



 VarFish Beta

Search


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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

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Project Overview

Cases

Timeline

Background Jobs

Members

Update Project

Home / Public Data / Corpasome Data

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)			



Search Results for "isdbm322017"



Cases (1)

Filter

Name	Project	Individuals
ISDBM322015	Corpasome Data	ISDBM322015, ISDBM322017, IS...

No results found:

- Projects

Manuel Holtgrewe
holtgrem@CHARITE

- ⓘ Import Release Info
- 🔍 API Tokens
- 👤 User Profile

➔ Log Out

☆ VarFish Beta

Search term

New Features!

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API Tokens

+ Create Token

#	Created	Expires	Key	
1	2020-03-04 17:59	Never	27651a02	<div></div>
2	2020-05-06 05:12	Never	b1c818dd	<div></div>

 VarFish Beta

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

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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

The screenshot shows the 'Advanced' tab of the IGV settings dialog. Red circles and boxes highlight specific elements: a red box around the 'Enable port' checkbox and its value '60151' (with a red circle '1' on the 'Advanced' tab), a red circle '2' around the 'Enable port' checkbox, and a red circle '3' around the '60151' text field.

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](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 reshown 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

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[Help](#)

Upload VCF File to VarFish Kiosk

Use the form below to upload a VCF into the VarFish Kiosk. For first-time users, we recommend the [Kiosk tutorial](#). You can download some example data [below](#).

VCF File*

Browse...

pfeiffer-singleton.vcf.gz

Select the VCF file to upload (compressed or uncompressed).

PED File

Browse...

No file selected.

Optional PED file with pedigree information, you can also type the text below.

PED Text

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

Alternatively, type the PED file here. If both are given, the file has precedence.

Submit

Example Data

Example data taken from the [the Corposome](#) (via the [Exomiser](#) sample data).

- [pfeiffer.vcf.gz](#) (healthy individual with variant associated with Pfeiffer syndrome)
- [pfeiffer.vcf.gz](#) / [pfeiffer.ped](#) (healthy quartet with spike-in of a variant associated with Pfeiffer syndrome).

Important and Legal Information

VarFish Kiosk is a password-less service.
Your case will get a long, random ID that is virtually impossible to guess. Anyone who knows this ID will be able to access your data.

Data protection and data retention.
VarFish Kiosk is currently a password-less online service. Your case will get a long, random ID that is virtually impossible to guess. Keep the case ID and address safe, anyone who knows this ID will be able to access your data. All data transfers to and from [varfish.kiosk.bihealth.org](#) use secure protocols. However, your data will be temporarily stored on our server. Only upload data if you have sufficient consent to do so. We do not inspect, make available or use uploaded data except for debugging the VarFish software. All uploads will be deleted automatically after 2 months.

Custom VarFish installation.
If your data and/or consents do not allow for online processing with VarFish Kiosk you might want to consider an installation on your own server and run VarFish in "Classic" mode. This allows to create password protection and unlock the VarFish collaboration features. See the [VarFish manual](#) for more information.

For research use only.
The software is provided "as is", without warranty of any kind, express or implied, including but not limited to the warranties of merchantability, fitness for a particular purpose and non infringement. In no event shall the authors or copyright holders be liable for any claim, damages or other liability, whether in an action of contract, tort or otherwise, arising from, out of or in connection with the software or the use or other dealings in the software.

Citation and Github project.
If you use VarFish Kiosk in your research please cite.

Holtgrewe, M., Stolpe, O., Nieminen, M., Knaus, A., Segebrecht, L., Spielmann, M., Kornak, U., Mundlos, S., Seelow, D., Fischer-Zirnsack, B., Boschann, F., Scholl, U., Ehmke, N., and Beule, D. **VarFish: Collaborative and Comprehensive Exome Analysis for Clinic and Research**. In preparation.

You can find the project source code on [Github](#).

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) Beta

ManualHelp

Annotating ...

