

### NextGENe Reference and Track Manager

Importing information from dbSNP or ClinVar



#### Track Manager Tool

NextGENe				
File Process	Tools Help			
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	Barcode Sorting		Total and the	
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5		actgactga	Software PowerTools for Genetics A	nalysis
US Patent No.	8,271,206			A DESCRIPTION OF THE OWNER OF THE



#### **Reference & Track Manager Tool**

Shows all imported preloaded references and tracks

enome Build: All		•				
reloaded References					Build Reference	Import Referen
Reference	Genome Build		Annotation DB	Comments		
Human_v37p10_dbsnp135	Human_GRCh	37	sg_grch37p10			
Human_v37p10_MajorChr_dbsnp135	Human_GRCh	37	sg_grch37p10			
Human_v37p13	Human_GRCh	37	sg_grch37p10			
Human_v37_3_dbsnp135_dna	Human_GRCh	37	sg_v37_3_dbsnp135_human_dna	a		
C.elegans_ws170_dna	reference::C.	elegans_ws170_dna	sg_ws170_c_elegans_dna			
c_elegans_WS226	reference::c_	elegans_WS226	N/A			
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racks 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	Default Query 7 No 77 Yes 77 No 77 Yes	Last Modified 06/08/16 08:30:19 05/23/16 15:11:24 02/09/16 11:30:54 01/29/16 16:45:25	Location Tracks Tracks Human_v37p10_1 Human_v37p10_1	Import Track > MajorChr_dbsnp MajorChr_dbsnp



#### Importing ClinVar/dbSNP

#### Click "Import Track" > "dbSNP/ClinVar"

nome Build: All		•					
eloaded References						Build Reference	Import Reference
Reference	Genome Build		Annotation DB		Comments		
Human_v37p10_dbsnp135	Human_GRCh3	37	sg_grch37p10				
luman_v37p10_MajorChr_dbsnp135	Human_GRCh3	37	sg_grch37p10				
Human_v37p13	Human_GRCh3	37	sg_grch37p10				
Human_v37_3_dbsnp135_dna	Human_GRCh3	37	sg_v37_3_dbsnp1	135_human_dna			
C.elegans_ws170_dna	reference::C.e	legans_ws170_dna	sg_ws170_c_elega	ans_dna			
c_elegans_WS226	reference::c_e	legans_WS226	N/A				
racks	Type	Genome Build		Default Query	Tas		Import Track > dbSNP/Clinvar
racks Track disconsult 0	Туре	Genome Build	7	Default Query	Las		Import Track > dbSNP/Clinvar
racks Track dbscsnv\1.0 dbscnvlinvar_20160502.vcf	Type dbscSNV dbSNP	Genome Build Human_GRCh3 Human_GRCh3	7	Default Query No Yes	Las 06/00/10 00:30:15	Tracks	Import Track > dbSNP/Clinvar
racks Track dbscsnv\1.0 dbsnp\dinvar_20160502.vcf custom\dbSNP b146	Type dbscSNV dbSNP VCF	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3	7 7 7	Default Query No Yes No	Las 06/00/10 00:30:15 05/23/16 15:11:24 02/09/16 11:30:54	Tracks Human v37010	Import Track > dbSNP/Clinvar contuc dbNSFP
racks Track dbscsnv\1.0 dbsnp\clinvar_20160502.vcf custom\dbSNP_b146 dbNSFP12.9	Type dbscSNV dbSNP VCF dbNSFP	Genome Build Human_GRCh3 Human_GRCh3 Human_GRCh3 Human_GRCh3	7 7 7 7	Default Query No Yes No Yes	Las 06/00/10 00:30:13 05/23/16 15:11:24 02/09/16 11:30:54 01/29/16 16:45:25	Tracks Human_v37p10_N Human_v37p10_N	Import Track > dbSNP/Clinvar dbNSFP dbscSNV
racks 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	7 7 7 7 7	Default Query No Yes No Yes	Las 06/00/10 00:30:13 05/23/16 15:11:24 02/09/16 11:30:54 01/29/16 16:45:25	Tracks Human_v37p10_N Human_v37p10_N	Import Track > dbSNP/Clinvar cock trc dbNSFP dbscSNV Custom Variant Track
racks 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	7 7 7 7 7	Default Query No Yes Yes	Las 06/00/10 00:30:15 05/23/16 15:11:24 02/09/16 11:30:54 01/29/16 16:45:25	Tracks Tracks Human_v37p10_N Human_v37p10_N	Import Track > dbSNP/Clinvar cockurc dbNSFP dbscSNV Custom Variant Track Gene Annoation Track



#### Importing ClinVar/dbSNP

Use File Explorer or an FTP Client such as FileZilla:

