1000 Genomes Browser Orientation

http://browser.1000genomes.org

Based on Project Phase I Data

European Bioinformatics Institute

11 May, 2011

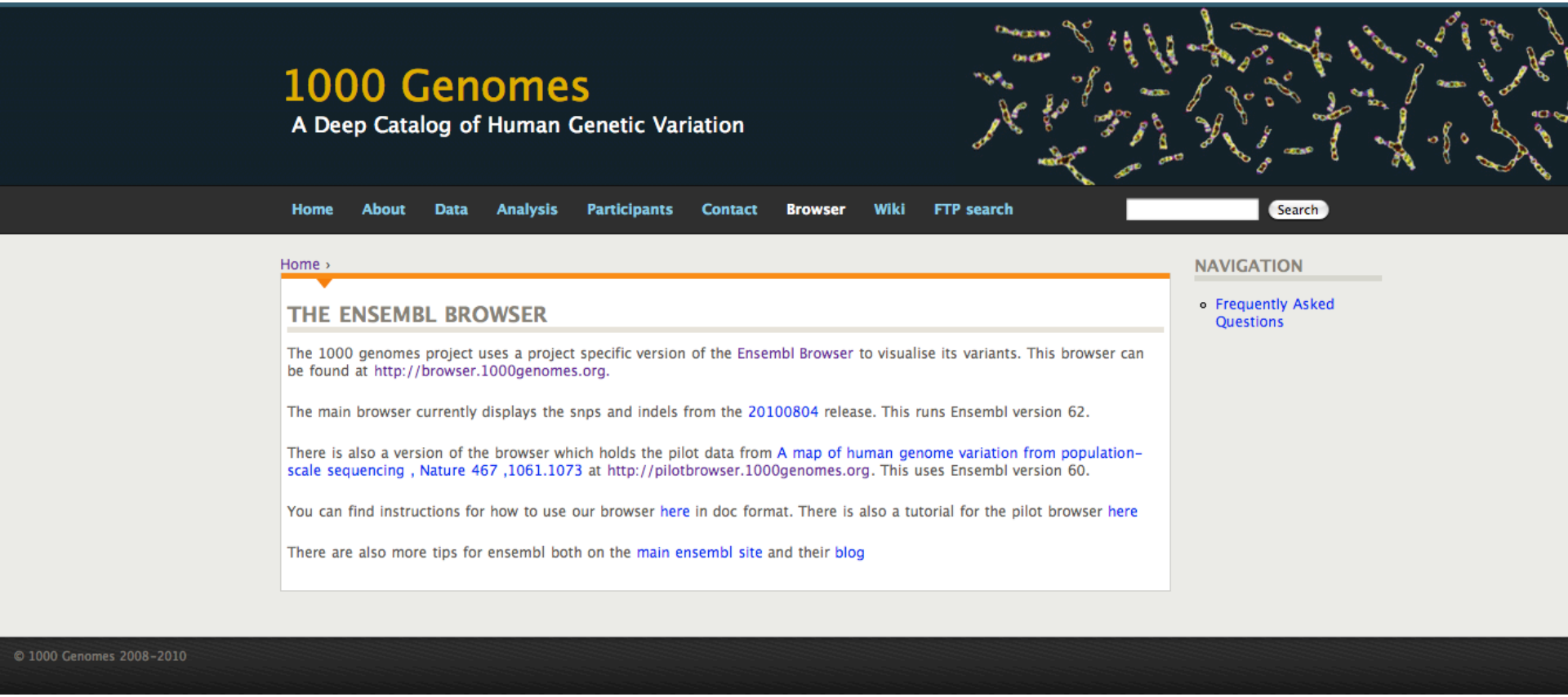
Overview

- Based on version 62 of Ensembl code and NCBI37 version of the human genome assembly
- Contains all of the gene information normally present in Ensembl
  - Gene and transcript annotation, external references, sequence data
- Incorporates essentially all of the 1000 Genomes Phase I data
- 1000 Genomes pilot project data are available on a separate browser –

