# **SUIVE VOIT** 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

reloaded References			3			Build Reference	Import Refer
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Human_v37p10_dbsnp135	Human_GRCh	37	sg_grch37p10				
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### **Importing** Reference Files

### • Click on the Import Reference link

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### **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

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### **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.

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#### Click "Import dbNSFP"

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

Genome Build: 📶

Preloaded References			Build Reference	<u>e Import Reference</u>
Reference	Genome Build	Annotation DB	Comments	
Human_v37p10_dbsnp135	Human_GRCh37	sg_grch37p10		
Human_GRCh38_106_dbSNP141	Human_GRCh38	sg_grch38r106		
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Import Track > Tracks dbSNP/Clinvar Track Genome Build Default Query Last Modified Type Location COSMIC dinvar\20170801 dbSNP Human GRCh37 08/22/17 16:54:57 Tracks Yes dinvar\20171029 dbSNP Human GRCh37 Yes 2017-11-08 14:32:02 Tracks dbNSFP dbsnp\b150 dbSNP Human\_GRCh37 05/09/17 13:37:16 Tracks Yes 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 Custom Variant Track ClinVar\20170501 Human\_GRCh37 dbSNP No 05/08/17 08:20:23 Human\_v37p10\_d Cosmic\68 Human\_GRCh37 Tuesday, January ... Human\_v37p10\_d Gene Annoation Track Cosmic Yes 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 Human GRCh38 106 dbSNP141 ClinVar\142 dbSNP Human GRCh38 No Monday, February ... dbNSFP\2.5 Human GRCh38 106 dbSNP141 dbNSFP Human GRCh38 No Thursday, Februar... dbSNP\142 dbSNP Human\_GRCh38 No Monday, February ... Human\_GRCh38\_106\_dbSNP141

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

Import dbNSFP		
	Open dbNSFP website	
		Add
		Remove
		Remove All
Group dbNSFP		
Name		
	About	Cancel

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

Import dbNSEP	×	1	Jpopgen	Search this site
Open dbNSFP website Group dbNSFP Name About OK	Add Remove Remove All Cancel		Navigation Jpopen - a collection of java programs for population genetic cnaspsis Sitemap About the maintained by: Xiaoming Liu, Ph.D. Assistant Professor, Human Genetics Center, School of Public Health, Science Center at Houston. Contact: xmliu.uth(at]gmail.com	dbNSFP         NTRODUCTION:         dbNSFP is a database developed for functional prediction and annotation of all potential non-synonymous single-nucleotide variants (nSNVs) 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 nSNVs and 2,270,742 essential splice site SNVs. It completes prediction scores from six prediction algorithms (SIFT, Polyphen2, LRT, MutationTaster, MutationAssessor and FATHMM), three conservation scores (PhyloP, GERP++ and SiPhyl) and other related information including allele frequencies observed in the 1000 Genomes Project phase 1 data and the IN-ILE Excore Sequencing Project, various gene 10s from different databases, functional descriptions of genes, gene expression and gene interaction information, etc.         Some dbNSPP contents (may not be up-to-date though) can alise be accessed through yariant tods, ANNOVAR, KGGSeg, UCSC Genome Browser's Variant Annotation Integrator, Ensembl Variant Effect Predictor and HGMD. Please cite our papers (see below) if you used dbNSPP contents toron-acdemic 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 (xmiu utifat)gmail.com).         CITATION:         1. Lux, Jian X, and Boerwinkle E. 2011. dbNSFP: a lightweight database of human non-synonymous SNPs and their functional predictions. Human Matation: 34 E2393-E2402.         H you used dbNSFP v1.x, please cite our papers 1 & 2.

- 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.

