



MUTATION
Surveyor®
DNA Variant Analysis Software

**Importing External Database
Annotation**

dbSNP, COSMIC, dbNSFP

SOFTGENETICS®

Software PowerTools for Genetic Analysis

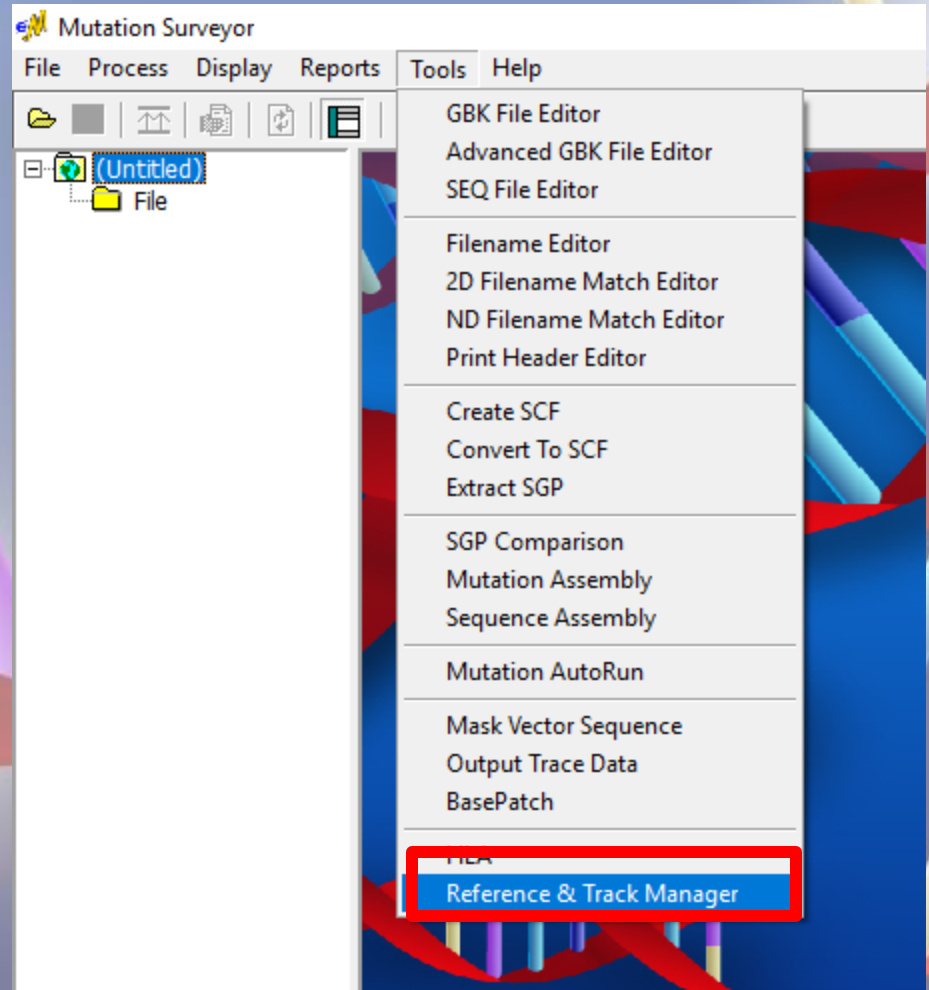
www.softgenetics.com / info@softgenetics.com

Mutation Surveyor Track Manager

- Mutation Surveyor software (5.0 and above) is able to import data from the dbNSFP, dbSNP, and COSMIC databases. Custom variation files may also be imported.
- Importing database information first requires the import of a whole human genome reference. Databases are installed specific to this reference.
- NextGENe software users may import and query data from same directory as NextGENe track manager.

Importing Reference Files

- To import a whole human genome reference, right click Mutation Surveyor icon and select “Run as Administrator”.
- Navigate to the tools menu and select the “Reference & Track Manager” option.



Selecting Reference Files

- If references have previously been imported by NextGENe software, you may select the directory

Storage Path: C:\Users\Public\Documents\SoftGenetics\NextGENe\References

Genome Build:

[Build Reference](#) [Import Reference](#)

Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	

[Import Track >](#)

Track	Type	Genome Build	Default Query	Last Modified	Location
ClinVar\142	dbSNP	Human_GRCh37	No	Thursday, Decemb...	Human_v37p10_dbsnp135
ClinVar\20160119	dbSNP	Human_GRCh37	No	01/19/16 07:42:58	Human_v37p10_dbsnp135
ClinVar\144	dbSNP	Human_GRCh37	Yes	02/05/16 14:55:38	Human_v37p10_dbsnp135
Cosmic\v67	Cosmic	Human_GRCh37	Yes	Wednesday, April 0...	Human_v37p10_dbsnp135
custom\clinvar_00-latest	custom	Human_GRCh37	No	01/19/16 07:47:32	Human_v37p10_dbsnp135
dbNSFP\2-5_commercial	dbNSFP	Human_GRCh37	Yes	Tuesday, Decembe...	Human_v37p10_dbsnp135
ESP\6500-V2-137	EspVCF	Human_GRCh37	No	Thursday, June 19,...	Human_v37p10_dbsnp135
ESP6500\137	custom	Human_GRCh37	Yes	05/13/15 11:16:48	Human_v37p10_dbsnp135

Importing Reference Files

- Click on the Import Reference link

SoftGenetics Reference & Track Manager

Storage Path: C:\Users\Public\Documents\SoftGenetics\NextGENE\References

Genome Build: Human_GRCh37

[Build Reference](#) [Import Reference](#)

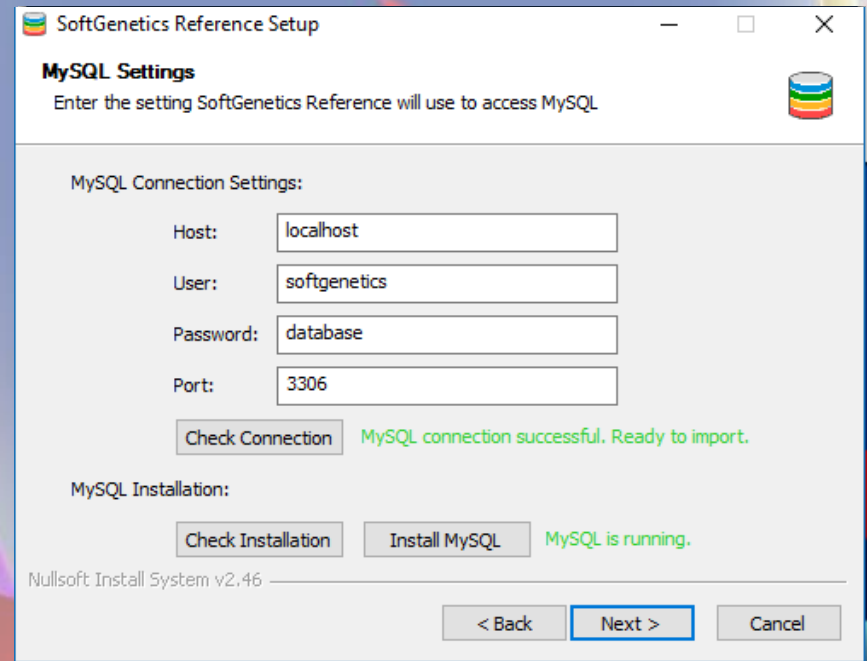
Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	

Tracks [Import Track >](#)

Track	Type	Genome Build	Default Query	Last Modified	Location
ClinVar\142	dbSNP	Human_GRCh37	No	Thursday, Decemb...	Human_v37p10_dbsnp135
ClinVar\20160119	dbSNP	Human_GRCh37	No	01/19/16 07:42:58	Human_v37p10_dbsnp135
ClinVar\144	dbSNP	Human_GRCh37	Yes	02/05/16 14:55:38	Human_v37p10_dbsnp135
Cosmic\v67	Cosmic	Human_GRCh37	Yes	Wednesday, April 0...	Human_v37p10_dbsnp135
custom\clinvar_00-latest	custom	Human_GRCh37	No	01/19/16 07:47:32	Human_v37p10_dbsnp135
dbNSFP\2-5-commercial	dbNSFP	Human_GRCh37	Yes	Tuesday, Decembe...	Human_v37p10_dbsnp135
ESP\6500-V2-137	EspVCF	Human_GRCh37	No	Thursday, June 19,...	Human_v37p10_dbsnp135
ESP6500\137	custom	Human_GRCh37	Yes	05/13/15 11:16:48	Human_v37p10_dbsnp135