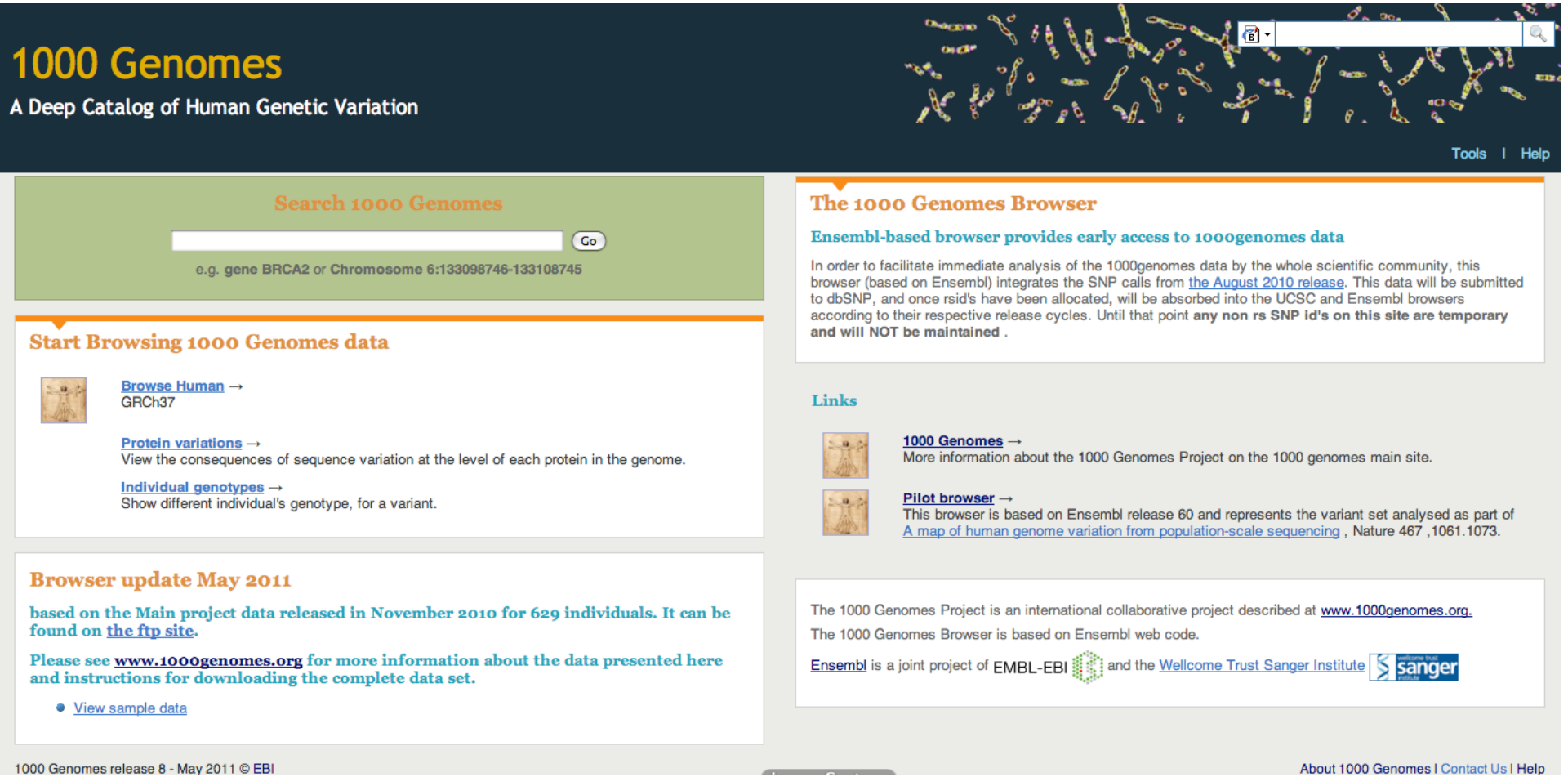
  http://pilotbrowser.1000genomes.org

- Please send questions to info@1000genomes.org

1000 Genomes Browser Guide Page



1000 Genomes Browser Home Page



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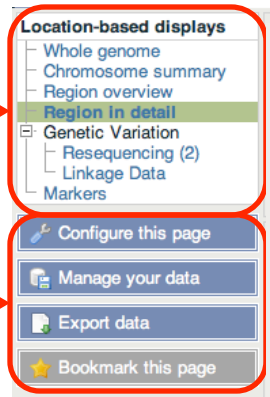
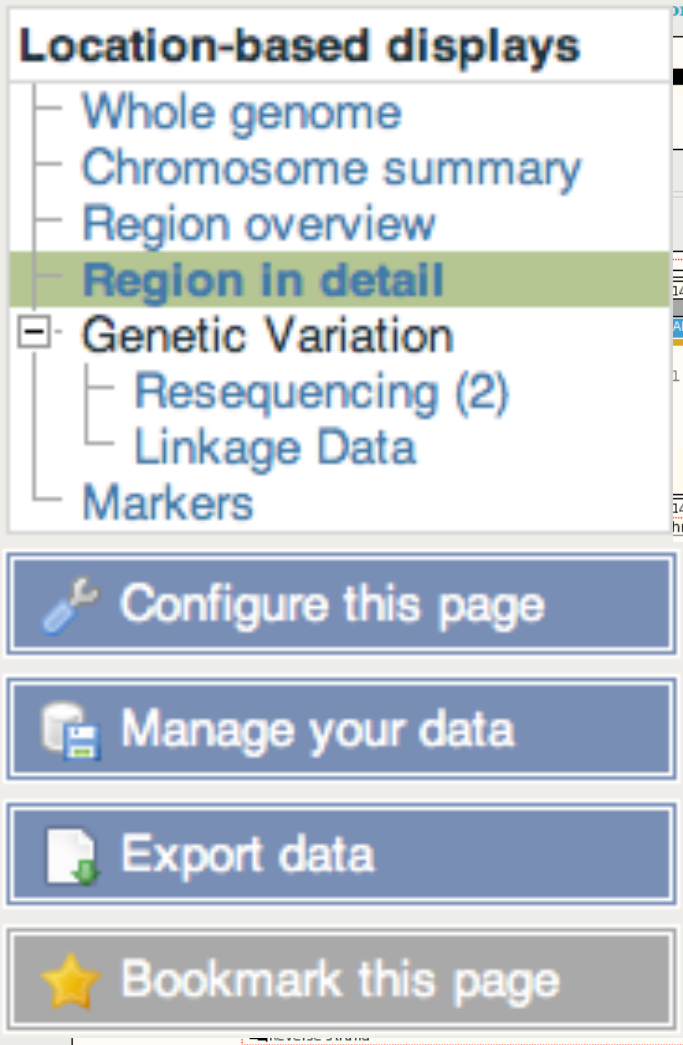
Main location view





- Built on Ensembl
- Navigation is on the left hand side
- Page configuration and data expert links
- Includes only human data in current release

