

The 1000 Genomes Project

Laura Clarke



- “After the first complete sequence of a human genome is obtained, the next challenge will be to discover and understand the function and variation of genes and, ultimately, to understand how such qualities affect health and disease. A key to this undertaking will be the availability of methods for efficient and accurate identification of genetic variation ...”
- Sydney Brenner et al. (June 2000) Nature Biotechnology



Glossary

- **Pilot** : The 1000 Genomes project ran a pilot study between 2008 and 2010
- **Phase 1**: The initial round of exome and low coverage sequencing of 1000 individuals
- **Phase 2**: Expanded sequencing of 1700 individuals and method improvement
- **SAM/BAM**: Sequence Alignment/Map Format, an alignment format
- **VCF**: Variant Call Format, a variant format
- **Date Formats**: In 1000 genomes file/directory names dates are mostly represented as YYYYMMDD

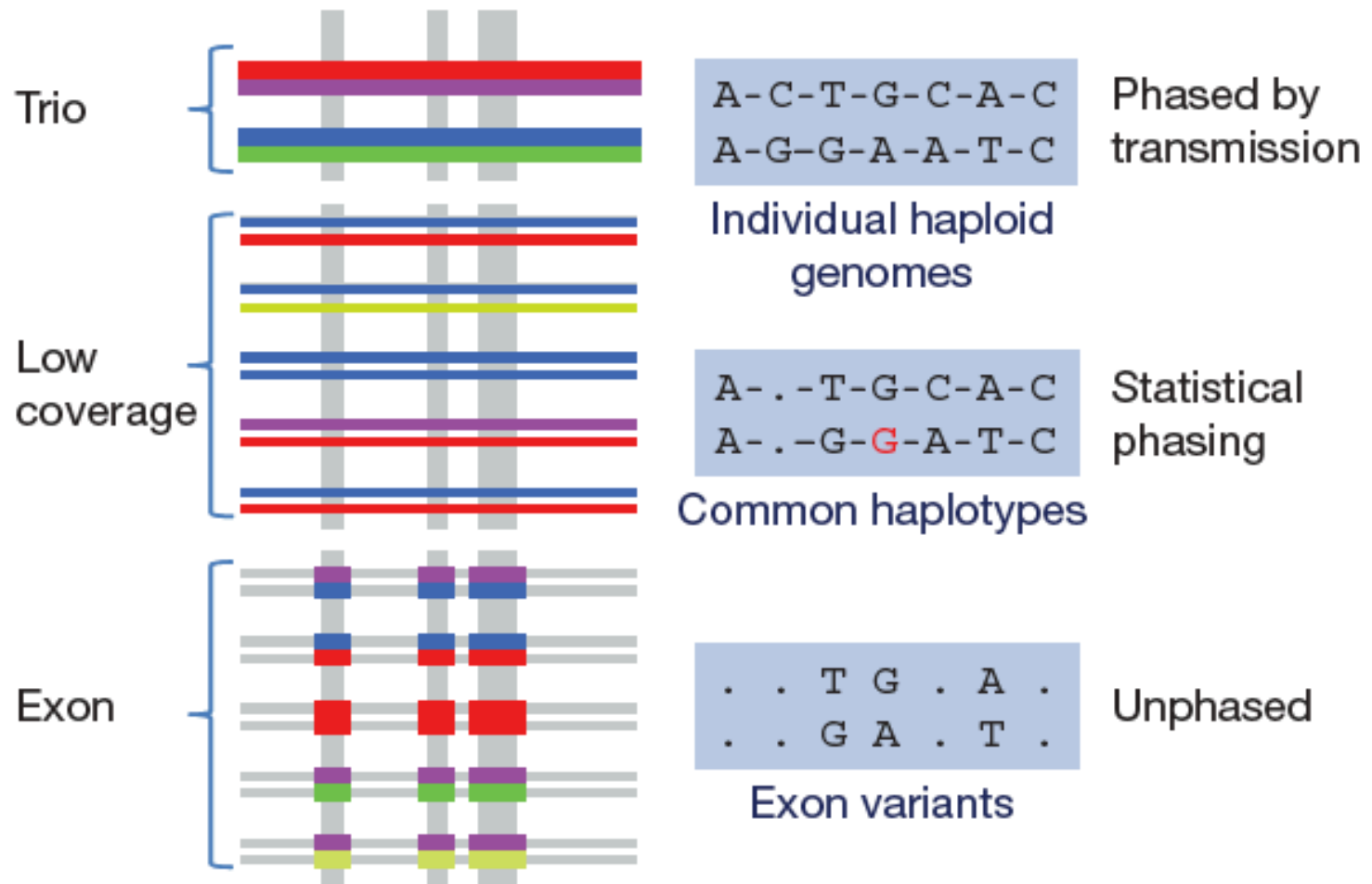


The 1000 Genomes Project: Overview

- International project to construct a foundational data set for human genetics
 - Discover virtually all common human variations by investigating many genomes at the base pair level
 - Consortium with multiple centers, platforms, funders
- Aims
 - Discover population level human genetic variations of all types (95% of variation $>$ 1% frequency)
 - Define haplotype structure in the human genome
 - Develop sequence analysis methods, tools, and other reagents that can be transferred to other sequencing projects



3 pilot coverage strategies



Main Project Design

- Based on the result of the pilot project, we decided to collect data on more than 2,500 samples from 5 continental groupings
 - Whole-genome low coverage data (>4x)
 - Full exome data at deep coverage (>20x)
 - 500 deep coverage genomes to be sequenced
 - High density genotyping at subsets of sites using both Illumina Omni and Affymetrix Axiom
- Phase 1 Release Integrated Variant Release has been made.



Phase I (1,150)

Phase II (1,721)

Phase III (2,500)

CDX
17S



CLM (70T); DNA from LCL



CHS (100T); DNA from LCL



PUR (70T); DNA from Blood



FIN (100S); DNA from LCL



GBR (96/100S); DNA from



IBS (84/100T); DNA from LCL



GWD



GWD



GWD



GWD (target - 100T); DNA from LCL



CDX (100S); DNA: 17 DNA from Bld, 83 from LCL

KHV (82/100) - 15 trios; DNA Bld



ACB (28/79T) - 14 trios; DNA Bld



PEL (70T); DNA from Blood



3



16 (8T)



PJL (target - 100T); DNA from Blood



Nigeria (target - 100T); DNA from
Sierra Leone (target - 100T); DNA from LCL




MAB (target - 100T); DNA from LCL



AJM (target - 80T); DNA from Bld



GIH vs. Sindhi (target - 100T)




Tamil (target -



Sri Lankan (target - 100T)



Bengalee (target - 100T)




Hapmap, The Pilot Project and The Main Project

- **Hapmap**
 - Starting in 2002
 - Last release contained ~3m snps
 - 1400 individuals
 - 11 populations
 - High Throughput genotyping chips
- **1000 Genomes Pilot project**
 - Started in 2008
 - Paper release contained ~14 million snps
 - 179 individuals
 - 4 populations
 - Low coverage next generation sequencing
- **1000 Genomes Phase 1**
 - Started in 2009
 - Phase 1 release has 36.6million snps, 1.5million indels and 14K deletions
 - 1092 individuals
 - 14 populations
 - Low coverage and exome next generation sequencing
- **1000 Genomes Phase 2**
 - Started in 2011
 - 1721 individuals
 - 19 Populations
 - Low coverage and exome next generation sequencing



Timeline

- **September 2007:** 1000 Genomes project formally proposed Cambridge, UK
- **April 2008:** First Submission of Data to the Short Read Archive.
- **May 2008:** First public data release.
- **October 2008:** SAM/BAM Format Defined.
- **December 2008:** First High Coverage Variants Released.
- **December 2008:** First 1000 genomes browser released
- **May 2009:** First Indel Calls released.
- **July 2009:** VCF Format defined
- **August 2009:** First Large Scale Deletions released.
- **December 2009:** First Main Project Sequence Data Released.
- **March 2010:** Low Coverage Pilot Variant Release made
- **July 2010:** Phased genotypes for 159 Individuals released.
- **October 2010:** A Map of Human Variation from population scale sequencing is published in Nature.
- **January 2011:** Final Phase 1 Low coverage alignments are released
- **May 2011:** @1000genomes appears on Twitter
- **May 2011:** First Variant Release made on more than 1000 individuals
- **October 2011:** Phase 1 integrated variant release made

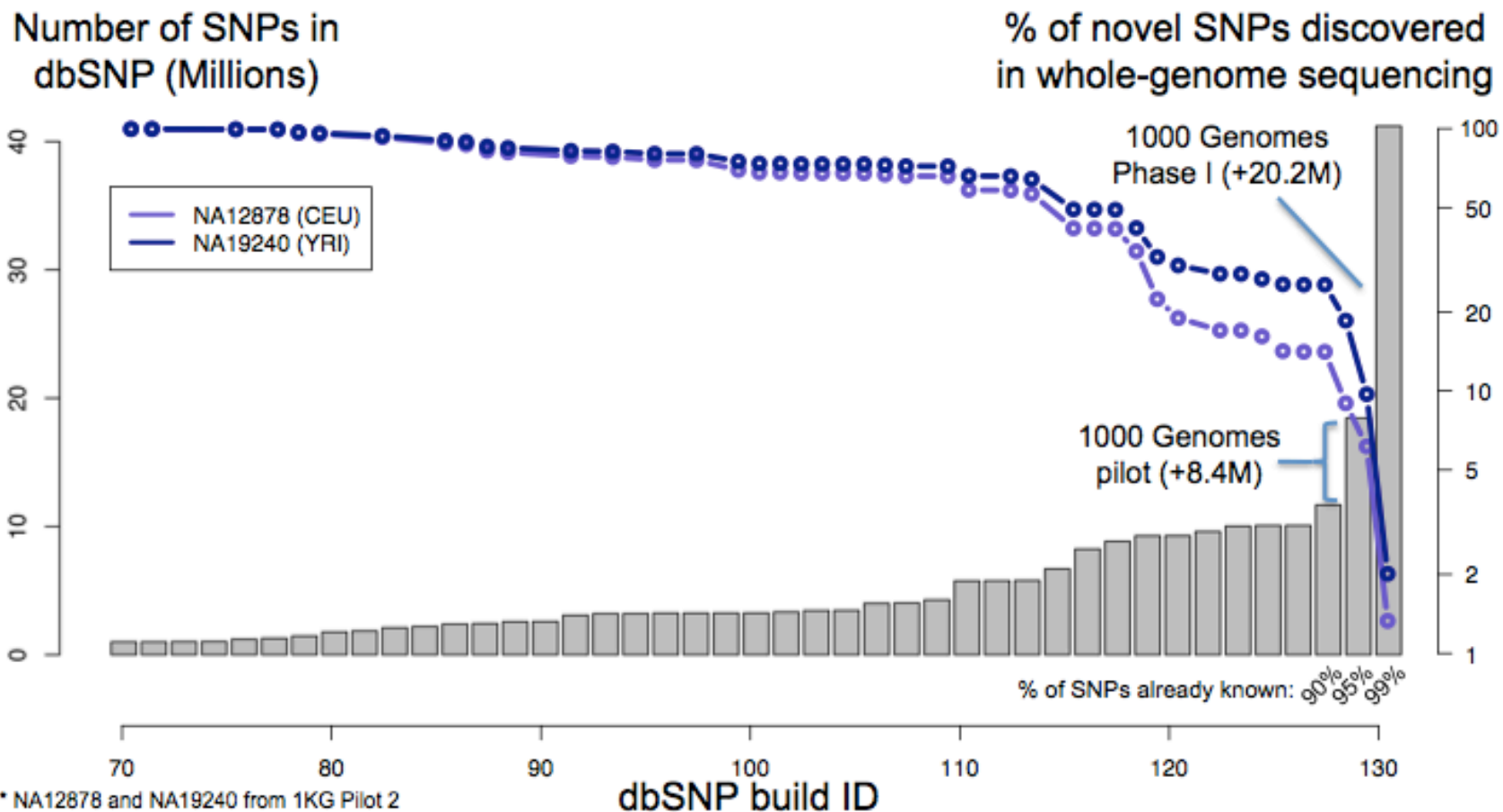


Sequencing Data Evolution

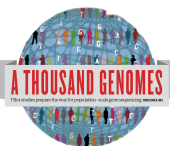
- The Project contains data from 3 different providers and multiple platforms

Platform	Min Read Length (bp)	Max Read Length (bp)
454 Roche GS FLX Titanium	70	400
Illumina GA	30	81
Illumina GA II	26	160
Illumina HiSeq	50	102
ABI Solid System 2.0	25	35
ABI Solid System 2.5	50	50
ABI Solid System 3.0	50	50

1000 Genomes discovered 29M new SNPs; now ~99% of variation in each person is already known



* NA12878 and NA19240 from 1KG Pilot 2 deep whole genome sequencing



1000 Genomes Project: Present & Future

- First Phase 2 sequence release 14th November 2011
- First Phase 2 alignment release 12th March 2012
- First Phase 2 variant site release Summer 2012

- Sample collected expected end to June 2012
- Final Phase 3 Sequence release expected December 2012
- 2013 will represent finalization of 1000 genomes analysis results and final data releases

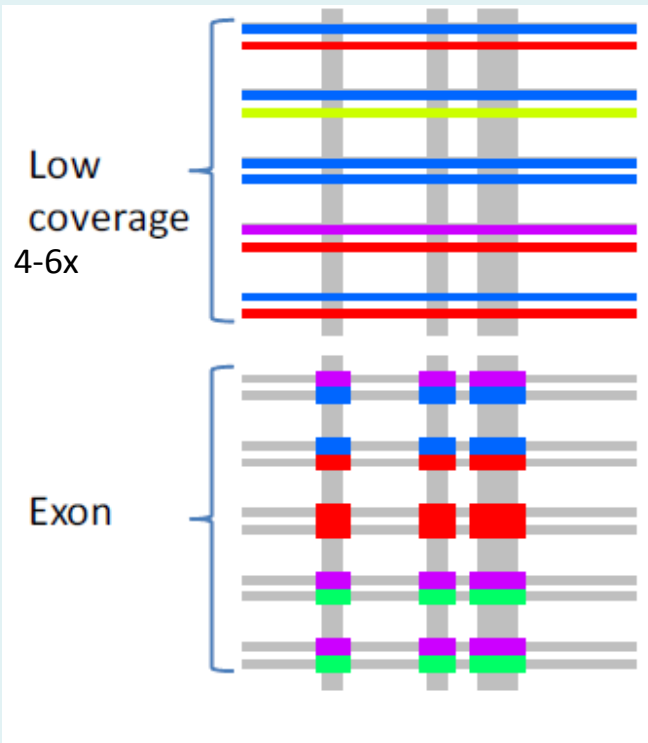


Pipelines for data processing and variant calling

- Tens of analysis groups have contributed
- Individual pipelines and component tools vary
- Typical main steps:
 - Read mapping
 - Duplicate filtering
 - Base quality score recalibration
 - INDEL realignment
 - Variant Site Discovery
 - Individual Genotype Assignment (sometimes part of site discovery)
 - Variant filtering / call set refinement
 - Variant reporting

Approach to the Analysis

Data



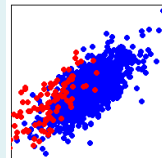
Multiple centres
Multiple technologies

Variant discovery

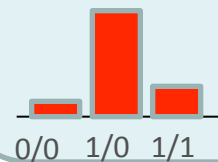
- SNPs
- ▲ Short indels
- ▭ Structural variants

Multiple Methodologies

Variant integration

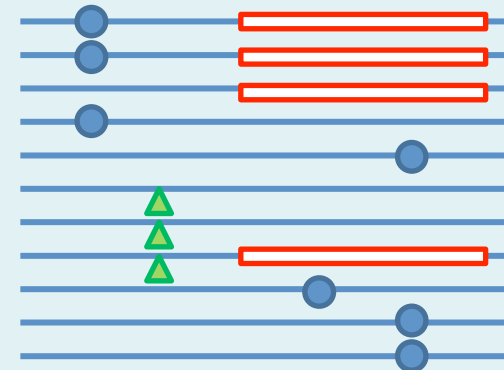


Adjudication



Likelihoods

Haplotype integration



Single release VCF

Alignment Data

- The project has made more than 10 releases of Alignment Data
- Pilot Project
 - Aligned to NCBI36
 - Maq and Corona
 - Base Quality Recalibration done
- Phase 1
 - Aligned to GRCh37
 - BWA and Bfast
 - Indel Realignment
- Phase 2
 - Aligned to extended GRCh37
 - Improvements to Base Quality Recalibration



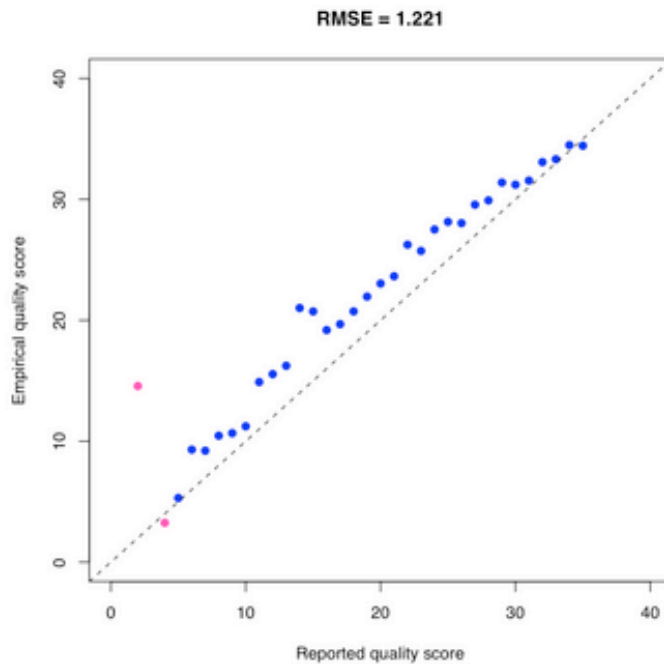
Base Quality Score Recalibration

- 1000 Genomes Sequence Data is sourced from many different machines across many different institutes
- Each machine may assign Base Quality Values differently
- Base Quality Score Recalibration tests empirical error rates
 - Run alignment
 - Compare mismatches to know variation
- Base Qualities adjusted on basis of empirical measurements

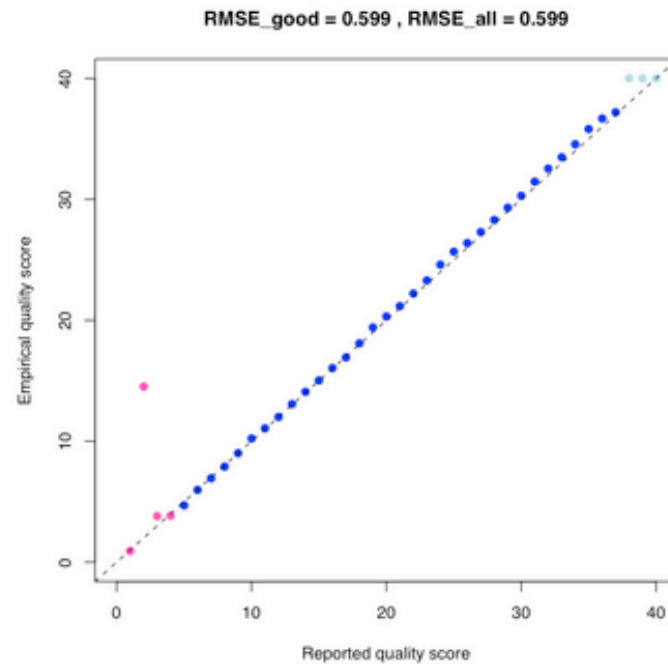


Base Quality Score Recalibration

Reported Quality vs. Empirical Quality



Original Data



After GATK Recalibration

Methods for integrated genotypes

Components		SNPs	INDELs	SVs
Low-Pass Genomes	Call Sets	BC, BCM, BI NCBI, SI, UM	BC, BI, DI OX, SI	BI, EBI, EMBL UW, Yale
	Consensus	VQSR	VQSR	GenomeSTRiP
Deep Exomes	Call Sets	BC, BCM, BI UM, WCMC	N/A	N/A
	Consensus	SVM	N/A	N/A
Likelihood		BBMM	GATK	GenomeSTRiP
Site Models		Variants are linearly ordered as point mutations		
Haplotyper		MaCH/Thunder with BEAGLE's initial haplotypes		