[2019-12-19 15:45:54] Kiosk annotate started
[2019-12-19 15:45:54] Writing to /tmp/bcftools-sort.n7DKey
[2019-12-19 15:45:56] Merging 13 temporary files
[2019-12-19 15:45:57] Cleaning
[2019-12-19 15:45:57] Done
[2019-12-19 15:45:58] OpenJDK 64-Bit Server VM warning: Option UseConcMarkSweepGC was deprecated in version 9.0 and will likely be removed in a future release.
[2019-12-19 15:45:58] annotate: AnnotateArgs(help=false, refseqSerPath="/vol/local/data/varfish-db-downloader/hg19_refseq_curated.ser", ensemblSerPath="/vol/local/data/varfish-db-downloader/hg19_refseq_curated.ser", ensemblSerPath="/vol/local/data/varfish-db-downloader/hg19_refseq_curated.ser", ensemblSerPath="/vol/local/data/varfish-db-downloader/hg19_refseq_curated.ser")
[2019-12-19 15:45:58] Running annotate; args: AnnotateArgs(help=false, refseqSerPath="/vol/local/data/varfish-db-downloader/hg19_refseq_curated.ser", ensemblSerPath="/vol/local/data/varfish-db-downloader/hg19_refseq_curated.ser", ensemblSerPath="/vol/local/data/varfish-db-downloader/hg19_refseq_curated.ser", ensemblSerPath="/vol/local/data/varfish-db-downloader/hg19_refseq_curated.ser")
[2019-12-19 15:45:58] Deserializing Jannovar file...
[2019-12-19 15:45:58] log4j:WARN No appenders could be found for logger (de.charite.compbio.jannovar.data.JannovarDataSerializer).
[2019-12-19 15:45:58] log4j:WARN Please initialize the log4j system properly.
[2019-12-19 15:45:58] log4j:WARN See http://logging.apache.org/log4j/1.2/faq.html#noconfig for more info.
[2019-12-19 15:46:11] Now on contig 1
[2019-12-19 15:46:18] Now on contig 2
[2019-12-19 15:46:22] Now on contig 3
[2019-12-19 15:46:25] Now on contig 4
[2019-12-19 15:46:28] Now on contig 5
[2019-12-19 15:46:30] Now on contig 6
[2019-12-19 15:46:33] Now on contig 7
[2019-12-19 15:46:35] Now on contig 8
[2019-12-19 15:46:38] Now on contig 9
[2019-12-19 15:46:40] Now on contig 10
[2019-12-19 15:46:42] Now on contig 11

Developed by BIH CUBI. For support and feedback, please contact [Oliver Stolpe](#). VarFish (Kiosk) v0.18.0+36.gf86f003

varfish-kiosk.bihealth.org

Case pfeiffer

Filter VariantsEdit Pedigree

OverviewQuality ControlVariant AnnotationQueries & Jobs

Details

Case Name	pfeiffer	Individuals	pfeiffer
Created At	2019/12/19 15:47	Last Modified	2019/12/19 15:47
Status, Notes & Tags	<div>initial</div> <div>No notes taken (yet).</div>		
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

Variant Filter pfeiffer

Back to Case

Quick Presets

Inheritance

Frequency

Impact

Quality

Chromosomes

Flags etc.

Load Presets

dominant / de novo

dominant strict (default)

AA change, splicing (default)

strict (default)

whole genome (default)

defaults

Genotype

Prioritization

Frequency

More ...

#	Family	Individual	Trio Role	Father	Mother	Sex	Affected	Genotype
1	pfeiffer	pfeiffer	index	0	0	♂	✓	0/1

RefSeqEnsEMBL

Filter & Display

Results

Details

Coordinates

Frequency

ExAC

Constraint

gnomAD pLI

First 126 of 126 records (case has a total of 313,285 variants) 0. Using RefSeq transcripts.

Show logs

Coordinates				ExAC			gnomAD				
		position	ref	alt	frequency	#hom.	pLI	gene	effect	pfeiffer	
>	#1	chr1:18,809,461	C	CG	0.00002	0	0.000	KLHDC7A	p.S665Qfs*19	0/1	MT IGV
>	#2	chr1:26,161,677	C	T	0.00004	0	0.000	AUNIP	p.G294D	0/1	MT IGV
>	#3	chr1:27,686,399	G	T	0.00000	0	0.000	MAP3K6	p.Q757K	0/1	MT IGV
>	#4	chr1:145,606,274	C	T	0.00006	0	0.000	POLR3C	c.678+1G>A	0/1	MT IGV
>	#5	chr1:150,830,831	C	T	0.00001	0	0.996	ARNT	p.R44Q	0/1	MT IGV
>	#6	chr1:154,901,497	T	C	0.00000	0	0.000	PMVK	c.312+3A>G	0/1	MT IGV
>	#7	chr1:159,284,334	G	C	0.00000	0	0.000	OR10J3	p.T39S	0/1	MT IGV
>	#8	chr1:159,804,970	C	G	0.00000	0	0.000	SLAMF8	c.782-4C>G	0/1	MT IGV

1	2	3	4	position	ref	5	alt	6	frequency	#hom.	pLI	7	gene	8	9	effect	10	pfeiffer	11
>	#1	chr1:18,809,461	C	CG	0.00002	0	0.000	KLHDC7A	p.S665Qfs*19	0/1	MT IGV								

126

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


























































































[← Back to Case](#)

Results Details Coordinates ExAC Constraint gnomAD pLI First 126 of 126 records (case has a total of 313,285 variants) 0. Using RefSeq transcripts.