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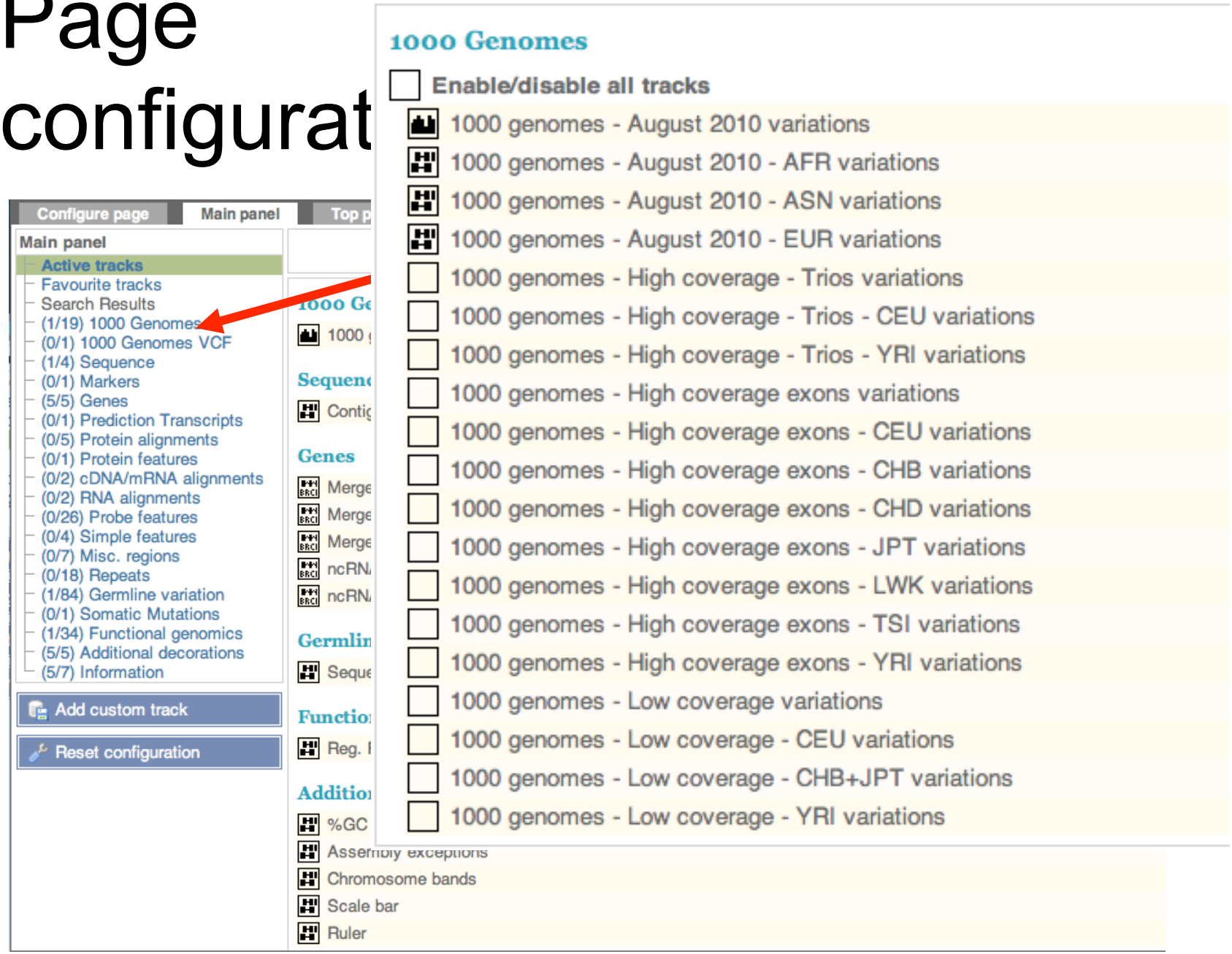
Page configurat



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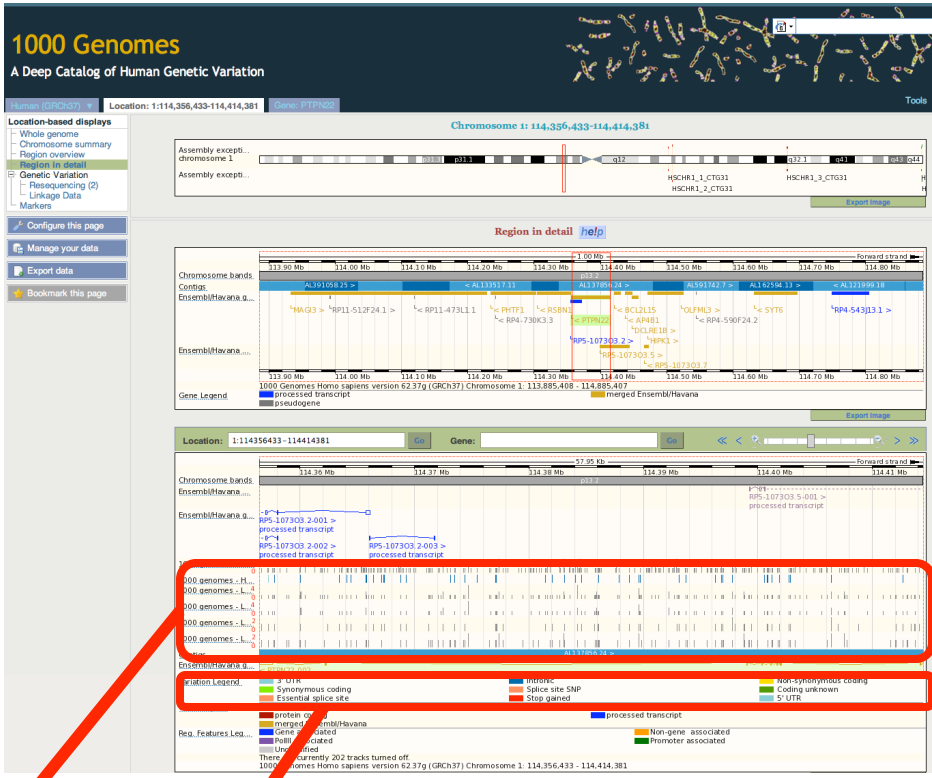
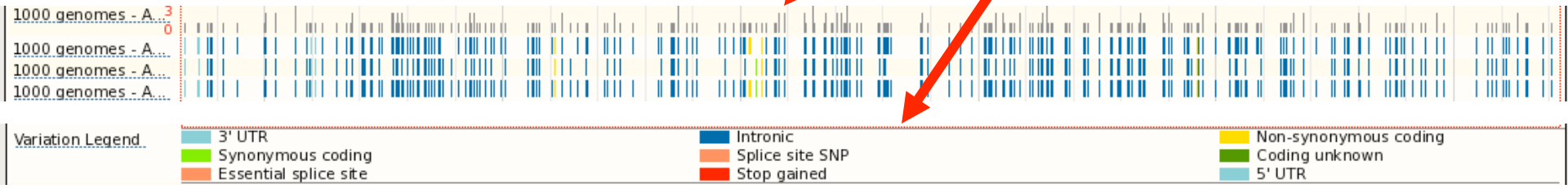
1000 Genomes SNPs

- The variation data of the August 2010 release and pilot project data can be viewed in specific tracks on location pages.
- These are selected from the “Configure this page” menu.
- Track can be displayed as “density” or “compact”.
- In “compact” view, variations are coloured by their functions.
- Tracks for all SNPs from dbSNP build 132 are also available