Variant Calling

- Early call sets used a single variant caller
- Intersect approach developed during pilot
- Variant Quality Score Recalibration (VQSR) developed for Phase 1
- Genotype Likelihoods assigned to help with genotype calling
- Integrated genotype calling based on individual variant call sets
- Phase 2 looks to make site discovery more integrated



Variant Quality Score Recalibration

- Multiple Different Variant Callers are used as part of the 1000 Genomes
- Variant Quality Score Recalibration used to define high quality variants from large input set
- Variants as points in a point cloud can be modeled using a Gaussian mixture model
- Model compared to various statistical models to define best set of variants



VQSR consensus out performs previous merging strategy

Called In	Total # variants	dbSNP % (129)	# novels	Novel ti/tv	Omni poly sensitivity	Omni mono false discovery
Union	46.26M	19.39%	37.29M	1.998	98.94% 2.09M / 2.12M	16.31% 9,739 / 59,721
2 of 6	39.11M	22.24%	30.41M	2.153	98.55% 2.09M / 2.12M	11.23% 6,707 / 59,721
3 of 6	35.69M	23.62%	27.26M	2.219	98.09% 2.08M / 2.12M	3.66% 2,184 / 59,721
4 of 6	32.55M	24.82%	24.48M	2.263	97.39% 2.06M / 2.12M	1.82% 1,085 / 59,721
5 of 6	28.45M	26.72%	20.85M	2.286	95.93% 2.03M / 2.12M	1.06% 634 / 59,721
Intersection	24.02M	27.57%	17.40M	2.317	89.23% 1.89M / 2.12M	0.76% 457 / 59,721
VQSR Project Consensus	38.88M	21.92%	30.36M	2.154	98.41% 2.08M / 2.12M	2.11% 1,261 / 59,721

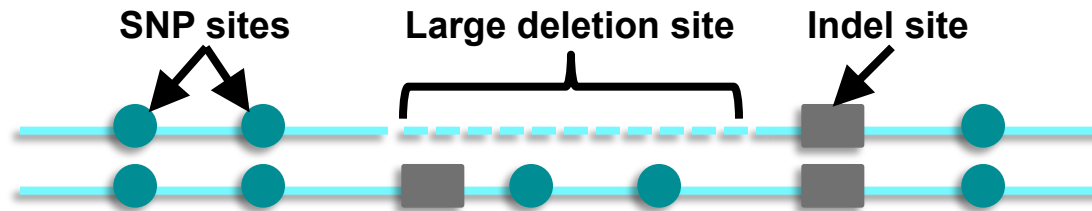


Methods for integrated genotypes

Components		SNPs	INDELs	SVs
Low-Pass Genomes	Call Sets	BC, BCM, BI NCBI, SI, UM	BC, BI, DI OX, SI	BI, EBI, EMBL UW, Yale
	Consensus	VQSR	VQSR	GenomeSTRiP
Deep Exomes	Call Sets	BC, BCM, BI UM, WCMC	N/A	N/A
	Consensus	SVM	N/A	N/A
Likelihood		BBMM	GATK	GenomeSTRiP
Site Models		Variants are linearly ordered as point mutations		
Haplotyper		MaCH/Thunder with BEAGLE's initial haplotypes		



Strategies for integrating deletions with other types of variation



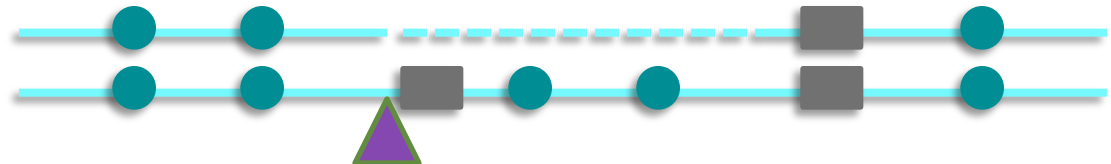
Previous Approach

Remove SNPs under SVs for imputation
(1000G pilot, Handsaker et al., 2010)



Current Approach

Treat SVs as point events
(1000 Genomes phase 1)



From PILOT to PHASE1

PILOT

- 14.8M SNPs
- Ts/Tv 2.01
- Includes
97.8% HapMap3

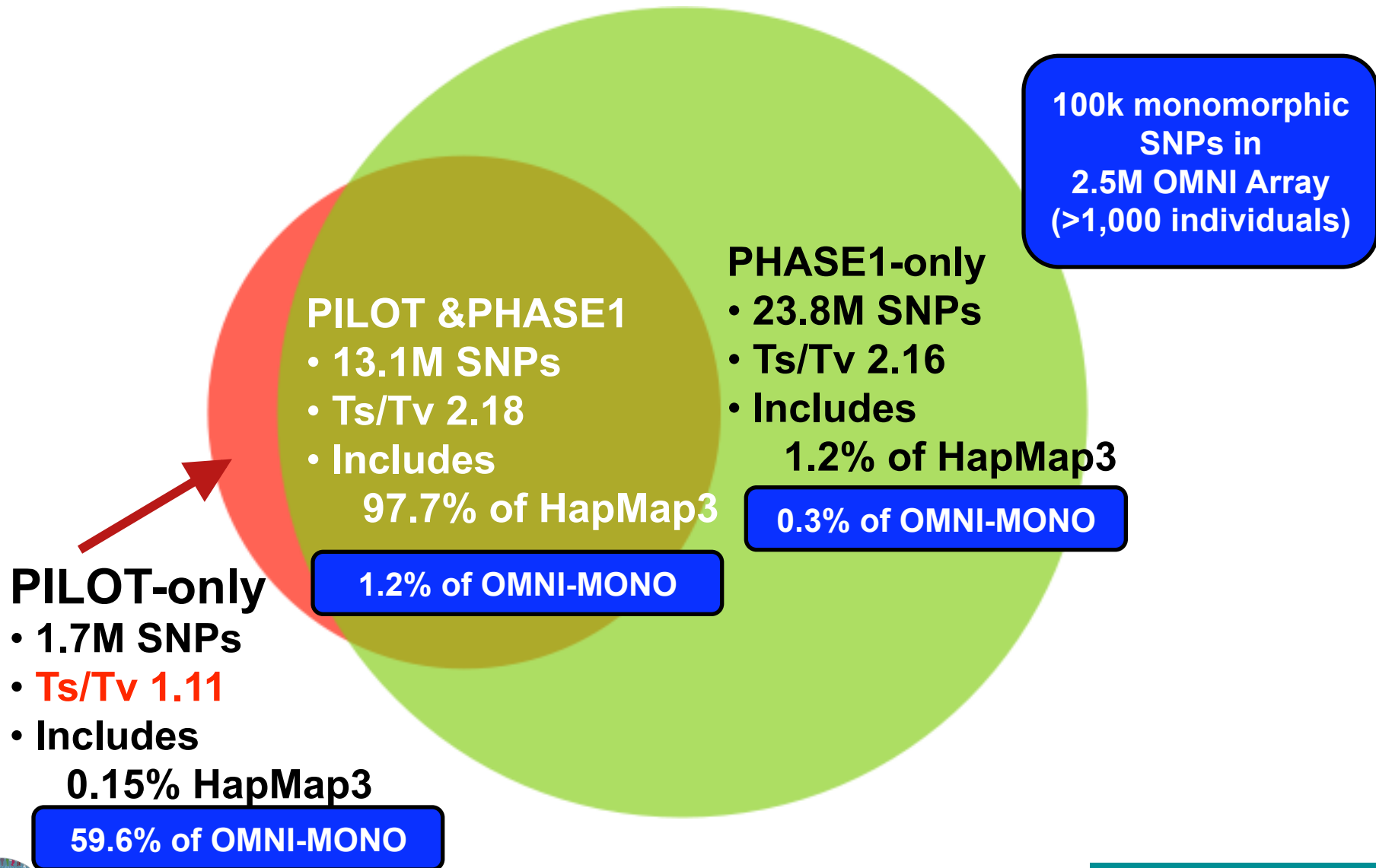
PHASE1

- 36.8M SNPs
- Ts/Tv 2.17
- Includes
98.9% HapMap3

Autosomal chromosomes only

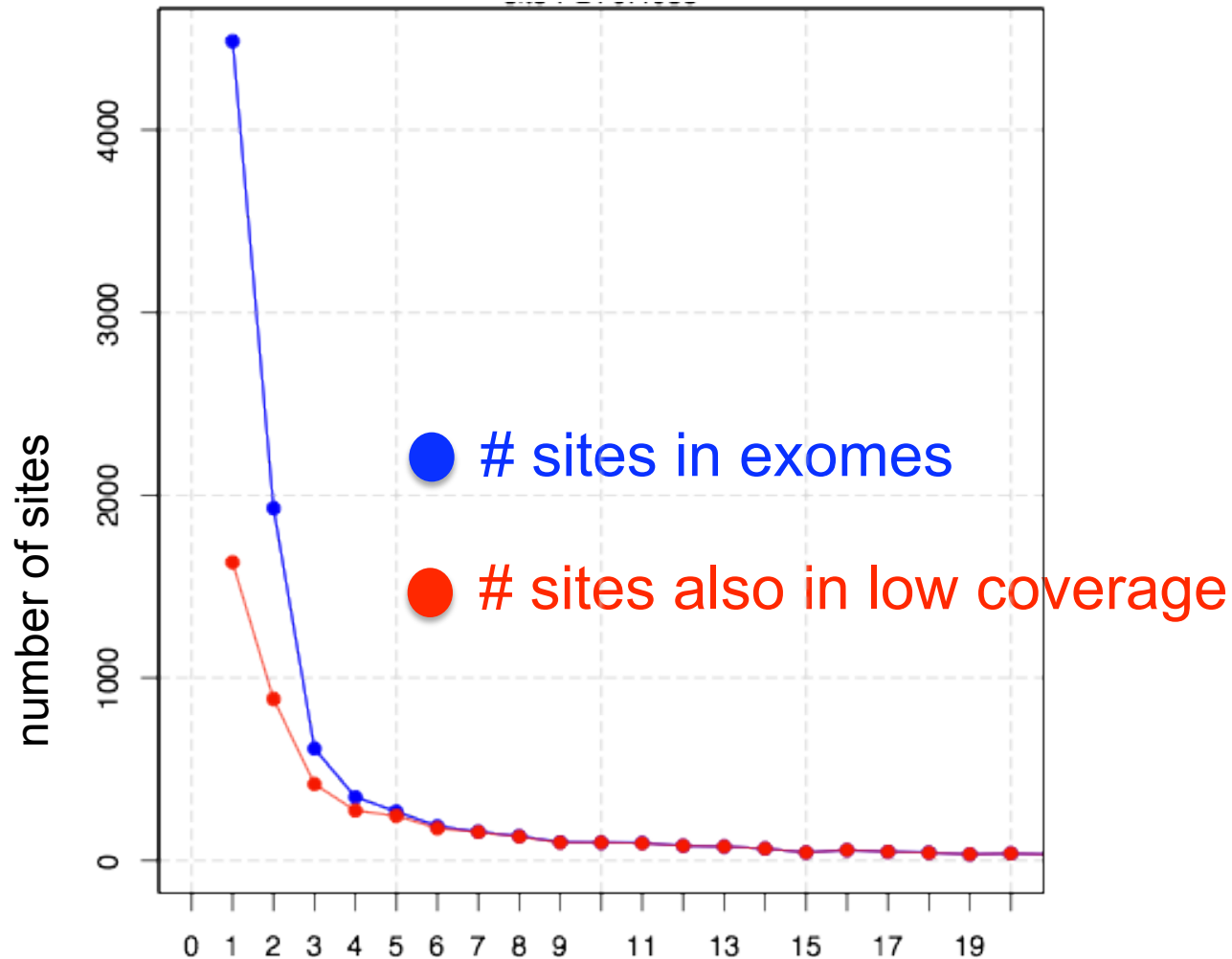


From PILOT to PHASE1 : Improved SNP calls



OMNI-MONO information was not used in making phase1 variant calls

Deep coverage exome data is more sensitive to low-frequency variants



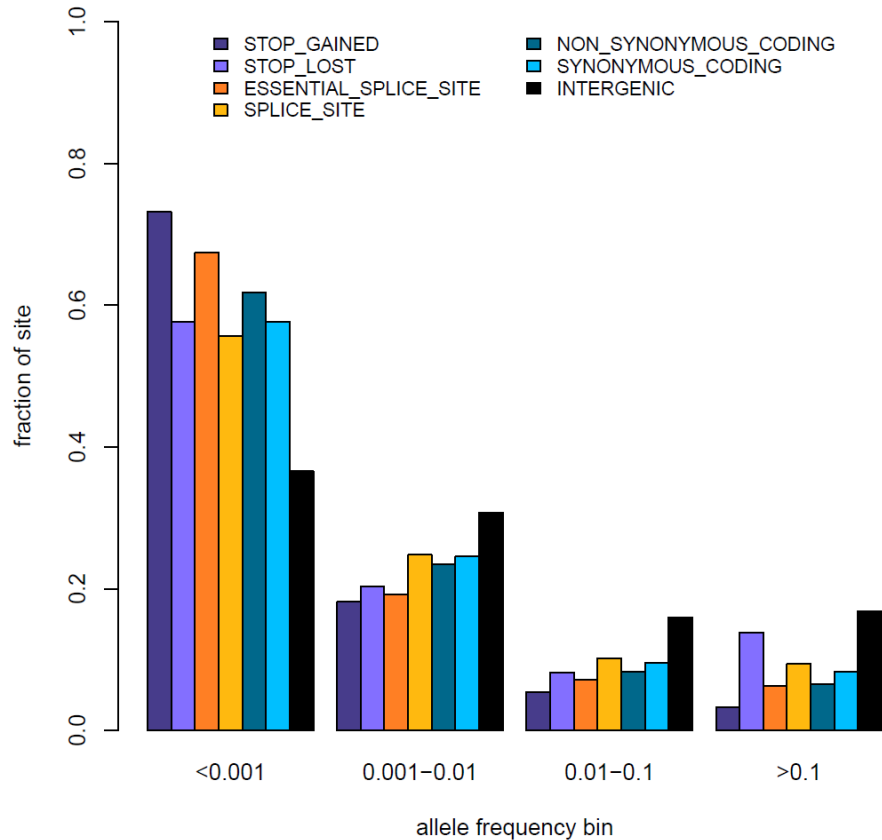
Allele count in 766 exomes (chr. 20, exons only)

Erik Garrison

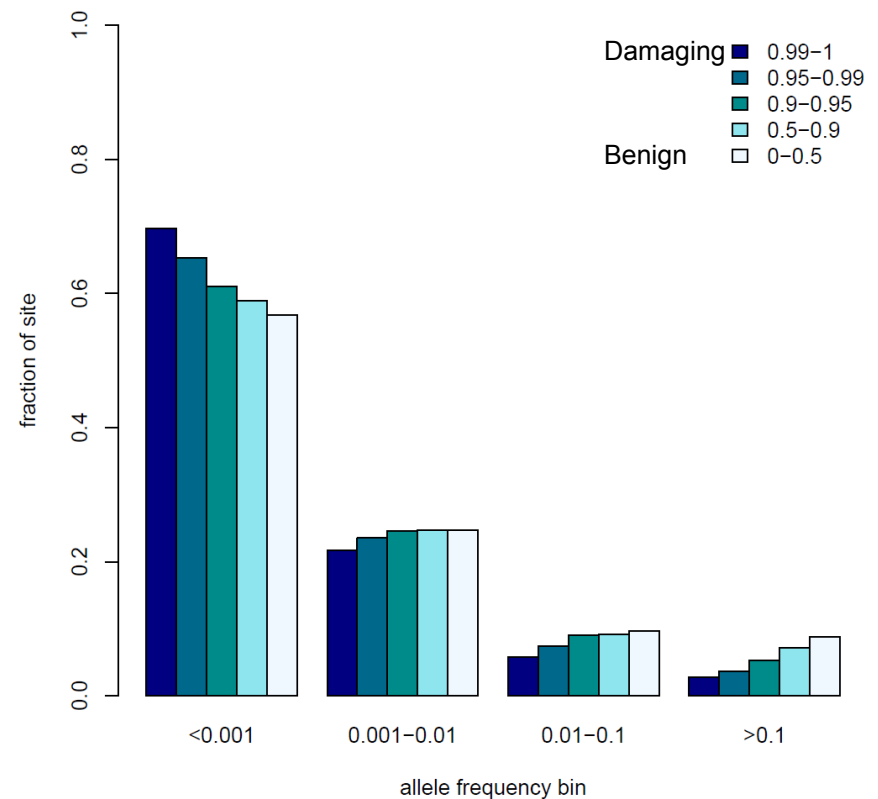


Newly discovered SNPs are mostly at low frequency and enriched for functional variants