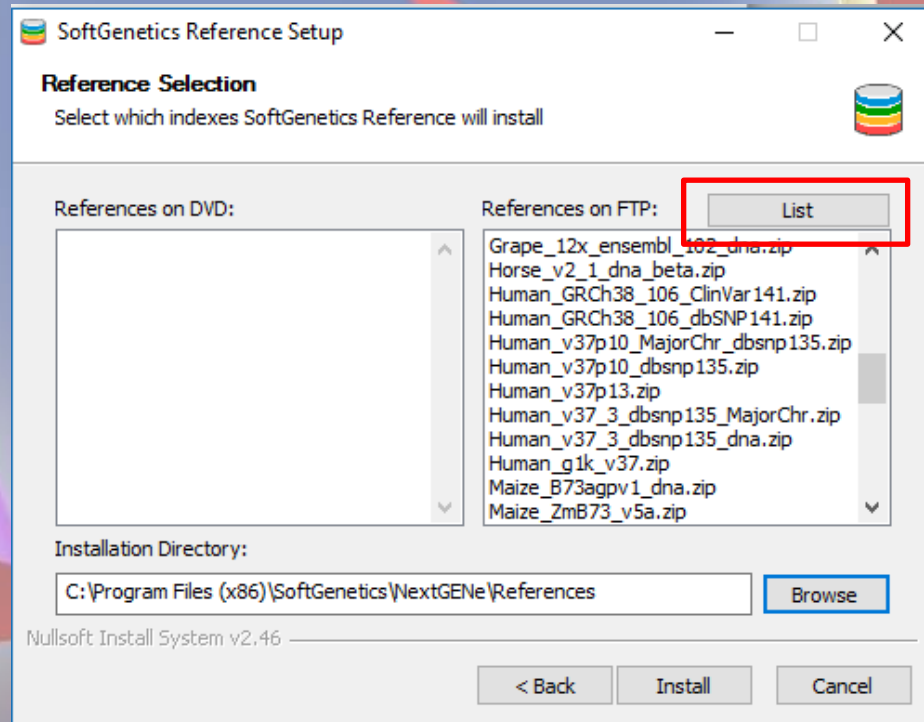
Import Reference Wizard

- Check MySQL connection. If connection failed because MySQL is not installed, click Install MySQL and proceed to next step.
- Messages will be green if the reference is ready for import.
- Click “Next” to continue to reference selection step



Import Reference Wizard

- To select and download a reference from our FTP server, select the “List” button.
- We recommend downloading Human_v37p13_105.zip.
- If you would like a DVD of a specific reference mailed to you, contact tech_support@softgenetics.com.
- After reference is selected, enter the installation directory at the bottom of the window
- Click install to install reference to directory. This process may take a few hours to complete.



Importing External Databases

- After selecting the reference directory, return to the Track Manager window to import external databases.
- To begin the process, select the Import Tracks link.

SoftGenetics Reference & Track Manager

Storage Path: C:\Users\Public\Documents\SoftGenetics\NextGENE\References

Genome Build: Human_GRCh37

Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	

Build Reference Import Reference

Track	Type	Genome Build	Default Query	Last Modified	Location
ClinVar_142	dbSNP	Human_GRCh37	No	Thursday, Decemb...	Human_v37p10_dbsnp135
ClinVar_20160119	dbSNP	Human_GRCh37	No	01/19/16 07:42:58	Human_v37p10_dbsnp135
ClinVar_144	dbSNP	Human_GRCh37	Yes	02/05/16 14:55:38	Human_v37p10_dbsnp135
Cosmic_v67	Cosmic	Human_GRCh37	Yes	Wednesday, April 0...	Human_v37p10_dbsnp135
custom\clinvar_00-latest	custom	Human_GRCh37	No	01/19/16 07:47:32	Human_v37p10_dbsnp135
dbNSFP 2-5_commercial	dbNSFP	Human_GRCh37	Yes	Tuesday, Decembe...	Human_v37p10_dbsnp135
ESP 6500-v2-137	EspVCF	Human_GRCh37	No	Thursday, June 19, ...	Human_v37p10_dbsnp135
ESP6500 137	custom	Human_GRCh37	Yes	05/13/15 11:16:48	Human_v37p10_dbsnp135

Import Track >

Importing dbNSFP

- Click “Import dbNSFP”

Storage Path: C:\Program Files (x86)\SoftGenetics\NextGENE\References

Genome Build:

Preloaded References Build Reference Import Reference

Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	
Human_GRCh38_106_dbSNP141	Human_GRCh38	sg_grch38r106	
Human_v37p13	reference::Human_v37p13	sg_grch37p10	

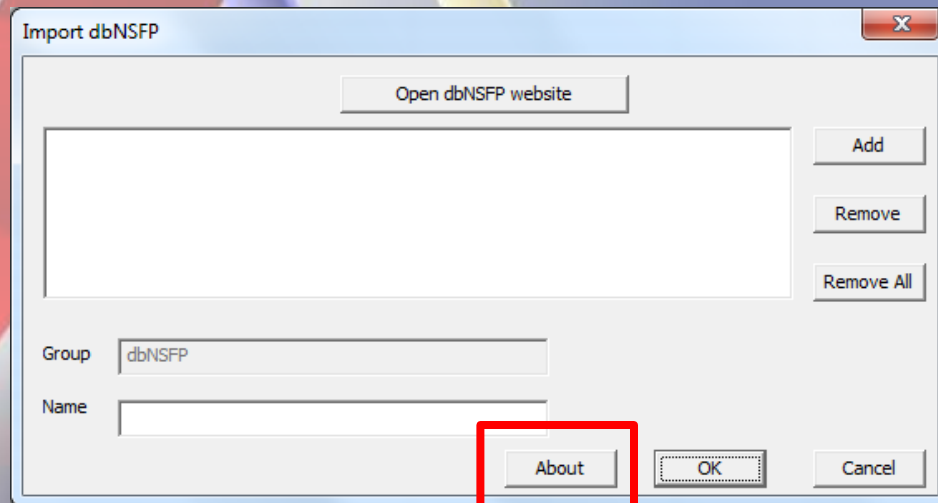
Tracks Import Track >

Track	Type	Genome Build	Default Query	Last Modified	Location
clinvar\20170801	dbSNP	Human_GRCh37	Yes	08/22/17 16:54:57	Tracks
clinvar\20171029	dbSNP	Human_GRCh37	Yes	2017-11-08 14:32:02	Tracks
dbsnp\b150	dbSNP	Human_GRCh37	Yes	05/09/17 13:37:16	Tracks
UKDB\Artifact	custom	Human_GRCh37	No	2017-12-18 10:55:22	Tracks
UKDB\False Positive	custom	Human_GRCh37	No	2017-12-18 10:55:24	Tracks
ClinVar\20170501	dbSNP	Human_GRCh37	No	05/08/17 08:20:23	Human_v37p10_c
Cosmic\68	Cosmic	Human_GRCh37	Yes	Tuesday, January ...	Human_v37p10_c
dbNSFP\2.9_commercial	dbNSFP	Human_GRCh37	Yes	04/01/15 15:50:47	Human_v37p10_dbsnp135
dbSNP\149	dbSNP	Human_GRCh37	No	01/30/17 18:22:55	Human_v37p10_dbsnp135
ExAC\0.3.1	custom	Human_GRCh37	Yes	06/24/16 13:57:39	Human_v37p10_dbsnp135
ClinVar\142	dbSNP	Human_GRCh38	No	Monday, February ...	Human_GRCh38_106_dbSNP141
dbNSFP\2.5	dbNSFP	Human_GRCh38	No	Thursday, Februar...	Human_GRCh38_106_dbSNP141
dbSNP\142	dbSNP	Human_GRCh38	No	Monday, February ...	Human_GRCh38_106_dbSNP141

dbSNP/Clinvar
COSMIC
dbNSFP
dbSNP
Custom Variant Track
Gene Annotation Track

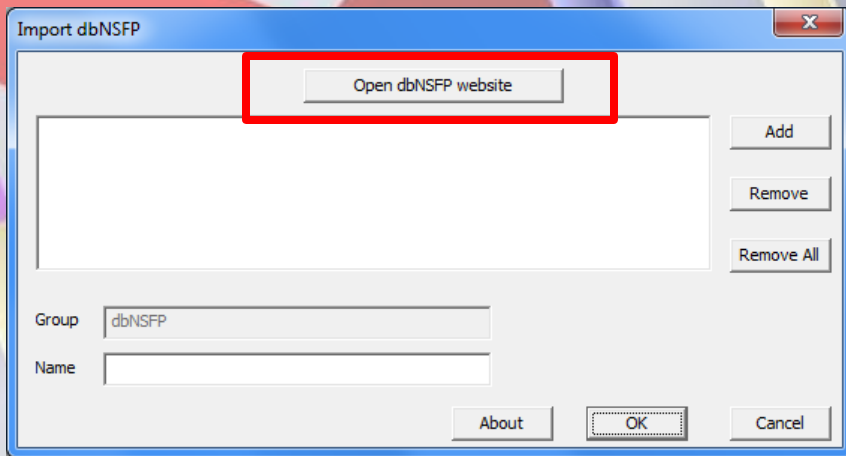
Importing dbNSFP

- A link to the paper for dbNSFP can be found by clicking the “About” button in the Import dbNSFP dialog



Importing dbNSFP

- Click the “Open dbNSFP website” button to download the database from the dbNSFP web page.
- The latest support version is v2.9.