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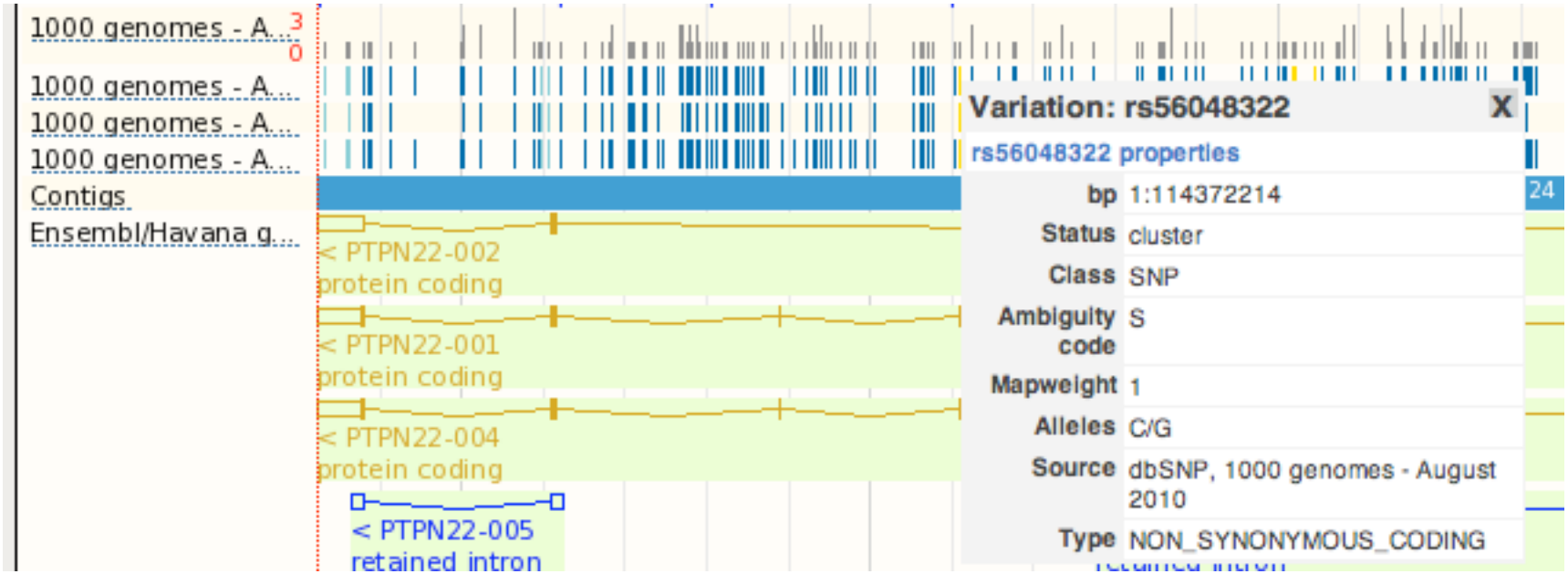
SNP Information

- SNPs are clickable which brings up a small window with basic information
- The “SNP properties” link leads to a dedicated page for the SNP with detailed information (including information imported from dbSNP)



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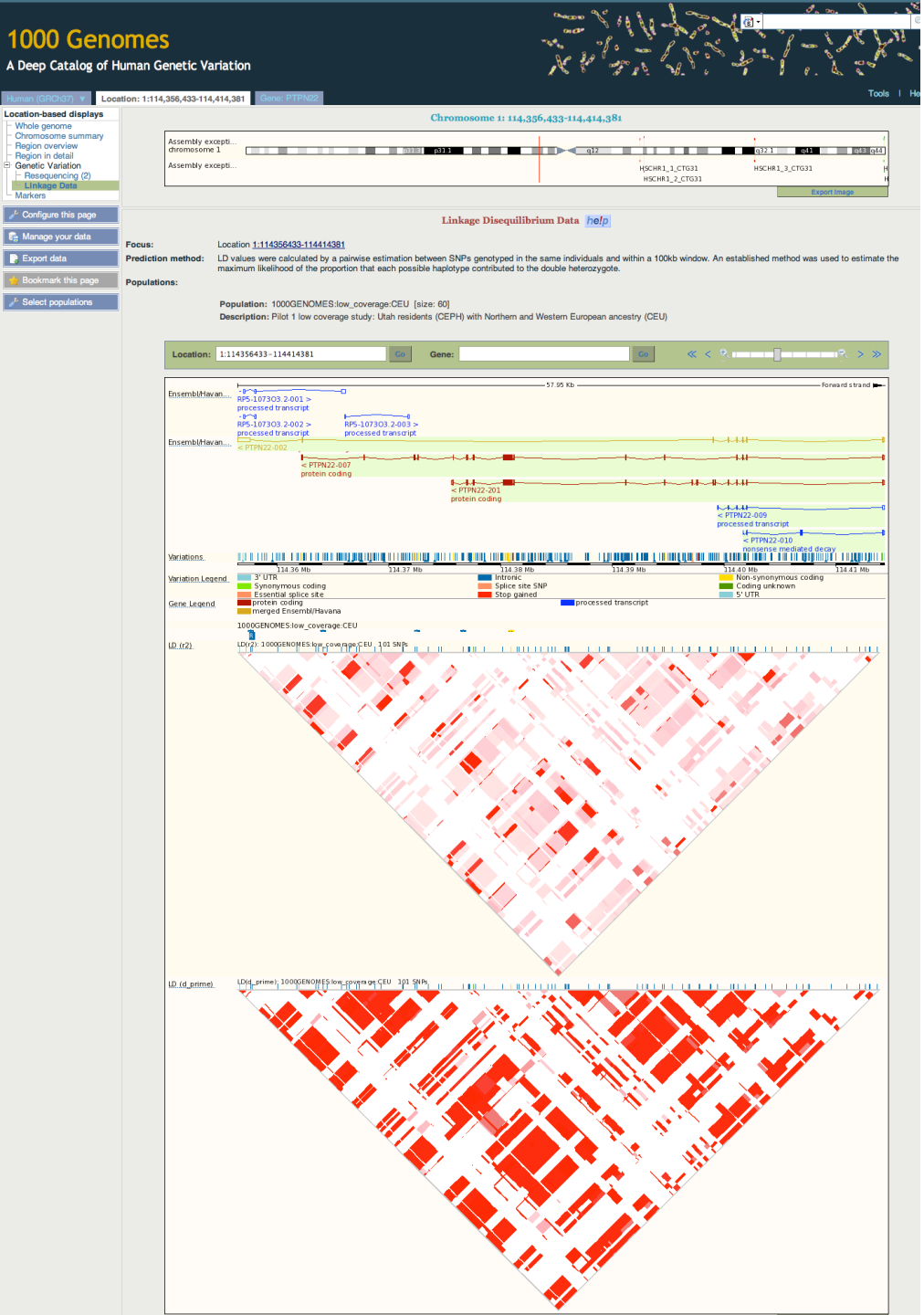
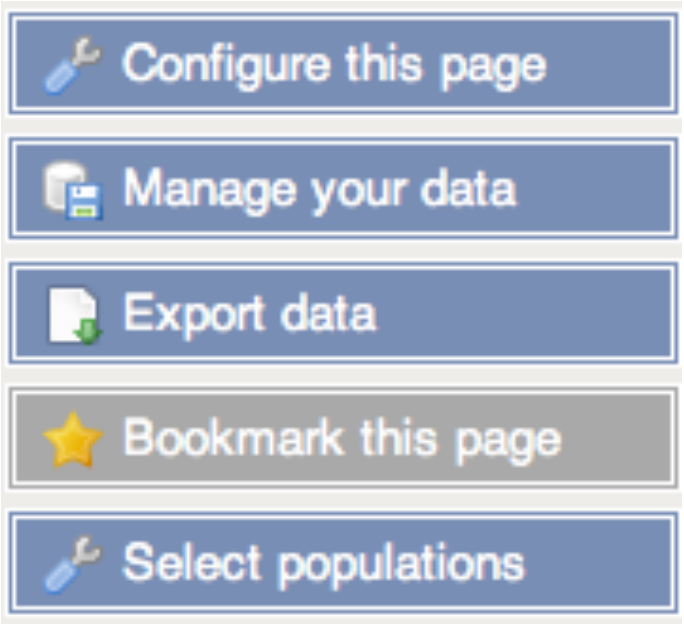