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	About OK	Cancel

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

dbNSFP Install Type	
<ul> <li>Install all databases available for research use.</li> </ul>	
O Install databases limited for commercial use.	
*Please see the dbNSFP readme file for more details.	
OK	

## Importing COSMIC

### Click "Import COSMIC"

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Human_v37p10_dbsnp135	Human_GRCh	37	sg_grch37p10				
Human_GRCh38_106_dbSNP141	Human_GRCh	38	sg_grch38r106				
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## Importing COSMIC

Oracle database dum

- Expand the VCF files list on the <u>Data Download</u> page.
- You can <u>download</u> COSMIC information for coding variants and/or non coding variants



### 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	Add
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### Click "Import dbSNP/ClinVar"

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 Download files using these links or by clicking the "Open FTP Folder to Download VCF" button

#### GRCh37:

dbSNP: <u>ftp://ftp.ncbi.nlm.nih.gov/snp/organisms/human\_9606\_b151\_GRCh37p13/VCF/00-All.vcf.gz</u> ClinVar: <u>ftp://ftp.ncbi.nlm.nih.gov/pub/clinvar/vcf\_GRCh37/clinvar.vcf.gz</u>

#### **GRCh38:**

dbSNP: <u>ftp://ftp.ncbi.nlm.nih.gov/snp/organisms/human\_9606/VCF/00-All.vcf.gz</u> ClinVar: <u>ftp.ncbi.nih.gov/pub/clinvar/vcf\_GRCh38/clinvar.vcf.gz</u>

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	All_20150605_papu.vcf.gz.tbi	33.4 kB	6/8/15, 12:00:00 AM
	GRCh38_VCF_files	0 B	10/13/15, 6:39:00 PM
	README.txt	101 B	10/9/15, 5:42:00 PM
	archive/		6/2/15, 12:00:00 AM
	clinical_vcf_set	0 B	6/2/15, 12:00:00 AM
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	common_all_20150605.vcf.gz.tbi	1.9 MB	6/8/15, 12:00:00 AM
	common_all_20150605_papu.vcf.gz	2.3 kB	6/8/15, 12:00:00 AM
	common_all_20150605_papu.vcf.gz.md5	66 B	6/8/15, 12:00:00 AM
	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 <u>Human Variation Sets in VCF</u> Format

- 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"

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

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

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Human_v37p13	Human_GRCh	37	sg_grch37p10			
Human_v37_3_dbsnp135_dna	Human_GRCh	37	sg_v37_3_dbsnp135_human_o	na		
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#### Click "Import Track" > "Custom Variant Track"