The image shows a web browser window displaying the Jpopgen website. The page title is "Jpopgen". The main heading is "dbNSFP". The page content includes an introduction, a citation list, and contact information for Xiaoming Liu.

Jpopgen

Navigation
Jpopgen - a collection of java programs for population genetic analysis
Sitemap

About the maintainer
This site is maintained by:
Xiaoming Liu, Ph.D.
Assistant Professor,
Human Genetics Center,
School of Public Health,
The University of Texas Health
Science Center at Houston.
Contact:
xmliu.uth[at]gmail.com

dbNSFP

INTRODUCTION:

dbNSFP is a database developed for functional prediction and annotation of all potential non-synonymous single-nucleotide variants (nsSNVs) in the human genome. Its current version (ver 2.0) is based on the Gencode release 9 / Ensembl version 64 and includes a total of 87,347,043 nsSNVs and 2,270,742 essential splice site SNVs. It compiles prediction scores from six prediction algorithms (SIFT, Polyphen2, LRT, MutationTaster, MutationAssessor and FATHMM), three conservation scores (PhyloP, GERP++ and SiPhy) and other related information including allele frequencies observed in the 1000 Genomes Project phase 1 data and the NHLBI Exome Sequencing Project, various gene IDs from different databases, functional descriptions of genes, gene expression and gene interaction information, etc.

Some dbNSFP contents (may not be up-to-date though) can also be accessed through [variant tools](#), [ANNOVAR](#), [KGGSeq](#), UCSC Genome Browser's [Variant Annotation Integrator](#), [Ensembl Variant Effect Predictor](#) and [HGMD](#). Please cite our papers (see below) if you used dbNSFP contents through those tools.

Please note some component scores/content of dbNSFP has specific requirements or licence for non-academic usage. dbNSFP does not grant the non-academic usage of those scores/contents, so please contact the original score/content provider for that purpose.

We welcome developers of functional prediction methods to provide their predictions and scores to the database. Please contact Dr. Liu (xmliu.uth[at]gmail.com).

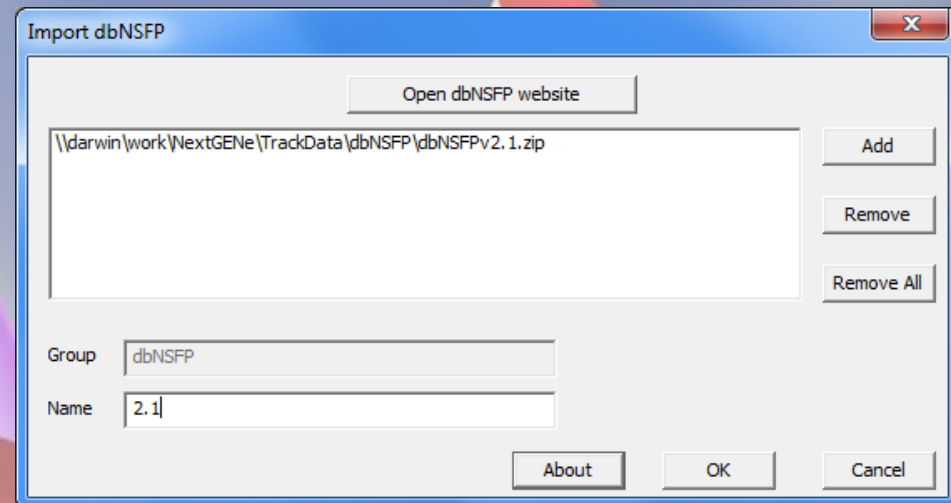
CITATION:

1. Liu X, Jian X, and Boerwinkle E. 2011. [dbNSFP: a lightweight database of human non-synonymous SNPs and their functional predictions](#). *Human Mutation*. 32:894-899.
2. Liu X, Jian X, and Boerwinkle E. 2013. [dbNSFP-v2.0: A Database of Human Non-synonymous SNVs and Their Functional Predictions and Annotations](#). *Human Mutation*. 34:E2393-E2402.

If you use dbNSFP v1.x, please cite our paper 1. If you use dbNSFP v2.x, please cite our papers 1 & 2.

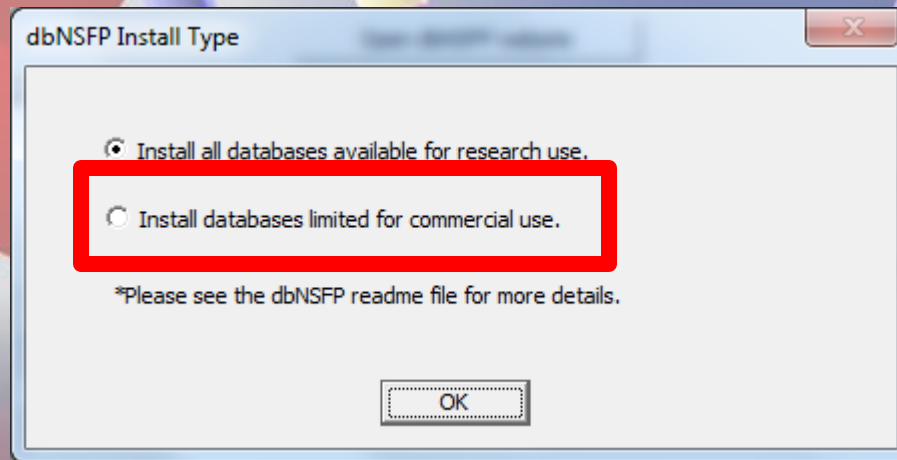
Importing dbNSFP

- Click “Add” to load the file.
- Enter Name of dbNSFP version.
- Click the “OK” button.
- Note: The original files can be deleted once the tracks have finished importing.



Importing dbNSFP

- Some databases are intended for research use only. Commercial users should install the commercial database.



Importing COSMIC

- Click "Import COSMIC"

Storage Path: C:\Program Files (x86)\SoftGenetics\NextGENE\References

Genome Build:

Preloaded References [Build Reference](#) [Import Reference](#)

Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	
Human_GRCh38_106_dbsnp141	Human_GRCh38	sg_grch38r106	
Human_v37p13	reference::Human_v37p13	sg_grch37p10	

Tracks [Import Track >](#)

Track	Type	Genome Build	Default Query	Last Modified	Location	
dinvar\20170801	dbSNP	Human_GRCh37	Yes	08/22/17 16:54:57	Tracks	dbSNP/Clinvar
dinvar\20171029	dbSNP	Human_GRCh37	Yes	2017-11-08 14:32:02	Tracks	COSMIC
dbsnp\b150	dbSNP	Human_GRCh37	Yes	05/09/17 13:37:16	Tracks	dbNSFP
UKDB\Artifact	custom	Human_GRCh37	No	2017-12-18 10:55:22	Tracks	dbscSNV
UKDB\False Positive	custom	Human_GRCh37	No	2017-12-18 10:55:24	Tracks	Custom Variant Track
ClinVar\20170501	dbSNP	Human_GRCh37	No	05/08/17 08:20:23	Human_v37p10_d	Gene Annoation Track
Cosmic\68	Cosmic	Human_GRCh37	Yes	Tuesday, January ...	Human_v37p10_d	
dbNSFP\2.9_commercial	dbNSFP	Human_GRCh37	Yes	04/01/15 15:50:47	Human_v37p10_dbsnp135	
dbSNP\149	dbSNP	Human_GRCh37	No	01/30/17 18:22:55	Human_v37p10_dbsnp135	
ExAC\0.3.1	custom	Human_GRCh37	Yes	06/24/16 13:57:39	Human_v37p10_dbsnp135	
ClinVar\142	dbSNP	Human_GRCh38	No	Monday, February ...	Human_GRCh38_106_dbsnp141	
dbNSFP\2.5	dbNSFP	Human_GRCh38	No	Thursday, Februar...	Human_GRCh38_106_dbsnp141	
dbSNP\142	dbSNP	Human_GRCh38	No	Monday, February ...	Human_GRCh38_106_dbsnp141	

Importing COSMIC

- Registered/Licensed users must [login](#) to COSMIC site prior to download and in accordance with the [terms and conditions](#) of the [licensing policy](#).
- Expand the VCF files list on the [Data Download](#) page.
- You can [download](#) COSMIC information for coding variants and/or non coding variants

COSMIC
Catalogue of somatic mutations in cancer

GRCh37

Home ▾ About ▾ Licensing ▾ Data Download ▾ News ▾ Help ▾ Enter search here...

Download

The following section contains descriptions and locations of data files in the [SFTP site](#) from the current COSMIC release v75. As part of COSMIC's growth and development plan, we have implemented a new licensing strategy and we encourage users to be aware of the [terms and conditions](#). Please contact cosmic_translation@sanger.ac.uk for more information.