LD Information

- Currently based on data from HapMap and 1000 genomes project populations
- Populations selectable from drop down tab

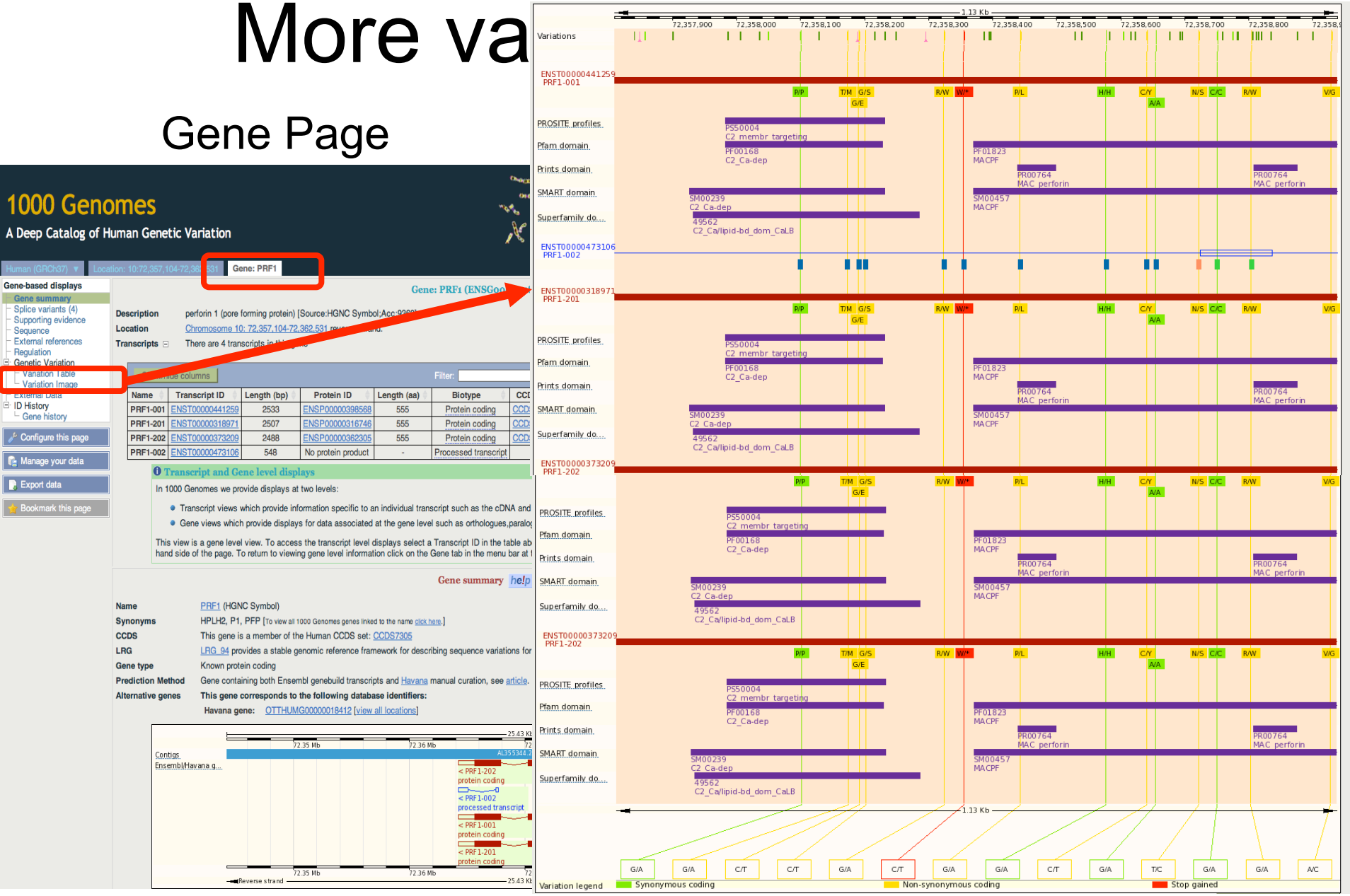
Data Export



- Summary data from the region being viewed can be exported
- Export format (such as html, excel, text) can be specified