Coordinates	ExAC	gnomAD
-------------	------	--------

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

Applied Phenotype Terms:

☰ HPO Terms

Results De

☰ Show logs

> #1

> #2

> #3

> #4

> #5

> #6

Flags & Comments

🚩 Flags ☒ ★ ☐ ⚗️ ☐ ❤️ ☐ 🚩
☐ ⚙️ ☐ 👍 ☐ 👎

👁 Visual	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
🔥 Molecular	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
⚗️ Validation	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
👤 Pheno./Clinic	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>
🖋 Summary	<input checked="" type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input type="radio"/>	<input checked="" type="radio"/>	<input type="radio"/>	<input type="radio"/>

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

can be arbitrary and doe

RefSeq

EnsEM

Applied Phenotyp

ResultsDetails

Show logs

ACMG Criteria

Pathogenic

VERY STRONG EVIDENCE

☒

PVS1

null variant

☐

PS1

Located in a mutational hot spot and/or critical and well-established functional domain (e.g., active site of an enzyme) without benign variation

☐

PS2

☐

PS3

☐

PS4

☒

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%

SUPPORTING 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

Applied Phenotype Terms:

HPO Terms

ResultsDetailsCoordinatesFrequencyExACConstraintgnomAD pLIFirst 126 of 126 records (case has a total of 313,285 variants) 0. Using RefSeq transcripts.

Show logs

Coordinates			ExAC			gnomAD		gene	effect	phenotype		pathogenicity		pheno. & patho.	
position	ref	alt	frequency	#hom.	pLI					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	ALDH3A1		p.D60*	0.500	#11	37.0	#1	18.5	#3

VarFish (Kiosk)Beta

New Features!ManualHelp

Casepfeiffer

Filter VariantsEdit PedigreeFix Sex

OverviewQuality ControlVariant AnnotationExport Jobs

Annotated Variants

Variant	Gene(s)	ACMG Rating	Flags							Comments
			Generic	Visual	Molecular	Validation	Phenotype	Summary		
chr10:123,256,215-T-G	-	4	★👤💬📄👍👎	✖	✖	✖	✖	!	kiosk_user 2020/03/25 17:59: This is a known pathogenic variant. <div>IGV</div>	

VarFish (Kiosk)Beta

ManualHelp

Casepfeiffer

Filter VariantsEdit Pedigree

OverviewQuality ControlVariant AnnotationQueries & Jobs

Details

Case Name	pfeiffer	Individuals	pfeiffer
Created At	2019/12/20 09:02	Last Modified	2019/12/20 09:02
Status, Notes & Tags	<div>initial</div> <div>initialactiveclosed as unsolvedclosed as uncertainclosed as solved</div>		
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

Case Comments

No case comments yet.

Enter comment here

Add Comment

Flag & Comment Summary

ACMG-Classified Variants	1	V:1IV:0III:0II:0I:0
Flagged Variants	1	🚫:0❤️:0
Commented Variants	1	

Filter & Display

Download as File

Submit to MutationDistiller

VarFish (Kiosk) Beta

ManualHelp

Created background job for your file download. After the file has been generated, you will be able to download it here.

Background File Creation Job

Create xlsx file for case pfeiffer

Back to FilterOperation

Results & Resubmit

Download Result

File is not there yet, please refresh page: Refresh page

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.

Base Details

Created	Dec. 20, 2019, 9:23 a.m.
Updated	Dec. 20, 2019, 9:23 a.m.
Creator	kiosk_user
Title	Create xlsx file for case pfeiffer
Description	-
Status	running

Export Job Details

Case	pfeiffer
------	----------

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

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

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Update Project

Home / Public Data / Corposome 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