Note: If you need any help downloading files, please [click here](#).

- Classification Information
- Complete COSMIC data
- Complete mutation data
- Structural Genomic rearrangements
- Complete Fusion Export
- All Mutation in census genes
- Non coding variants
- Copy Number Variants
- Gene Expression
- Methylation
- Cancer Gene Census
- Cosmic sample features
- VCF files (coding and non-coding mutations)**

CosmicCodingMuts.vcf.gz -> VCF file of all coding mutations in cosmic, location for this file is here : [CosmicCodingMuts.vcf.gz](#)

CosmicNonCodingVariants.vcf.gz -> VCF file of all non coding mutations in cosmic, location for this file is here : [CosmicNonCodingVariants.vcf.gz](#)

- Fasta file (genes)
- Oracle database dump

Importing COSMIC

- Click “Add” to load file(s).
- Specify the COSMIC version number or other name.
- Select appropriate genome build from dropdown
- Click “Next”.

Import track - COSMIC

Guidelines on Use of COSMIC Data Register for COSMIC download

Load Coding Variants: CosmicCodingMuts_vXX_DDMMYYYY_nolimit.vcf.gz
Load NonCoding Variants: CosmicNonCodingVariants_vXX_DDMMYYYY_noLimit.vcf.gz

Group: Cosmic

Name:

Genome Build:

Add Remove Remove All

Next Cancel

Import ClinVar/dbSNP

- Click “Import dbSNP/ClinVar”

Storage Path: C:\Program Files (x86)\SoftGenetics\NextGENE\References

Genome Build:

Preloaded References [Build Reference](#) [Import Reference](#)

Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	
Human_GRCh38_106_dbSNP141	Human_GRCh38	sg_grch38r106	
Human_v37p13	reference::Human_v37p13	sg_grch37p10	

Tracks [Import Track >](#)

Track	Type	Genome Build	Default Query	Last Modified	Location
clinvar\20170801	dbSNP	Human_GRCh37	Yes	08/22/17 16:54:57	Tracks
clinvar\20171029	dbSNP	Human_GRCh37	Yes	2017-11-08 14:32:02	Tracks
dbsnp\b150	dbSNP	Human_GRCh37	Yes	05/09/17 13:37:16	Tracks
UKDB\Artifact	custom	Human_GRCh37	No	2017-12-18 10:55:22	Tracks
UKDB\False Positive	custom	Human_GRCh37	No	2017-12-18 10:55:24	Tracks
ClinVar\20170501	dbSNP	Human_GRCh37	No	05/08/17 08:20:23	Human_v37p10_d
Cosmic\68	Cosmic	Human_GRCh37	Yes	Tuesday, January ...	Human_v37p10_d
dbNSFP\2.9_commercial	dbNSFP	Human_GRCh37	Yes	04/01/15 15:50:47	Human_v37p10_dbsnp135
dbSNP\149	dbSNP	Human_GRCh37	No	01/30/17 18:22:55	Human_v37p10_dbsnp135
ExAC\0.3.1	custom	Human_GRCh37	Yes	06/24/16 13:57:39	Human_v37p10_dbsnp135
ClinVar\142	dbSNP	Human_GRCh38	No	Monday, February ...	Human_GRCh38_106_dbSNP141
dbNSFP\2.5	dbNSFP	Human_GRCh38	No	Thursday, Februar...	Human_GRCh38_106_dbSNP141
dbSNP\142	dbSNP	Human_GRCh38	No	Monday, February ...	Human_GRCh38_106_dbSNP141

dbSNP/Clinvar

Cosmic

dbNSFP

dbscSNV

Custom Variant Track

Gene Annoation Track

Import ClinVar/dbSNP

- Download files using these links or by clicking the “Open FTP Folder to Download VCF” button

GRCh37:

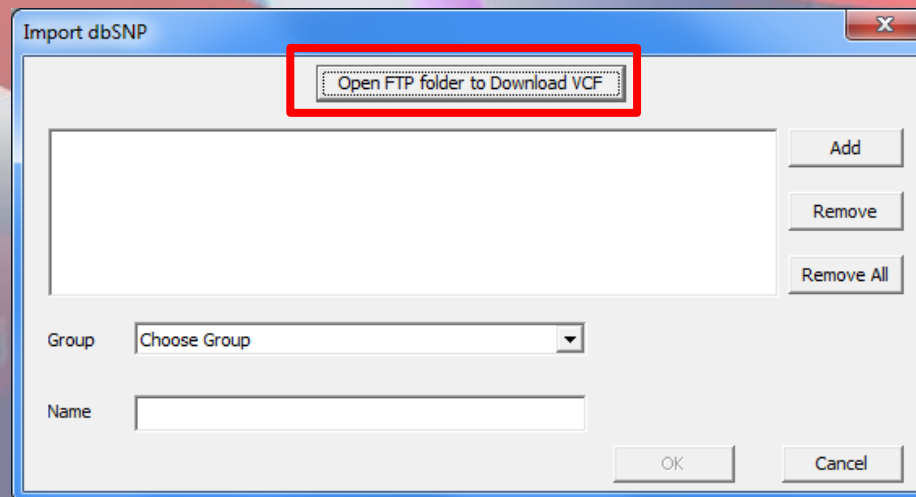
dbSNP: ftp://ftp.ncbi.nlm.nih.gov/snp/organisms/human_9606_b151_GRCh37p13/VCF/00-All.vcf.gz

ClinVar: ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/vcf_GRCh37/clinvar.vcf.gz

GRCh38:

dbSNP: ftp://ftp.ncbi.nlm.nih.gov/snp/organisms/human_9606/VCF/00-All.vcf.gz

ClinVar: ftp.ncbi.nih.gov/pub/clinvar/vcf_GRCh38/clinvar.vcf.gz



Import ClinVar/dbSNP

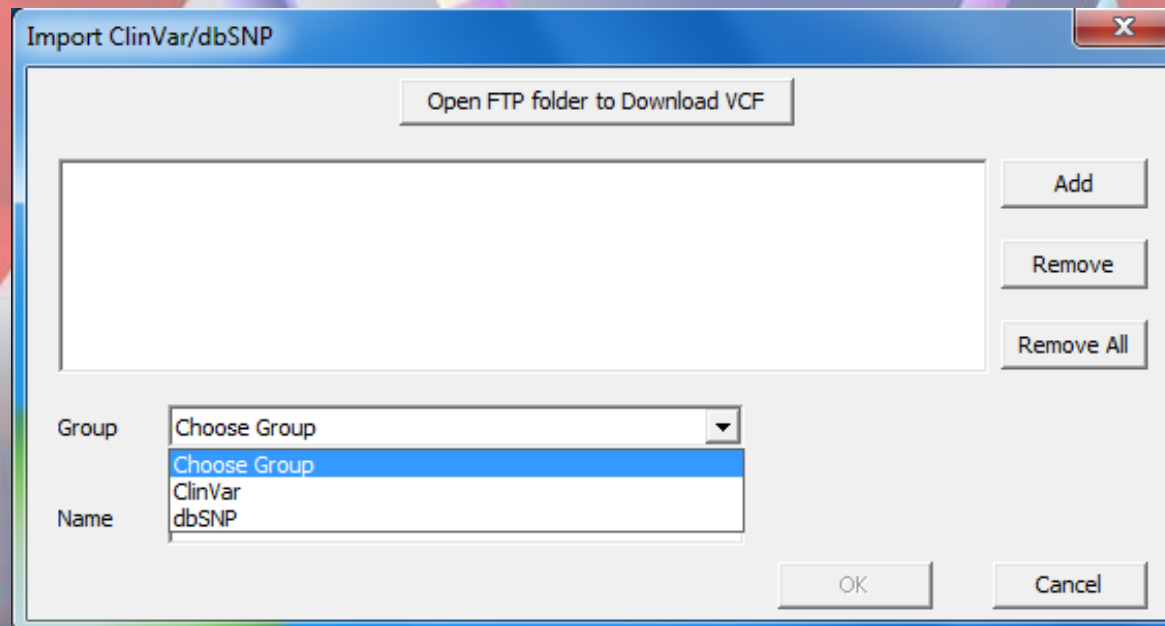
Index of /snp/organisms/human_9606_b144_GRCh37p13/VCF/