eloaded References						Build Reference	Import Reference
Reference	Genome Build		Annotation DB		Comments		
Human_v37p10_dbsnp135	Human_GRCh3	37	sg_grch37p10				
Human_v37p10_MajorChr_dbsnp135	Human_GRCh37 sg Human_GRCh37 sg		sg_grch37p10				
Human_v37p13			sg_grch37p10				
Human_v37_3_dbsnp135_dna	Human_GRCh	Human_GRCh37 sg		5_human_dna			
C.elegans_ws170_dna	reference::C.elegans_ws170_dna sg		sg_ws170_c_elegans	s_dna			
_elegans_WS226	reference::c_e	elegans_WS226	N/A				
acks							Import Track >
acks Track	Туре	Genome Build	De	efault Query L	ast Modified	Location	Import Track > dbSNP/Clinvar
acks Track dbscsnv\1.0	Type dbscSNV	Genome Build Human_GRCh3	D4	efault Query L	ast Modified 16/08/16 08:30:19	Location Tracks	Import Track > dbSNP/Clinvar COSMIC
acks Track Jbscsnv\1.0 Jbsnp\dinvar_20160502.vcf	Type dbscSNV dbSNP	Genome Build Human_GRCh3 Human_GRCh3	2 Dr 7 Nr 7 Ye	efault Query L o C	ast Modified 16/08/16 08:30:19 15/23/16 15:11:24	Location Tracks Tracks	Import Track > dbSNP/Clinvar COSMIC dbNSEP
acks Track dbscsnv\1.0 dbsny\dinvar_20160502.vcf custom\dbSNP_b146	Type dbscSNV dbSNP VCF	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3	7 Ni 7 Ni 7 Ni	efault Query L o C es C o C	ast Modified /6/08/16 08:30:19 /5/23/16 15:11:24 /2/09/16 11:30:54	Location Tracks Tracks Human_v37p10_N	Import Track > dbSNP/Clinvar COSMIC dbNSFP
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acks Track dbscsnv\1.0 dbsnp\dinvar_20160502.vcf custom\dbSNP_b146 dbNSFP\2.9	Type dbscSNV dbSNP VCF dbNSFP	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3	Di 7 Ni 7 Ye 7 Ni 7 Ye	efault Query L o ( es ( o ( es (	ast Modified 16/08/16 08:30:19 15/23/16 15:11:24 12/09/16 11:30:54 1/29/16 16:45:25	Location Tracks Tracks Human_v37p10_N Human_v1 10_1	Import Track > dbSNP/Clinvar COSMIC dbNSFP dbscSNV
acks Track dbscsnv\1.0 dbsnv\dinvar_20160502.vcf custom\dbSNP_b146 dbNSFP\2.9	Type dbscSNV dbSNP VCF dbNSFP	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3	Di 7 Ni 7 Ye 7 Ni 7 Ye	efault Query L o ( es ( es ( es (	ast Modified 16/08/16 08:30:19 15/23/16 15:11:24 12/09/16 11:30:54 11/29/16 16:45:25	Location Tracks Tracks Human_v37p10_N Human_v2_10_N	Import Track > dbSNP/Clinvar COSMIC dbNSFP dbscSNV Custom Variant Tra
acks Track dbscsnv\1.0 dbsnp\clinvar_20160502.vcf custom\dbSNP_b146 dbNSFP\2.9	Type dbscSNV dbSNP VCF dbNSFP	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3	D4 7 N4 7 Y2 7 N4 7 Y6	efault Query L o C es C o C es C	ast Modified 6/08/16 08:30:19 5/23/16 15:11:24 12/09/16 11:30:54 11/29/16 16:45:25	Location Tracks Tracks Human_v37p10_V Human_v1 10_1	Import Track > dbSNP/Clinvar COSMIC dbNSFP dbscSNV Custom Variant Tra Gene Annoation In
acks Track Ibscsnv\1.0 Ibscsnv\dnvar_20160502.vcf custom\dbSNP_b146 IbNSFP\2.9	Type dbscSNV dbSNP VCF dbNSFP	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3	Da 7 Na 7 Ye 7 Na 7 Ye	efault Query L o ( es ( es ( es (	ast Modified 16/08/16 08:30:19 15/23/16 15:11:24 2/09/16 11:30:54 11/29/16 16:45:25	Location Tracks Tracks Human_v37p10_N Human_v1 10_N	Import Track > dbSNP/Clinvar COSMIC dbNSFP dbscSNV Custom Variant Tra Gene Annoation Tr
acks Track Ibscsnv\1.0 Ibsny\clinvar_20160502.vcf custom\dbSNP_b146 IbNSFP\2.9	Type dbscSNV dbSNP VCF dbNSFP	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3	Da 7 Ne 7 Ye 7 No 7 Ye	efault Query L o (25) o (25) es (27) es (27)	ast Modified 16/08/16 08:30:19 15/23/16 15:11:24 12/09/16 11:30:54 1/29/16 16:45:25	Location Tracks Tracks Human_v37p10_N Human_v3_10_1	Import Track > dbSNP/Clinvar COSMIC dbNSFP dbscSNV Custom Variant Tra Gene Annoation Tr

Click "Add" to browse to and select the downloaded database file

Figure 1 - Custom Variant Track	
	Add
	Remove
	Remove All
Group custom	
Name	
Genome Build 🗸	
	OK Cancel

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

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".

😑 Import track - Custom Variant Track	×
	Add
	Remove
	Remove All
Group custom	
Name	
Genome Build	
	OK Cancel

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".