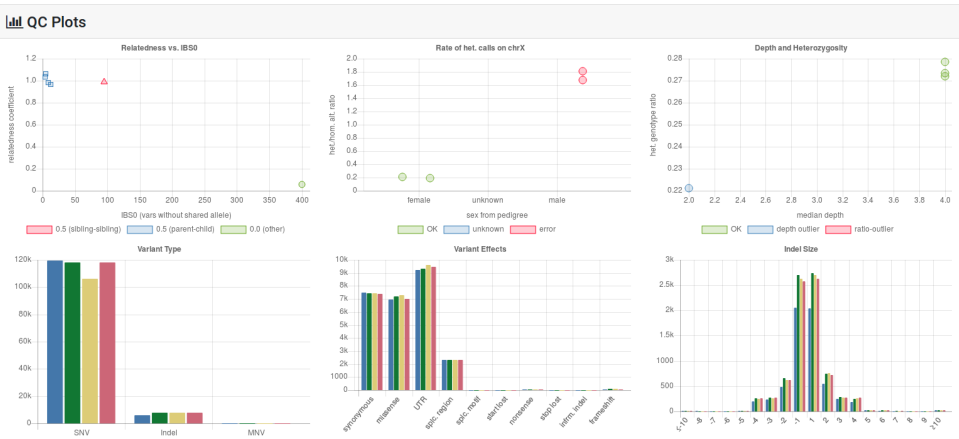
Variant	Comments
---------	----------

Background Jobs Overview

📁

VarFish App Overview

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



--

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
<input checked="" type="checkbox"/> 1000 Genomes	10	Maximal het. count in 1000 genomes	0.01
<input checked="" type="checkbox"/> ExAC	20	Maximal het. count in ExAC	0.01
<input type="checkbox"/> gnomAD exomes	30	Maximal het. count in gnomAD exome	0.01
<input type="checkbox"/> gnomAD genomes	20	Maximal het. count in gnomAD genome	0.01

Leave field empty to exclude from query.

RefSeq

Ensembl

Filter & Display

10
0.01

GenotypeFrequencyVariants & EffectsQualityGene ListsFlags & CommentsMore ...

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

RefSeqEnsEMBL

Filter & Display

GenotypeFrequencyVariants & EffectsQualityGene ListsFlags & CommentsMore ...

Load Presets

Individual	Father	Mother	min DP het.	min DP hom.	min AB	min GQ	min AD	on FAIL
ISDBM322015	ISDBM322016	ISDBM322018	10	5	0.3	30	3	drop variant
ISDBM322017	ISDBM322016	ISDBM322018	10	5	0.3	30	3	drop variant
ISDBM322016	0	0	10	5	0.3	30	3	drop variant
ISDBM322018	0	0	10	5	0.3	30	3	drop variant

RefSeqEnsEMBL

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

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Validation

Phenotype Match

Summary

bookmarked

candidate

final causative

for validation

no simple flag

positive

uncertain

negative

empty

positive

uncertain

negative

empty

positive

uncertain

negative

empty

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Ensembl

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"ISDBM322015" – Variant Filter

Genotype

Frequency

Variants & Effects

Quality

Gene Lists

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More ...

Load Presets

HGMD Public Membership

☐ HGMD public membership required

Require variant to be present in HGMD public (ENSEMBL track). Please note that this data is several years old!

ClinVar Membership

☐ Clinvar membership required

ClinVar Status

Only applied if membership required.

☐ benign

☐ likely benign

☐ uncertain significance

☐ likely pathogenic

☐ pathogenic

RefSeq

Ensembl

Filter & Display

Developed by BIH CUBL. For support and feedback, please contact [Oliver Stolpe](#). VarFish v0.6.1

(# Bookmark for the variant bookmark popup #)

Genotype

Frequency

Variants & Effects

Quality

Gene Lists

Flags & Comments

More ...

Load Presets

Configure Downloads

The settings in this tab are only used when using the **Download** button and not when using **Filter** button below.

File Type

Excel (.xlsx)

Donors to Export

☒ ISDBM322015

☒ ISDBM322017

☒ ISDBM322016

☒ ISDBM322018

Flags & Comments

☒ Export flags

Export flags and label rows by summary.

☒ Export comments

Include comments in export.

RefSeq

Ensembl

Filter & Display

Genotype

Frequency

Variants & Effects

Quality

Gene Lists

Flags & Comments

More ...

Load Presets

Miscellaneous Settings

Result row limit*

80

Maximal number of rows displayed when rendering on the website. This setting is not used when creating a file for export.