Name	Size	Date Modified
[parent directory]		
<input checked="" type="checkbox"/> 00-All.vcf.gz	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-All.vcf.gz.md5	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-All.vcf.gz.tbi	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-All_papu.vcf.gz	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-All_papu.vcf.gz.md5	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-All_papu.vcf.gz.tbi	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-common_all.vcf.gz	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-common_all.vcf.gz.md5	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-common_all.vcf.gz.tbi	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-common_all_papu.vcf.gz	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-common_all_papu.vcf.gz.md5	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> 00-common_all_papu.vcf.gz.tbi	0 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> All_20150605.vcf.gz	3.0 GB	6/8/15, 12:00:00 AM
<input type="checkbox"/> All_20150605.vcf.gz.md5	54 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> All_20150605.vcf.gz.tbi	2.3 MB	6/8/15, 12:00:00 AM
<input type="checkbox"/> All_20150605_papu.vcf.gz	3.9 MB	6/8/15, 12:00:00 AM
<input type="checkbox"/> All_20150605_papu.vcf.gz.md5	59 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> All_20150605_papu.vcf.gz.tbi	33.4 kB	6/8/15, 12:00:00 AM
<input type="checkbox"/> GRCh38_VCF_files	0 B	10/13/15, 6:39:00 PM
<input type="checkbox"/> README.txt	101 B	10/9/15, 5:42:00 PM
archive/		6/2/15, 12:00:00 AM
<input checked="" type="checkbox"/> clinical_vcf_set	0 B	6/2/15, 12:00:00 AM
<input type="checkbox"/> common_all_20150605.vcf.gz	888 MB	6/8/15, 12:00:00 AM
<input type="checkbox"/> common_all_20150605.vcf.gz.md5	61 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> common_all_20150605.vcf.gz.tbi	1.9 MB	6/8/15, 12:00:00 AM
<input type="checkbox"/> common_all_20150605_papu.vcf.gz	2.3 kB	6/8/15, 12:00:00 AM
<input type="checkbox"/> common_all_20150605_papu.vcf.gz.md5	66 B	6/8/15, 12:00:00 AM
<input type="checkbox"/> common_all_20150605_papu.vcf.gz.tbi	72 B	6/8/15, 12:00:00 AM

- Scroll down the list of organisms and select the preferred organism and database build.
- Click on the VCF folder option
- Select “00-All.vcf.gz” to begin download of the whole dbSNP database.
- To download ClinVar database, click “clinical_vcf_set” and select the file “clinvar.vcf.gz”
- For information regarding the different VCF files generated in each build, please see NCBI’s [Human Variation Sets in VCF Format](#)

Import ClinVar/dbSNP

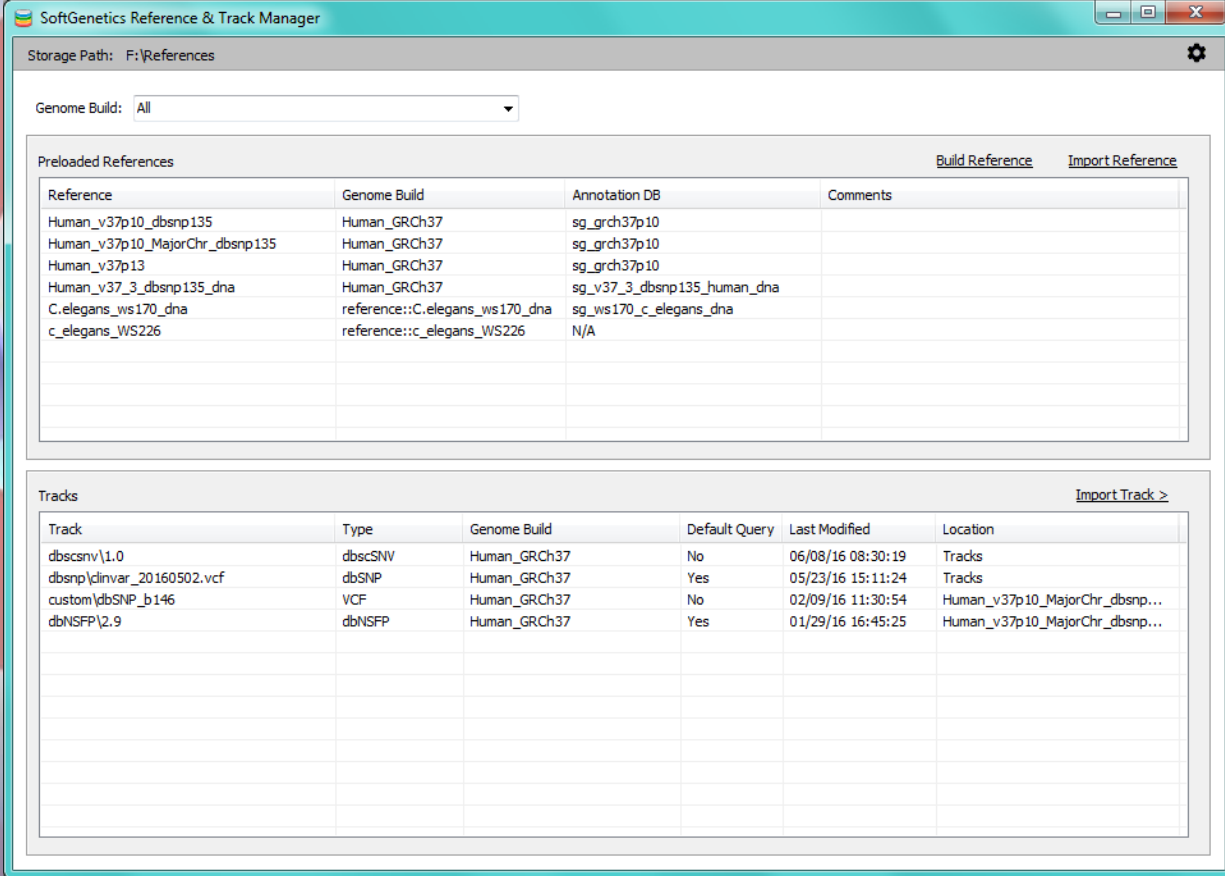
- After download is complete, select Group from dropdown and click “Add” to add database
 - Select ClinVar for clinically relevant SNPs (if downloaded)
 - Select dbSNP for common SNPs (and for 00-All.vcf.gz)
- Enter dbSNP build number in name field
- Click “OK”



Importing a Custom Variant Database

The Track Manager Tool includes specialized support for the dbNSFP, dbSNP, COSMIC and ClinVar/dbSNP databases.

To import a custom database, database files should first be downloaded outside of Mutation Surveyor



The screenshot displays the 'SoftGenetics Reference & Track Manager' application window. The 'Storage Path' is set to 'F:\References' and the 'Genome Build' is set to 'All'. The interface is divided into two main sections: 'Preloaded References' and 'Tracks'.

Preloaded References

Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	
Human_v37p10_MajorChr_dbsnp135	Human_GRCh37	sg_grch37p10	
Human_v37p13	Human_GRCh37	sg_grch37p10	
Human_v37_3_dbsnp135_dna	Human_GRCh37	sg_v37_3_dbsnp135_human_dna	
C.elegans_ws170_dna	reference::C.elegans_ws170_dna	sg_ws170_c_elegans_dna	
c_elegans_WS226	reference::c_elegans_WS226	N/A	

Tracks

Track	Type	Genome Build	Default Query	Last Modified	Location
dbscsnv\1.0	dbscSNV	Human_GRCh37	No	06/08/16 08:30:19	Tracks
dbsnp\clinvar_20160502.vcf	dbSNP	Human_GRCh37	Yes	05/23/16 15:11:24	Tracks
custom\dbSNP_b146	VCF	Human_GRCh37	No	02/09/16 11:30:54	Human_v37p10_MajorChr_dbsnp...
dbNSFP\2.9	dbNSFP	Human_GRCh37	Yes	01/29/16 16:45:25	Human_v37p10_MajorChr_dbsnp...

Importing a Custom Variant Database

Click “Import Track” > “Custom Variant Track”

Storage Path: F:\References

Genome Build: All

Preloaded References

Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	
Human_v37p10_MajorChr_dbsnp135	Human_GRCh37	sg_grch37p10	
Human_v37p13	Human_GRCh37	sg_grch37p10	
Human_v37_3_dbsnp135_dna	Human_GRCh37	sg_v37_3_dbsnp135_human_dna	
C.elegans_ws170_dna	reference::C.elegans_ws170_dna	sg_ws170_c_elegans_dna	
c_elegans_WS226	reference::c_elegans_WS226	N/A	

