOME
```

```
# use $GENOME and $RELEASE from above
$ wget --no-check-certificate \
    https://file-public.cubi.bihealth.org/transient/varfish/anthenea/varfish-test-
↪data-v1-20211125.tar.gz{,.sha256}
$ sha256sum --check varfish-test-data-v1-20211125.tar.gz.sha256
varfish-test-data-v1-20211125.tar.gz: OK
$ tar -xf varfish-test-data-v1-20211125.tar.gz
varfish-test-data-v1-20211125/
...
varfish-test-data-v1-20211125/GRCh37/vcf/HG00107-N1-DNA1-WES1/bwa.gatk_hc.HG00107-
↪N1-DNA1-WES1.vcf.gz
...
```

varfish-annotatorINPUT.vcf.gz

```
# Use the path to the FASTA file that you used for alignment.
$ REFERENCE=path/to/hs37fa.fa--or--hs38.fa
# use $GENOME and $RELEASE from above
$ varfish-annotator \
    -XX:MaxHeapSize=10g \
    -XX:+UseConcMarkSweepGC \
    annotate \
    --db-path varfish-annotator-20210728-$GENOME/varfish-annotator-db-$RELEASE-
↪$GENOME.h2.db \
    --ensembl-ser-path varfish-annotator-20210728-$GENOME/ensembl*.ser \
    --refseq-ser-path varfish-annotator-20210728-$GENOME/refseq_curated*.ser \
    --ref-path $REFERENCE \
    --input-vcf "INPUT.vcf.gz" \
    --release "$GENOME" \
    --output-db-info "FAM_name.db-infos.tsv" \
    --output-gts "FAM_name.gts.tsv" \
    --case-id "FAM_name"
```

java

```
$ varfish-annotator \
    -XX:MaxHeapSize=10g \
    -XX:+UseConcMarkSweepGC \
```

annotate.h2.db.ser.fachr

```
--db-path varfish-annotator-20210728-$GENOME/varfish-annotator-db-$RELEASE-$GENOME.
↪h2.db \    --ensembl-ser-path varfish-annotator-20210728-$GENOME/ensembl*.ser \ ␣
↪  --refseq-ser-path varfish-annotator-20210728-$GENOME/refseq_curated*.ser \    -
↪-ref-path $REFERENCE \
```

GRCh37

```
--input-vcf "INPUT.vcf.gz" \    --release "GRCh37" \    --case-id "index" \
```

```
--output-db-info "FAM_name.db-info.tsv" \    --output-gts "FAM_name.gts.tsv"
```

```
$ gzip -c FAM_name.db-info.tsv >FAM_name.db-info.tsv.gz
$ md5sum FAM_name.db-info.tsv.gz >FAM_name.db-info.tsv.gz.md5
$ gzip -c FAM_name.gts.tsv >FAM_name.gts.tsv.gz
$ md5sum FAM_name.gts.tsv.gz >FAM_name.gts.tsv.gz.md5
```

```
0
0
120
120
```

```
FAM_index   index       father  mother  2       2
FAM_index   father      0       0       1       1
FAM_index   mother      0       0       2       1
```

```
FAM_index   index       0       0       2       1
```

```
FAM_index   sister      father  mother  2       2
FAM_index   broth       father  mother  2       2
FAM_index   father      0       0       1       1
FAM_index   mother      0       0       2       1
```

```
# use $GENOME from above
$ varfish-annotator \
    annotate-svs \
    -XX:MaxHeapSize=10g \
    -XX:+UseConcMarkSweepGC \
    \
    --default-sv-method=YOURCALLERvVERSION"
    --release $GENOME \
    \
    --db-path varfish-annotator-20210728-$GENOME/varfish-annotator-db-$RELEASE-
↪$GENOME.h2.db \
    --ensembl-ser-path varfish-annotator-20210728-$GENOME/ensembl*.ser \
    --refseq-ser-path varfish-annotator-20210728-$GENOME/refseq_curated*.ser \
    \
    --input-vcf FAM_sv_calls.vcf.gz \
```

```
    --output-db-info FAM_sv_calls.db-info.tsv \
    --output-gts FAM_sv_calls.gts.tsv
    --output-feature-effects CASE_SV_CALLS.feature-effects.tsv
```

---

varfish-annotator annotate-svsINFO/SVMETHOD--default-sv-methodINFO/SVMETHOD --default-sv-method

INFO/SVMETHODYOURCALLERvVERSION

```
cat >$TMPDIR/header.txt <<"EOF"
##INFO=<ID=SVMETHOD,Number=1,Type=String,Description="Type of approach used to␣
↪detect SV">
EOF

bcftools annotate \
    --header-lines $TMPDIR/header.txt \
    INPUT.vcf.gz \
| awk -F $'\t' '
    BEGIN { OFS = FS; }
    /^#/ { print $0; }
    /^[^#]/ { $8 = $8 ";SVMETHOD=YOURCALLERvVERSION"; print $0; }
    ' \
| bgzip -c \
> OUTPUT.vcf.gz
tabix -f OUTPUT.vcf.gz
```

---

gzip

```
$ gzip -c FAM_sv_calls.db-info.tsv >FAM_sv_calls.db-info.tsv.gz
$ md5sum FAM_sv_calls.db-info.tsv.gz >FAM_sv_calls.db-info.tsv.gz.md5
$ gzip -c FAM_sv_calls.gts.tsv >FAM_sv_calls.gts.tsv.gz
$ md5sum FAM_sv_calls.gts.tsv.gz >FAM_sv_calls.gts.tsv.gz.md5
$ gzip -c FAM_sv_calls.feature-effects.tsv >FAM_sv_calls.feature-effects.tsv.gz
$ md5sum FAM_sv_calls.feature-effects.tsv.gz >FAM_sv_calls.feature-effectstsv.gz.
↪md5
```

varfish-clipip install varfish-cliconda install varfish-cli

~/.varfishrc.toml

```
[global]
varfish_server_url = "https://varfish.example.com/"
varfish_api_token = "XXX"
```

test-data