RefSeq

Ensembl

Filter & Display

[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",
  "ovar_enabled": true
}
```

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 » SettingsDownload JSON

RefSeqEnsEMBL

Filter & Display

VarFish Beta

Search term

Search

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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
>	chr1:120,336,890	C	G	0.00000	0	REG4	p.=	0/1	0/1	0/1	0/0
>	chr1:120,508,991	G	A	0.00088	0	NOTCH2	p.?	0/1	0/1	0/1	0/0
>	chr1:144,828,910	A	T	0.00000	0	NBPF9	p.=	0/1	0/1	0/1	0/1
>	chr1:145,606,274	C	T	0.00026	0	POLR3C	p.?	0/1	0/1	0/1	0/0
>	chr1:153,431,506	C	G	0.00423	0	S100A7	p.=	0/1	0/1	0/0	0/1
>	chr1:154,901,497	T	C	0.00000	0	PMVK	p.?	0/1	0/1	0/1	0/0
>	chr1:166,826,752	A	G	0.00000	0	TADA1	p.=	0/1	0/1	0/1	0/0
>	chr1:171,561,002	G	A	0.00196	0	PRRC2C	p.=	0/1	0/1	0/1	0/0
>	chr1:235,602,095	G	C	0.00009	0	TBCE	p.E376D	0/1	0/1	0/1	0/0
>	chr10:3,141,542	G	A	0.00000	0	PFKP	p.S53N	0/1	0/1	0/0	0/1
>	chr10:5,011,081	A	T	0.00026	0	AKR1C1	p.Q172L	0/1	0/1	0/1	0/0
>	chr10:26,880,240	A	T	0.00000	0	FAM238A	-	0/1	0/1	0/1	0/0
>	chr10:29,813,396	C	G	0.00000	0	SVIL	p.?	0/1	0/1	0/1	0/0
>	chr10:119,043,458	T	G	0.00799	3	PDZD8	p.N929T	0/1	0/1	0/0	0/1
>	chr10:135,342,118	C	T	0.00030	0	CYP2E1	p.P104L	0/1	0/1	0/1	0/0
>	chr10:135,342,118	C	T	0.00030	0	CYP2E1	p.P104L	0/1	0/1	0/1	0/0
>	chr11:433,417	C	T	0.00120	0	ANO9	p.D83N	0/1	0/1	0/1	0/0
>	chr11:18,047,154	C	T	0.00291	2	TPH1	p.A300T	0/1	0/1	0/1	0/0
>	chr11:18,740,269	G	A	0.00110	0	IGSF22	p.R235W	0/1	0/1	0/1	0/0
>	chr11:45,891,977	T	G	0.00000	0	CRY2	p.I420M	0/1	0/1	0/1	0/0
>	chr11:57,886,280	C	T	0.00937	5	OR9I1	p.V213I	0/1	0/1	0/1	0/0

60151

	#9							chr1:196,963,210
	#10							chr1:235,602,095
	#11							chr1:249,149,660

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	<div><div></div><div></div><div>4</div></div>	<div><div></div><div></div><div></div></div>	chr1:235,602,095
>	#11	<div><div></div><div></div><div></div></div>	<div><div></div><div></div><div></div></div>	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.

	#9							chr1:196,963,210
	#10							chr1:235,602,095
	#11							chr1:249,149,660

ACMG Criteria

Pathogenic

VERY STRONG EVIDENCE

☒ **PVS1** null variant

STRONG EVIDENCE

☐ **PS1** Located in a mutational hot spot and/or critical and well-established functional domain (e.g., active site of an enzyme) without benign variation

☐ **PS2**

☐ **PS3**

☐ **PS4**

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					chr1:235,602,095
>	#11					chr1:249,149,660

Overview

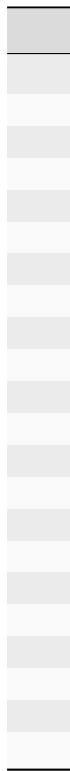
Quality Control

Variant Annotation

Export Jobs

Annotated Variants

Variant	Gene(s)	ACMG Rating	Flags							Comments
			Generic	Visual	Molecular	Validation	Phenotype	Summary		
chr1:235,602,095-G-C	-	<div></div>	<div><div></div><div></div><div></div><div></div><div></div><div></div></div>	<div></div>	<div></div>	<div></div>	<div></div>	<div></div>	<div></div>	<div>kiosk_user 2020/03/25 15:49: This variant lies in a known disease causing gene but is not described in literature yet.</div>





<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/varfish-org/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-v1-20210728b-grch37.tar.gz{,.sha256}
$ sha256sum --check varfish-site-data-v1-20210728b-grch37.tar.gz.sha256
$ tar xf varfish-site-data-v1-20210728b-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 varfish-org/varfish-server:0.22.1-0
↪"docker-entrypoint.s..." 17 hours ago Up 31 seconds 8080/tcp
↪ compose_varfish-celeryd-import_1
313afb611ab1 varfish-org/varfish-server:0.22.1-0
↪"docker-entrypoint.s..." 17 hours ago Up 30 seconds 8080/tcp
↪ compose_varfish-celerybeat_1
4d865726e83b varfish-org/varfish-server:0.22.1-0
↪"docker-entrypoint.s..." 17 hours ago Up 31 seconds 8080/tcp
```

```

compose_varfish-celeryd-query_1
a5f90232c4da  varfish-org/varfish-server:0.22.1-0
compose_varfish-celeryd-default_1
96cec7caebe4  varfish-org/varfish-server:0.22.1-0
compose_varfish-web_1
8d1f310c9b48  postgres:12
compose_postgres_1
8f12e16e20cd  minio/minio
compose_minio_1
03e877ac11db  quay.io/biocontainers/jannovar-cli:0.33--0
compose_jannovar_1
6af09b819e59  traefik:v2.3.1
compose_traefik_1
514cb4386224  redis:6
compose_redis_1
5678b9e6797b  quay.io/biocontainers/exomiser-rest-prioritiser:12.1.0--1
compose_exomiser-rest-prioritiser_1

```

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Log In

Please log in.