😝 Column Properties Settin	g				
					1
Track Titles	Preview	Status	Numeric	Description	
CHROM	11	Chr	String	AN IDENTIFIER FROM THE	Set as primary keys
POS	93500	Pos	String	THE REFERENCE POSITION	
ID	rs377032113	Display and Filter	String	ID	
REF	С	Reference Allele	String	REFERENCE BASE(S)	
ALT	т	Mutation Allele	String	NON-REFERENCE ALLELES	String
QUAL		Display and Filter	Interger	PHRED-SCALED QUALITY	
INDEL5		Display and Filter	String	FILTER "NEARBY 1000 GEN =	
SVM		Display and Filter	String	FILTER "FAILED SVM-BASE	Integer
PASS		Display and Filter	String	FILTER PASS - THE VARIAN.	
INFO_DBSNP	dbSNP_138	Display only	String	INFO "DBSNP VERSION WH	Decimal
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	Interger	INFO "AVERAGE SAMPLE R	
INFO_AA	С	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. 👻	
•				•	

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

😝 Column Properties Settin	g				
Track Titles	Preview	Status	Numeric	Description	Cat an arimony laws
CHROM	11	Chr	String	AN IDENTIFIER FROM THE	Set as primary keys
POS	93500	Pos	String	THE REFERENCE POSITION	
ID	rs377032113	Display and Filter	String	ID	
REF	С	Reference Allele	String	REFERENCE BASE(S)	
ALT	т	Mutation Allele	String	NON-REFERENCE ALLELES	String
QUAL		Display and Filter	Interger	PHRED-SCALED QUALITY	
INDEL5		Display and Filter	String	FILTER "NEARBY 1000 GEN =	
SVM		Display and Filter	String	FILTER "FAILED SVM-BASE	Integer
PASS		Display and Filter	String	FILTER PASS - THE VARIAN.	
INFO_DBSNP	dbSNP_138	Display only	String	INFO "DBSNP VERSION WH	Decimal
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	Interger	INFO "AVERAGE SAMPLE R	
INFO_AA	С	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. 👻	
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Next>>

## **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"

eloaded References						Build Re	ference	Import Refere
Reference	Genome Build		Annotation DB		Comments			
Human_v37p10_dbsnp135	Human_GRCh	37	sg_grch37p10					
arke								Import Track
adıs								Import Track
acks	Туре	Genome Build		Default Query	Last Modified	Locatio	'n	Import Track
acks Track ClinVar\142	Type dbSNP	Genome Build Human_GRCh3	7	Default Query No	Last Modified Thursday, Decemb	Locatio	n _v37p10_dbs	Import Track
acks Track ClinVar \142 ClinVar \20160 119	Type dbSNP dbSNP	Genome Build Human_GRCh3 Human_GRCh3	7	Default Query No	Last Modified Thursday, Decemb	Locatio	n _v37p10_dbs v37p10_dbs	Import Track mp 135 mp 135
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acks Track ClinVar\142 ClinVar\142 ClonVar\144 CosmicV\67	Type dbSNP dbSNP dbSNP Cosmic	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3	7 7 7 7 7	Default Query No Ye Ye Ye Set	Last Modified Thursday, Decemb 2016/16 14:55:32 Default Query	Locatio Human	n _v37p10_dbs v37p10_dbs v23p10_dbs Yes	Import Track
racks Track ClinVar\142 ClinVar\20160119 ClinVar\20160119 Cosmic\v67 custom\clinvar_00-latest	Type doSNP doSNP doSNP cosmic custom	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3	7 7 7 7 7	Default Query No No Ye Ye Set No Set	Last Modified Thursday, Decemb Data (c) 14:55-39 Default Query Genome Build	Locatio Human	n _v37p10_dbs v37p10_dbs V37p10_dbs Yes No	Import Track
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adks Track ClinVar\142 ClinVar\142 Comic\v67 Custom\clinVar_00-latest dMSFPV-2-Sommercial ESP\6500-V2-137	Type dbSNP dbSNP Cosmic custom dbNSPP EspVCF	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3	7 7 7 7 7 7 7 7	Default Query No Ye Ye No Set Yes No Del	Last Modified Thursday, Decemb Dollar (16 14: E1-32 Default Query Genome Build ete Track	Locatio Human	n _v37p10_dbs v37p10_dbs v37p10_dbs Ys No v37p10_dbs v37p10_dbs	Import Track
acks Track ClinVar\142 ClinVar\20160119 ClinVar\20160119 ClinVar\144 Cosmic\v67 custom\clinvar_00-latest db/SFP[2-5_commercial ESP(5500-127) ESP(5500.137	Type dbSNP dbSNP dbSNP Cosmic custom dbNSFP EspVCF custom	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3	7 7 7 7 7 7 7 7 7 7	Default Query No Ye Ye Yes Del Yes Sho	Last Modified Thursday, Decemb Difference and Decemb Difference and Decemb Default Query Genome Build ete Track ww Detail	Locatio Human	n 	Import Track