Functional category

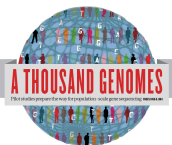


Non-synonymous: Condel score

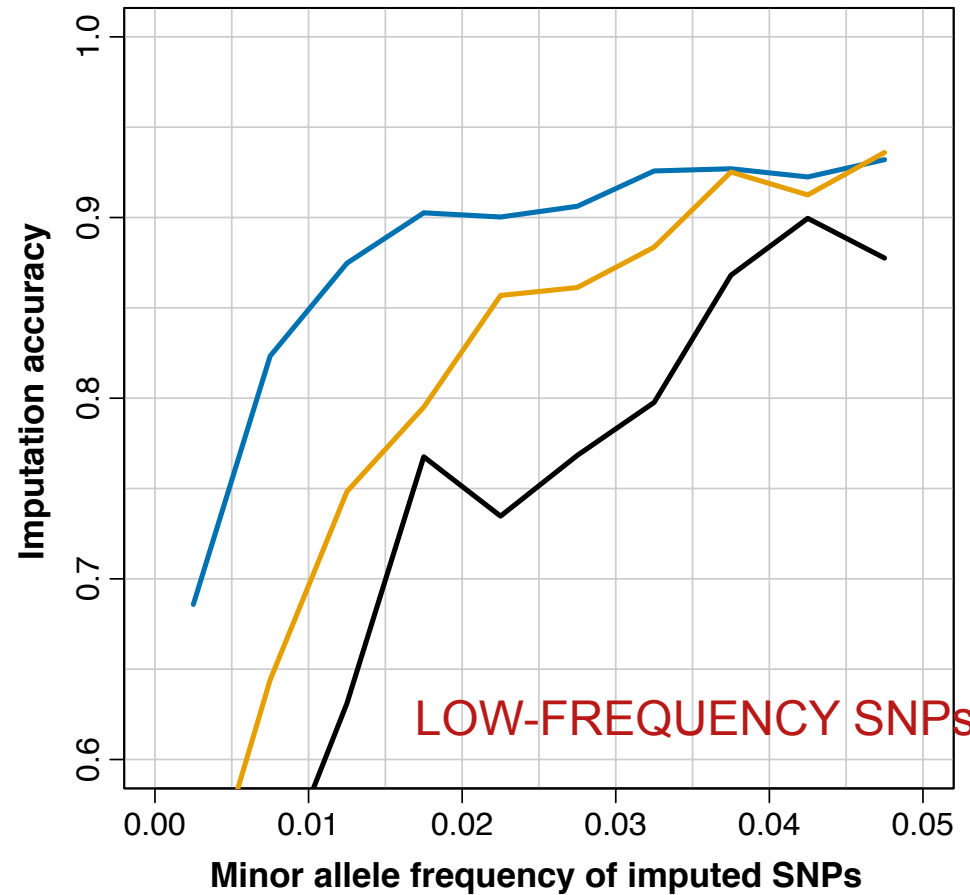
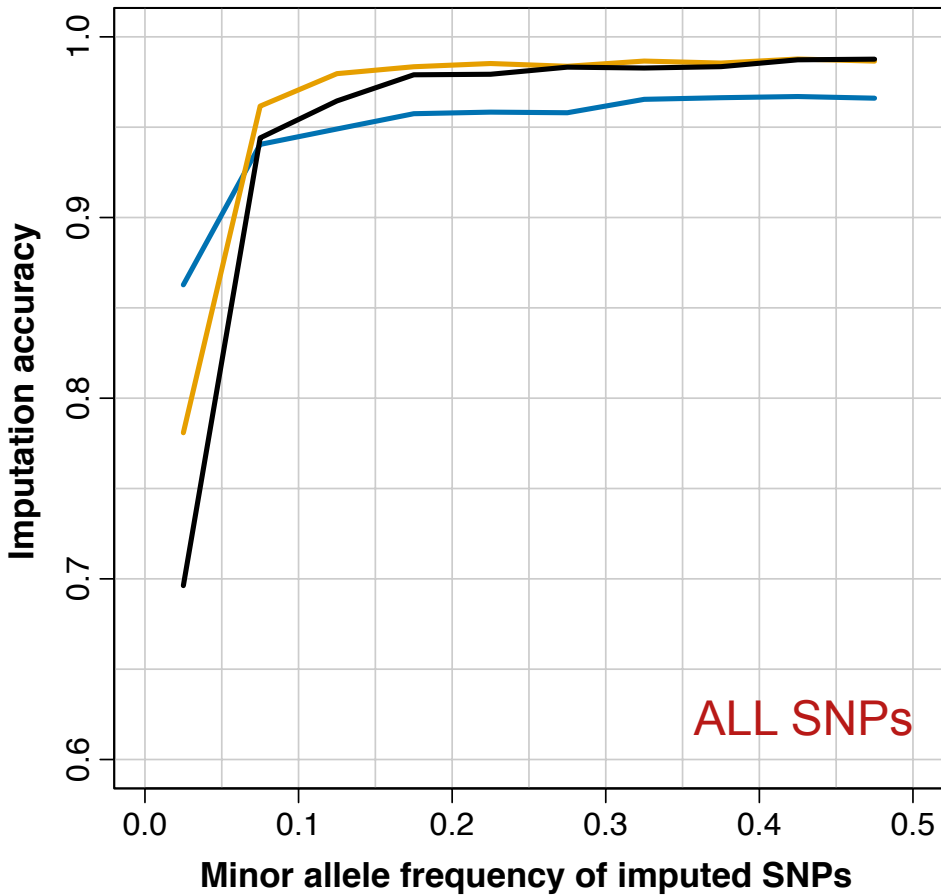


Presentation on using the data for GWAS by Brian Howie

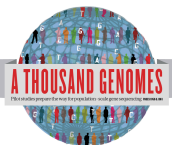
Enza Colonna, Yuan Chen, Yali Xue



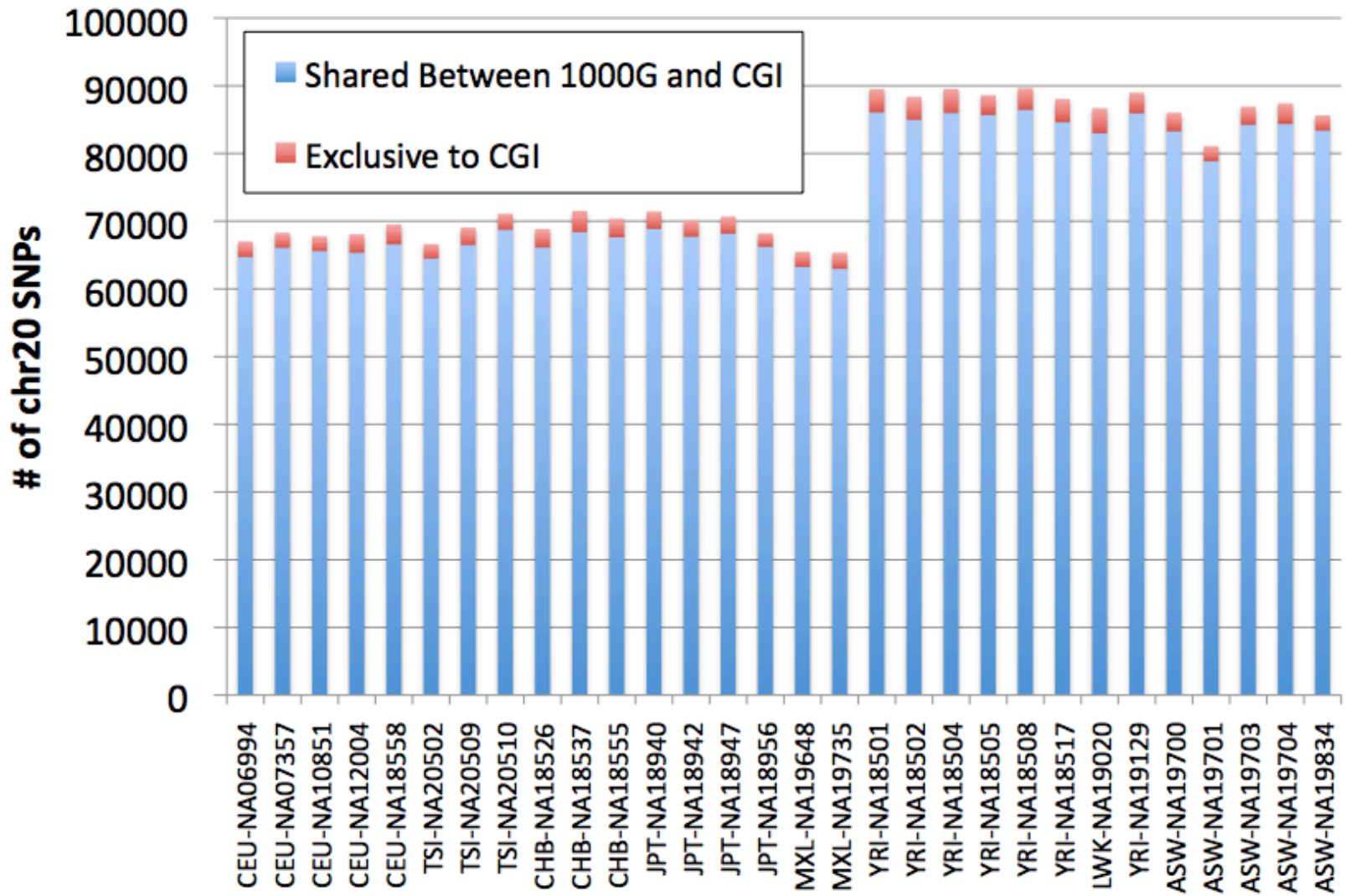
1,000 Genomes haplotypes are highly accurate



- European ancestry
- African ancestry
- Admixed (Americas)



>96% SNPs are detected compared to deep genomes



Data Availability and the FTP site



File Formats

- Sequence in Fastq
- Alignments in SAM/BAM
- Variant Calls in VCF
- Other data
 - ped
 - gff/gtf
 - bed



More Information About BAM Files

- <http://samtools.sourceforge.net/>
- samtools-help@lists.sourceforge.net

BIOINFORMATICS APPLICATIONS NOTE

Vol. 25 no. 16 2009, pages 2078–2079
doi:10.1093/bioinformatics/btp352

Sequence analysis

The Sequence Alignment/Map format and SAMtools

Heng Li^{1,†}, Bob Handsaker^{2,†}, Alec Wysoker², Tim Fennell², Jue Ruan³, Nils Homer⁴, Gabor Marth⁵, Goncalo Abecasis⁶, Richard Durbin^{1,*} and 1000 Genome Project Data Processing Subgroup⁷

¹Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Cambridge, CB10 1SA, UK, ²Broad Institute of MIT and Harvard, Cambridge, MA 02141, USA, ³Beijing Institute of Genomics, Chinese Academy of Science, Beijing 100029, China, ⁴Department of Computer Science, University of California Los Angeles, Los Angeles, CA 90095, ⁵Department of Biology, Boston College, Chestnut Hill, MA 02467, ⁶Center for Statistical Genetics, Department of Biostatistics, University of Michigan, Ann Arbor, MI 48109, USA and ⁷<http://1000genomes.org>

Received on April 28, 2009; revised on May 28, 2009; accepted on May 30, 2009

Advance Access publication June 8, 2009

Associate Editor: Alfonso Valencia

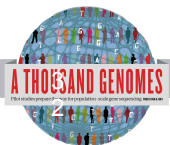
ABSTRACT

Summary: The Sequence Alignment/Map (SAM) format is a generic alignment format for storing read alignments against reference sequences, supporting short and long reads (up to 128 Mbp) produced by different sequencing platforms. It is flexible in style,

2 METHODS

2.1 The SAM format

2.1.1 Overview of the SAM format The SAM format consists of one header section and one alignment section. The lines in the header section start with character '@', and lines in the alignment section do not. All lines



More Information About VCF Files

<http://vcftools.sourceforge.net/>
vcftools-help@lists.sourceforge.net

BIOINFORMATICS APPLICATIONS NOTE Vol. 27 no. 15 2011, pages 2156–2158
doi:10.1093/bioinformatics/btr330

Sequence analysis

Advance Access publication June 7, 2011

The variant call format and VCFtools

Petr Danecek^{1,†}, Adam Auton^{2,†}, Goncalo Abecasis³, Cornelis A. Albers¹, Eric Banks⁴, Mark A. DePristo⁴, Robert E. Handsaker⁴, Gerton Lunter², Gabor T. Marth⁵, Stephen T. Sherry⁶, Gilean McVean^{2,7}, Richard Durbin^{1,*} and 1000 Genomes Project Analysis Group[‡]

¹Wellcome Trust Sanger Institute, Wellcome Trust Genome Campus, Cambridge CB10 1SA, ²Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford OX3 7BN, UK, ³Center for Statistical Genetics, Department of Biostatistics, University of Michigan, Ann Arbor, MI 48109, ⁴Program in Medical and Population Genetics, Broad Institute of MIT and Harvard, Cambridge, MA 02141, ⁵Department of Biology, Boston College, MA 02467, ⁶National Institutes of Health National Center for Biotechnology Information, MD 20894, USA and ⁷Department of Statistics, University of Oxford, Oxford OX1 3TG, UK

Associate Editor: John Quackenbush

VCF variant files

All indexed for fast retrieval

BIOINFORMATICS APPLICATIONS NOTE Vol. 27 no. 5 2011, pages 718–719
doi:10.1093/bioinformatics/btq671

Sequence analysis

Advance Access publication January 5, 2011

Tabix: fast retrieval of sequence features from generic TAB-delimited files

Heng Li

Program in Medical Population Genetics, The Broad Institute of Harvard and MIT, Cambridge, MA 02142, USA

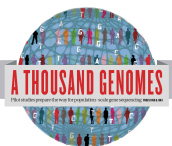
Associate Editor: Dmitrij Frishman

ABSTRACT

Summary: Tabix is the first generic tool that indexes position sorted files in TAB-delimited formats such as GFF, BED, PSL, SAM and SQL export, and quickly retrieves features overlapping specified regions. Tabix features include few seek function calls per query, data

2 METHODS

Tabix indexing is a generalization of BAM indexing for generic TAB-delimited files. It inherits all the advantages of BAM indexing, including data compression and efficient random access in terms of few seek function calls per query.



FTP Site

- Two mirrored ftp sites
 - <ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp>
 - <ftp://ftp-trace.ncbi.nih.gov/1000genomes/ftp>
- NCBI site is direct mirror of EBI site
- Can be up to 24 hours out of date
- Both also accessible using aspera
- <http://asperasoft.com/>
- EBI site has http mirror
 - <http://ftp.1000genomes.ebi.ac.uk/vol1/ftp>



















ftp://ftp.1000genomes.ebi.ac.uk

ftp://ftp-trace.ncbi.nih.gov/1000genomes/ftp

Index of ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/

 Up to higher level directory

Name	Size	Last Modified
 CHANGELOG	118 KB	05/01/2012 5/01/2012 12:40:00
 README.alignment_data	12 KB	26/01/2011 26/01/2011 12:00:00
 README.ftp_structure	9 KB	04/04/2011 4/04/2011 12:00:00
 README.pilot_data	3 KB	14/07/2011 14/07/2011 12:00:00
 README.populations	2 KB	18/02/2010 18/02/2010 12:00:00
 README.sequence_data	7 KB	23/07/2011 23/07/2011 19:03:00
 alignment_indices		14/07/2011 14/07/2011 10:53:00
 changelog_details		05/01/2012 05/01/2012 12:40:00
 current.tree	29933 KB	05/01/2012 05/01/2012 12:37:00
 data		04/07/2011 04/07/2011 8:50:00
 phase1		14/07/2011 14/07/2011 14:03:00
 pilot_data		27/07/2011 27/07/2011 12:00:00
 release		12/10/2011 12/10/2011 13:18:00
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 sequence_indices		14/11/2011 14/11/2011 10:10:00
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Documentation

Raw Data

Phase 1 Data

Pilot Data

Release Data

Technical Data



The FTP Site: Data

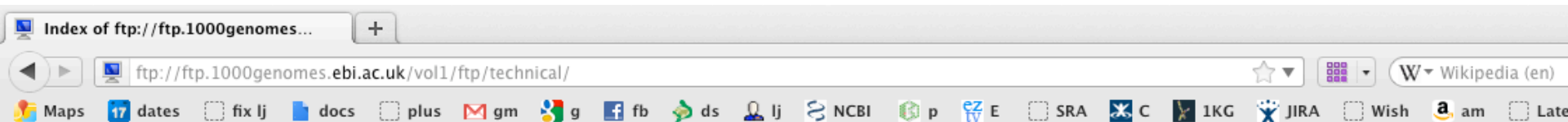
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ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data/

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HG00104	14/12/2011	14/12/2011	12:06:00
HG00105	13/12/2011	13/12/2011	12:45:00
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HG00107	13/12/2011	13/12/2011	12:40:00
HG00108	13/12/2011	13/12/2011	12:43:00
HG00109	13/12/2011	13/12/2011	12:43:00
HG00110	13/12/2011	13/12/2011	12:43:00
HG00111	13/12/2011	13/12/2011	12:36:00
HG00112	13/12/2011	13/12/2011	12:41:00
HG00113	13/12/2011	13/12/2011	12:41:00
HG00114	13/12/2011	13/12/2011	12:41:00
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HG00125	13/12/2011	13/12/2011	12:39:00
HG00126	13/12/2011	13/12/2011	12:06:00
HG00127	13/12/2011	13/12/2011	12:06:00
HG00128	13/12/2011	13/12/2011	12:46:00
HG00129	13/12/2011	13/12/2011	12:44:00
HG00130	13/12/2011	13/12/2011	12:44:00
HG00131	13/12/2011	13/12/2011	12:44:00

Sample Level Files
sequence_read
alignment



FTP Site: Technical



Index of ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/

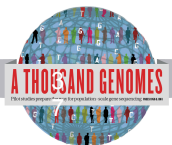
[Up to higher level directory](#)

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ncbi_varpipe_data		
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qc		
reference		
retired_reference		
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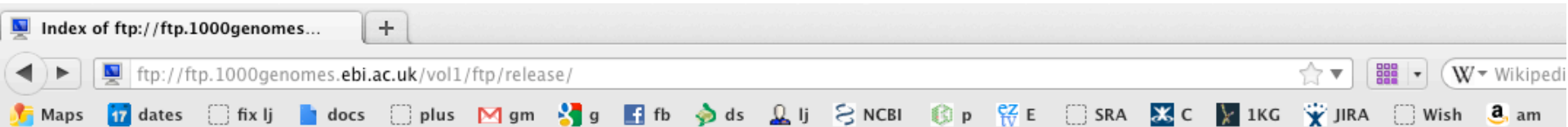
Alternative Alignments

Reference Data Sets

Experimental Data



FTP Site: Release



Index of ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/

[Up to higher level directory](#)

Name

Size

Last Modified

2008_12					
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2009_05		08/06/2009	08/06/2009	12 :00:00	
2009_08		10/08/2009	10/08/2009	12 :00:00	
20100804					00:00
20101123					00:00
2010_11		16/02/2011	16/02/2011	12 :00:00	
20110521		16/12/2011	16/12/2011	10 :09:00	
20110521.sequence.index	23693 KB	19/07/2011	19/07/2011	12 :00:00	
20110521.sequence.index.exome.stats	48 KB	19/07/2011	19/07/2011	12 :00:00	
20110521.sequence.index.low_coverage.stats	53 KB	21/05/2011	21/05/2011	12 :00:00	
20110521_20110719.exome.stats.csv	2 KB	19/07/2011	19/07/2011	12 :00:00	
20110521_20110719.low_coverage.stats.csv	2 KB	19/07/2011	19/07/2011	12 :00:00	
20110719.sequence.index	23961 KB	19/07/2011	19/07/2011	12 :00:00	
20110719.sequence.index.exome.stats	52 KB	10/10/2011	10/10/2011	10 :10:00	
20110719.sequence.index.low_coverage.stats	54 KB	10/10/2011	10/10/2011	10 :13:00	
20110719_20110920.exome.stats.csv	1 KB	10/10/2011	10/10/2011	19 :45:00	
20110719_20110920.low_coverage.stats.csv	2 KB	10/10/2011	10/10/2011	19 :45:00	

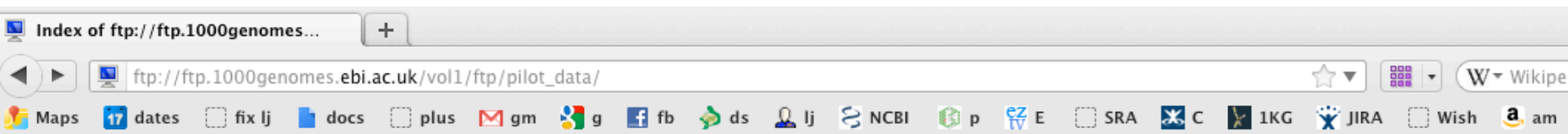
Date Format YYYYMMDD

Older Release Dirs

Sequence Index Dates



FTP Site: Pilot Data

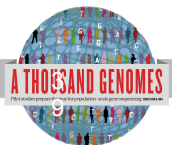


Index of ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/pilot_data/

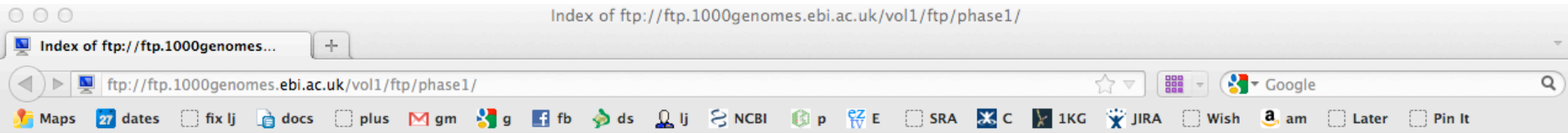
[Up to higher level directory](#)