Build Reference Import Reference

Tracks

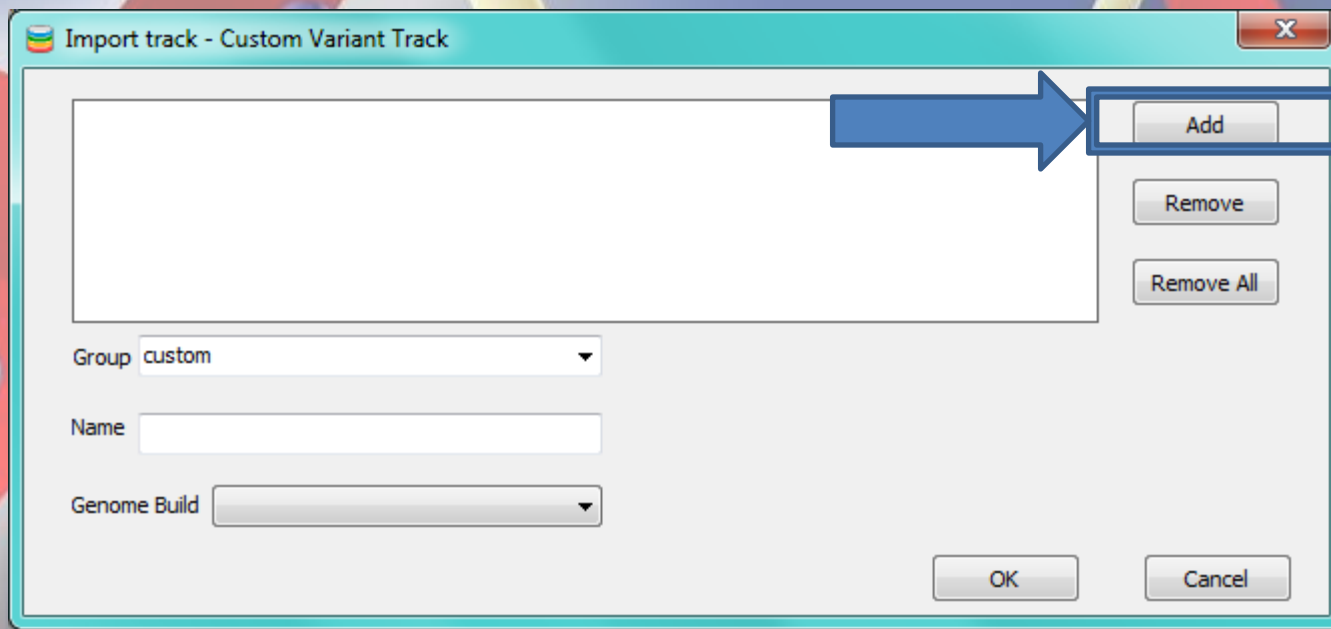
Track	Type	Genome Build	Default Query	Last Modified	Location
dbscsnv\1.0	dbscSNV	Human_GRCh37	No	06/08/16 08:30:19	Tracks
dbsnp\clinvar_20160502.vcf	dbSNP	Human_GRCh37	Yes	05/23/16 15:11:24	Tracks
custom\dbsnp_b146	VCF	Human_GRCh37	No	02/09/16 11:30:54	Human_v37p10_M
dbNSFP\2.9	dbNSFP	Human_GRCh37	Yes	01/29/16 16:45:25	Human_v37p10_M

Import Track >

- dbSNP/Clinvar
- COSMIC
- dbNSFP
- dbscSNV
- Custom Variant Track**
- Gene Annotation Track

Importing a Custom Variant Database

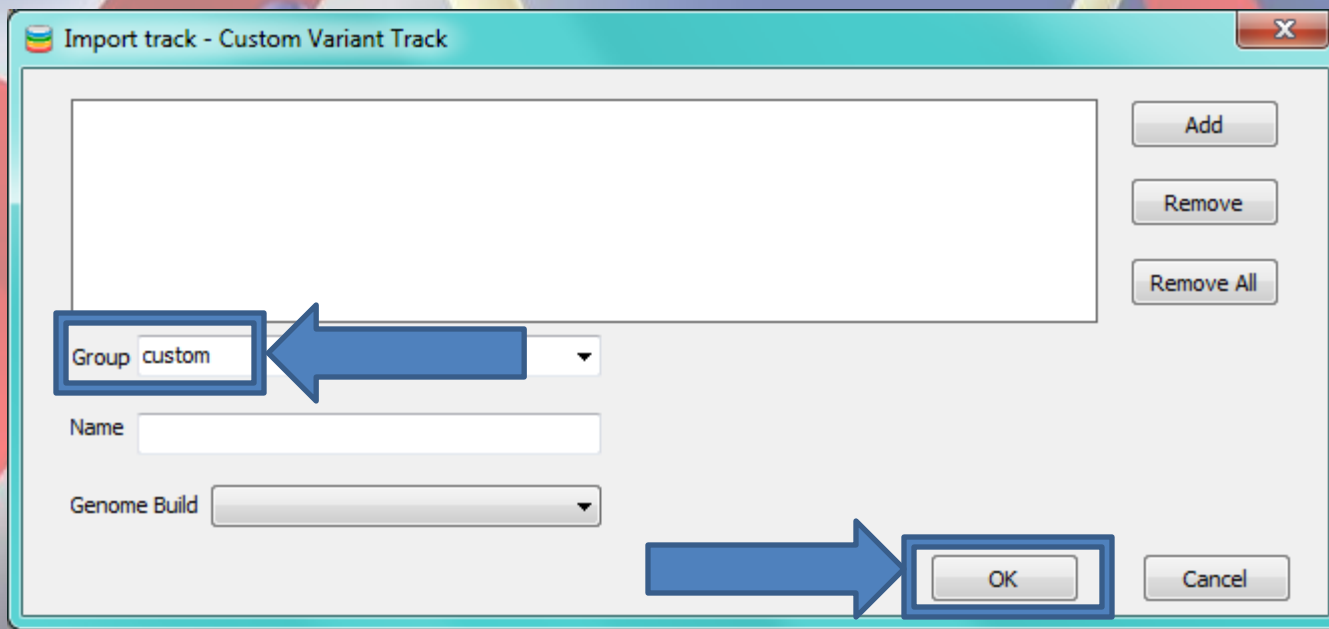
Click “Add” to browse to and select the downloaded database file



Note: To import a custom database, database files should first be downloaded outside of Mutation Surveyor

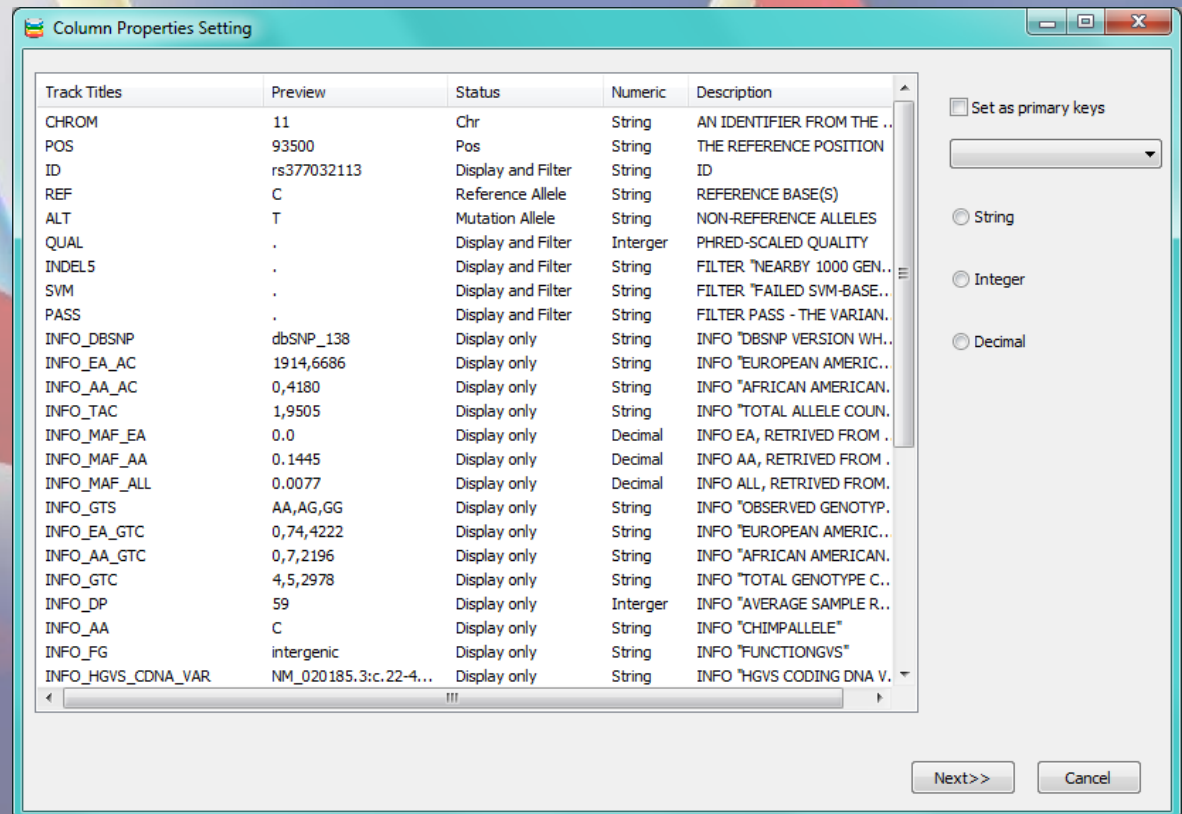
Importing a Custom Variant Database

Group can be renamed from Custom. Enter a name including version for the database track and select the appropriate genome build from the dropdown list. Then click “OK”.