More va

Gene Page



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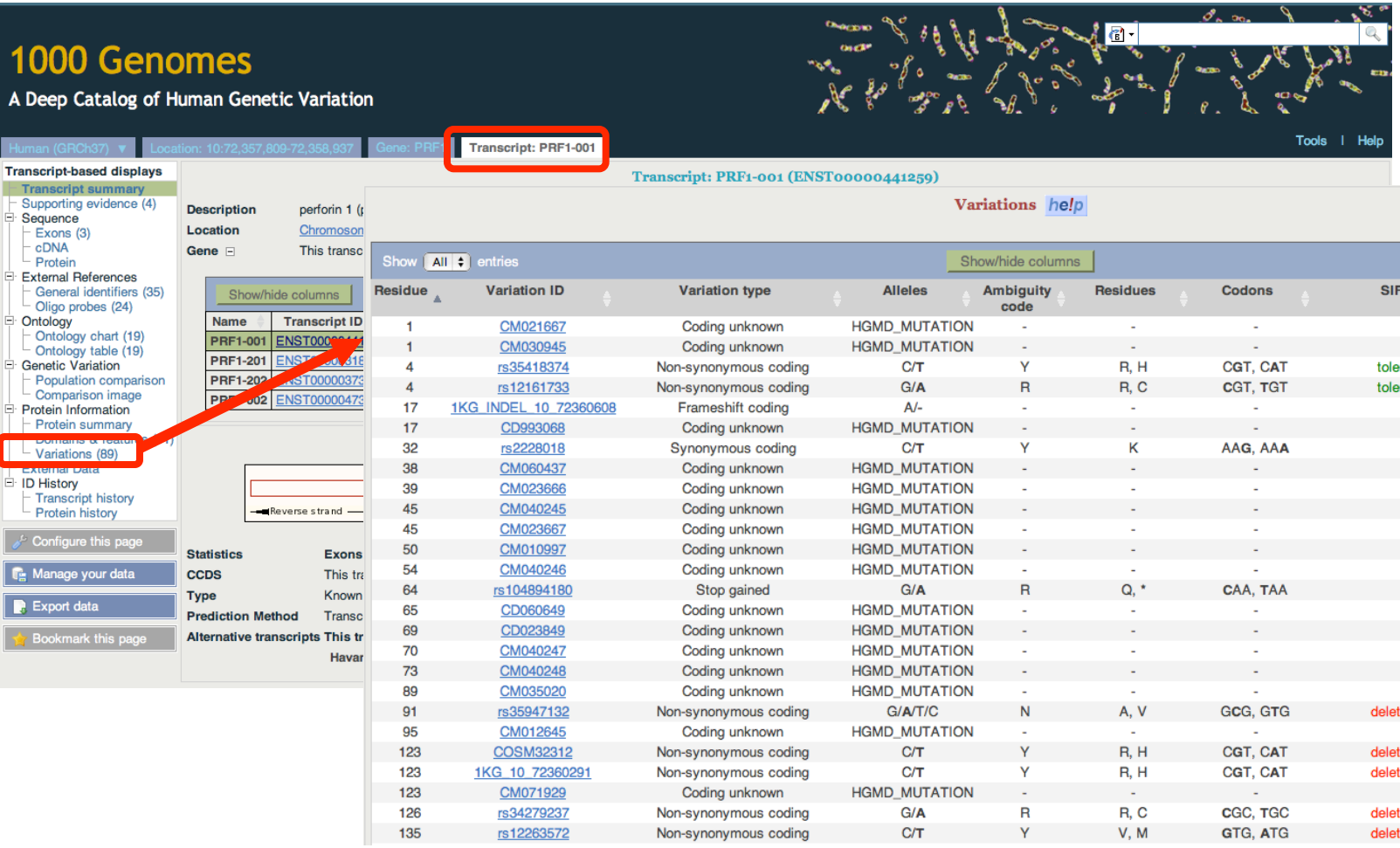
More variation displays

Transcript Page



Residue	Variation ID	Variation type	Alleles	Ambiguity code	Residues	Codons	SIFT	PolyPhen
1	CM021667	Coding unknown	HGMD_MUTATION	-	-	-	-	-
1	CM030945	Coding unknown	HGMD_MUTATION	-	-	-	-	-
4	rs35418374	Non-synonymous coding	C/T	Y	R, H	CGT, CAT	tolerated	probably damaging
4	rs12161733	Non-synonymous coding	G/A	R	R, C	CGT, TGT	tolerated	probably damaging
17	1KG_INDEL_10_72360608	Frameshift coding	A/-	-	-	-	-	-
17	CD993068	Coding unknown	HGMD_MUTATION	-	-	-	-	-
32	rs2228018	Synonymous coding	C/T	Y	K	AAG, AAA	-	-
38	CM060437	Coding unknown	HGMD_MUTATION	-	-	-	-	-
39	CM023666	Coding unknown	HGMD_MUTATION	-	-	-	-	-
45	CM040245	Coding unknown	HGMD_MUTATION	-	-	-	-	-
45	CM023667	Coding unknown	HGMD_MUTATION	-	-	-	-	-
50	CM010997	Coding unknown	HGMD_MUTATION	-	-	-	-	-
54	CM040246	Coding unknown	HGMD_MUTATION	-	-	-	-	-
64	rs104894180	Stop gained	G/A	R	Q, *	CAA, TAA	-	-
65	CD060649	Coding unknown	HGMD_MUTATION	-	-	-	-	-
69	CD023849	Coding unknown	HGMD_MUTATION	-	-	-	-	-
70	CM040247	Coding unknown	HGMD_MUTATION	-	-	-	-	-
73	CM040248	Coding unknown	HGMD_MUTATION	-	-	-	-	-
89	CM035020	Coding unknown	HGMD_MUTATION	-	-	-	-	-
91	rs35947132	Non-synonymous coding	G/A/T/C	N	A, V	GCG, GTG	deleterious	probably damaging
95	CM012645	Coding unknown	HGMD_MUTATION	-	-	-	-	-
123	COSM32312	Non-synonymous coding	C/T	Y	R, H	CGT, CAT	deleterious	benign
123	1KG_10_72360291	Non-synonymous coding	C/T	Y	R, H	CGT, CAT	deleterious	benign
123	CM071929	Coding unknown	HGMD_MUTATION	-	-	-	-	-
126	rs34279237	Non-synonymous coding	G/A	R	R, C	CGC, TGC	deleterious	probably damaging
135	rs12263572	Non-synonymous coding	C/T	Y	V, M	GTG, ATG	deleterious	probably damaging