Name	Size	Last Modified	
README.alignment.index	2 KB	26/08/2009	26/08/2009 12:00:00
README.bas	3 KB	27/08/2009	27/08/2009 12:00:00
README.sequence.index	2 KB	22/07/2009	22/07/2009 12:00:00
SRP000031.sequence.index	7365 KB	12/07/2010	12/07/2010 12:00:00
SRP000032.sequence.index	2181 KB	12/07/2010	12/07/2010 12:00:00
SRP000033.sequence.index	480 KB	12/07/2010	12/07/2010 12:00:00
data			
paper_data_sets		03/02/2011	3/02/2011 12:00:00
pilot_data.alignment.index	795 KB	06/05/2010	6/05/2010 12:00:00
pilot_data.alignment.index.bas.gz	1740 KB	14/06/2010	14/06/2010 12:00:00
pilot_data.sequence.index	10025 KB	12/07/2010	12/07/2010 12:00:00
release		20/07/2010	20/07/2010 12:00:00
technical		29/07/2010	29/07/2010 12:00:00

Pilot Paper Data



FTP Site: Phase 1



Index of ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/phase1/

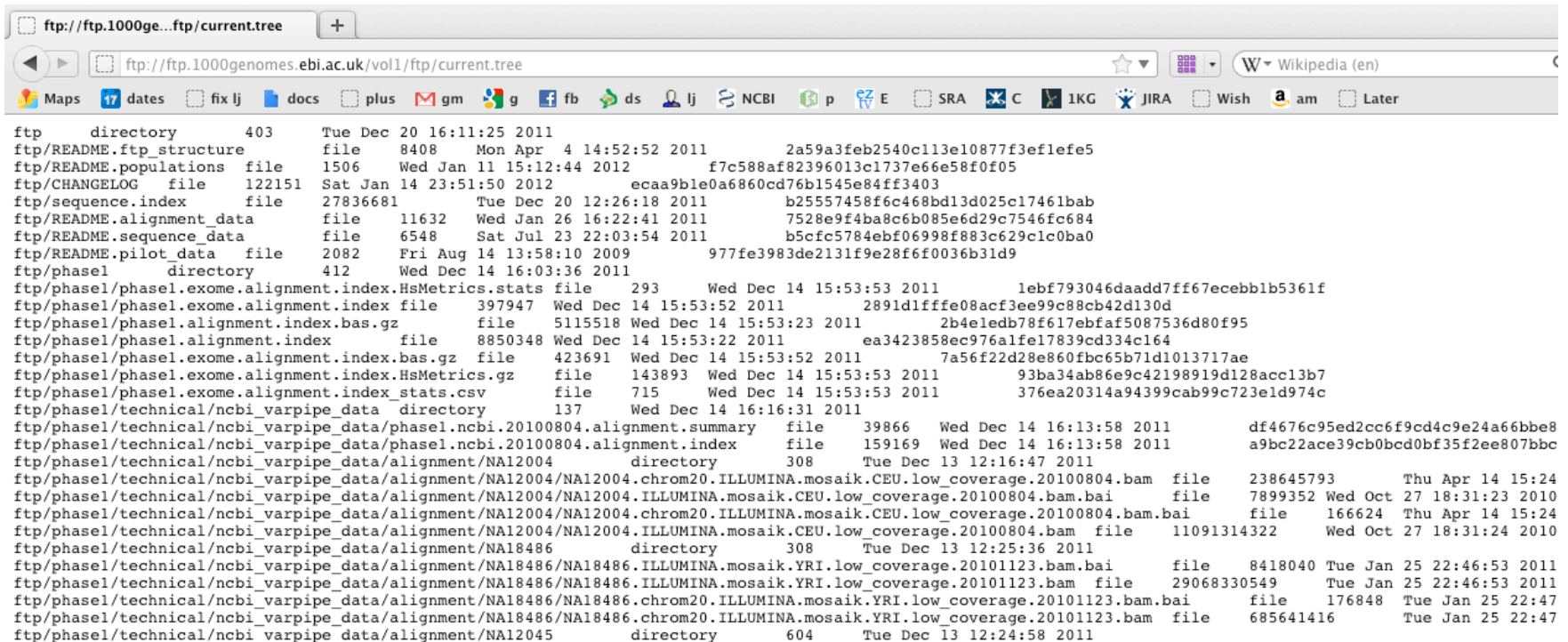
[Up to higher level directory](#)

Name	Size		
README.phase1_alignment_data	11 KB	08	
data		13/12/2011	13/12/2011 12:54:00
phase1.alignment.index	8643 KB	14/12/2011	14/12/2011 13:53:00
phase1.alignment.index.bas.gz	4996 KB	14/12/2011	14/12/2011 13:53:00
phase1.exome.alignment.index	389 KB	14/12/2011	14/12/2011 13:53:00
phase1.exome.alignment.index.HsMetrics.gz	141 KB	14/12/2011	14/12/2011 13:53:00
phase1.exome.alignment.index.HsMetrics.stats	1 KB	14/12/2011	14/12/2011 13:53:00
phase1.exome.alignment.index.bas.gz	414 KB	14/12/2011	14/12/2011 13:53:00
phase1.exome.alignment.index_stats.csv	1 KB	14/12/2011	14/12/2011 13:53:00
technical		14/12/2011	14/12/2011 14:11:00

Frozen Phase1
Alignments

Finding Data

- Current.tree file
- <ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/current.tree>
- Current Tree is updated nightly so can be upto 24 hours out of date



```
ftp directory 403 Tue Dec 20 16:11:25 2011
ftp/README.ftp_structure file 8408 Mon Apr 4 14:52:52 2011 2a59a3feb2540c113e10877f3ef1efe5
ftp/README.populations file 1506 Wed Jan 11 15:12:44 2012 f7c588af82396013c1737e66e58f0f05
ftp/CHANGELOG file 122151 Sat Jan 14 23:51:50 2012 ecaa9b1e0a6860cd76b1545e84ff3403
ftp/sequence.index file 27836681 Tue Dec 20 12:26:18 2011 b2557458f6c468bd13d025c17461bab
ftp/README.alignment_data file 11632 Wed Jan 26 16:22:41 2011 7528e9f4ba8c6b085e6d29c7546fc684
ftp/README.sequence_data file 6548 Sat Jul 23 22:03:54 2011 b5cfc5784ebf06998f883c629c10ba0
ftp/README.pilot_data file 2082 Fri Aug 14 13:58:10 2009 977fe3983de2131f9e28f6f0036b31d9
ftp/phase1 directory 412 Wed Dec 14 16:03:36 2011
ftp/phase1/phase1.exome.alignment.index.HsMetrics.stats file 293 Wed Dec 14 15:53:53 2011 1ebf793046daadd7ff67ececbb1b5361f
ftp/phase1/phase1.exome.alignment.index file 397947 Wed Dec 14 15:53:52 2011 2891d1ffffe08acf3ee99c88cb42d130d
ftp/phase1/phase1.alignment.index.bas.gz file 5115518 Wed Dec 14 15:53:23 2011 2b4e1edb78f617ebfaf5087536d80f95
ftp/phase1/phase1.alignment.index file 8850348 Wed Dec 14 15:53:22 2011 ea3423858ec976a1fe17839cd334c164
ftp/phase1/phase1.exome.alignment.index.bas.gz file 423691 Wed Dec 14 15:53:52 2011 7a56f22d28e860fbc65b71d1013717ae
ftp/phase1/phase1.exome.alignment.index.HsMetrics.gz file 143893 Wed Dec 14 15:53:53 2011 93ba34ab86e9c42198919d128acc13b7
ftp/phase1/phase1.exome.alignment.index_stats.csv file 715 Wed Dec 14 15:53:53 2011 376ea20314a94399cab99c723e1d974c
ftp/phase1/technical/ncbi_varpipe_data directory 137 Wed Dec 14 16:16:31 2011
ftp/phase1/technical/ncbi_varpipe_data/phase1.ncbi.20100804.alignment.summary file 39866 Wed Dec 14 16:13:58 2011 df4676c95ed2cc6f9cd4c9e24a66bbe8
ftp/phase1/technical/ncbi_varpipe_data/phase1.ncbi.20100804.alignment.index file 159169 Wed Dec 14 16:13:58 2011 a9bc22ace39cb0bcd0bf35f2ee807bbc
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA12004 directory 308 Tue Dec 13 12:16:47 2011
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA12004/NA12004.chrom20.ILLUMINA.mosaik.CEU.low_coverage.20100804.bam file 238645793 Thu Apr 14 15:24
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA12004/NA12004.chrom20.ILLUMINA.mosaik.CEU.low_coverage.20100804.bam.bai file 7899352 Wed Oct 27 18:31:23 2010
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA12004/NA12004.chrom20.ILLUMINA.mosaik.CEU.low_coverage.20100804.bam.bai file 166624 Thu Apr 14 15:24
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA12004/NA12004.ILLUMINA.mosaik.CEU.low_coverage.20100804.bam file 11091314322 Wed Oct 27 18:31:24 2010
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA18486 directory 308 Tue Dec 13 12:25:36 2011
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA18486/NA18486.ILLUMINA.mosaik.YRI.low_coverage.20101123.bam.bai file 8418040 Tue Jan 25 22:46:53 2011
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA18486/NA18486.ILLUMINA.mosaik.YRI.low_coverage.20101123.bam file 29068330549 Tue Jan 25 22:46:53 2011
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA18486/NA18486.chrom20.ILLUMINA.mosaik.YRI.low_coverage.20101123.bam.bai file 176848 Tue Jan 25 22:47
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA18486/NA18486.chrom20.ILLUMINA.mosaik.YRI.low_coverage.20101123.bam file 685641416 Tue Jan 25 22:47
ftp/phase1/technical/ncbi_varpipe_data/alignment/NA12045 directory 604 Tue Dec 13 12:24:58 2011
```



Finding Data

- Current tree file

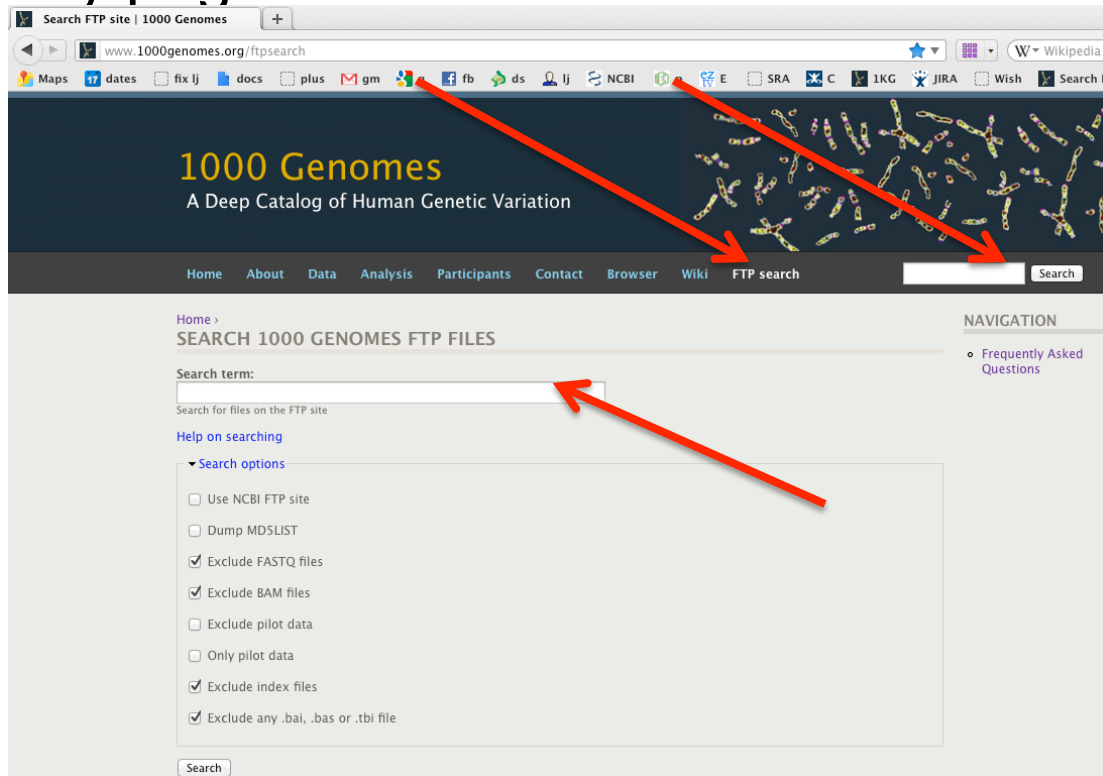
Description	Example
Relative Path	ftp/data/NA21091/alignment/ NA21091.chrom20.ILLUMINA.bwa.GIH.low_coverage. 20111114.bam
Type (file/directory)	file
Size in bytes	297914382
Last Updated Time Stamp	Thu Jan 26 00:26:52 2012
MD5 checksum	3fd679acc8c92cdc838aa0e5c1849d58

- Relative path does not contain the complete ftp path
- <ftp://ftp.1000genomes.ebi.ac.uk/vol1/>
- <ftp://ftp-trace.ncbi.nih.gov/1000genomes/>



Finding Data

- FTP search
- <http://www.1000genomes.org/ftpsearch>
- Search on the current.tree file
- Provides full ftp paths and md5 checksums
- Every page also has a website search box



The screenshot shows a web browser window at the URL www.1000genomes.org/ftpsearch. The page features a dark blue header with the text "1000 Genomes" and "A Deep Catalog of Human Genetic Variation". Below the header is a navigation menu with links for Home, About, Data, Analysis, Participants, Contact, Browser, Wiki, and FTP search. A search box is located in the top right corner of the navigation menu. The main content area is titled "SEARCH 1000 GENOMES FTP FILES" and contains a "Search term:" input field. Below the input field are search options, including checkboxes for "Use NCBI FTP site", "Dump MD5LIST", "Exclude FASTQ files", "Exclude BAM files", "Exclude pilot data", "Only pilot data", "Exclude index files", and "Exclude any .bai, .bas or .tbi file". A "Search" button is located at the bottom of the search options section. Red arrows point to the search box in the navigation menu and the search term input field.



Data Availability

- FTP site: <ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/>
 - Raw Data Files
- Web site: <http://www.1000genomes.org>
 - Release Announcements
 - Documentation
- Ensembl Style Browser: <http://browser.1000genomes.org>
 - Browse 1000 Genomes variants in Genomic Context
 - Variant Effect Predictor
 - Data Slicer
 - Other Tools



Exercises

- 1a. Find what Omni VCF files we have on our ftp site using the website ftp search. (Omni is a high throughput genotyping platform from Illumina on which all 1000 genomes samples are being genotyped)
- 1b. Find the most recent Omni VCF file on GRCh37 from the 31st January 2012
2. Use the Website search box found in the top right hand corner of all pages to find the FAQ question about getting subsections of VCF files.

Hints: You can use <http://www.1000genomes.org/ftpsearch> or <ftp://ftp.1000genomes.ebi.ac.uk/vol1/current.tree> and grep to answer 1a and b



Exercise Answers

1a. Put omni*vcf into the ftp site search box

Home >
SEARCH 1000 GENOMES FTP FILES

Search term:

Search for files on the FTP site

[Help on searching](#)

– ▶ [Search options](#)

RESULTS

52 files found

File
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20120131_omni_genotypes_and_intensities/Omni25_genotypes_2141_samples.b36.vcf.gz
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20120131_omni_genotypes_and_intensities/Omni25_genotypes_2141_samples.b37.vcf.gz



Exercise Answers, Finding Data

1b. Use `31*omni*vcf` to get results. This should return 2 files. One is labeled b36 and it in NCBI36 coordinates. The other is labeled b37 and is on GRCh37

Search term:
31*omni*vcf
Search for files on the FTP site

[Help on searching](#)

– [Search options](#)

Search

RESULTS

2 files found

File
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20120131_omni_genotypes_and_intensities/Omni25_genotypes_2141_samples.b36.vcf.gz
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/technical/working/20120131_omni_genotypes_and_intensities/Omni25_genotypes_2141_samples.b37.vcf.gz

Exercise Answers, Finding Data

2. Using the box that is in the top right hand corner of every page of 1000genomes.org with the term sub-section and vcf should return the appropriate FAQ page

[Home](#) > [Search](#) >

Content

Users

Enter your keywords:

vcf sub-section

Search

– [▶ Advanced search](#)

Search results

[How do I get a sub-section of a vcf file?](#)

... (Data Access, tabix, tools, variants, vcf) ...

FAQ Question – [ripley](#) – 2011-10-28 13:43 – 0 comments – 0 attachments

Update to 20110521 Release

... SNPs, short indels and large deletions. Files are in VCF format, The sites file represents all the autosomes and chrX but the ... as haploid. The .tbi file associated with each gzipped vcf file can be used to extract data for arbitrary chromosome subintervals.

... FAQ <http://www.1000genomes.org/faq/how-do-i-get-sub-section-vcf-file> The VCF File is in format 4.1 ...



The 1000 Genomes Browser

<http://browser.1000genomes.org>

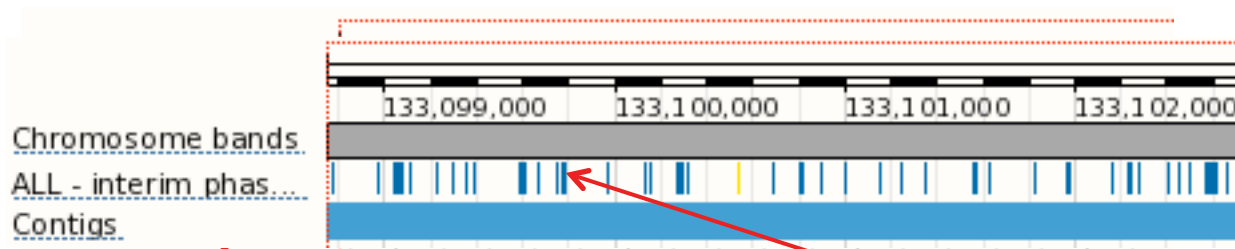
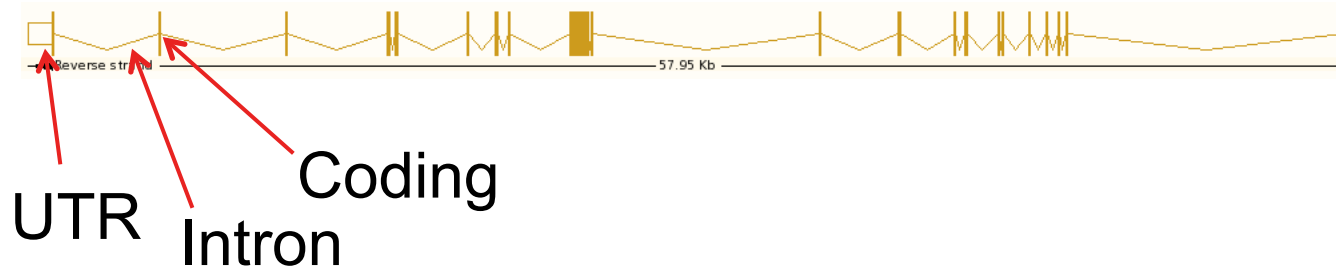


Caveats

- 1000 Genomes and Ensembl always define variants on the forward strand
- Allele strings are always reported ref/alt



Genes and SNPs



Line indicates number of SNPS

Each Line is One SNP

1000 Genomes
A Deep Catalog of Human Genetic Variation

Home About Data Analysis Participants Contact **Browser** Wiki FTP search Search

LATEST ANNOUNCEMENTS

WEDNESDAY OCTOBER 12, 2011

October 2011 Integrated Variant Set release #ICHG2011

This **October 2011** release represents an integrated set of variant calls and phased genotypes including SNPS, short INDELS and Deletions based on low coverage and exome sequencing data across 1092 individuals.