```
$ varfish-cli --no-verify-ssl case create-import-info --resubmit \
    94777783-8797-429c-870d-c12bec2dd6ea \
    test-data/tsv/HG00102-N1-DNA1-WES1/*.{tsv.gz,.ped}
```

.ped

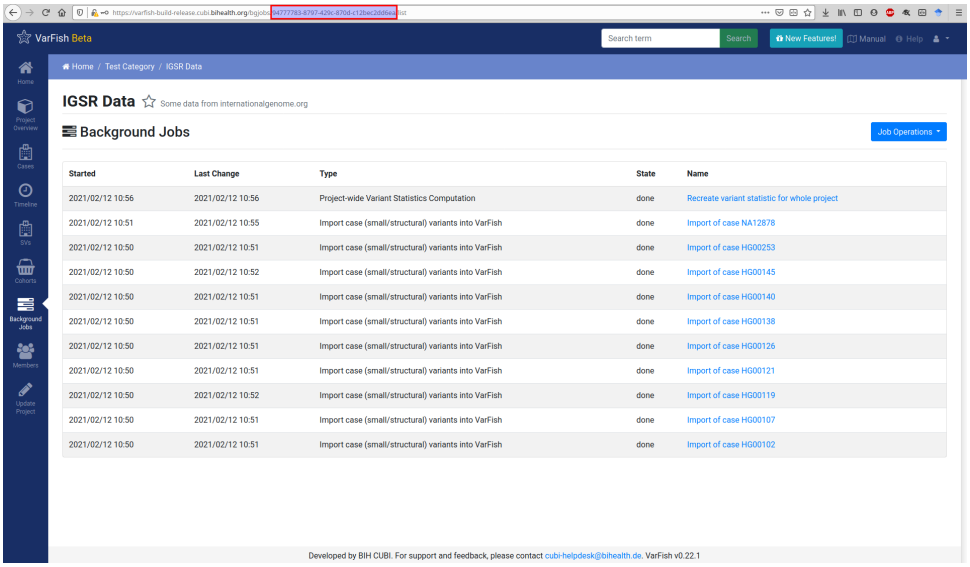
varfish-annotator.gts.tsv.gz

---

varfish-annotator.db-info.tsv.gz

.bam-qc.tsv.gz

.feature-effects.tsv.gz.gts.tsv.gz



.bam-qc.tsv.gz

case_id	set_id	bam_stats

."""""`

bamstats samtools stats

min_cov_target "0""200"number

min_cov_base min_cov_target

summary

idxstats samtools idxstats


```
{
  "index": {
    "bamstats": {
      "raw total sequences": 154189250,
      "filtered sequences": 0,
      "sequences": 154189250,
      "is sorted": 1,
      "1st fragments": 77094625,
      "last fragments": 77094625,
      "reads mapped": 153919815,
```

```
      "reads mapped and paired": 153863370,
      "reads unmapped": 269435,
      "reads properly paired": 153071356,
      "reads paired": 154189250,
      "reads duplicated": 7273644,
      "reads MQ0": 2701485,
      "reads QC failed": 0,
      "non-primary alignments": 129724,
      "total length": 19427845500,
      "total first fragment length": 9713922750,
      "total last fragment length": 9713922750,
      "bases mapped": 19393896690,
      "bases mapped (cigar)": 19238950186,
      "bases trimmed": 0,
      "bases duplicated": 916479144,
      "mismatches": 61093079,
      "error rate": 0.003175489,
      "average length": 126,
      "average first fragment length": 126,
      "average last fragment length": 126,
      "maximum length": 126,
      "maximum first fragment length": 126,
      "maximum last fragment length": 126,
      "average quality": 35,
      "insert size average": 192.6,
      "insert size standard deviation": 54.3,
      "inward oriented pairs": 73269191,
      "outward oriented pairs": 3391556,
      "pairs with other orientation": 12579,
      "pairs on different chromosomes": 258359,
      "percentage of properly paired reads (%)": 99.3
    },
    "min_cov_target": {
      "0": 100,
      "10": 87.59,
      "190": 12.31,
      "200": 10.74
    },
    "min_cov_base": {
      "0": 100,
      "10": 95.89,
      "190": 46.55,
      "200": 43.88
    },
    "summary": {
      "mean coverage": 206.69,
      "target count": 232447,
      "total target size": 57464133
    },
    "idxstats": {
      "1": {
        "mapped": 14553406,
        "unmapped": 5166
      },
      "MT": {
        "mapped": 10058,
        "unmapped": 7
      },
      "*": {
        "mapped": 0,
        "unmapped": 212990
      }
```

```
    }
  },
  "father": {
    "bamstats": {
```

sda

postgresql.conf

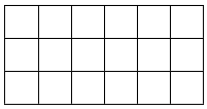
full_page_writesoff

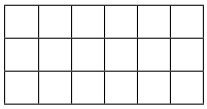
```
full_page_writes = off  # only do this on ZFS (!)
```

random_page_cost1.1

```
random_page_cost = 1.1  # optimized for SSD
```

```
conservation_knowngeneaa
dbsnp_dbsnp
frequencies_*
extra_annos_*
```





20210728

20210728b20210728

/datavarfish-web

```
$ docker exec -it varfish-docker-compose_varfish-web_1 python /usr/src/app/manage.
↪py \
    import_tables --tables-path /data --truncate --force
```

varfish-docker-composedocker-compose up -d

varfish-docker-composedocker-compose up -d

FIELD_ENCRYPTION_KEY

maintenanceexport

VARFISH_ENABLE_SPANR_SUBMISSION1

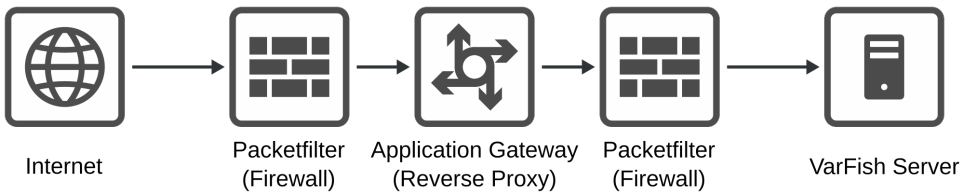
---

cadd-rest-api-servercadd-rest-api-celeryd-worker-?

varfish-docker-compose.ymlvarfish-celeryd-maintenancevarfish-celeryd-export
docker-compose up

FIELD_ENCRYPTION_KEY

.env