Log In

VarFish v0.22.1 - For research use only.

VarFish Beta

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Case List

Back to ProjectJoint Filtration

This is the project case list. Click on a case's name below to see its detail or click the button in its row to start filtering with a single click. Use the button on the top right to analyze all cases of this project together. The "Quality Control" tab shows the quality metrics for all cases in the project.

Case ListQuality ControlVariant Annotation

Progress10/10 (100%) cases

Case List

Filterinitial, active, closed as so

#	Updated	Status	Name	Individuals	Small Vars
1	6 minutes ago		NA12878	NA12878, NA12891, NA12892	1,862,277
2	7 minutes ago		HG00145	HG00145	743,861
3	7 minutes ago		HG00119	HG00119	1,046,059
4	7 minutes ago		HG00253	HG00253	199,467
5	8 minutes ago		HG00140	HG00140	221,114
6	8 minutes ago		HG00138	HG00138	204,543
7	8 minutes ago		HG00126	HG00126	159,947
8	8 minutes ago		HG00121	HG00121	263,372
9	8 minutes ago		HG00107	HG00107	188,251
10	8 minutes ago		HG00102	HG00102	188,579

Developed by BIH CUBI. For support and feedback, please contact cubi-helpdesk@duke.edu. VarFish v0.22.1

2021072820210728b
v1.2.120210728c20210728b20210728c

```
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/varfish-org/varfish-server:VERSION  
  env_file:  
    - .env  
  networks:  
    - varfish  
  restart: unless-stopped
```

```
volumes:
  - "/root/varfish-server-background-db-20210728b:/data:ro"
[...]
```

```
volumes:
  - "/root/varfish-server-background-db-20210728b:/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				
<div>Search term <input type="text"/> <input type="button" value="Search"/> <input type="button" value="New Features!"/> <input type="button" value="Manual"/> <input type="button" value="Help"/> <input type="button" value="User"/></div>				
Home				
Import Release Info				
Date of Import	Genomebuild	Table	Release	Comment
2019/12/18 22:11	GRCh37	EnsemblGenesensembl	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
$ cd volumes/cadd-rest-api
$ git clone https://github.com/kircherlab/CADD-scripts .
$ git checkout 7502f47
$ mkdir -p db
```

install.sh

```
$ docker run -it -e CADD=/opt/miniconda3/share/cadd-scripts-1.6-0 \
-v $PWD/volumes/cadd-rest-api:/data varfish-org/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 guide 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

```

cadd-rest-api.env

```

HTTP_PROXY=http://proxy.compa.ny:8080/
HTTPS_PROXY=http://proxy.compa.ny:8080/

```

.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: varfish-org/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: varfish-org/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
docker-compose.yml

upvarfish-docker-compose

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_CLIENT_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/pro/](https://my.qiagendigitalinsights.com/bbp/view/hgmd/pro

traefik

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

80808443

webwebsecure

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

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

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

```
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

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

```
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
$ H2_RELEASE=20210728b
$ JV_RELEASE=20210728
$ mkdir varfish-annotator-$RELEASE-$GENOME
$ cd varfish-annotator-$RELEASE-$GENOME
$ wget --no-check-certificate \
  https://file-public.cubi.bihealth.org/transient/varfish/anthenea/varfish-
↪annotator-db-$H2_RELEASE-$GENOME.h2.db.gz{,.sha256} \
  https://file-public.cubi.bihealth.org/transient/varfish/anthenea/jannovar-db-
↪$JV_RELEASE-$GENOME.tar.gz{,.sha256}
$ sha256sum --check varfish-annotator-db-$H2_RELEASE-$GENOME.h2.db.gz.sha256
varfish-annotator-db-$H2_RELEASE-grch37.h2.db.gz: OK
$ sha256sum --check jannovar-db-$JV_RELEASE-$GENOME.tar.gz.sha256
jannovar-db-$JV_RELEASE-grch37.tar.gz: OK
$ gzip -d varfish-annotator-db-$H2_RELEASE-$GENOME.h2.db.gz
$ tar xf jannovar-db-$JV_RELEASE-$GENOME.tar.gz
```