Our [FAQ](#) contains instructions on how to get [smaller subsections](#) of these files

Data access links: [EBI](#) / [NCBI](#)

Link to additional information: [README file](#)

THURSDAY JUNE 23, 2011

June 2011 Data Release

Genotypes for 1094 individuals for the [May 2011 snp calls](#) from the 20101123 sequence and alignment release of the 1000 genomes project has now been made. This release is based on the GRCh37 assembly of the human genome and is released in the format [VCF 4.0](#)

Our [FAQ](#) contains instructions on how to get [smaller subsections](#) of these files

Data access links: [EBI](#) / [NCBI](#)

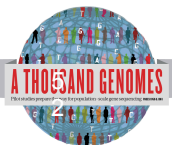
Link to additional information: [README file](#)

NAVIGATION

- [Frequently Asked Questions](#)

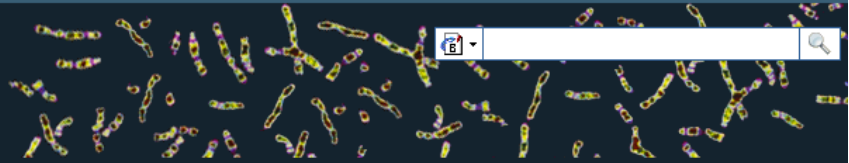
LINKS

- [All Project Announcements](#)
- [Sample and Project Information](#)
- [Media Archive](#)
- [Download the 1000 Genomes Pilot Paper](#)
- [Project Contacts](#)



1000 Genomes

A Deep Catalog of Human Genetic Variation



Tools | Help

Search 1000 Genomes

e.g. gene BRCA2 or Chromosome 6:133098746-133108745

Start Browsing 1000 Genomes data



[Browse Human](#) →
GRCh37

[Protein variations](#) →
View the consequences of sequence variation at the level of each protein in the genome.

[Individual genotypes](#) →
Show different individual's genotype, for a variant.

Browser update September 2011

based on interim Main project data from 20101123 for 1094 individuals and ensembl release 63. The data can be found on [the ftp site](#).

Please see www.1000genomes.org for more information about the data presented here and instructions for downloading the complete data set.

- [View sample data](#)

The 1000 Genomes Browser

Ensembl-based browser provides early access to 1000genomes data

In order to facilitate immediate analysis of the 1000 Genomes Project data by the whole scientific community, this browser (based on Ensembl) integrates the SNP calls from an [interim release 20101123](#). This data has been submitted to dbSNP, and once rsid's have been allocated, will be absorbed into the UCSC and Ensembl browsers according to their respective release cycles. Until that point any non rs SNP id's on this site are temporary and will NOT be maintained.

Links



[1000 Genomes](#) →
More information about the 1000 Genomes Project on the 1000 genomes main site.



[Pilot browser](#) →
This browser is based on Ensembl release 60 and represents the variant set analysed as part of [A map of human genome variation from population-scale sequencing](#), Nature 467, 1061.1073.



[Tutorial](#) →
The 1000 Genomes Browser Tutorial.

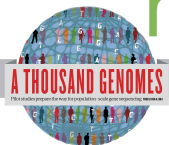
The 1000 Genomes Project is an international collaborative project described at www.1000genomes.org.

The 1000 Genomes Browser is based on Ensembl web code.

Ensembl is a joint project of EMBL-EBI  and the [Wellcome Trust Sanger Institute](#)



<http://browser.1000genomes.org>



Searching the Browser

- <http://browser.1000genomes.org>



1000 Genomes
A Deep Catalog of Human Genetic Variation

Search 1000 Genomes

PTPN22

e.g. gene BRCA2 or Chromosome 6:133098746-133108745

- Search for PTPN22
- Click 'Region in Detail'

You searched for 'PTPN22'

Gene or Gene Product

6 entrie(s) matched your search strings.

1. Gene: [ENSG00000134242](#) [\[Region in detail\]](#)
PTPN22 - protein tyrosine phosphatase, non-receptor type 22 (lymphoid) [Source:HGNC Symbol;Acc:9652]
2. Variations in gene [ENSG00000134242](#): [\[Variations in gene\]](#)
3. Transcript: [ENST00000359785](#) [\[Region in detail\]](#)
4. Peptide: [ENSP00000435176](#) [\[Region in detail\]](#)
PTPN22
5. Peptide: [ENSP00000352833](#) [\[Region in detail\]](#)
PTPN22
6. Peptide: [ENSP00000346621](#) [\[Region in detail\]](#)
PTPN22



Region in Detail

Ensembl genome browser 9: Homo sapiens - Region in detail - Chromosome 1: 114,356,433-114,414,381

browser.1000genomes.org/Homo_sapiens/Location/View?db=core;g=ENSG00000134242;r=1:114356433-114414381

1000 Genomes
A Deep Catalog of Human Genetic Variation

Human (GRCh37) Location: 1:114,356,433-114,414,381 Gene: PTPN22

Location-based displays

- Whole genome
- Chromosome summary
- Region overview
- Region in detail**
- Genetic Variation
 - Resequencing (20)
 - Linkage Data
 - Markers

Configure this page

Manage your data

Export data

Get VCF data

Bookmark this page

View in Ensembl

Chromosome 1: 114,356,433-114,414,381

Assembly excepti... chromosome 1

Assembly excepti...

H5CHR1_1_CTG31 H5CHR1_2_CTG31 H5CHR1_3_CTG31

Export Image

Region in detail help

Chromosome bands

Contigs.

Ensembl/Havana g...

Ensembl/Havana....

Gene Legend

- processed transcript
- pseudogene

1000 Genomes Homo sapiens version 63.37 (GRCh37) Chromosome 1: 113,885,408 - 114,885,407

merged Ensembl/Havana

Location: 1:114356433-114414381 Go

Gene: Go

Ensembl/Havana g...

Contigs.

Ensembl/Havana g...

PTPN22-002 protein coding

PTPN22-001 protein coding

PTPN22-004

Turning on Tracks

Configure this page



Configure Region Image | **Configure Overview Image** | Custom Data

Configure view

- Image options
 - Active tracks
 - Favourite tracks
 - Track order
 - Search results
 - 1000 Genomes (2/5)**
 - 1000 Genomes VCF (0/1)
 - Sequence (2/4)
 - Markers (1/1)
 - Genes (5/5)
 - Prediction transcripts (0/1)
 - Protein alignments (0/5)
 - Protein features (4/5)
 - cDNA/mRNA alignments (0/2)
 - RNA alignments (0/2)

1000 Genomes

- Enable/disable all tracks
- ALL - interim phase 1 - 1000 Genomes variations
- AFR - interim phase 1 - 1000 Genomes variations
- AMR - interim phase 1 - 1000 Genomes variations
- ASN - interim phase 1 - 1000 Genomes variations
- EUR - interim phase 1 - 1000 Genomes variations



Configure Region Image | **Configure Overview Image** | Custom Data

1000 Genomes
A Deep

Human (G...)
Location b...

- Whole g...
- Chromos...
- Region c...

Configure view

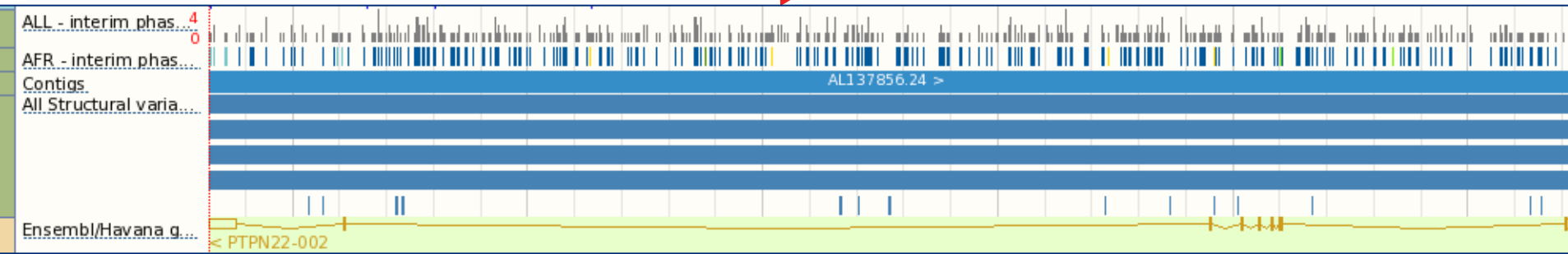
- Image options
- Active tracks
- Favourite tracks
- Track order
- Search results**
- 1000 Genomes (2/5)
- 1000 Genomes VCF (0/1)
- Sequence (1/4)
- Markers (0/1)
- Genes (5/5)
- Prediction transcripts (0/1)

Germline variation

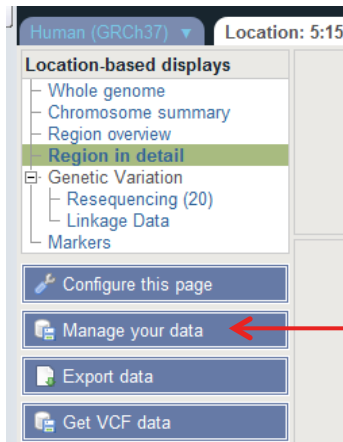
- Structural variants (all sources)
- DGVa structural variations

Key

- Track style



File upload to view with 1000 Genomes data



Human (GRCh37) Location: 5:15

Location-based displays

- Whole genome
- Chromosome summary
- Region overview
- Region in detail
- Genetic Variation
 - Resequencing (20)
 - Linkage Data
 - Markers

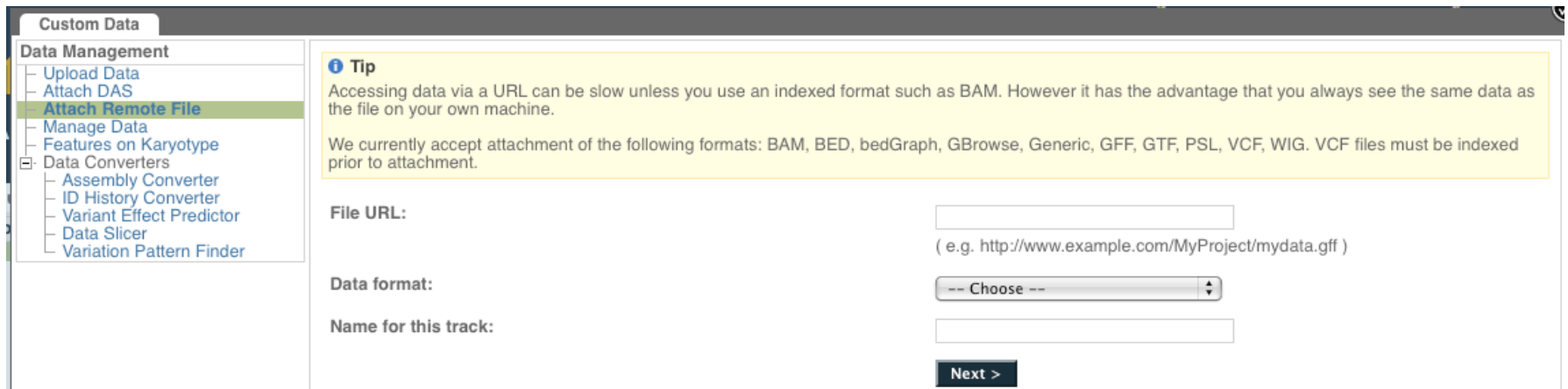
Configure this page

Manage your data

Export data

Get VCF data

Manage your data



Custom Data

Data Management

- Upload Data
- Attach DAS
- Attach Remote File
- Manage Data
- Features on Karyotype
- Data Converters
 - Assembly Converter
 - ID History Converter
 - Variant Effect Predictor
 - Data Slicer
 - Variation Pattern Finder

Tip
Accessing data via a URL can be slow unless you use an indexed format such as BAM. However it has the advantage that you always see the same data as the file on your own machine.

We currently accept attachment of the following formats: BAM, BED, bedGraph, GBrowse, Generic, GFF, GTF, PSL, VCF, WIG. VCF files must be indexed prior to attachment.

File URL:
(e.g. http://www.example.com/MyProject/mydata.gff)

Data format:

Name for this track:

Next >

- Supports popular file types:
 - BAM, BED, bedGraph, BigWig, GBrowse, Generic, GFF, GTF, PSL, VCF*, WIG
- * VCF must be indexed



Uploaded VCF

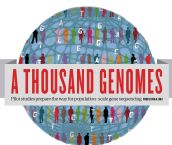
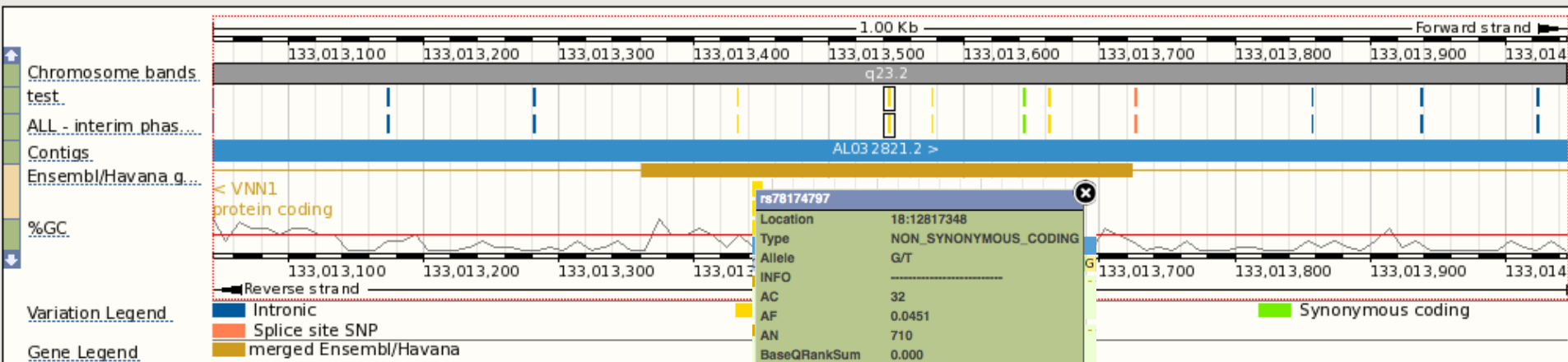

Example:

<ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20110521/>

ALL.wgs.phase1_release_v2.20101123.snps_indels_sv.sites.vcf.gz

Location:

Gene:



Uploaded BAM

Example:

http://ftp.1000genomes.ebi.ac.uk/vol1/ftp/data/HG01375/alignment/HG01375.mapped.ILLUMINA.bwa.CLM.low_coverage.20111114.bam



Gene View

Click the Gene tab, then 'Variation Table' or 'Variation Image'

Gene Tab

Human (GRCh37) Location: 1:114,362,205-114,362,276 Gene: PTPN22

Gene: PTPN22 (ENSG00000134242)

Description: protein tyrosine phosphatase, non-receptor type 22 (lymphoid) [Source:HGNC Symbol;Acc:9652]
Location: [Chromosome 1: 114,356,433-114,414,381](#) reverse strand.
Transcripts: There are 12 transcripts in this gene
Click the plus to show the transcript table

Variation Table [help](#)

Summary of variations in ENSG00000134242 by consequence type

Show entries

Number of variants	Type	Description
19 Show	Essential splice site	In the first 2 or the last 2 basepairs of an intron
9 Show	Stop gained	In coding sequence, resulting in the gain of a stop codon
0 -	Stop lost	In coding sequence, resulting in the loss of a stop codon
0 -	Complex in/del	Insertion or deletion that spans an exon/intron or coding sequence/UTR border
0 -	Frameshift coding	In coding sequence, resulting in a frameshift
160 Show	Non-synonymous coding	In coding sequence and results in an amino acid change in the encoded peptide sequence
65 Show	Splice site	1-3 bps into an exon or 3-8 bps into an intron
0 -	Partial codon	Located within the final, incomplete codon of a transcript whose end coordinate is unknown
83 Show	Synonymous coding	In coding sequence, not resulting in an amino acid change (silent mutation)

Get VCF data

Download as csv

Get in vcf format

Structural variation (in the Gene tab)

Human (GRCh37) Location: 1:114,356,433-114,414,381 Gene: PTPN22

Gene: **PTPN22 (ENSG00000134242)**

Description: protein tyrosine phosphatase, non-receptor type 22 (lymphoid) [Source:HGNC Symbol;Acc:9652]
 Location: [Chromosome 1: 114,356,433-114,414,381](#) reverse strand.
 Transcripts: There are 12 transcripts in this gene

Structural Variation

Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
PTPN22-001	ENST00000359785	3654	ENSP00000352833	807	Protein coding	CCDS863
PTPN22-002	ENST00000460620	1794	ENSP00000433141	179	Protein coding	-
PTPN22-004	ENST00000528414	3424	ENSP00000435176	752	Protein coding	-
PTPN22-006	ENST00000420377	2726	ENSP00000388229	795	Protein coding	-
PTPN22-007	ENST00000525799	2118	ENSP00000432674	668	Protein coding	-
PTPN22-201	ENST00000354605	2347	ENSP00000346621	691	Protein coding	CCDS864
PTPN22-202	ENST00000538253	2414	ENSP00000439372	563	Protein coding	-
PTPN22-008	ENST00000532224	2421	ENSP00000431249	135	Nonsense mediated decay	-
PTPN22-010	ENST00000529045	527	ENSP00000434932	92	Nonsense mediated decay	-
PTPN22-009	ENST00000534519	565	No protein product	-	Processed transcript	-
PTPN22-003	ENST00000484147	2258	No protein product	-	Retained intron	-
PTPN22-005	ENST00000469077	562	No protein product	-	Retained intron	-

All Structural varia...

Structural variants

Name	Chr:bp	Genomic size (bp)	Class	Source Study	Study description
nsv435973	1.81610203-127449918	45,839,716	SV	DGVA:nstd16	Database of Genomic Variants Archive: Korbel 2007 "Paired-end mapping reveals extensive structural variation in the human genome." PMID:17901297 [remapped from build NCBI36]
esv705	1.113157135-116741372	3,584,238	SV	DGVA:estd1	Database of Genomic Variants Archive: Redon 2006 "Global variation in copy number in the human genome." PMID:17122850 [remapped from build NCBI35]
esv21206	1.113862952-114901117	1,038,166	SV	DGVA:estd20	Database of Genomic Variants Archive: Conrad 2009 "Origins and functional impact of copy number variation in the human genome." PMID:19812545 [remapped from build NCBI36]
esv23869	1.113862952-114901117	1,038,166	SV	DGVA:estd20	Database of Genomic Variants Archive: Conrad 2009 "Origins and functional impact of copy number variation in the human genome." PMID:19812545 [remapped from build NCBI36]
CN_447814	1.114360689-114360713	25	CNV_PROBE	Affy	Copy Number Variation (CNV) probes from the Affymetrix Genome-Wide Human SNP Array 6.0

Structural Variants as boxes

Table



Variation Image

- Gene variation zoom

Human (GRC37) Location: 13:32,890,598-32,890,664 Gene: BRCA2

Gene-based displays

- Gene summary
- Splice variants (6)
- Supporting evidence
- Sequence
- External references
- Regulation
- Genetic Variation
- Variation Table
- Variation Image**
- External Data
- ID History
- Gene history

Gene: BRCA2a (ENSG00000139618)

Description: breast cancer 2, early onset [Source:HGNC Symbol;Acc:1101]

Location: Chromosome 13: 32,889,811-32,973,805 forward strand.