Importing a Custom Variant Database

Custom Database Import – Exome Variant Server used for example
Choose how to handle all fields in the database. Highlight any field and choose “Skip”, “Display Only”, or “Display and Filtering”.



The screenshot shows a window titled "Column Properties Setting" with a table of variant fields. The table has five columns: Track Titles, Preview, Status, Numeric, and Description. The fields listed include CHROM, POS, ID, REF, ALT, QUAL, INDEL5, SVM, PASS, and various INFO fields (INFO_DBSNP, INFO_EA_AC, INFO_AA_AC, INFO_TAC, INFO_MAF_EA, INFO_MAF_AA, INFO_MAF_ALL, INFO_GTS, INFO_EA_GTC, INFO_AA_GTC, INFO_GTC, INFO_DP, INFO_AA, INFO_FG, INFO_HGVS_CDNA_VAR). The Status column indicates how each field is handled, such as "Chr", "Pos", "Display and Filter", "Display only", or "Mutation Allele". The Numeric column shows the data type, such as "String", "Integer", or "Decimal". The Description column provides a brief explanation of each field.

Track Titles	Preview	Status	Numeric	Description
CHROM	11	Chr	String	AN IDENTIFIER FROM THE ..
POS	93500	Pos	String	THE REFERENCE POSITION
ID	rs377032113	Display and Filter	String	ID
REF	C	Reference Allele	String	REFERENCE BASE(S)
ALT	T	Mutation Allele	String	NON-REFERENCE ALLELES
QUAL	.	Display and Filter	Integer	PHRED-SCALED QUALITY
INDEL5	.	Display and Filter	String	FILTER "NEARBY 1000 GEN..
SVM	.	Display and Filter	String	FILTER "FAILED SVM-BASE..
PASS	.	Display and Filter	String	FILTER PASS - THE VARIAN.
INFO_DBSNP	dbSNP_138	Display only	String	INFO "DBSNP VERSION WH..
INFO_EA_AC	1914,6686	Display only	String	INFO "EUROPEAN AMERIC..
INFO_AA_AC	0,4180	Display only	String	INFO "AFRICAN AMERICAN.
INFO_TAC	1,9505	Display only	String	INFO "TOTAL ALLELE COUN.
INFO_MAF_EA	0.0	Display only	Decimal	INFO EA, RETRIVED FROM .
INFO_MAF_AA	0.1445	Display only	Decimal	INFO AA, RETRIVED FROM .
INFO_MAF_ALL	0.0077	Display only	Decimal	INFO ALL, RETRIVED FROM.
INFO_GTS	AA,AG,GG	Display only	String	INFO "OBSERVED GENOTYP.
INFO_EA_GTC	0,74,4222	Display only	String	INFO "EUROPEAN AMERIC..
INFO_AA_GTC	0,7,2196	Display only	String	INFO "AFRICAN AMERICAN.
INFO_GTC	4,5,2978	Display only	String	INFO "TOTAL GENOTYPE C..
INFO_DP	59	Display only	Integer	INFO "AVERAGE SAMPLE R..
INFO_AA	C	Display only	String	INFO "CHIMPALLELE"
INFO_FG	intergenic	Display only	String	INFO "FUNCTIONGVGS"
INFO_HGVS_CDNA_VAR	NM_020185.3:c.22-4...	Display only	String	INFO "HGVS CODING DNA V..

On the right side of the dialog, there is a checkbox labeled "Set as primary keys" which is unchecked. Below it is a dropdown menu. Further down are three radio buttons labeled "String", "Integer", and "Decimal". At the bottom right, there are "Next >>" and "Cancel" buttons.

Importing a Custom Variant Database

Custom Database Import – Exome Variant Server used for example
After modifying the Import Settings as needed, click “Next” to begin import

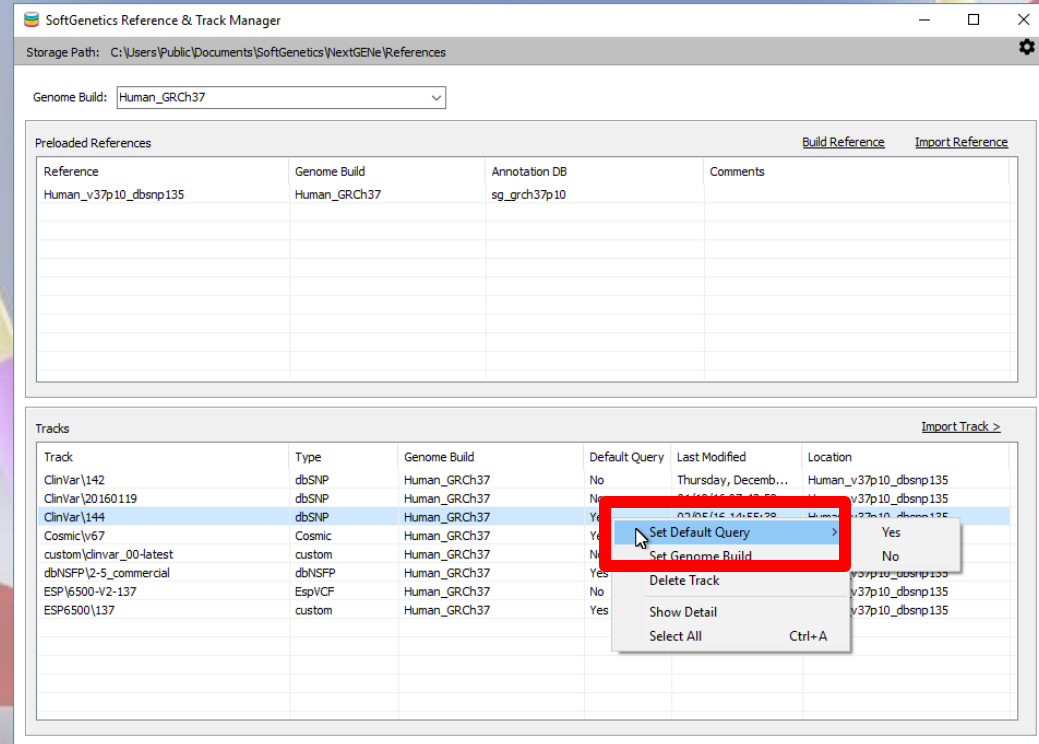
The screenshot shows the 'Column Properties Setting' dialog box. The main area contains a table with the following columns: Track Titles, Preview, Status, Numeric, and Description. The table lists various columns such as CHROM, POS, ID, REF, ALT, QUAL, INDEL5, SVM, PASS, and INFO_* columns. The 'Next >>' button is highlighted with a blue arrow.

Track Titles	Preview	Status	Numeric	Description
CHROM	11	Chr	String	AN IDENTIFIER FROM THE ..
POS	93500	Pos	String	THE REFERENCE POSITION
ID	rs377032113	Display and Filter	String	ID
REF	C	Reference Allele	String	REFERENCE BASE(S)
ALT	T	Mutation Allele	String	NON-REFERENCE ALLELES
QUAL	.	Display and Filter	Integer	PHRED-SCALED QUALITY
INDEL5	.	Display and Filter	String	FILTER "NEARBY 1000 GEN..
SVM	.	Display and Filter	String	FILTER "FAILED SVM-BASE..
PASS	.	Display and Filter	String	FILTER PASS - THE VARIAN.
INFO_DBSNP	dbSNP_138	Display only	String	INFO "DBSNP VERSION WH..
INFO_EA_AC	1914,6686	Display only	String	INFO "EUROPEAN AMERIC..
INFO_AA_AC	0,4180	Display only	String	INFO "AFRICAN AMERICAN.
INFO_TAC	1,9505	Display only	String	INFO "TOTAL ALLELE COUN.
INFO_MAF_EA	0.0	Display only	Decimal	INFO EA, RETRIVED FROM .
INFO_MAF_AA	0.1445	Display only	Decimal	INFO AA, RETRIVED FROM .
INFO_MAF_ALL	0.0077	Display only	Decimal	INFO ALL, RETRIVED FROM.
INFO_GTS	AA,AG,GG	Display only	String	INFO "OBSERVED GENOTYP.
INFO_EA_GTC	0,74,4222	Display only	String	INFO "EUROPEAN AMERIC..
INFO_AA_GTC	0,7,2196	Display only	String	INFO "AFRICAN AMERICAN.
INFO_GTC	4,5,2978	Display only	String	INFO "TOTAL GENOTYPE C..
INFO_DP	59	Display only	Integer	INFO "AVERAGE SAMPLE R..
INFO_AA	C	Display only	String	INFO "CHIMPALLELE"
INFO_FG	intergenic	Display only	String	INFO "FUNCTIONGVS"
INFO_HGVS_CDNA_VAR	NM_020185.3:c.22-4...	Display only	String	INFO "HGVS CODING DNA V..