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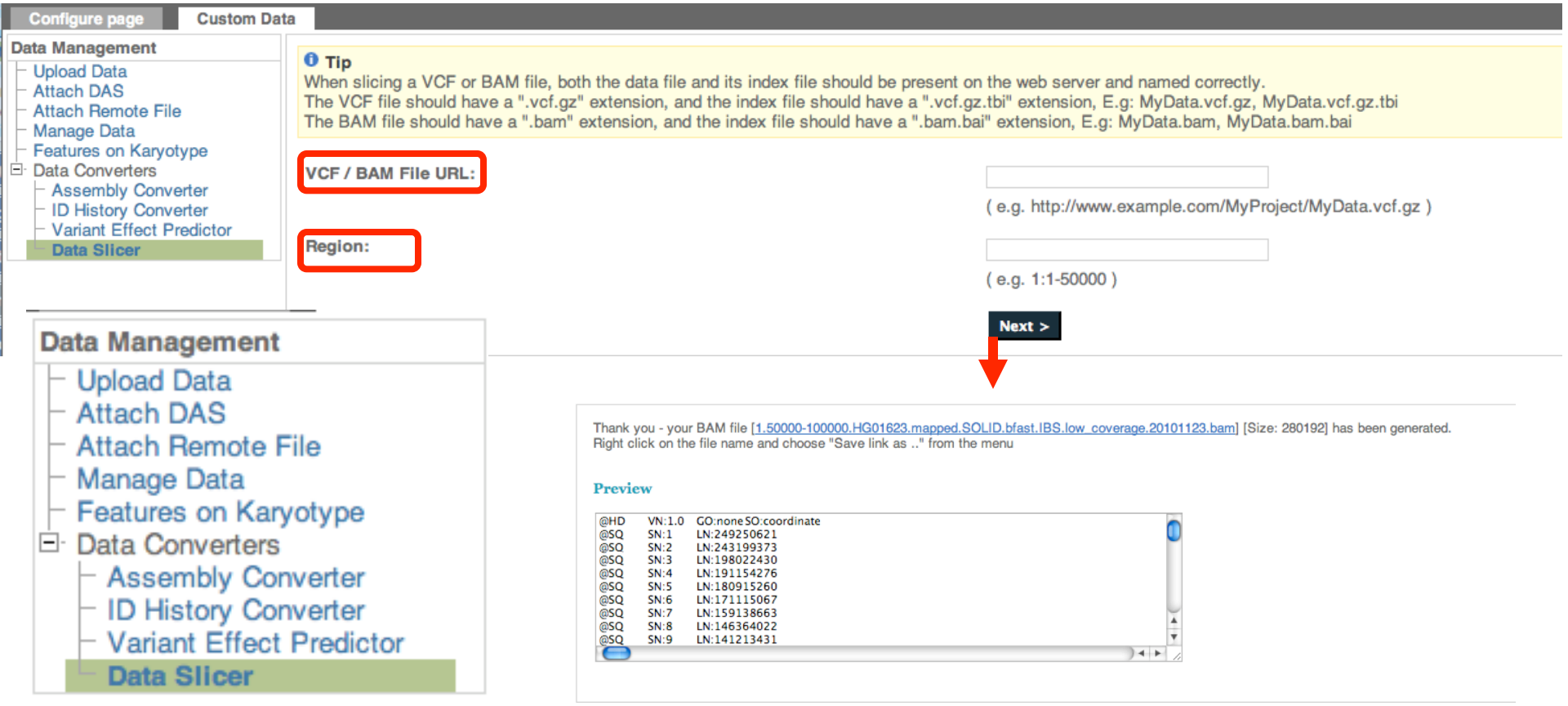
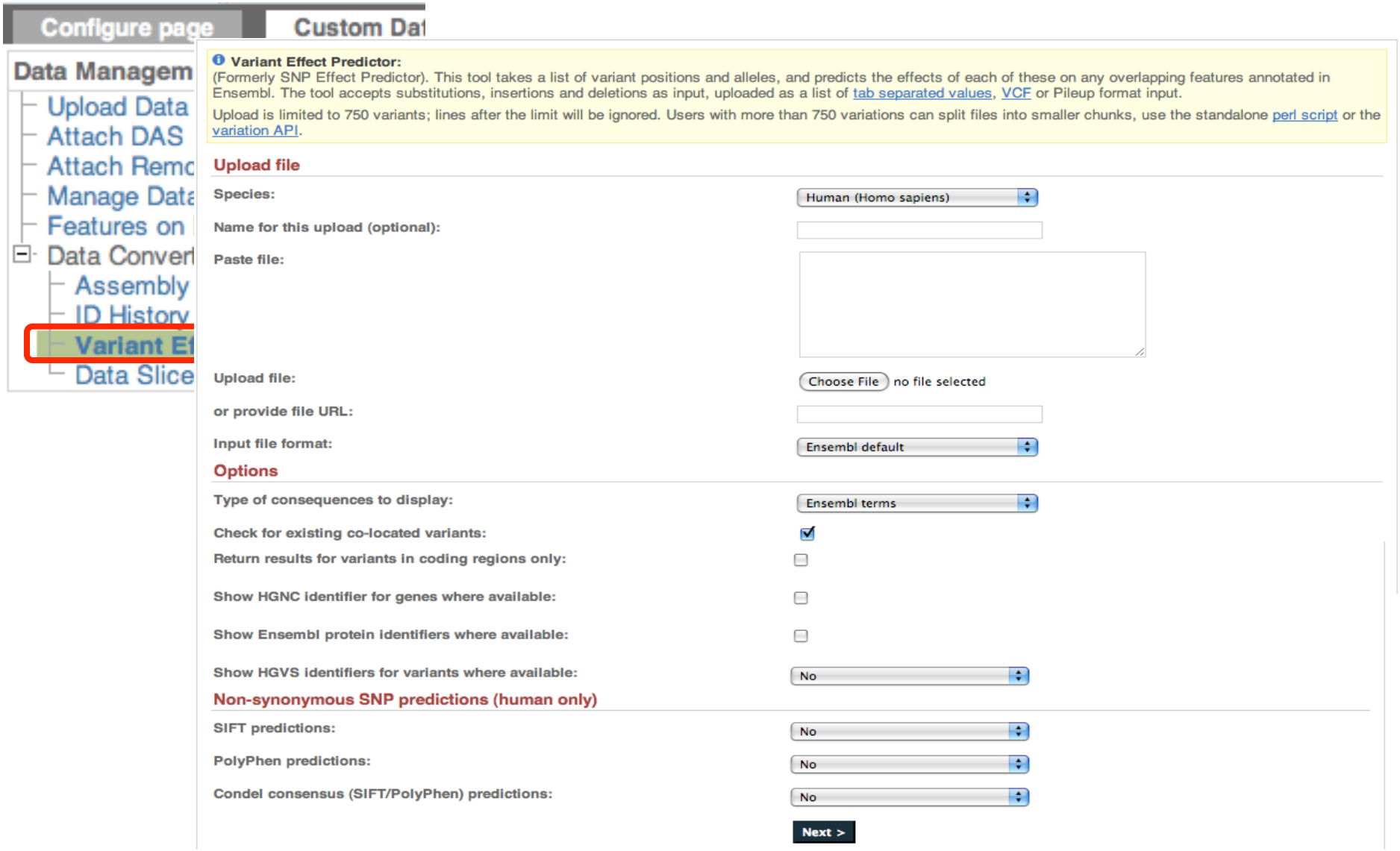
Data Slicer