```
10.0.10.10

    1.2.3.0/301.2.3.11.2.3.2
1.2.3.2
192.168.0.110.0.10.10
```

1.2.3.2

varfish-ext.example.com1.2.3.28.8.8.8

varfish-int.example.com

2.3.4.0/283.4.5.6

```
# mkdir -p /etc/reverse-proxy
# mkdir -p /etc/reverse-proxy/var/traefik
# mkdir -p /etc/reverse-proxy/etc/trafik
# mkdir -p /etc/reverse-proxy/etc/trafik/conf.d
```

/etc/reverse-proxy/docker-compose.yaml

```
version: "2"

services:
  traefik:
    image: traefik:latest
    restart: always
    ports:
      - "443:443"
    networks:
      - web
    volumes:
      - ./var/traefik:/var/traefik:rw
      - ./etc/traefik:/etc/traefik:ro
    container_name: traefik

networks:
  web:
```

traefik/etc/reverse-proxy/traefik/etc/traefik/etc/reverse-proxy/var/traefik
/var/traefik

/etc/reverse-proxy/etc/traefik/traefik.yaml/etc/reverse-proxy/etc/traefik/
conf.d/dynamic_config.yaml

```
entryPoints:
  websecure:
    address: ":443"

providers:
  file:
    directory: /etc/traefik/conf.d
  docker:
    exposedByDefault: false
```

```
certificatesResolvers:
  le:
    acme:
      email: youremail@example.com
      storage: /var/traefik/acme.json
      tlsChallenge: true
```

ipwhitelist

```
# (1) TLS store
tls:
  stores:
    default: {}

http:
  # (2) set routing source for reverse proxy
  routers:
    varfish:
      middlewares:
        - varfish-add-prefix
        - varfish-ip-allowlist
      entryPoints:
        - websecure
      service: varfish
      rule: "Host(`varfish-ext.example.com`)"
      tls:
        certresolver: le
  # (3) routing destination for the reverse proxy
  services:
    varfish:
      loadBalancer:
        servers:
          - url: "https://varfish-int.bihealth.org"

  middlewares:
    # (4) expose only beaconsite endpoint
    varfish-add-prefix:
      addprefix:
        prefix: "/beaconsite/endpoint"
    varfish-ip-allowlist:
      ipwhitelist:
        sourcerange: "2.3.4.0/28,3.4.5.6"
```

/beaconsite/endpoint

```
# cd /etc/reverse-proxy
# docker-compose up -d
```

```
docker logs --tail=100 --follow traefiktraefik.yaml
```

```yaml
log:
  level: DEBUG
```

pg_dump

```
# docker exec -it varfish-docker-compose_varfish-web_1 \
    python /usr/src/app/manage.py pg_dump --mode=MODE
```

python /usr/src/app/manage.py pg_dump --mode=MODE

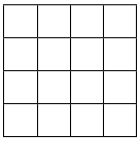
full

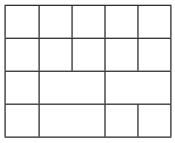
backup-large

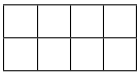
backup-small

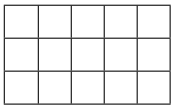
varfish-${day_of_week}.sql.gz

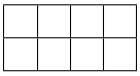
```
# docker exec -it varfish-docker-compose_varfish-web_1 \
    python /usr/src/app/manage.py pg_dump --mode=MODE \
  | gzip -c \
  > varfish-$(date +%a).sql.gz
```

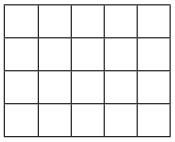




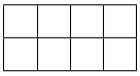


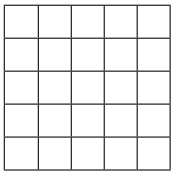


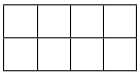


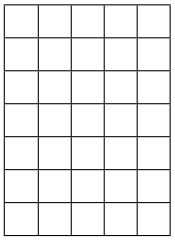


chrX









chrX

Authorization

```
Authorization: token␣
↪90c2483172515bc8f6d52fd608e5031db3fcdc06d5a83b24bec1688f39b72bcd
```

Accept

sodar_uuid

sodar_uuid

detail

```
Accept: application/vnd.bihealth.sodar-core+json; version=0.10.7
```

```
Accept: application/vnd.bihealth.varfish+json; version=0.23.9
```

```
Accept: application/vnd.bihealth.varfish+json; version=0.23.9
```

detail

sodar_uuid

class variants.views_api.**CaseListApiView**()

/variants/api/case/{project.sodar_uid}/

GET

CaseRetrieveApiView

class variants.views_api.**CaseRetrieveApiView**()

/variants/api/case/{project.sodar_uuid}/{case.sodar_uuid}/

GET

date_createdstr

date_modifiedstr

indexstr

namestr

notesstrnull

num_small_varsintnull

num_svsintnull

pedigreelistdictdict

sexint

fatherstr

motherstr

namestr

affectedint

has_gt_entriesboolean

projectstr

releasestr["GRCh37", "GRCh37"]

sodar_uuidstr

statusstr"initial""active""closed-unsolved""closed-uncertain" "closed-solved"

tagsliststr

class variants.views_api.**SmallVariantQueryListApiView**()

/variants/api/query-case/list/{case.sodar_uuid}

GET

page1

page_size10100

countint

nextstrnull

previousstrnull

resultslistSmallVariantQuery

class variants.views_api.**SmallVariantQueryCreateApiView**()

/variants/api/query-case/create/{case.sodar_uuid}

POST

form_idstr"variants.small_variant_filter_form"

form_versionint1

query_settingsdict

namestrNone

publicboolFalse

SmallVariantQuery
class variants.views_api.**SmallVariantQueryRetrieveApiView**()

/variants/api/query-case/retrieve/{query.sodar_uuid}
GET

SmallVariantQuery
class variants.views_api.**SmallVariantQueryStatusApiView**()

/variants/api/query-case/status/{query.sodar_uuid}
GET

dictstatusstr
class variants.views_api.**SmallVariantQueryUpdateApiView**()

/variants/api/query-case/update/{query.sodar_uuid}
PUTPATCH

name
public

SmallVariantQuery
class variants.views_api.**SmallVariantQueryFetchResultsApiView**()

/variants/api/query-case/results/{query.sodar_uuid}
GET
page1
page_size10100

countint
nextstrnull
previousstrnull
resultslistdict

class variants.views_api.**SmallVariantQuerySettingsShortcutApiView**()

/variants/api/query-case/settings-shortcut/{case.uuid}
GET

database"refseq""ensembl"
quick_preset
defaults
de_novo
dominant
homozygous_recessive
compound_heterozygous
recessive
x_recessive
clinvar_pathogenic
mitochondrial
whole_exomes
inheritance
any
dominant
homozygous_recessive
compound_heterozygous
recessive
x_recessive
mitochondrial
custom
frequency
dominant_super_strict
dominant_strict
dominant_relaxed
recessive_strict
recessive_relaxed
custom
impact
null_variant
aa_change_splicing
all_coding_deep_intronic
whole_transcript
any_impact