Transcripts: There are 6 transcripts in this gene

Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
BRCA2-001	ENST00000380152	10930	ENSP00000369407	3418	Protein coding	CCDS9344
BRCA2-003	ENST00000530893	2009	ENSP00000435689	602	Protein coding	-
BRCA2-201	ENST00000544455	10984	ENSP00000439202	3418	Protein coding	CCDS9344
BRCA2-002	ENST00000470094	842	ENSP00000434988	188	Nonsense mediated decay	-
BRCA2-005	ENST00000530792	495	ENSP00000433188	64	Nonsense mediated decay	-
BRCA2-006	ENST00000533776	523	No protein product	-	Retained intron	-

Transcript and Gene level displays

In 1000 Genomes we provide displays at two levels:

- Transcript views which provide information specific to an individual transcript such as the cDNA and CDS sequences and protein domain annotation.
- Gene views which provide displays for data associated at the gene level such as orthologues, paralogues, regulatory regions and splice variants.

This view is a gene level view. To access the transcript level displays select a Transcript ID in the table above and then navigate to the information you want using the menu at the left hand side of the page. To return to viewing gene level information click on the Gene tab in the menu bar at the top of the page.



Configuring the display

Tip: use the 'Configure this page' link on the left to customise the protein domains and types of variations displayed above. Please note the default 'Context' settings will probably filter out some intronic SNPs. 5 of the 20 variations in this region have been filtered out by the Source, Class and Type filters. None of the intronic variations are removed by the Context filter.

1000 Genomes release 8 - May 2011 © EBI

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Transcript Tab: Variations

Effect on Protein:

- SIFT
- PolyPhen

1000 Genomes A Deep Catalog of Human Genetic Variation

Human (GRCh37) Location: 1:114,359,433-114,414,381 Gene: PTPN22 Transcript: PTPN22-001

Transcript: PTPN22-001 (ENST00000359785)

Description: protein tyrosine phosphatase, non-receptor type 22 (lymphoid) [Source:HGNC Symbol;Acc:9652]
 Location: Chromosome 1:114,359,433-114,414,381 reverse strand.
 Gene: This transcript is a product of gene [ENSG00000134242](#) - There are 12 transcripts in this gene

Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
PTPN22-001	ENST00000359785	3654	ENSP00000352833	807	Protein coding	CCDS8883
PTPN22-002	ENST00000460620	1794	ENSP00000433141	179	Protein coding	-
PTPN22-004	ENST00000528414	3424	ENSP00000435176	752	Protein coding	-
PTPN22-006	ENST00000420377	2726	ENSP00000388229	795	Protein coding	-
PTPN22-007	ENST00000525799	2118	ENSP00000432674	668	Protein coding	-
PTPN22-201	ENST00000354605	2347	ENSP00000346621	691	Protein coding	CCDS884
PTPN22-202	ENST00000538253	2414	ENSP00000439372	563	Protein coding	-
PTPN22-008	ENST00000532224	2421	ENSP00000431249	135	Nonsense mediated decay	-
PTPN22-010	ENST00000529045	527	ENSP00000434932	92	Nonsense mediated decay	-
PTPN22-009	ENST00000534519	565	No protein product	-	Processed transcript	-
PTPN22-003	ENST00000484147	2258	No protein product	-	Retained intron	-
PTPN22-005	ENST00000469077	562	No protein product	-	Retained intron	-

Transcript and Gene level displays

Viewers in 1000 Genomes are separated into gene based views and transcript based views according to which level the information is more appropriately associated with. This view is a transcript level view. To flip between the two sets of views you can click on the Gene and Transcript tabs in the menu bar at the top of the page.

Variations [help](#)

Download view as CSV

Residue	Variation ID	Variation type	Alleles	Ambiguity code	Residues	Codons	SIFT	PolyPhen
16	rs74163639	Synonymous coding	G/A	R	S	AGC, AGT	-	-
49	rs61745743	Synonymous coding	A/G	R	A	GCT, GCC	-	-
71	rs74163642	Non-synonymous coding	A/G	R	V, A	GTA, GCA	deleterious	probably damaging
141	rs115552198	Non-synonymous coding	G/A	R	R, C	CGC, TGC	deleterious	probably damaging
177	1KG_1_114399013	Synonymous coding	C/T	Y	K	AAG, AAA	-	-
183	rs34590413	Stop gained	G/A	R	R, *	CGA, TGA	-	-
201	rs74163647	Non-synonymous coding	G/A	R	S, F	TCT, TTT	deleterious	probably damaging
206	rs61738614	Non-synonymous coding	A/C	M	L, R	CTT, CGT	deleterious	probably damaging
232	rs78195073	Synonymous coding	T/C	Y	G	GGA, GGG	-	-
247	rs35910094	Synonymous coding	T/G	K	L	CTA, CTC	-	-
263	rs33996649	Non-synonymous coding	C/T	Y	R, Q	CGG, CAG	tolerated	benign
266	rs72650670	Non-synonymous coding	G/A	R	R, W	CGG, TGG	deleterious	probably damaging
277	rs72483511	Stop gained, Splice site	C/A	M	E, *	GAA, TAA	-	-
324	rs113984534	Synonymous coding	A/G	R	Y	TAT, TAC	-	-
366	rs74163654	Synonymous coding	C/T	Y	E	GAG, GAA	-	-
370	rs72650671	Non-synonymous coding	G/T	K	H, N	CAC, AAC	deleterious	possibly damaging
388	rs77913785	Non-synonymous coding	G/T	K	D, E	GAC, GAA	deleterious	benign
413	1KG_1_114380784	Non-synonymous coding	T/G	K	Q, P	CAA, CCA	deleterious	benign
414	1KG_1_114380780	Synonymous coding	A/G	R	S	AGT, AGC	-	-
427	rs112873647	Non-synonymous coding	-/ATT	-	-, N	-, AAT	-	-
444	rs74163655	Non-synonymous coding	T/A	W	I, L	ATA, TTA	tolerated	benign
447	rs112191110	Non-synonymous coding	G/A	R	T, I	ACC, ATC	deleterious	probably damaging
452	rs56174946	Synonymous coding	A/G	R	F	TTT, TTC	-	-
456	rs72650672	Non-synonymous coding	G/C	S	Q, E	CAG, GAG	deleterious	possibly damaging
477	rs74163656	Synonymous coding	A/G	R	L	CAT, CAC	-	-

Start again- search for a variation (rs31685)

1000 Genomes
A Deep Catalog of Human Genetic Variation

Search 1000 Genomes

rs31685

e.g. gene BRCA2 or Chromosome 6:133098746-133108745

- The Variation tab- left hand links take you to more information

Human (GRCh37) Location: 5:159,283,673-159,284,673 Variation: rs31685

Variation displays

- Flanking sequence
- Gene/Transcript (1)
- Population genetics (117)
- Individual genotypes (4343)
- Genomic context
- Phenotype Data
- Phylogenetic Context
- External Data

Variation: rs31685

Variation class SNP ([rs31685](#) source [dbSNP_132](#) - Variants (including SNPs and indels) imported from dbSNP [<http://www.ncbi.nlm.nih.gov/projects/SNP/>])

Synonyms Affy GeneChip 100K Array SNP_A-1683078
Affy GeneChip 500K Array SNP_A-4265358
Affy GenomeWideSNP_6.0 AFFY_6_1M_SNP_A-4265358, SNP_A-4265358
dbSNP [rs17746160](#), [rs60752908](#), [rs713581](#), [rs58941657](#)
ENSEMBL ENSSNP12948257, ENSSNP9597299

Present in This feature is present in **1000 genomes** and 3 other sets - click the plus to show all sets

Alleles G/A (Ambiguity code: R)

Ancestral allele A

Location This feature maps to 5:159284173 (forward strand) | [View in location tab](#)

Validation status Proven by **cluster, frequency, doublehit, 1000Genome HapMap variant**

HGVS names This feature has 2 HGVS names - click the plus to show

- Population

1000 Genomes

A Deep Catalog of Human Genetic Variation

Human (GRCh37) Location: 6:74,125,388-74,126,388 Variation: rs311685

Variation: rs311685

Variation displays

- Flanking sequence
- Gene/Transcript (3)
- Population genetics (46)**
- Individual genotypes (2769)
- Genomic context
- Phenotype Data
- Phylogenetic Context
- External Data

Variation class SNP (rs311685 source dbSNP_132 - Variants (including SNPs and indels) imported from dbSNP [http://www.ncbi.nlm.nih.gov/projects/SNP/])

Synonyms Affy GeneChip 100K Array SNP_A-1679873
Affy GenomeWideSNP_6.0 AFFY_6_1M_SNP_A-8668494, SNP_A-8668494
dbSNP rs58378291, rs17756820, rs52794514, rs524803, rs3173186, rs11567000, rs17421786
ENSEMBL ENSNP9062281
Illumina_Human1M-duoV3 rs311685
Uniprot VAR_057235

Present in 1000 genomes - High coverage - Trios (1000 genomes - High coverage - Trios - CEU, 1000 genomes - High coverage - Trios - YRI), 1000 genomes - Low coverage (1000 genomes - Low coverage - CEU, 1000 genomes - Low coverage - CHB+JPT, 1000 genomes - Low coverage - YRI), ALL - interim phase 1 - 1000 Genomes (AFR - interim phase 1 - 1000 Genomes, AMR - interim phase 1 - 1000 Genomes, ASN - interim phase 1 - 1000 Genomes, EUR - interim phase 1 - 1000 Genomes), ENSEMBL:Venter,HapMap

Alleles A/G (Ambiguity code: R)

Ancestral allele A

Location This feature maps to 6:74125888 (forward strand) | [View in location tab](#)

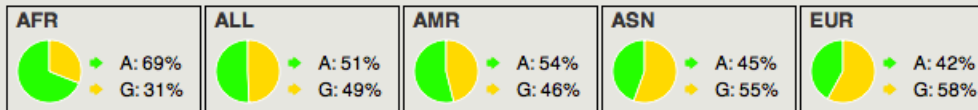
Validation status Proven by cluster, frequency, doublehit, 1000Genome HapMap variant

HGVS names This feature has 4 HGVS names - click the plus to show

[Population genetics help](#)



1000 genomes alleles frequencies



1000 genomes

Show/hide columns Filter

Population	Alleles A	Alleles G	Genotypes A/A	Genotypes A/G	Genotypes G/G	Count
1000GENOMES:AFR	0.689	0.311	0.463	0.451	0.085	114
1000GENOMES:ALL	0.507	0.493	0.269	0.477	0.254	294
1000GENOMES:AMR	0.539	0.461	0.293	0.492	0.215	53
1000GENOMES:ASN	0.446	0.554	0.199	0.493	0.308	57
1000GENOMES:EUR	0.421	0.579	0.184	0.475	0.341	70

1000 genomes pilot

Show/hide columns Filter

Population	ssID	Submitter	Alleles A	Alleles G	Count
1000GENOMES:pilot 1 CEU low coverage panel	ss233534774	1000GENOMES	0.458	0.542	
1000GENOMES:pilot 1 CHB+JPT low coverage panel	ss240577229	1000GENOMES	0.400	0.600	
1000GENOMES:pilot 1 YRI low coverage panel	ss222470667	1000GENOMES	0.729	0.271	

Phenotype for one variant

2,027 Variation: rs420259

Variation: rs420259

Variation class (source [dbSNP](#))

Synonyms Affy GeneChip 500K Array SNP_A-2248415
Affy GenomeWideSNP_6.0 SNP_A-2248415

Alleles A/G (Type: Unknown)
Ancestral allele: G

Location This feature maps to 1 genomic location(s). [show locations](#)

« Context **Phenotype Data** Evolutionary or Phylogenetic

Disease/Trait	Source	Study	Associated Gene(s)	Strongest risk allele	Associated variant	P value
Bipolar Disorder (BD)	[EGA]				rs420259	
	[NHGRI_GWAS_catalog]	pubmed/17554300	PALB2,NDUFAB1,DCTN5	rs420259-A	rs420259	6.00E-08

EGA

<http://www.ebi.ac.uk/ega>

NHGRI

<http://www.genome.gov/gwastudies/>

Open GWAS DB

<http://www.biomedcentral.com/1471-2350/10/6>

COSMIC

<http://www.sanger.ac.uk/genetics/CGP/cosmic/>

OMIM

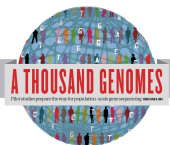
<http://www.ncbi.nlm.nih.gov/omim>

HGMD-Public

<http://www.hgmd.cf.ac.uk/ac/index.php>

UniProt

<http://www.uniprot.org/>



Coming Soon Ensembl 65

e!Ensembl BLAST/BLAT | BioMart | Tools | Downloads | Help & Documentation | Blog | Mirrors Login · Register

Human (GRCh37) | Location: 9:22,125,003-22,126,003 | Variation: rs1333049

Variation displays

- Explore this variation
- Genomic context
 - Gene/Transcript (2)
- Population genetics (28)
- Individual genotypes (1737)
- Linkage disequilibrium
- Phenotype Data (8)
- Phylogenetic Context (4)
- Flanking sequence
- External Data

rs1333049 SNP

Source [dbSNP 134](#) - Variants (including SNPs and indels) imported from [dbSNP](#)

Alleles Reference/Alternative: **G/C** | Ancestral: **C** | Ambiguity code: **S** | MAF: **0.40** (C)

Location Chromosome **9:22125503** (forward strand) | [View in location tab](#)

Validation status This variation is validated by **1000 Genomes**, **HapMap** and also cluster, doublehit, frequency, precious, submitter

Synonyms This feature has **7** synonyms - click the plus to show

HGVS name [g.22125503G>C](#)

[Configure this page](#)
[Manage your data](#)
[Export data](#)
[Bookmark this page](#)

Explore this variation [help](#)

- Genomic context**
- Gene / Transcript**
- Population genetics**
- Individual genotypes**
- Linkage disequilibrium**
- Phenotype data**
- Phylogenetic context**
- Flanking sequence**

Help with variations

YouTube videos

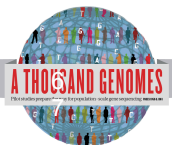
- [SNPs and other Variations - 1 of 2](#)
- [SNPs and other Variations - 2 of 2](#)
- [Clip: Genome Variation](#)
- [BioMart: Variation IDs to HGNC Symbols](#)

Reference materials

- [Ensembl variation data: background and terminology](#)
- [Variation Quick Reference card](#)

Additional resources

- [Accessing variation data with the Variation API](#)
- [Genomes and SNPs in Malaria](#)



Should arrive in May



Exercise, Browser

3. Find the variant rs45562238 using <http://browser.1000genomes.org>.
4. In what 1000 Genomes Super Population is this variant detected?
5. What are its global allele frequencies in the 1000 Genomes Data set?
6. In which gene is the variant found?



Exercise Answers, Browser

3



SNP

1 entrie(s) matched your search strings.

1. dbSNP SNP: [rs45562238](#)

Interpro Domain

0 entrie(s) matched your search strings.

Exercise Answers

4. In what 1000 Genomes Super Population is this variant detected?

American and European

5. What are its global allele frequencies in the 1000 Genomes Data set?

0.02 is the global allele frequency, this is also the American Allele Frequency but it rises to 0.04 in the Europeans. The absence of Asians or Africans in this chart means that the variant was not found in any of our Asian or African individuals.

6. In which gene is the variant found?

ENSG00000112299, Vanin 1

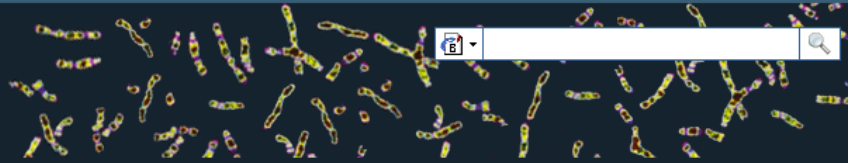


1000 Genomes Tools



1000 Genomes

A Deep Catalog of Human Genetic Variation



Tools | Help

Search 1000 Genomes

e.g. gene BRCA2 or Chromosome 6:133098746-133108745

Start Browsing 1000 Genomes data



[Browse Human](#) →
GRCh37

[Protein variations](#) →
View the consequences of sequence variation at the level of each protein in the genome.

[Individual genotypes](#) →
Show different individual's genotype, for a variant.

Browser update September 2011

based on interim Main project data from 20101123 for 1094 individuals and ensembl release 63. The data can be found on [the ftp site](#).

Please see www.1000genomes.org for more information about the data presented here and instructions for downloading the complete data set.

- [View sample data](#)

The 1000 Genomes Browser

Ensembl-based browser provides early access to 1000genomes data

In order to facilitate immediate analysis of the 1000 Genomes Project data by the whole scientific community, this browser (based on Ensembl) integrates the SNP calls from an [interim release 20101123](#). This data has been submitted to dbSNP, and once rsid's have been allocated, will be absorbed into the UCSC and Ensembl browsers according to their respective release cycles. Until that point any non rs SNP id's on this site are temporary and will NOT be maintained.

Links



[1000 Genomes](#) →
More information about the 1000 Genomes Project on the 1000 genomes main site.



[Pilot browser](#) →
This browser is based on Ensembl release 60 and represents the variant set analysed as part of [A map of human genome variation from population-scale sequencing](#), Nature 467, 1061.1073.

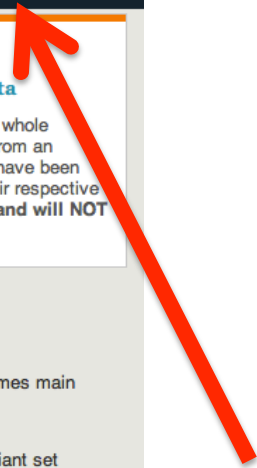


[Tutorial](#) →
The 1000 Genomes Browser Tutorial.

The 1000 Genomes Project is an international collaborative project described at www.1000genomes.org.

The 1000 Genomes Browser is based on Ensembl web code.

Ensembl is a joint project of EMBL-EBI  and the [Wellcome Trust Sanger Institute](#)



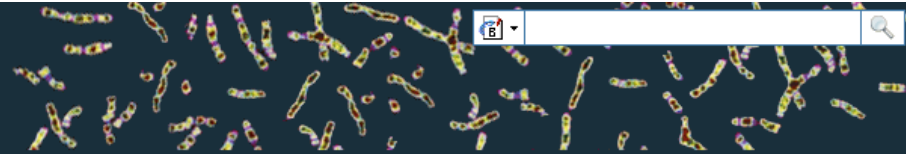
<http://browser.1000genomes.org>



Tools page

1000 Genomes

A Deep Catalog of Human Genetic Variation



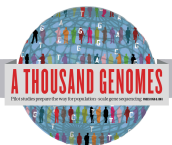
Tools | Help

We provide a number of ready-made tools for processing your data. At the moment, small datasets can be uploaded to our servers and processed online; for larger datasets, we provide an API script that can be downloaded (you will also need to [install our Perl API](#) to use these).

In the near future we aim to offer an intermediate service, whereby medium-to-large data sets can be submitted to a queue, similar to BLAST.