```
$ rm jannovar-db-$JV_RELEASE-$RELEASE.tar.gz{,.sha256} \
    varfish-annotator-db-$RELEASE-$GENOME.h2.db.gz.sha256
$ mv jannovar-db-$JV_RELEASE-$GENOME/* .
$ rmdir jannovar-db-$JV_RELEASE-$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 \
    --

    ./varfish-annotator-$RELEASE-$GENOME/varfish-annotator-db-$RELEASE-$GENOME.h2.
    ↪db \
    --ensembl-ser-path varfish-annotator-$RELEASE-$GENOME/ensembl*.ser \
    --refseq-ser-path varfish-annotator-$RELEASE-$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-$RELEASE-$GENOME/varfish-annotator-db-$RELEASE-  
↪$GENOME.h2.db \  
--ensembl-ser-path varfish-annotator-$RELEASE-$GENOME/ensembl*.ser \  
--refseq-ser-path varfish-annotator-$RELEASE-$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 $GENOME \
    \
    --db-path ./varfish-annotator-$RELEASE-$GENOME/varfish-annotator-db-$RELEASE-
    ↪$GENOME.h2.db \
    --ensembl-ser-path varfish-annotator-$RELEASE-$GENOME/ensembl*.ser \
    --refseq-ser-path varfish-annotator-$RELEASE-$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}
```

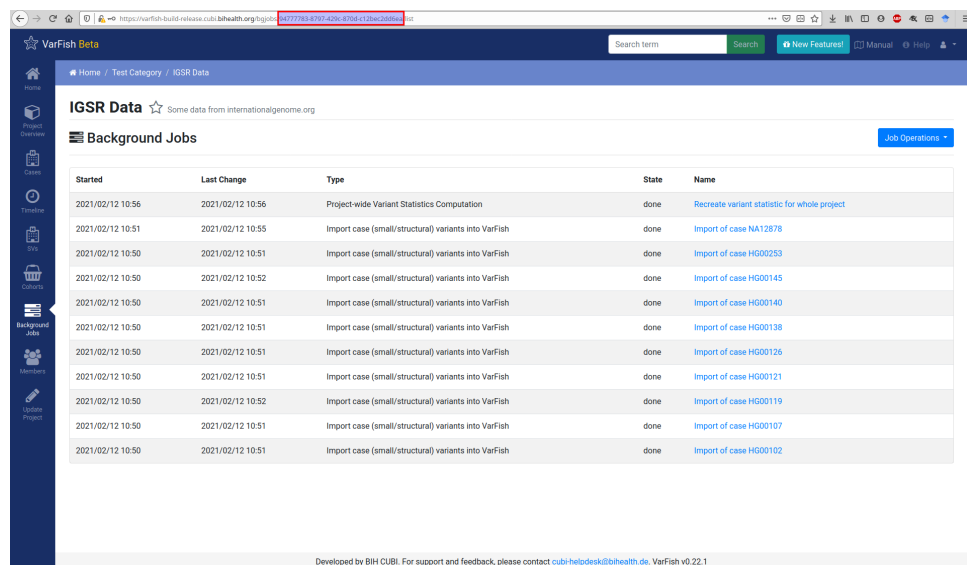
.ped

varfish-annotator.gts.tsv.gz

varfish-annotator.db-info.tsv.gz

.bam-qc.tsv.gz

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



The screenshot shows the VarFish Beta web interface. The browser address bar displays the URL: <https://varfish-build-release.cubi.bihhealth.org/igscr/94777783-8797-429c-870d-c12bec2dd6ea>. The page title is "IGSR Data" with a subtitle "Some data from internationalgenome.org". The main content area is titled "Background Jobs" and contains a table with the following data:

Started	Last Change	Type	State	Name
2021/02/12 10:56	2021/02/12 10:56	Project-wide Variant Statistics Computation	done	Recreate variant statistic for whole project
2021/02/12 10:51	2021/02/12 10:55	Import case (small/structural) variants into VarFish	done	Import of case NA12878
2021/02/12 10:50	2021/02/12 10:51	Import case (small/structural) variants into VarFish	done	Import of case HG00253
2021/02/12 10:50	2021/02/12 10:52	Import case (small/structural) variants into VarFish	done	Import of case HG00145
2021/02/12 10:50	2021/02/12 10:51	Import case (small/structural) variants into VarFish	done	Import of case HG00140
2021/02/12 10:50	2021/02/12 10:51	Import case (small/structural) variants into VarFish	done	Import of case HG00138
2021/02/12 10:50	2021/02/12 10:51	Import case (small/structural) variants into VarFish	done	Import of case HG00126
2021/02/12 10:50	2021/02/12 10:51	Import case (small/structural) variants into VarFish	done	Import of case HG00121
2021/02/12 10:50	2021/02/12 10:52	Import case (small/structural) variants into VarFish	done	Import of case HG00119
2021/02/12 10:50	2021/02/12 10:51	Import case (small/structural) variants into VarFish	done	Import of case HG00107
2021/02/12 10:50	2021/02/12 10:51	Import case (small/structural) variants into VarFish	done	Import of case HG00102

At the bottom of the page, it says: "Developed by BIH CUBI. For support and feedback, please contact cubi-helpdesk@bihhealth.de. VarFish v0.22.1"

.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,
  "100": 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

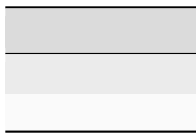
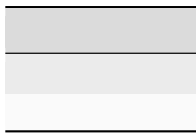
full_page_writesoff

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

random_page_cost1.1

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