```
    custom
quality
    super_strict
    strict
    relaxed
    any
    custom
chromosomes
    whole_genome
    autosomes
    x_chromosome
    y_chromosome
    mt_chromosome
    custom
flags_etc
    defaults
    clinvar_only
    user_flagged
    custom

presetsdict
    quick_presetsquick_presets
    inheritanceinheritance
    frequencyfrequency
    impactimpact
    qualityquality
    chromosomeschromosomes
    flags_etcflags_etc
query_settingsdict
```

	true		
		exac_enabledexac_frequencynull	

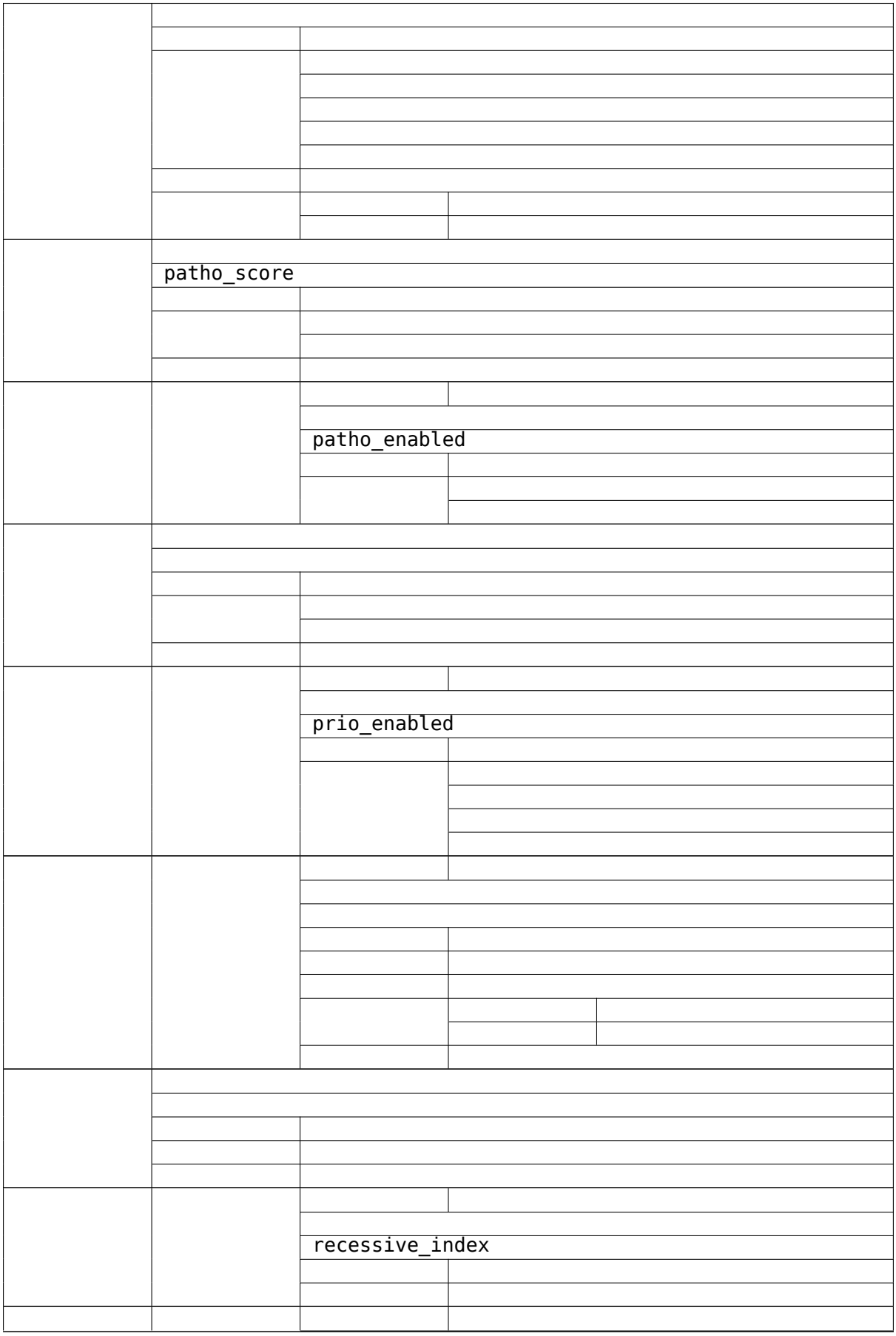
		exac_enabledexac_heterozygousnull
		exac_enabledexac_homozygousnull
		exac_enabledexac_hemizygousnull
	true	
		gnomad_exomes_enabledgnomad_exomes_frequencynull
		gnomad_exomes_enabledgnomad_exomes_heterozygousnull
		gnomad_exomes_enabledgnomad_exomes_homozygousnull
		gnomad_exomes_enabledgnomad_exomes_hemizygousnull

	true		
	gnomad_genomes_enabledgnomad_genomes_frequencynull		
	gnomad_genomes_enabledgnomad_genomes_heterozygous null		
	gnomad_genomes_enabledgnomad_genomes_homozygousnull		
	gnomad_genomes_enabledgnomad_genomes_hemizygousnull		
	true		
	thousand_genomes_enabledthousand_genomes_frequency null		

		thousand_genomes_enabledthousand_genomes_heterozygous null	
		thousand_genomes_enabledthousand_genomes_homozygous null	
		thousand_genomes_enabledthousand_genomes_hemizygous null	
	true		
		inhouse_enabledinhouse_carriersnull	
		inhouse_enabledinhouse_heterozygousnull	
		inhouse_enabledinhouse_homozygousnull	
		inhouse_enabledinhouse_hemizygousnull	
	true		

		mtdb_enabledmtdb_countnull	
		mtdb_enabledmtdb_frequencynull	
	true		
		helixmtdb_enabledhelixmtdb_frequencynull	
		helixmtdb_enabledhelixmtdb_het_countnull	
		helixmtdb_enabledhelixmtdb_hom_countnull	
	true		
		mitomap_enabledmitomap_countnull	

		mitomap_enablednull	
	true		
	trueCGA>CC>CGA		
	trueCG>TT		
		max_exon_dist	


patho_score
patho_enabled
prio_enabled
recessive_index

X-Beacon-User

X-Beacon-UserDate

X-Beacon-User

X-Beacon-User

Date Header