Currently available:

Tool	Description		
Assembly converter	Map your data to the current assembly. Accepted file formats: GFF , GTF , BED , PSL . N.B. Export is currently in GFF only	Online version	API script
ID History converter	Convert a set of Ensembl IDs from a previous release into their current equivalents.	Online version (max 30 ids)	API script
Variant Effect Predictor	(Formerly SNP Effect Predictor). Upload a set of SNPs in our standard format and export a file containing consequence types. Uploaded tracks can also be viewed on Location pages.	Online version (max 750 SNPs)	API script
Data Slicer	Get a subset of data from a BAM or VCF file.	Online version (max 10K region)	
Variation Pattern Finder	Identify variation patterns in a chromosomal region of interest for different individuals. Only variations with functional significance such non-synonymous coding, splice site will be reported by the tool. Click here for more extensive documentation.	Online version	API script
VCF to PED converter	The VCF to PED converter allows users to parse a vcf file to create a linkage pedigree file (.ped) and a marker information file, which together may be loaded into Id visualization tools like Haploview. Click here for more extensive documentation.	Online version	API script



Data Slicer

- Remote Bam or VCF files
- Genomic Location
- Returns subsection of given file
- VCF files can be subset by
 - Population
 - Individual
 - Must provide a panel file to map individual to population



Data Slicing

Custom Data

Data Management

- Upload Data
- Attach DAS
- Attach Remote File
- Manage Data
- Features on Karyotype
- Data Converters
 - Assembly Converter
 - ID History Converter
 - Variant Effect Predictor
 - **Data Slicer**
 - Variation Pattern Finder
 - VCF to PED converter

i Data Slicer:

When slicing a VCF or BAM file, both the data file and its index file should be present on the web server and named correctly. The VCF file should have a ".vcf.gz" extension, and the index file should have a ".vcf.gz.tbi" extension, E.g: MyData.vcf.gz, MyData.vcf.gz.tbi. The BAM file should have a ".bam" extension, and the index file should have a ".bam.bai" extension, E.g: MyData.bam, MyData.bam.bai

Click [here](#) for more extensive documentation.

Upload files

VCF File URL:

```
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr1.phase1.projectConsensus.genotypes.vcf.gz
```

[Clear box](#)

e.g. ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr1.phase1.projectConsensus.genotypes.vcf.gz

Region:

```
6:46620015-46620998
```

e.g. 1:1-50000

Use VCF filters (this doesn't apply to BAM files):

- None
- By individual(s)
- By population(s) *

(to filter by populations please provide URL to a Sample-Population Mapping File in the box below)

Sample-Population Mapping File URL:

```
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel
```

[Clear box](#)

e.g. ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel

Next >



Data Slicer Example screens

VCF filter by population(s)

Select one or more populations from the scrollable list:

- ASW
- CEU
- CHB
- CHS
- CLM
- FIN
- GBR
- IBS
- JPT
- LWK

< Back

Next >

Thank you - your VCF file [\[filtered_6.31830969-31846823.ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz\]](#) [Size: 7529] has been generated. Right click on the file name and choose "Save link as .." from the menu

Preview

```
##fileformat=VCFv4.0
##source=BCM:SNPTools:hapfuse
##reference=1000Genomes-NCBI37
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
##FORMAT=<ID=AP,Number=2,Type=Float,Description="Allelic Probability, P(Allele=1)"/>
##source_20120302.1=/nfs/public/rw/ensembl/vcftools/bin/vcf-subset -c HG01112,HG01113
#CHROM POS ID REF ALT QUAL FILTER INFO FORMAT HG01112 I
6 31831159 rs3869144 C T 100 PASS . C
```

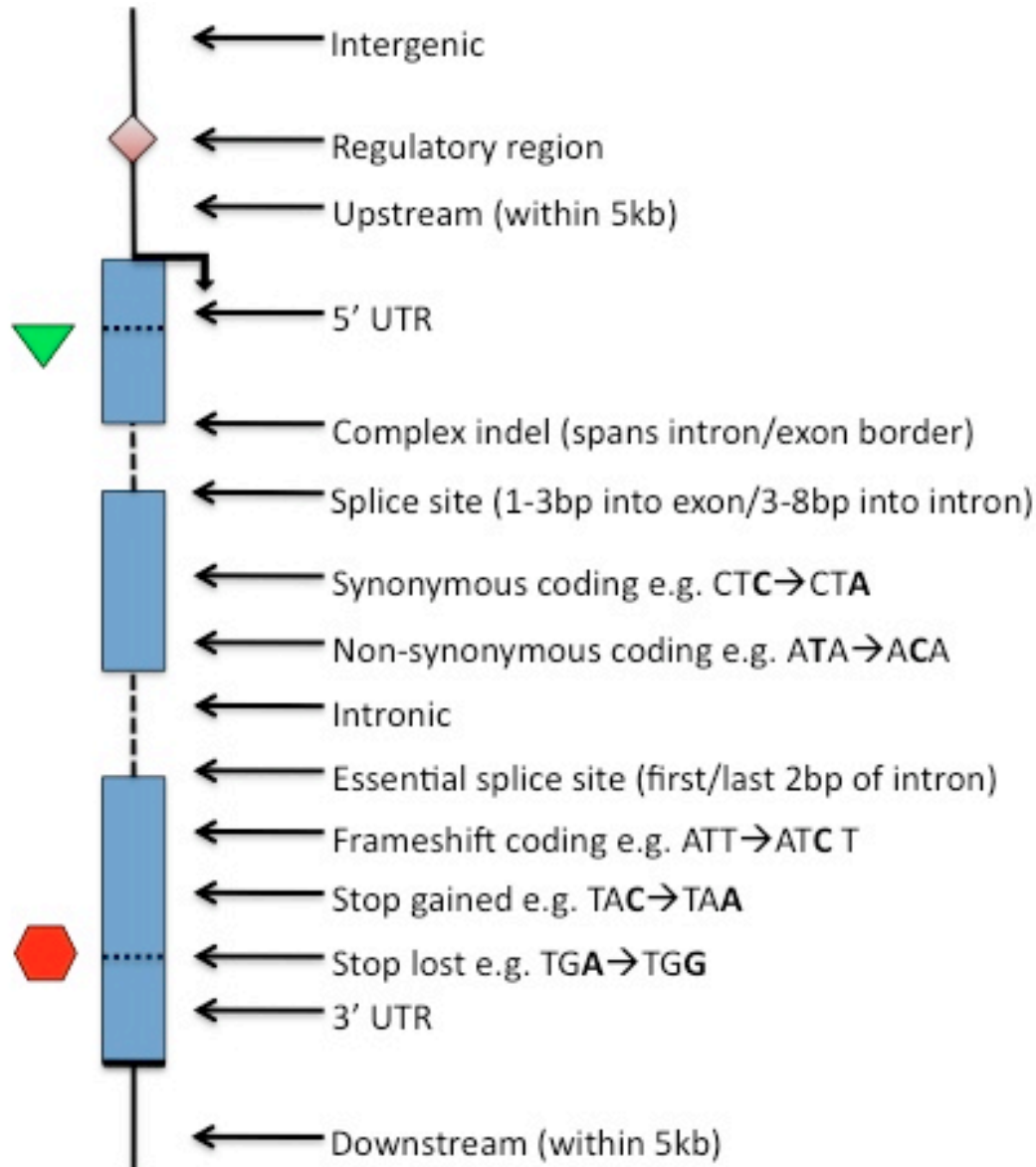


Variant Effect Predictor

- Predicts Functional Consequences of Variants
- Both Web Front end and API script
- Can provide
 - sift/polyphen/condel consequences
 - Refseq gene names
 - HGVS output
- Can run from a cache as well as Database
- Convert from one input format to another
- Script available for download from:
- ftp://ftp.ensembl.org/pub/misc-scripts/Variant_effect_predictor/
- http://browser.1000genomes.org/Homo_sapiens/UserData/UploadVariations



Variant Effect Predictor



Others: Within non-coding gene, Within mature miRNA, NMD transcript

Data Management

- Upload Data
- Attach DAS
- Attach Remote File
- Manage Data
- Features on Karyotype
- Data Converters
 - Assembly Converter
 - ID History Converter
 - Variant Effect Predictor**
 - Data Slicer
 - Variation Pattern Finder

Variant Effect Predictor:

This tool takes a list of variant positions and alleles, and predicts the effects of each of these on overlapping transcripts and regulatory regions annotated in Ensembl. The tool accepts substitutions, insertions and deletions as input, uploaded as a list of [tab separated values](#), [VCF](#) or Pileup format input.

Upload is limited to 750 variants; lines after the limit will be ignored. Users with more than 750 variations can split files into smaller chunks, use the standalone [perl script](#) or the [variation API](#). See also [full documentation](#)

Input file

Species:

Human (Homo sapiens): GRCh37

Name for this upload (optional):

Paste file:

Upload file:

Choose File no file selected

or provide file URL:

Input file format:

Ensembl default

Options

Get regulatory region consequences:

Type of consequences to display:

Ensembl terms

Check for existing co-located variants:

Yes

Return results for variants in coding regions only:

Show HGNC identifier for genes where available:

Show Ensembl protein identifiers where available:

Show HGVS identifiers for variants where available:

No

Non-synonymous SNP predictions (human only)

SIFT predictions:

No

PolyPhen predictions:

No

Condel consensus (SIFT/PolyPhen) predictions:

No

Frequency filtering of existing variants (human only)

Filter variants by frequency:

NB: Enabling frequency filtering may be very slow for large datasets

Filter: Exclude variants with MAF greater than 0.1 in any 1KG low coverage population

Next >

Variation Effect Predictor Output

6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000480384	Transcript	UPSTREAM	-	-	-	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000491768	Transcript	UPSTREAM	-	-	-	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000375631	Transcript	UPSTREAM	-	-	-	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000479533	Transcript	UPSTREAM	-	-	-	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000229729	Transcript	NON_SYNONYMOUS_CODING	1625	1604	535	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious; PolyPhen=probably_damaging
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000375562	Transcript	NON_SYNONYMOUS_CODING	1544	1478	493	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious; PolyPhen=possibly_damaging
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000544672	Transcript	NON_SYNONYMOUS_CODING	1673	1376	459	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious; PolyPhen=probably_damaging
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000487680	Transcript	UPSTREAM	-	-	-	-	-	1KG 6 31833357	-
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000414427	Transcript	DOWNSTREAM	-	-	-	-	-	1KG 6 31833357	-
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000479777	Transcript	DOWNSTREAM	-	-	-	-	-	1KG 6 31833357	-
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000475563	Transcript	DOWNSTREAM	-	-	-	-	-	1KG 6 31833357	-
0204386	ENST00000491768	Transcript	UPSTREAM	-	-	-	-	-	-	-	-	1KG 6 31833357	-
0204386	ENST00000375631	Transcript	UPSTREAM	-	-	-	-	-	-	-	-	1KG 6 31833357	-
0204386	ENST00000479533	Transcript	UPSTREAM	-	-	-	-	-	-	-	-	1KG 6 31833357	-
0204385	ENST00000229729	Transcript	NON_SYNONYMOUS_CODING	1625	1604	535	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious; PolyPhen=probably_damaging			
0204385	ENST00000375562	Transcript	NON_SYNONYMOUS_CODING	1544	1478	493	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious; PolyPhen=possibly_damaging			
0204385	ENST00000544672	Transcript	NON_SYNONYMOUS_CODING	1673	1376	459	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious; PolyPhen=probably_damaging			
0204385	ENST00000487680	Transcript	UPSTREAM	-	-	-	-	-	-	-	-	1KG 6 31833357	-
0204385	ENST00000414427	Transcript	DOWNSTREAM	-	-	-	-	-	-	-	-	1KG 6 31833357	-
0204385	ENST00000479777	Transcript	DOWNSTREAM	-	-	-	-	-	-	-	-	1KG 6 31833357	-
0204385	ENST00000475563	Transcript	DOWNSTREAM	-	-	-	-	-	-	-	-	1KG 6 31833357	-



Variation Pattern Finder

- Remote or local tabix indexed VCF input
- Discovers patterns of Shared Inheritance
- Variants with functional consequences considered by default
- Web output with CSV and Excel downloads
- [http://browser.1000genomes.org/Homo_sapiens/
UserData/VariationsMapVCF](http://browser.1000genomes.org/Homo_sapiens/UserData/VariationsMapVCF)



Variation Pattern Finder

Variation Pattern Finder:

The Variation Pattern Finder allows one to look for patterns of shared variation between individuals in the same vcf file. The finder looks for distinct variation combinations within the region, as well as individuals associated with each variation combination pattern. Only variants which have potentially functional consequences are considered, both intergenic and intronic snps are excluded. Click [here](#) for more extensive documentation.

The search will be performed on any VCF file you provided. It should be a URL for the file location. Please refer to <http://vcftools.sourceforge.net/specs.html> for VCF format specification. A URL for the latest VCF file for variation calls and genotypes released by the 1000 Genomes Project is displayed as an example below the input box. A mapping file between individual sample and population is required as well. The latest mapping file between individual sample and population released by the 1000 Genomes Project is displayed as well below the input box.

Upload files

VCF File URL:

```
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz
```

[Clear box](#)

e.g. ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz

Sample-Population Mapping File URL:

```
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel
```

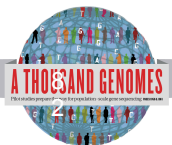
[Clear box](#)

e.g. ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel

Region:

e.g. 6:46620015-46620998

Next >



Variation Pattern Finder Output

Variation Pattern Finder

Export data: [CSV](#) [Excel](#)

Go to collapsed view

CEU	CI Freq	rs12661281:T/A	6:31843711:C/T	6:31845340:C/T	rs2075798:C/A
		6:31842598	6:31843711	6:31845340	6:31846741
		DING:N/S ENST00000229729 NON_SYNONYMOUS_CODING:D/V	ENST00000229729 SPLICE_SITE	ENST00000544672 SPLICE_SITE	ENST00000229729 NON_SYNONYMOU
		DING:N/S ENST00000544672 NON_SYNONYMOUS_CODING:D/V	ENST00000375562 SPLICE_SITE	ENST00000544672 5PRIME_UTR	ENST00000375562 NON_SYNONYMOU
		DING:N/S ENST00000414427 NON_SYNONYMOUS_CODING:D/V	ENST00000544672 SPLICE_SITE		ENST00000414427 NON_SYNONYMOU
			ENST00000414427 SPLICE_SITE		
			ENST00000465707 SPLICE_SITE		
			ENST00000462671 SPLICE_SITE		
NA12872, NA07000 and 1 other(s)	N 0.032	TIA	CIC	CIC	CIC
NA12874, NA12717	N 0.028	TIT	CIC	CIC	AIC
NA07346	N 0.027	TIT	CIC	CIC	CIA
	N 0.027	TIT	CIC	CIC	CIC
NA10851, NA12342 and 5 other(s)	N 0.024	AIT	CIC	CIC	CIC
NA12058, NA12273 and 1 other(s)	N 0.020	AIA	CIC	CIC	CIC
	N 0.018	TIT	CIC	CIC	CIC
	N 0.015	AIT	CIC	CIC	CIA
	N 0.014	TIT	CIC	CIC	AIA
	N 0.013	TIT	CIC	CIC	CIC
NA10847	N 0.011	TIA	CIC	CIC	AIC
NA12286, NA11892 and 2 other(s)	N 0.009	TIT	CIC	CIC	CIC

VCF to PED

- LD Visualization tools like Haploview require PED files
- VCF to PED converts VCF to PED
- Will a file divide by individual or population
- http://browser.1000genomes.org/Homo_sapiens/UserData/Haploview



VCF to PED

Custom Data

Data Management

- Upload Data
- Attach DAS
- Attach Remote File
- Manage Data
- Features on Karyotype
- Data Converters
 - Assembly Converter
 - ID History Converter
 - Variant Effect Predictor
 - Data Slicer
 - Variation Pattern Finder
 - VCF to PED converter**

VCF to PED converter:

When providing a VCF file, both the data file and its index file should be present on the web server and named correctly. The VCF file should have a ".vcf.gz" extension, and the index file should have a ".vcf.gz.tbi" extension, E.g: MyData.vcf.gz, MyData.vcf.gz.tbi. Click [here](#) for more extensive documentation.

Upload files

VCF File URL:

`ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz`

[Clear box](#)

e.g. `ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz`

Sample-Population Mapping File URL:

`ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel`

[Clear box](#)

e.g. `ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel`

Region:

e.g. 6:46620015-46620998

[Next >](#)



VCF to PED example output

VCF filter by population(s)

Select one or more populations from the scrollable list:

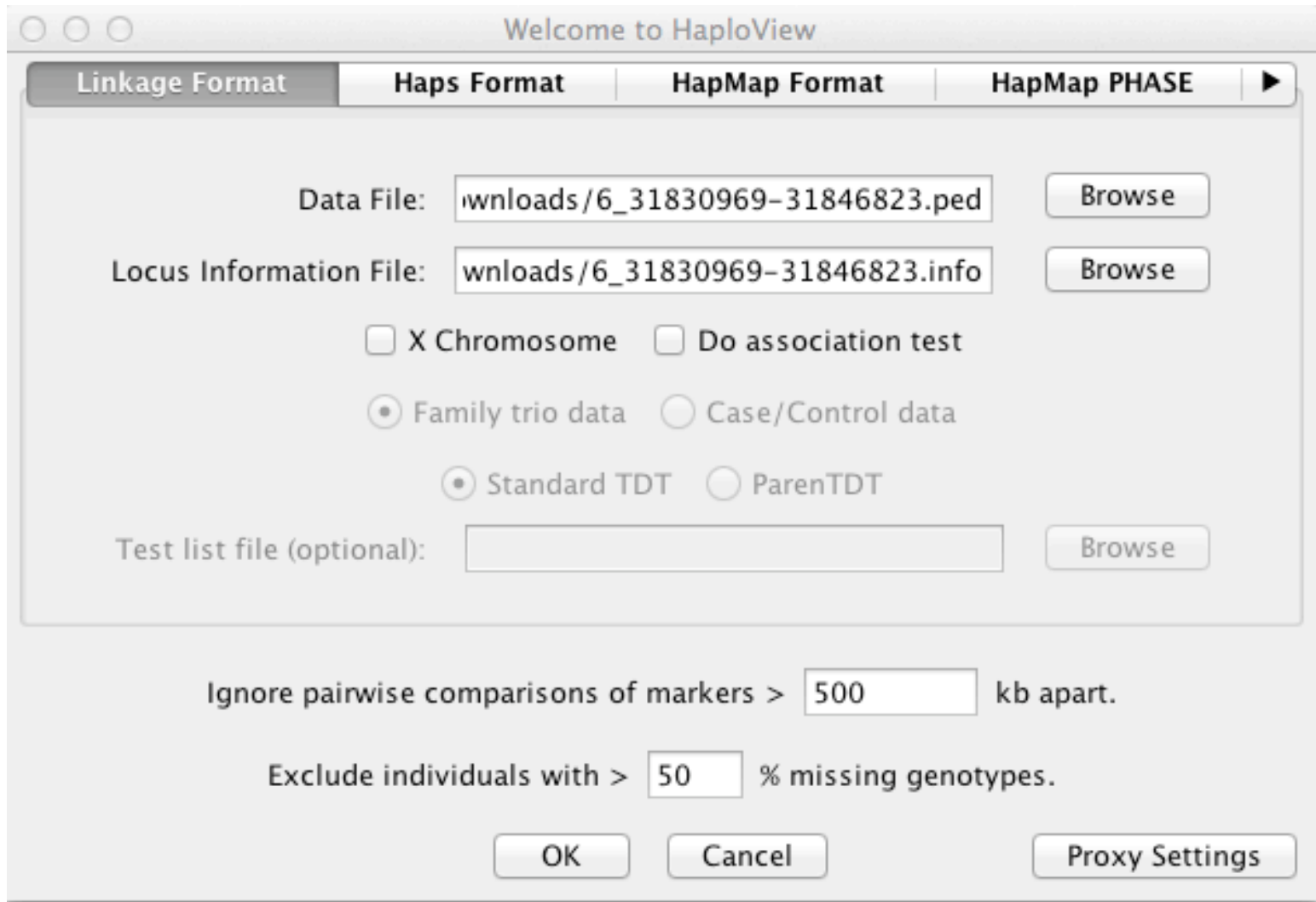
CHS
CLM
FIN
GBR
IBS
JPT
LWK
MXL
PUR
TSI

Next >

Your linkage pedigree and marker information files have been generated:
Right click on the file name and choose "Save link as .." from the menu:
[Marker Information File](#) [Linkage Pedigree File](#)