```
conservation_knowngeneaa  
dbnp_dbnp  
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
```

python manage.py rebuild_variant_summaryvarfish-docker-compose

```
$ docker exec -it varfish-docker-compose_varfish-web_1 python /usr/src/app/manage.py \
rebuild_variant_summary
```

varfish-annotatorvarfish-cli

```
$ docker exec -it varfish-docker-compose_varfish-web_1 python /usr/src/app/manage.py \
svs_bg_sv_set_build
```

```
$ docker exec -it varfish-docker-compose_varfish-web_1 python /usr/src/app/manage.py \
svs_sv_fill_nulls
```

```
20210728bvarfish-docker-composedocker-compose down && docker-compose up -d
20210728c
```

```
/data/varfish-data/varfish-server-background-db-20210728c-grch37
import_versions.tsvdocker-compose.ymlvarfish-docker-composevarfish-webvol-
umes
```

```
volumes:
  - "/data:/data:ro"
```

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

```
docker exec -it varfish-docker-compose_varfish-web_1 python /usr/src/app/manage.py_
↳ \
    import_tables --force --truncate --tables-path /data/varfish-server-background-
↳ db-20210728c-grch37 \
    --threads=0
```

```
docker exec -it varfish-docker-compose_varfish-web_1 python /usr/src/app/manage.py_
↳ \
    import_tables --force --truncate --tables-path /data/varfish-server-background-
↳ db-20210728c-grch38 \
    --threads=0
```

```
v1.2.2
```

```
docker exec -it varfish-docker-compose_varfish-web_1 python /usr/src/app/manage.py_
↳ \
    rebuild_variant_summary
```

```
varfish-docker-composedocker-compose up -d
```

varfish-docker-composedocker-compose up -d

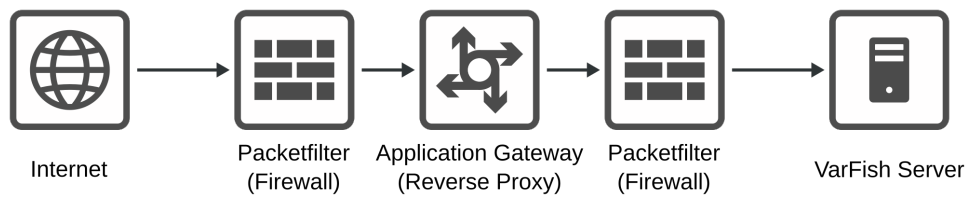
FIELD_ENCRYPTION_KEY
maintenanceexport
VARFISH_ENABLE_SPANR_SUBMISSION1

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
```

```
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

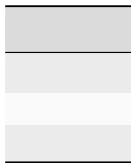
backup-large

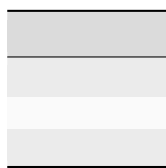
backup-small

varfish-`\${day_of_week}.sql.gz

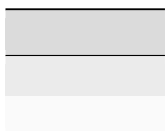
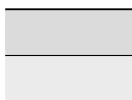
```
# docker exec -it varfish-docker-compose_varfish-web_1 \  
python /usr/src/app/manage.py pg_dump --mode=MODE \  
| gzip -c \  
> varfish-`${date +%a}.sql.gz
```

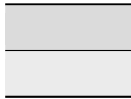
--

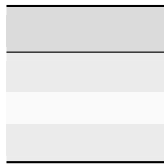




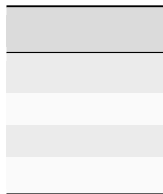
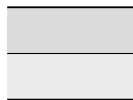
--

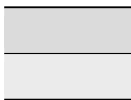






chrX







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