GRCh37

ClinVar: <u>ftp.ncbi.nih.gov/pub/clinvar/vcf\_GRCh37/clinvar.vcf.gz</u>

dbSNP:

ftp://ftp.ncbi.nlm.nih.gov/snp/.redesign/pre\_build152/organisms/human\_9606\_b151\_GRCh37p13/ VCF/All\_20180423.vcf.gz

#### GRCh38

ClinVar: <u>ftp.ncbi.nih.gov/pub/clinvar/vcf\_GRCh38/clinvar.vcf.gz</u> dbSNP: <u>ftp.ncbi.nih.gov/snp/organisms/human\_9606\_b144\_GRCh38p2/VCF/00-All.vcf.gz</u>



#### Importing ClinVar/dbSNP

- Click "Add" to load the file.
- Select "ClinVar" or "dbSNP" from the Group dropdown menu
- Enter a name, such as the dbSNP or ClinVar version number
- Select the appropriate genome build and click OK.

Import track - dbSNP		×
	Open FTP folder to Download VCF	
C:\Users\Kevin\Downloads\clir	ıvar.vcf.gz	Add
Com lines		Remove All
Name 20240416		
Genome Build Human_GRCh37	~	СК

#### Automatically Query Tracks

Following import, the Default Query should be set to "Yes" to automatically query the track information for all future projects

Right-click on a track and select "Set Default Query" to set to "Yes" or "No"

Sequencing Software for Biologists

reloaded References					Build Reference	Import Referen
Reference	Genome Build		Annotation DB	Comments		
Human_v37p10_dbsnp135	Human_GRCh3	7	sg_grch37p10			
Human_v37p10_MajorChr_dbsnp135	Human_GRCh3	7	sg_grch37p10			
Human_v37p13	Human_GRCh3	7	sg_grch37p10			
Human_v37_3_dbsnp135_dna	Human_GRCh3	7	sg_v37_3_dbsnp135_human_dna			
C.elegans_ws170_dna	reference::C.e	legans_ws170_dna	sg_ws170_c_elegans_dna			
c_elegans_WS226	reference::c_e	legans_WS226	N/A			
racks	Type	Genome Build	Default Query	Last Modified	location	Import Track >
racks Track	Туре	Genome Build	Default Query	Last Modified	Location	Import Track >
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racks Track dbscsnv\1.0 dbsnp\clinvar_20160502.vcf	Type dbscSNV dbSNP VCE	Genome Build Human_GRCh3 Human_GRCh3	Default Query 17 No 17 Yes	Last Modified 06/08/16 08:30:19 05/23/16 15:11:24 02/09/16 11:30:54	Location Tracks Tracks	Import Track >
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Tracks 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	IZ Default Query IZ No IZ Yes IZ No IZ Yes	Last Modified 06/08/16 08:30:19 05/23/16 15:11:24 02/09/16 11:30:54 01/29/16 16:45:25	Location Tracks Tracks Human_v37p10_1 Human_v37p10_1	Import Track > MajorChr_dbsnp MajorChr_dbsnp
Tracks 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	IZ Default Query IZ No IZ Yes IZ No IZ Yes	Last Modified 06/08/16 08:30:19 05/23/16 15:11:24 02/09/16 11:30:54 01/29/16 16:45:25	Location Tracks Tracks Human_v37p10_1 Human_v37p10_1	Import Track > MajorChr_dbsnp MajorChr_dbsnp
Tracks 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	IZ Default Query IZ No IZ Yes IZ No IZ Yes	Last Modified 06/08/16 08:30:19 05/23/16 15:11:24 02/09/16 11:30:54 01/29/16 16:45:25	Location Tracks Tracks Human_v37p10_1 Human_v37p10_1	Import Track > MajorChr_dbsnp MajorChr_dbsnp



#### Query ClinVar/dbSNP

# The database can be queried for existing projects from within the Viewer



## Variation Track Settings

Generation Sequencing Software for Biologists



Main dialog shows imported tracks in Left Pane,

Filtering options in Right Pane, and hyperlink to Report Display options

🖌 🔐 Tracks	clinvar\20220430	
<ul> <li>clinvar</li> <li>20220430</li> <li>dbNSFP</li> <li>v3.5a</li> </ul>	Show All Reported Unreported	Report Displ
♥ ∰ dbsnp b151	At least Criteria Satisfied (Total 3)	
	Pathogenic     Pathogenic     Likely_pathogenic     Drug_response     Risk_factor     Affects     Association     Protective     Uncertain_significance     Conflicting_data_from_submitters     Conflicting_interpretations_of_pathogenicity     Likely_benign     Benign     Other     Not_provided	
	Select All Unselect All	

Save Settings, so they can be used automatically



#### **Configure to Use Track Settings**

- Click Set
- Add Track Settings
- Click OK
- Save Process Settings

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