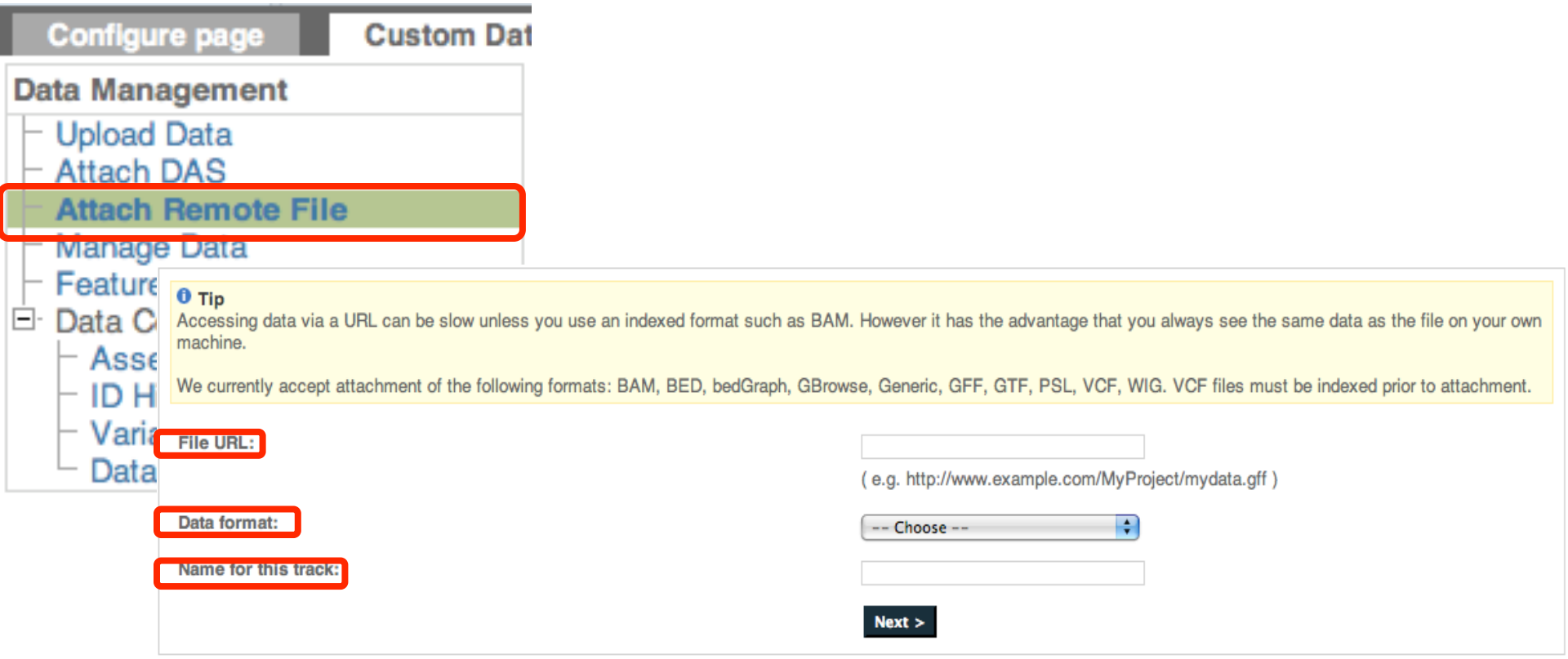
- Data Slicer allows you to get a fraction of the data within a chromosomal region you specified.
- Accessed from the Data Management page.
- Works on BAMs or VCFs accompanied by appropriate index files.

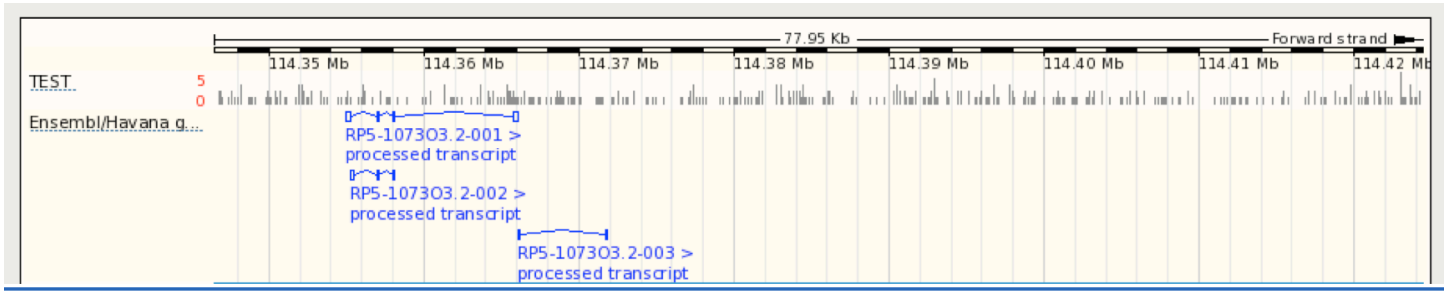


Variant Effect Predictor



Upload Variant or Alignment Data Into the Browser





Credits

- Eugene Kulesha, Glenn Proctor
- Natassa Spiridou
- Will McLaren, Fiona Cunningham
- Laura Clarke, Holly Zheng-Bradley, Rick Smith

- info@1000genomes.org