### **Querying Databases**

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

uman_GRCh37		
rack	Information	Location
clinvar\20170801	dbSNP	Tracks
clinvar\20171029	dbSNP	Tracks
✓ dbsnp\b150	dbSNP	Tracks
UKDB\Artifact	UserKnowledge	Tracks
UKDB\False Positive	UserKnowledge	Tracks
ClinVar\20170501	dbSNP	Human_v37p10_dbsnp135
Cosmic\68	Cosmic	Human_v37p10_dbsnp135
dbNSFP\2.9_commercial	dbNSFP	Human_v37p10_dbsnp135
dbSNP\149	dbSNP	Human_v37p10_dbsnp135
ExAC\0.3.1	VCF	Human_v37p10_dbsnp135

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### **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

🛃 Variation Tracks Settings		
Tracks	ClinVar\144	
G SNP G G AbSNP G G 146	INFO_CLNORIGIN UNKNOWN GERMLINE OTHER UNIPARENTAL I Clinical Significance UNCERTAIN SIGNIFICANCE NOT PROVIDED BENIGN	
Save Settings >		

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• 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

🗖 Display track name	
DBSNP VARIANT INFO 'VARIANT NAMES FROM HGVS. T INFO 'VARIANT ALLELES FROM REF OR INFO 'VARIANT CLINICAL CHANELS'' INFO 'VARIANT CLINICAL CHANNEL IDS INFO 'VARIANT CLINICAL SIGNIFICANCE INFO 'VARIANT DISEASE DATABASE IO INFO 'VARIANT DISEASE DATABASE INFO 'VARIANT DISEASE NAME'' INFO 'VARIANT DISEASE NAME'' INFO 'VARIANT DISEASE NAME'' INFO 'VARIANT DISEASE NAME'' INFO 'VARIANT ACCESSION AND VERSI	THE ORDER OF THESE VARIANTS ALT COLUMNS. 0 IS REF, 1 IS TH IF THE FOLLOWING VALUES MAY  , 0 - UNCERTAIN SIGNIFICANCE,  IPROVIDED, NO_CRITERIA - NO 4 ONS''
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	Display track name DBSNP VARIANT INFO 'VARIANT NAMES FROM HGVS INFO 'VARIANT ALLELES FROM REF OR INFO 'VARIANT CLINICAL CHANELS' INFO 'VARIANT CLINICAL CHANNEL IDS INFO 'VARIANT CLINICAL SIGNIFICANCE INFO 'VARIANT DISEASE DATABASE ID' INFO 'VARIANT DISEASE NAME'' INFO 'VARIANT DISEASE NAME'' INFO 'VARIANT DISEASE NAME'' INFO 'VARIANT DISEASE NAME'' INFO 'VARIANT ACCESSION AND VERSI