```
Signature keyId="org.bihealth.varfish",algorithm="rsa-sha512",headers="date x-
↪beacon-user",\
signature="mxY7+9vizRbO7mUJVyvxXm3VgpYycQWNulrAafMOWJ29WYQYMf2i5PBPP3jYBhIGd/
↪3zZ+x+mlQw8xEw\
M6UWvE3QRqzlzBE0ZHeWKgX4h11N1MhtXTnhXL9CL/VqbcgbBI9trkwB/
↪xxaXhUOpvavA37J1ljrdTbXhghCHZ65hMi\
04fUnKKkFhuwOzZ6N5/
↪amIuizc2JeDe73Pg+D5HA4AnE2bnCmf8AqhKLd434SdchcYAHqYTJaxBA2Pxngerg6oSenli\
rgukzrBdbdRpvnFFtQzZsQ56v9hS8cqF/phtl+isAT/dcwvO9/lCKaf3QE8YKCcQmDnPJiQLdtQ9mZKw==
↪",\
created="1646407724"'
```

keyId

algorithm

headersdate x-beacon-user

signature

```
rsa-sha256
rsa-sha512
ecdsa-sha256
ecdsa-sha256
```

```
sudo apt install postgresql-12
```

```
sudo apt install redis-server
```

```
$ wget https://repo.anaconda.com/miniconda/Miniconda3-latest-Linux-x86_64.sh
$ bash Miniconda3-latest-Linux-x86_64.sh -b -p ~/miniconda3
$ source ~/miniconda3/bin/activate
$ conda init
$ conda create -n varfish python=3.8 pip
$ conda activate varfish
```

```
$ git clone https://github.com/bihealth/varfish-server
$ cd varfish-server
```

```
$ sudo apt install libsasl2-dev python-dev libldap2-dev libssl-dev
```

```
$ for i in requirements/*; do pip install -r $i; done
```

utility/varfish

```
$ bash utility/setup_database.sh
```

utility/

```
$ sudo bash utility/install_vue_dev.sh
```

```
$ cd clinvar_export/vueapp
$ npm install
```

```
$ npm run serve
```

.env

```
export DATABASE_URL="postgres://varfish:varfish@127.0.0.1/varfish"
export CELERY_BROKER_URL=redis://localhost:6379/0
export PROJECTROLES_ADMIN_OWNER=root
export DJANGO_SETTINGS_MODULE=config.settings.local
```

```
export VARFISH_ENABLE_SVS=1
```

migrate

```
$ python manage.py migrate
```

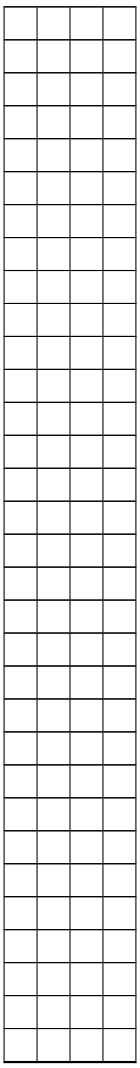
root.envPROJECTROLES_ADMIN_OWNER

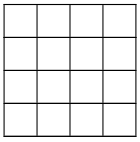
```
$ python manage.py createsuperuser
```

```
$ python manage.py geticons -c bi cil fa-regular fa-solid gridicons octicon
$ python manage.py collectstatic
```

```
terminal1$ make serve
terminal2$ make celery
```

```
$ cd /plenty/space
$ wget https://file-public.bihealth.org/transient/varfish/varfish-server-
↪background-db-20201006.tar.gz{,.sha256}
$ sha256sum -c varfish-server-background-db-20201006.tar.gz.sha256
$ tar xzvf varfish-server-background-db-20201006.tar.gz
```





import_versions.tsvtable_group#

```
build       table_group     version
GRCh37      acmg    v2.0
#GRCh37     clinvar 20200929
#GRCh37     dbSNP   b151
#GRCh37     dbVar   latest
GRCh37      DGV     2016
GRCh37      ensembl_genes   r96
GRCh37      ensembl_regulatory      latest
GRCh37      ensembltogenesymbol     latest
GRCh37      ensembltorefseq latest
GRCh37      ExAC_constraints        r0.3.1
#GRCh37     ExAC    r1
#GRCh37     extra-annos     20200704
GRCh37      gnomAD_constraints      v2.1.1
#GRCh37     gnomAD_exomes   r2.1
#GRCh37     gnomAD_genomes  r2.1
#GRCh37     gnomAD_SV       v2
GRCh37      HelixMTdb       20190926
GRCh37      hgmd_public     ensembl_r75
GRCh37      hgnc    latest
GRCh37      hpo     latest
GRCh37      kegg    april2011
#GRCh37     knowngeneaa     latest
GRCh37      mgi     latest
GRCh37      mim2gene        latest
GRCh37      MITOMAP 20200116
GRCh37      mtDB    latest
GRCh37      ncbi_gene       latest
GRCh37      refseq_genes    r105
GRCh37      refseqtoensembl latest
GRCh37      refseqtogenesymbol      latest
GRCh37      tads_hesc       dixon2012
GRCh37      tads_imr90      dixon2012
#GRCh37     thousand_genomes        phase3
GRCh37      vista   latest
#GRCh38     clinvar 20200929
#GRCh38     dbVar   latest
#GRCh38     DGV     2016
```