Buttons: Set as primary keys, String, Integer, Decimal, Next >>, Cancel

Querying Databases

- Following import, each database can be queried for Mutation Surveyor projects.
- To automatically query a database for projects, right click the database and set “Default Query” value to “Yes”



SoftGenetics Reference & Track Manager

Storage Path: C:\Users\Public\Documents\SoftGenetics\NextGENE\References

Genome Build: Human_GRCh37

Preloaded References

Reference	Genome Build	Annotation DB	Comments
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10	

Tracks

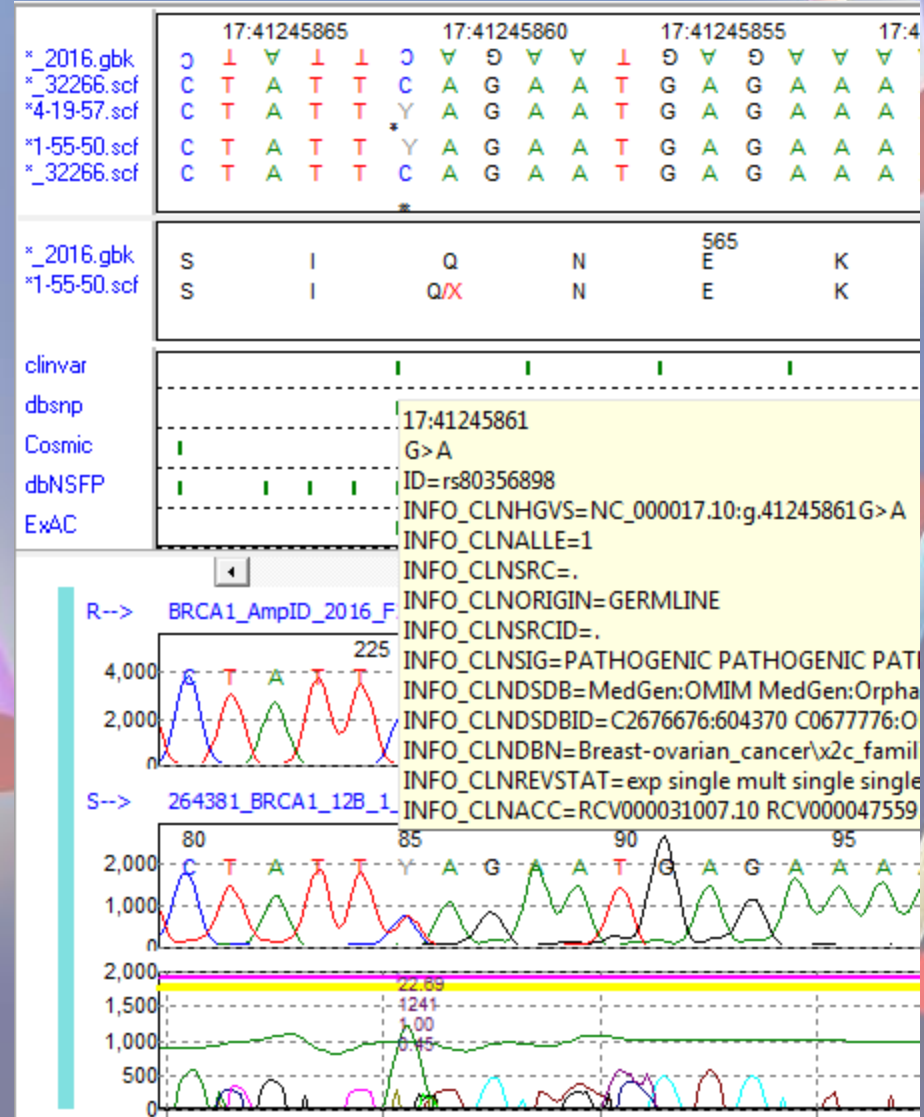
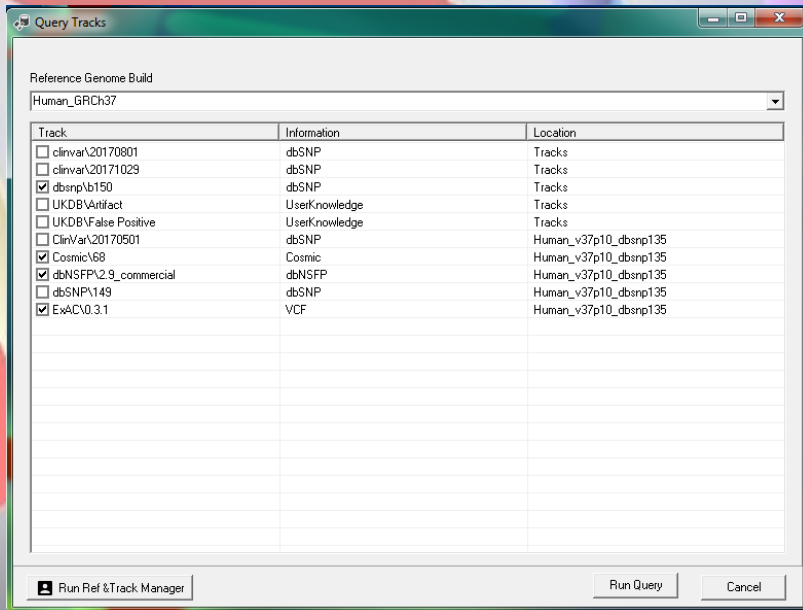
Track	Type	Genome Build	Default Query	Last Modified	Location
ClinVar_142	dbSNP	Human_GRCh37	No	Thursday, Decemb...	Human_v37p10_dbsnp135
ClinVar_20160119	dbSNP	Human_GRCh37	No	2/10/16 07:18:58	Human_v37p10_dbsnp135
ClinVar_144	dbSNP	Human_GRCh37	No	12/16/14 14:22:20	Human_v37p10_dbsnp135
Cosmic_v67	Cosmic	Human_GRCh37	Yes		Yes
custom\clinvar_00-latest	custom	Human_GRCh37	No		No
dbNSFP 2-5_commercial	dbNSFP	Human_GRCh37	Yes		Human_v37p10_dbsnp135
ESP 6500-V2-137	EspVCF	Human_GRCh37	No		Human_v37p10_dbsnp135
ESP6500 137	custom	Human_GRCh37	Yes		Human_v37p10_dbsnp135

Context Menu for ClinVar_144:

- Set Default Query
- Set Genome Build
- Delete Track
- Show Detail
- Select All (Ctrl+A)

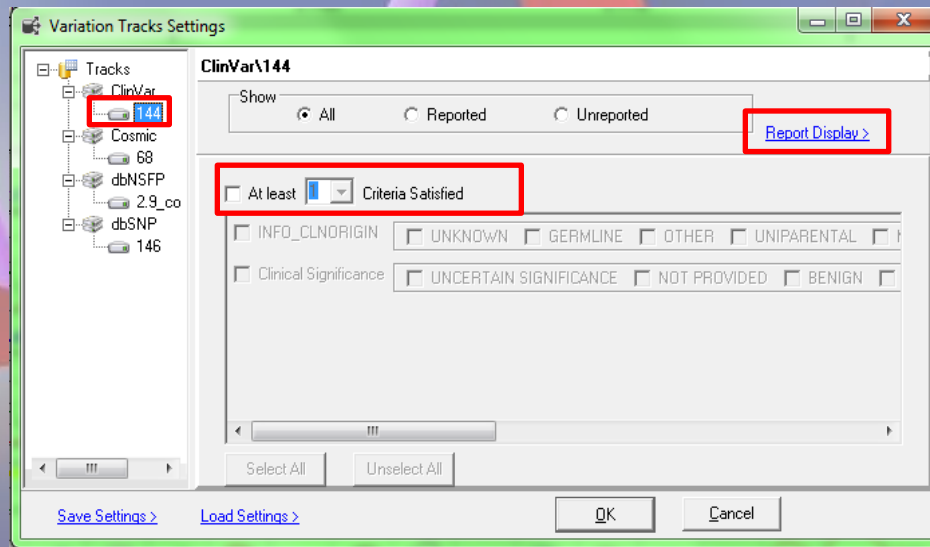
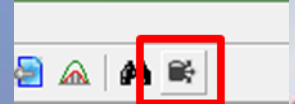
Querying Databases

- Select Process / Query Reference Tracks to customize the displayed tracks within each project



Adjust Track Settings

- Select the Tracks Settings icon on the GAD toolbar
- Click on the track name of interest
- Set filters based on track criteria
 - Check box next to “At least” and select the minimum number of criteria a variant must meet to pass filter
 - Specify required criteria
- Set filters based on track inclusion
 - Variants must first pass all other filters
 - Select to Show All variants, only variants Reported in track, or only variants Unreported in track
- Open Report Display dialog for annotation display options



Report Display Settings

- Select the information to be displayed for variant annotation
- After all selections have been made, click “OK” to return to the Variation Tracks Settings dialog