Haplotype example input

```
java -jar Haploview.jar
```



Welcome to HaploView

Linkage Format | **Haps Format** | HapMap Format | HapMap PHASE ▶

Data File:

Locus Information File:

X Chromosome Do association test

Family trio data Case/Control data

Standard TDT ParentDTT

Test list file (optional):

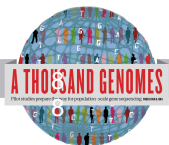
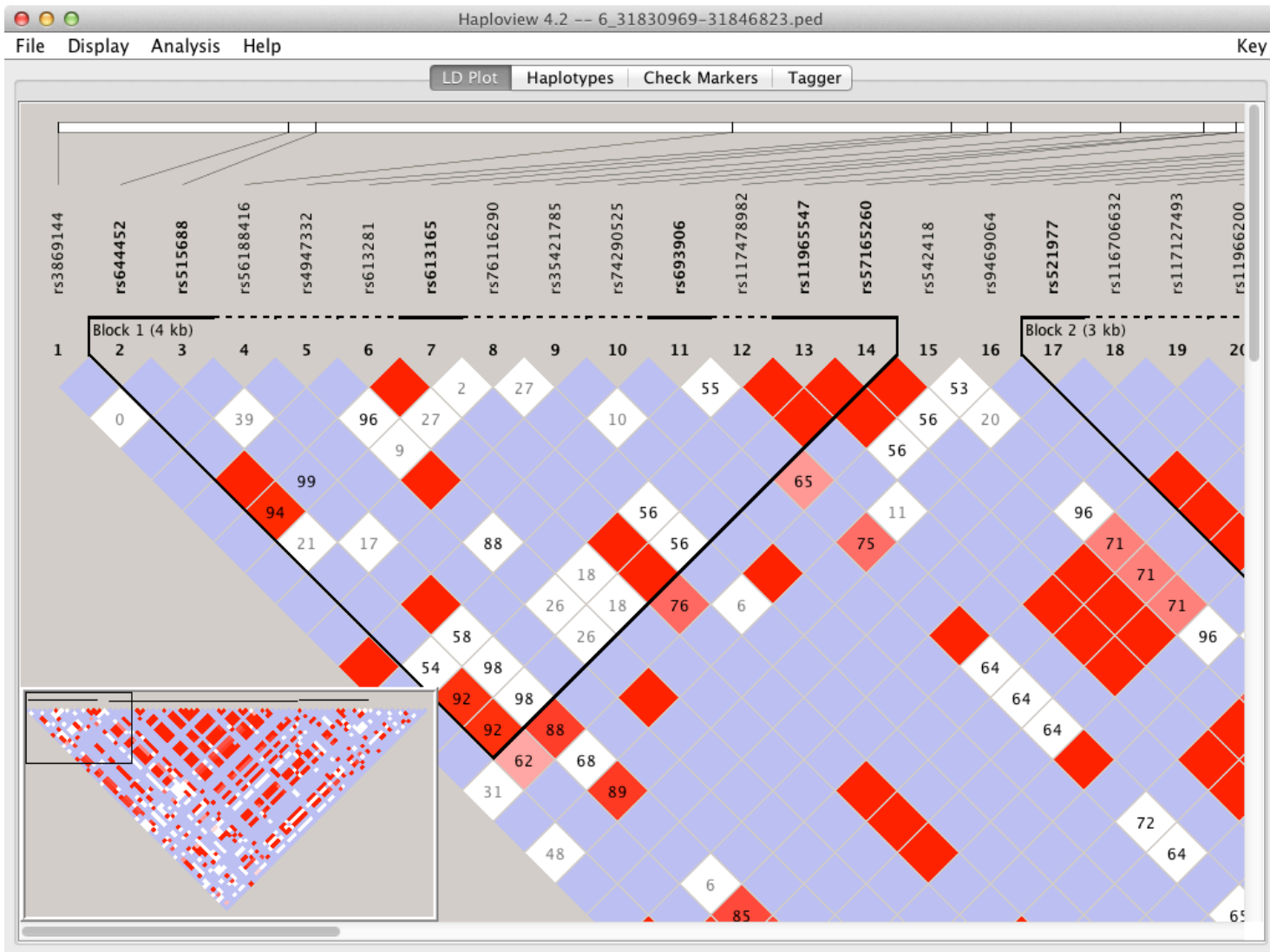
Ignore pairwise comparisons of markers > kb apart.

Exclude individuals with > % missing genotypes.



Haploview

- haploview



<http://www.broadinstitute.org/scientific-community/science/programs/medical-and-population-genetics/haploview>



Exercises

Use the browser to find the SLC44A4 gene.

7. Use the get VCF button in the left hand menu on the gene page to get a slice of a vcf file for this Gene.

8. Unzip this VCF file using a tool like winzip or Archive Utility.

9. Upload this VCF file to the Variant Effect Predictor.

http://browser.1000genomes.org/Homo_sapiens/UserData/UploadVariations

10. Do any of the variants have negative Sift or Polyphen predictions?

11. Using the example URLs on the Variation Pattern Finder tool menu look at the patterns of inheritance for this region: 6:31830700-31840700

http://browser.1000genomes.org/Homo_sapiens/UserData/VariationsMapVCF

12. For the same region use the VCF to PED tool to produce a ped and info file for the CEU population.

13. Look at these files in haploview.

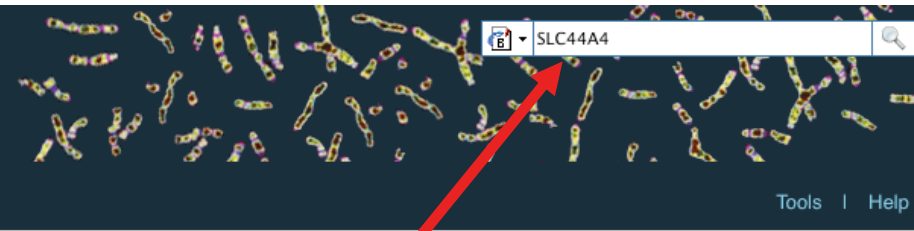
14. How many haplotype blocks does haploview think there are in this section?



Exercise Answers

1000 Genomes

A Deep Catalog of Human Genetic Variation



Search 1000 Genomes

The 1000 Genomes Browser

Ensembl-based browser provides early access to 1000 genomes data

1000 Genomes

A Deep Catalog of Human Genetic Variation



Human (GRCh37)

Search 1000 Genomes

New Search

Configure this page

Manage your data

Export data

Get VCF data

Bookmark this page

Results Summary

You searched for 'SLC44A4'

Gene or Gene Product

10 entrie(s) matched your search strings.

1. Gene: [ENSG00000204385](#) [Region in detail]
SLC44A4 -olute carrier family 44, member 4 [Source:HGNC Symbol;Acc:13941]
2. Transcript: [ENST00000229729](#) [Region in detail]
3. Peptide: [ENSP00000398764](#) [Region in detail]
SLC44A4
4. Peptide: [ENSP00000392054](#) [Region in detail]
SLC44A4
5. Peptide: [ENSP00000404572](#) [Region in detail]
SLC44A4
6. Peptide: [ENSP00000398901](#) [Region in detail]
SLC44A4
7. Peptide: [ENSP00000415708](#) [Region in detail]
SLC44A4
8. Peptide: [ENSP00000400263](#) [Region in detail]
SLC44A4
9. Peptide: [ENSP00000414296](#) [Region in detail]
SLC44A4
10. Peptide: [ENSP00000399161](#) [Region in detail]
SLC44A4



Exercise Answers

Human (GRCh37) ▾

Location: 6:31,830,969-31,846,823

Gene: SLC44A4

Gene: SLC44A4 (ENSG00000204385)

Gene-based displays

- Gene summary
- Splice variants (9)
- Supporting evidence
- Sequence
- External references
- Regulation
- Genetic Variation
 - Variation Table
 - Structural Variation
 - Variation Image
- External Data
- ID History
 - Gene history

Description solute carrier family 44, member 4 [Source:HGNC Symbol;Acc:13941]
Location [Chromosome 6: 31,830,969-31,846,823](#) reverse strand.
Transcripts There are 9 transcripts in this gene

Show/hide columns

Name	Transcript ID	Length (bp)	Protein ID	Length (aa)	Biotype	CCDS
SLC44A4-001	ENST00000229729	2580	ENSP00000229729	710	Protein coding	CCDS4724
SLC44A4-004	ENST00000229729					
SLC44A4-201	ENST00000229729					
SLC44A4-202	ENST00000229729					
SLC44A4-002	ENST00000229729					
SLC44A4-003	ENST00000229729					
SLC44A4-007	ENST00000229729					
SLC44A4-005	ENST00000229729					
SLC44A4-006	ENST00000229729					

VCF / BAM File URL:

Region:

(e.g. 1:1-50000)

Use VCF filters (this doesn't apply to BAM files):

- None
- By individual(s)
- By population(s) *

(to filter by populations please provide URL to a Sample-Population Mapping File in the box below)

-
-
-
-
-

Configure Page Custom Data

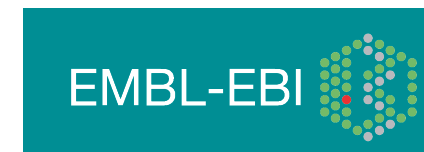
Data Management

- Upload Data
- Attach DAS
- Attach Remote File
- Manage Data
- Features on Karyotype
- Data Converters
 - Assembly Converter
 - ID History Converter
 - Variant Effect Predictor
 - Data Slicer
 - Variation Pattern Finder

Thank you - your VCF file [[6_31830969-31846823.ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz](#)] [Size: 83436] has been generated. Right click on the file name and choose "Save link as ..." from the menu

Preview

```
##fileformat=VCFv4.0
##source=BCM:SNPTools:hapfuse
##reference=1000Genomes-NCBI37
##FORMAT=<ID=GT,Number=1,Type=String,Description="Genotype">
##FORMAT=<ID=AP,Number=2,Type=Float,Description="Allelic Probability, P(Allele=1)>
#CHROM POS ID REF ALT QUAL FILTER INFO FORMAT HG00096
6 31831159 rs3869144 C T 100 PASS . GT:AP
6 31831167 . T C 100 PASS . GT:AP
```



Exercise Answers

- Custom Data
 - Assembly Converter
 - ID History Converter
 - Variant Effect Predictor**
 - Data Slicer
 - Variation Pattern Finder

Input file

Species:

Name for this upload (optional):

Paste file:

Upload file:

or provide file URL:

Input file format:

Options

Get regulatory region consequences:

Type of consequences to display:

Check for existing co-located variants:

Return results for variants in coding regions only:

Show HGNC identifier for genes where available:

Show Ensembl protein identifiers where available:

Show HGVS identifiers for variants where available:

Non-synonymous SNP predictions (human only)

SIFT predictions:

PolyPhen predictions:

Condel consensus (SIFT/PolyPhen) predictions:



Exercise Answers

6_31833249_A/G	6:31833249	G	ENSG00000204385	ENST00000487680	Transcript	UPSTREAM	-	-	-	-
6_31833249_A/G	6:31833249	G	ENSG00000204385	ENST00000414427	Transcript	DOWNSTREAM	-	-	-	-
6_31833249_A/G	6:31833249	G	ENSG00000204385	ENST00000479777	Transcript	DOWNSTREAM	-	-	-	-
6_31833249_A/G	6:31833249	G	ENSG00000204385	ENST00000475563	Transcript	DOWNSTREAM	-	-	-	-
6_31833357_C/T	6:31833357	T	-	ENSR00000487922	RegulatoryFeature	REGULATORY_REGION	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000495807	Transcript	UPSTREAM	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000480384	Transcript	UPSTREAM	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000491768	Transcript	UPSTREAM	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000375631	Transcript	UPSTREAM	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204386	ENST00000479533	Transcript	UPSTREAM	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000229729	Transcript	NON_SYNONYMOUS_CODING	1625	1604	535	R/H
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000375562	Transcript	NON_SYNONYMOUS_CODING	1544	1478	493	R/H
6_31833357_C/T	6:31833357	T	ENSG00000204385	ENST00000544672	Transcript	NON_SYNONYMOUS_CODING	1673	1376	459	R/H
6_31833357_C/T	6:31833357	T	ENSG00	-	-	1KG 6 31833357	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00	-	-	1KG 6 31833357	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00	-	-	1KG 6 31833357	-	-	-	-
6_31833357_C/T	6:31833357	T	ENSG00	535	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious;	PolyPhen=probably_damaging;	Condel=deleterious
6_31833612_C/G	6:31833612	G	ENSG00	-	-	-	-	-	-	-
6_31833612_C/G	6:31833612	G	ENSG00	493	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious;	PolyPhen=possibly_damaging;	Condel=deleterious
6_31833612_C/G	6:31833612	G	ENSG00	459	R/H	cGc/cAc	1KG 6 31833357	SIFT=deleterious;	PolyPhen=probably_damaging;	Condel=deleterious
				-	-	-	1KG 6 31833357	-	-	-
				-	-	-	1KG 6 31833357	-	-	-
				-	-	-	1KG 6 31833357	-	-	-



Exercise Answers

Custom Data

Data Management

- Upload Data
- Attach DAS
- Attach Remote File
- Manage Data
- Features on Karyotype
- Data Converters
 - Assembly Converter
 - ID History Converter
 - Variant Effect Predictor
 - Data Slicer
 - **Variation Pattern Finder**

i Variation Pattern Finder:

The Variation Pattern Finder allows one to look for patterns of shared variation between individuals in the same vcf file. The finder looks for distinct variation combinations within the region, as well as individuals associated with each variation combination pattern. Only variants which have potentially functional consequences are considered, both intergenic and intronic snps are excluded. Click [here](#) for more extensive documentation.

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Upload files

VCF File URL:

```
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz
```

[Clear box](#)

e.g. `ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz`

Sample-Population Mapping File URL:

```
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel
```

[Clear box](#)

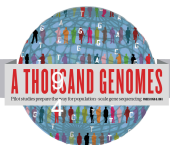
e.g. `ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel`

Region:

```
6:31830700-31840700
```

e.g. `6:46620015-46620998`

Next >



Exercise Answers

Custom Data

Data Management

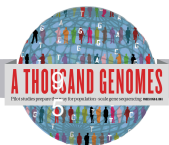
- Upload Data
- Attach DAS
- Attach Remote File
- Manage Data
- Features on Karyotype
- Data Converters
 - Assembly Converter
 - ID History Converter
 - Variant Effect Predictor
 - Data Slicer
- Variation Pattern Finder

Variation Pattern Finder

Export data: [CSV](#) [Excel](#)

Go to collapsed view

Population ASW	CEU	Freq	rs116706632:G/A	rs117127493:G/C	rs644827:T/C
			6:31836976	6:31837009	6:31838441
			ENST00000229729 NON_SYNONYMOUS_CODING:P/S	ENST00000229729 NON_SYNONYMOUS_CODING:Q/E	ENST00000229729 NON_SYNONYMOUS_CODING:Q/E
			ENST00000375562 NON_SYNONYMOUS_CODING:P/S	ENST00000375562 NON_SYNONYMOUS_CODING:Q/E	ENST00000375562 NON_SYNONYMOUS_CODING:Q/E
			ENST00000544672 NON_SYNONYMOUS_CODING:P/S	ENST00000544672 NON_SYNONYMOUS_CODING:Q/E	ENST00000544672 NON_SYNONYMOUS_CODING:Q/E
			ENST00000414427 NON_SYNONYMOUS_CODING:P/S	ENST00000414427 NON_SYNONYMOUS_CODING:Q/E	
NA20289, NA20296 and 13 other(s)	NA069	0.293	GIG	GIG	CIC
NA20127, NA19703 and 9 other(s)	NA125	0.203	GIG	GIG	CIT
NA20314, NA20317 and 6 other(s)	NA120	0.195	GIG	GIG	TIC
NA19920, NA19700 and 2 other(s)		0.032	GIG	GIG	CIC
NA19819, NA20281 and 2 other(s)		0.026	GIG	GIG	CIC
NA20291, NA20356 and 3 other(s)		0.016	GIG	GIG	TIC
NA19908	NA122	0.013	GIG	GIG	CIT
		0.008	GIG	CIG	CIC
		0.005	GIG	GIC	TIC
	NA119	0.005	GIG	GIC	CIC
NA19916		0.004	GIG	GIG	CIC
NA19711, NA20340		0.003	GIG	GIG	CIC
		0.003	GIG	GIG	CIT
	NA119	0.003	GIA	GIG	CIC
		0.003	GIG	CIG	CIT



Exercise Answers

i VCF to PED converter:

When providing a VCF file, both the data file and its index file should be present on the web server and named correctly. The VCF file should have a ".vcf.gz" extension, and the index file should have a ".vcf.gz.tbi" extension, E.g: MyData.vcf.gz, MyData.vcf.gz.tbi Click [here](#) for more extensive documentation.

Upload files

VCF File URL:

```
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz
```

[Clear box](#)

e.g. ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/ALL.chr6.phase1.projectConsensus.genotypes.vcf.gz

Sample-Population Mapping File URL:

```
ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel
```

[Clear box](#)

e.g. ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/release/20101123/interim_phase1_release/interim_phase1.20101123.ALL.panel

Region:

```
6:31830700-31840700
```

e.g. 6:46620015-46620998

Next >



Exercise Answers

VCF filter by population(s)

Select one or more populations from the scrollable list:

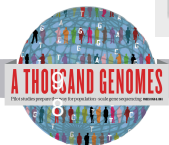
ASW
CEU
CHB
CHS
CLM
FIN
GBR
IBS
JPT
LWK

Next >

Your linkage pedigree and marker information files have been generated:
Right click on the file name and choose "Save link as .." from the menu:

[Marker Information File](#) [Linkage Pedigree File](#)

Exercise Answers



Exercise Answers



Data Availability

- FTP site: <ftp://ftp.1000genomes.ebi.ac.uk/vol1/ftp/>
 - Raw Data Files
- Web site: <http://www.1000genomes.org>
 - Release Announcements
 - Documentation
- Ensembl Style Browser: <http://browser.1000genomes.org>
 - Browse 1000 Genomes variants in Genomic Context
 - Variant Effect Predictor
 - Data Slicer
 - Other Tools



Announcements

- <http://1000genomes.org>
- 1000announce@1000genomes.org
- <http://www.1000genomes.org/1000-genomes-announcement-mailing-list>
- <http://www.1000genomes.org/announcements/rss.xml>
- <http://twitter.com/#!/1000genomes>



Questions

Please send any future questions about this presentation and any other material on our website to info@1000genomes.org



<http://www.1000genomes.org/using-1000-genomes-data>



1000 Genomes Community Meeting

- University of Michigan, Ann Arbor on the 12th and 13th of July 2012
- Showcase Advances made by the Project
- Generate Discussion about the next round of Human Genome Sequencing
- Registration closes May 15th
- <http://1000gconference.sph.umich.edu/>



Thanks

- The 1000 Genomes Project Consortium
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- Richard Smith
- Holly Zheng Bradley
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- David Richardson