```
$ python manage.py import_tables --tables-path /plenty/space/varfish-server-
↪background-db-20201006
```

--force

```
$ python manage.py import_tables --tables-path varfish-db-downloader --force
```

```
$ make test
```

```
$ make test-noselenium
```

```
$ python manage.py test -v2 --settings=config.settings.test variants.tests.test_ui.
↪TestVariantsCaseFilterView.test_variant_filter_case_multi_bookmark_one_variant
```

git guigitk

Fork

```
$ git pull --rebase
```

<ticket_number>-<ticket_title>

```
$ git checkout -b 123-adding-useful-feature
```

```
Fixed serious bug that prevented user from doing x.

Closes: #123
Related-Issue: #123
Projected-Results-Impact: none
```

```
$ git rebase -i main
```

git commit --amend

```
$ git checkout 123-adding-useful-feature
$ git rebase main
```

<<<<

```
$ git add conflicting.file
$ git rebase --continue
```

```
$ git rebase --abort
```

```
$ git push origin 123-adding-useful-feature
```

```
$ git push -f origin 123-adding-useful-feature
```

```
$ wget https://file-public.bihealth.org/transient/varfish/varfish-annotator-{,
↪transcripts-}20191129.tar.gz{,.sha256}
$ tar xzvf varfish-annotator-20191129.tar.gz
$ tar xzvf varfish-transcripts-20191129.tar.gz
```

.env

```
export VARFISH_KIOSK_MODE=1
export VARFISH_KIOSK_VARFISH_ANNOTATOR_REFSEQ_SER_PATH=/path/to/varfish-annotator-
↪transcripts-20191129/hg19_refseq_curated.ser
export VARFISH_KIOSK_VARFISH_ANNOTATOR_ENSEMBL_SER_PATH=/path/to/varfish-annotator-
↪transcripts-20191129/hg19_ensembl.ser
export VARFISH_KIOSK_VARFISH_ANNOTATOR_REFERENCE_PATH=/path/to/unpacked/varfish-
↪annotator-20191129/hs37d5.fa
export VARFISH_KIOSK_VARFISH_ANNOTATOR_DB_PATH=/path/to/unpacked/varfish-annotator-
↪20191129/varfish-annotator-db-20191129.h2.db
export VARFISH_KIOSK_CONDA_PATH=/path/to/miniconda/bin/activate
```

```
terminal1$ make serve
terminal2$ make celery
```

```
**Describe the bug**
A clear and concise description of what the bug is.

**To Reproduce**
Steps to reproduce the behavior:
1. Go to '...'
2. Click on '....'
3. Scroll down to '....'
4. See error

**Expected behavior**
A clear and concise description of what you expected to happen.

**Screenshots**
If applicable, add screenshots to help explain your problem.

**Desktop (please complete the following information):**
 - OS: [e.g. iOS]
 - Browser [e.g. chrome, safari]
 - Version [e.g. 22]

**Smartphone (please complete the following information):**
 - Device: [e.g. iPhone6]
 - OS: [e.g. iOS8.1]
 - Browser [e.g. stock browser, safari]
 - Version [e.g. 22]

**Additional context**
Add any other context about the problem here.
```

```
**Resolution Proposal**
e.g. The component X needs to be changed to Y so Z is not executed when M occurs.

**Affected Components**
e.g. VarFish server

**Affected Modules/Files**
e.g. variants module or queries.py

**Required Architectural Changes**
e.g. Function F needs to be moved to X.

**Required Database Changes**
i.e. name any model that needs changing, to be added and will lead to a migration

**Backport Possible?**
e.g., "Yes" if this is a bug fix or small change and should be backported to the␣
↪current stable version

**Resolution Sketch**
e.g. Change X in F. Then do Y.
```

```
Resolve some issue (#NUMBER)
```

Related-IssueNo-Related-Issue

```
Related-Issue: #123
No-Related-Issue: Short text reason
```

Projected-Results-Impactnonerequire-revalidation

```
Projected-Results-Impact: none
Projected-Results-Impact: require-revalidation
```

123-fix-for-issue

```
**Is your feature request related to a problem? Please describe.**
A clear and concise description of what the problem is. Ex. I'm always frustrated␣
↪when [...]

**Describe the solution you'd like**
A clear and concise description of what you want to happen.

**Describe alternatives you've considered**
A clear and concise description of any alternative solutions or features you've␣
↪considered.

**Additional context**
Add any other context or screenshots about the feature request here.
```

```
**Implementation Proposal**
e.g. The component X needs to be changed to Y so Z is not executed when M occurs.

**Affected Components**
e.g. VarFish server

**Affected Modules/Files**
e.g. variants module or queries.py

**Required Architectural Changes**
e.g. Function F needs to be moved to X.

**Implementation Sketch**
e.g. Change X in F. Then do Y.
```

```
Release for version vVERSION

- [ ] edit `HISTORY.rst` and ensure a proper section is added
- [ ] edit `admin_upgrade.rst` to reflect the upgrade instructions
- [ ] create a git tag `v.MAJOR.MINOR.PATCH` and `git push --tags`
- [ ] create a "Github release` based on the tag with the text

      ```
      All details can be found in the `HISTORY.rst` file.
      ```
```

varfish-build-release-{37,38}.cubi.bihealth.org

varfish-site-data-X.tar.gz

```
Validate data for:

- **VarFish:** vMAJOR.MINOR.PATCH
- **Site Data:** vVERSION (`sha256:CHECKSUM`)
- **Genome Build:** GRCh37 or GRCh38

Result Reports:

PASTE HERE
```

varfish-wf-validation

```
$ ./docker/build-docker.sh
```

varfish-docker-compose

varfish-docker-compose

```
$ git clone git@github.com:bihealth/varfish-docker-compose.git
$ cd varfish-docker-compose
$ ./init.sh
```

```
$ docker-compose up postgres
<Ctrl-C>
```

postgresql.conf

```
$ cp config/postgres/postgresql.conf volumes/postgres/data/postgresql.conf
```

```
$ docker-compose up
```

varfish-webVARFISH MIGRATIONS ENDrun-docker-compose-up.sh

```
$ cd /plenty/space
$ wget https://file-public.bihealth.org/transient/varfish/anthenea/varfish-server-
↪background-db-20201006.tar.gz{,.sha256}
$ sha256sum -c varfish-server-background-db-20201006.tar.gz.sha256
$ tar xzvf varfish-server-background-db-20201006.tar.gz
```

docker-compose.yml/plenty/space

```
volumes:
    - "/plenty/space:/data"
```

```
$ docker ps
CONTAINER ID   IMAGE                                                          COMMAND␣
↪                  CREATED          STATUS              PORTS                    ␣
↪                 NAMES
44be6ece102e   minio/minio                                                    "/usr/
↪bin/docker-ent…"   11 minutes ago   Up About a minute   9000/tcp                 ␣
↪                  varfish-docker-compose_minio_1
3b23113e5aa1   quay.io/biocontainers/exomiser-rest-prioritiser:12.1.0--1
↪"exomiser-rest-prior…"   11 minutes ago   Up About a minute                     ␣
↪                         varfish-docker-compose_exomiser-rest-prioritiser_1
b8c49e8c24a6   quay.io/biocontainers/jannovar-cli:0.33--0
↪"jannovar -Xmx6G -Xm…"   11 minutes ago   Up About a minute                     ␣
↪                         varfish-docker-compose_jannovar_1
409a535b9951   bihealth/varfish-server:0.22.1-0                               "docker-
↪entrypoint.s…"   12 minutes ago   Up About a minute   8080/tcp                 ␣
↪                 varfish-docker-compose_varfish-celerybeat_1
7eb7425c59e2   bihealth/varfish-server:0.22.1-0                               "docker-
↪entrypoint.s…"   12 minutes ago   Up About a minute   8080/tcp                 ␣
↪                 varfish-docker-compose_varfish-celeryd-import_1
020811fde306   bihealth/varfish-server:0.22.1-0                               "docker-
↪entrypoint.s…"   12 minutes ago   Up About a minute   8080/tcp                 ␣
↪                 varfish-docker-compose_varfish-celeryd-query_1
87b03ee0249b   bihealth/varfish-server:0.22.1-0                               "docker-
↪entrypoint.s…"   12 minutes ago   Up About a minute   8080/tcp                 ␣
↪                 varfish-docker-compose_varfish-celeryd-default_1
7a3fdb337fae   bihealth/varfish-server:0.22.1-0                               "docker-
↪entrypoint.s…"   12 minutes ago   Up About a minute   8080/tcp                 ␣
↪                 varfish-docker-compose_varfish-web_1
9295a101570f   postgres:12                                                    "docker-
↪entrypoint.s…"   12 minutes ago   Up About a minute   5432/tcp                 ␣
↪                 varfish-docker-compose_postgres_1
1c4d6e235074   traefik:v2.3.1                                                 "/
↪entrypoint.sh --pr…"   12 minutes ago   Up About a minute   0.0.0.0:80->80/tcp,␣
↪0.0.0.0:443->443/tcp   varfish-docker-compose_traefik_1
8d72fd096743   redis:6                                                        "docker-
↪entrypoint.s…"   12 minutes ago   Up About a minute   6379/tcp                 ␣
↪                 varfish-docker-compose_redis_1
```

docker-compose up varfish-web postgres redis

```
$ docker exec -it -w /usr/src/app varfish-docker-compose_varfish-web_1 python␣
↪manage.py import_tables --tables-path /data --threads 8
```

docker-compose upvolumes:varfish-web

jannovarexomiser

```
# tree volumes/jannovar volumes/exomiser
volumes/jannovar
├── hg19_ensembl.ser
├── hg19_refseq_curated.ser
└── hg19_refseq.ser
volumes/exomiser
├── 1909_hg19
│   ├── 1909_hg19_clinvar_whitelist.tsv.gz
.   .   [..]
│   └── 1909_hg19_variants.mv.db
└── 1909_phenotype
    ├── 1909_phenotype.h2.db
    ├── phenix
    │   ├── 10.out
    .   .   [..]
    │   ├── ALL_SOURCES_ALL_FREQUENCIES_genes_to_phenotype.txt
    │   ├── hp.obo
    │   └── phenotype_annotation.tab
    └── rw_string_10.mv

3 directories, 55 files
```

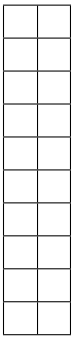
docker-compose up

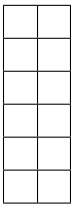
```
$ docker exec -it -w /usr/src/app varfish-docker-compose_varfish-web_1 python␣
↪manage.py createsuperuser
Username: root
Email address:
Password: <changeme>
Password (again): <changeme>
Superuser created successfully.
```

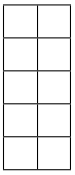
varfish-cli

```
$ varfish-cli --no-verify-ssl case create-import-info --resubmit \
    92f5d735-0967-4db2-a801-50fe96359f51 \
    $(find path/to/variant_export/work/*NA12878* -name '*.tsv.gz' -or -name '*.ped
↪')
```

```
tar -cf - volumes | pigz -c > varfish-site-data-v1-20210728-grch37.tar.gz &&␣
↪sha256sum varfish-site-data-v1-20210728-grch37.tar.gz >varfish-site-data-v1-
↪20210728-grch37.tar.gz.sha256 &
tar -cf - volumes | pigz -c > varfish-site-data-v1-20210728-grch38.tar.gz &&␣
↪sha256sum varfish-site-data-v1-20210728-grch38.tar.gz >varfish-site-data-v1-
↪20210728-grch38.tar.gz.sha256 &
tar -cf - test-data | pigz -c > varfish-test-data-v1-20211125.tar.gz && sha256sum␣
↪varfish-test-data-v1-20211125.tar.gz >varfish-test-data-v1-20211125.tar.gz.sha256
```





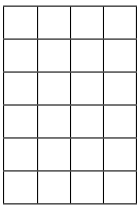


x.y.zx1Antheneay

    x.0.z

    x.1.z

    x.2.z



20210728` data release (#450). Includes instructions on how to apply patch to get ``20210728b

20210728` data release (#450). Includes instructions on how to apply patch to get ``20210728b

pg_dump

fa-solid:refresh

_ClosingWrapper

mdi

SmallVariantcase_idset_id

PROJECT

kiosk_user

```
set -xsettings.DEBUG
```

```
SmallVariantcase_idset_id
```

PROJECT

VARFISH_CADD_SUBMISSION_RELEASEVARFISH_CADD_SUBMISSION_VERSION
import_info.tsv202107281b03e97
Hgnc

VARFISH_QUERY_MAX_UNION=20

UNLOGGED

UNLOGGEDLOGGED

```
sudo apt-get update
```

PROJECTROLES_ADMIN_OWNERadminrootroot

nltk

SENTRY_DSN

ProjectExportTest
MetaCase

variants
check_installationdbsnp

maintenanceexport

ProjectExportTest
MetaCase

regmaps

variants
check_installationdbsnp

maintenanceexport

variant

synonymousall coding/deep intronic

SvAnnotationReleaseInfo

importedCaseImportInfo

cDNA effectprotein effecteffect textdistance to splicesite distance to splicesite

PROJECTROLES_EMAIL_

variant

synonymousall coding/deep intronic

intergenic_variant_\d+validation_warning_\d+

SvAnnotationReleaseInfo

StructuralVariant
stateVariantSetImportInfo
importedCaseImportInfo

cDNA effectprotein effecteffect textdistance to splicesite
distance to splicesite

PROJECTROLES_EMAIL_

Variants & Effects
HelixMTdb

molecular

molecular

Download as FileSubmit to MutationDistiller
ClinVar Pathogenic

Variants & Effects

HelixMTdb

molecular

molecular

Download as FileSubmit to MutationDistiller
ClinVar Pathogenic

Variant Annotation

AA change, splicing (default)Impactall coding, deep intronic

import_cases_bulk

Variant Annotation
AA change, splicing (default)Impactall coding, deep intronic

import_cases_bulk

HpoHpoNameHpoHpoNamehpo_idHP:0031988"unknown"None
ProjectCasesFilterView

mtDBHelixMTdbMITOMAP

Case
importer

```
django_su
```

```
ClinVar only
```

```
recessive
```

refseq_hgvs_crefseq_transcript_id

bootstrap-select

NcbiGeneRifpubmed_ids

django_su

ClinVar only

recessive

CaddPathogenicityScoreCacheUmdPathogenicityScoreCacheMutationtaster-PathogenicityScoreCache

ProjectCasesSmallVariantQueryVariantScores

Delete Case

remove if in dbSNPClinVar membership required
SmallVariantFlagsSmallVariantCommentExacConstraintsGnomadConstraints

urls

Delete CaseCaseSmallVariantStructuralVariantStructuralVariantGeneAnnotation

messages

remove if in dbSNPClinVar membership required

SmallVariantFlagsSmallVariantCommentExacConstraintsGnomadConstraints

RefseqToGeneSymbolEnsemblToGeneSymbol

chromosome_noSmallVariantCommentsSmallVariantFlagschromosome

info

somaticgermline
somaticgermline
gene

"scattergl"
UNLOGGED

somaticgermline
somaticgermline
genesign. & rating

human_entrez_idMgiMapping

notesstatus

active
indexc/h index

hgvs_p
index

notesstatusCase
status
CaseComments

MgiHomMouseHumanSequenceMgiMappingentrez_idMGI ID

annotation

active

rebuild_project_case_stats

indexc/h index

import_sv_dbsimport_tables

None

model_support.py

None

test_submit_filter.py

Case.index

Annotation
--force

```
--forceimport_tables
```

```
total
```

--forceimport_tables

Annotation

sodar_corev0.6.1

siteinfo
releaseKnownGeneAA

Annotation

bgjobs
containing_bins
svs_fixtures.py
startendpositionpositionbin
totaltotalafhet

import_tables

rebuild_variant_summary

import_case
import_case

rebuild_variant_summary

ImportInfo

GnomadConstraintExacConstraint

SmallVariant

SmallVariant

SmallVariantSummary

SmallVariantAnnotation

NcbiGeneInfoNcbiGeneInfogeneinfo

```
django_redis

pedigree_relatedness
```

```
var_qc_stats
```

ExportVcfFileFilterQuery

SmallVariant``s are now also identified by the ``ensembl_gene_id
flag_summarySmallVariantFlags

hgmdHGMD_PUBLIC
make blackMakefile

SmallVariantQuery

```
+/-1

file_export

VariantSmallCommentVariantFlags

"has_gt_fields"
"has_gt_fields"
"has_gt_fields"

variants

search_tokensCase
```

```
min(AD/DP, 1 - AD / DP)
```

AAB
ACGS
ACMG
AD

CADD
CaseListApiView
CaseRetrieveApiView
ClinVar

DP

ENSEMBL
Entrez
Exomiser

HiPhive
HTS

IGV

MEDLINE
MutationDistiller
MutationTaster

NCBI

OMIM

Phenix
Phive
PubMed

QC

SmallVariantQueryCreateApiView
SmallVariantQueryFetchResultsApiView
SmallVariantQueryListApiView
SmallVariantQueryRetrieveApiView
SmallVariantQuerySettingsShortcutApiView

SmallVariantQueryStatusApiView
SmallVariantQueryUpdateApiView
SNV
SOP

UCSC
UMD Predictor

Varsome

WES
WGS