

1000 Genomes Browser Quick start guide

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

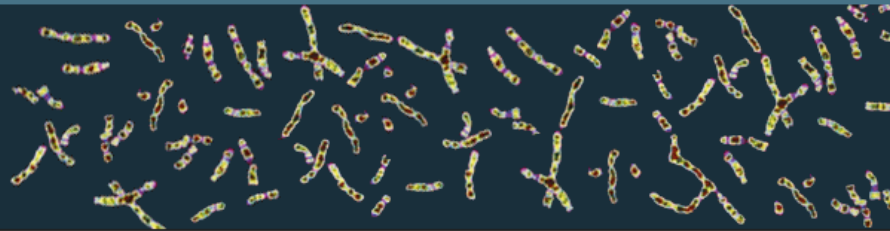
6 January 2008

Overview

- Based on version 50 (“old version”) of Ensembl code
- Contains all of the gene information normally present in Ensembl
 - Gene and transcript annotation, external references, sequence data
- There are things that don’t work and we have not transferred to “production” web hardware
 - Please send questions, problems or apparent errors to flicek@ebi.ac.uk

1000 Genomes

A Deep Catalog of Human Genetic Variation



Ensembl release 50 - Jul 2008

HOME



THE 1000 GENOMES BROWSER

Ensembl-based browser provides early access to 1000genomes data

In order to facilitate immediate analysis of the 1000genomes data by the whole scientific community, this browser (based on Ensembl) integrates the SNP calls and read coverage from this December 2008 release. All of 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 SNP id's on this site are temporary and will NOT be maintained.**

START BROWSING 1000 GENOMES DATA!!



[Browse Human](#) →
NCBI 36

[Transcript SNP view](#) →
View the consequences of sequence variation at the level of each transcript in the genome.

[SeqAlignView](#) →
Shows read-depth data alongside SNPs

[Other sites using Ensembl software...](#)

LINKS



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



[1000 Genomes Wiki](#) →
Browse the 1000 Genomes Wiki.

Search 1000 Genomes

Go

e.g. human gene BRCA2

PRESS RELEASE

December 2008

Browser displays SNP calls on CEU and YRI high coverage individuals from Pilot2

- ▶ [View sample data](#)
- ▶ [EBI Mirror](#)
- ▶ [NCBI Mirror](#)

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1000 Genomes Browser Home Page

1000 Genomes

A Deep Catalog of Human Genetic Variation

Ensembl release 50 - Jul 2008

HOME

Chromosome 9
21,957,751 - 21,984,490

- View of Chromosome 9
- Graphical view
- Graphical overview
- Resequencing alignment
- View region at UCSC
- View region at Ensembl

Export data

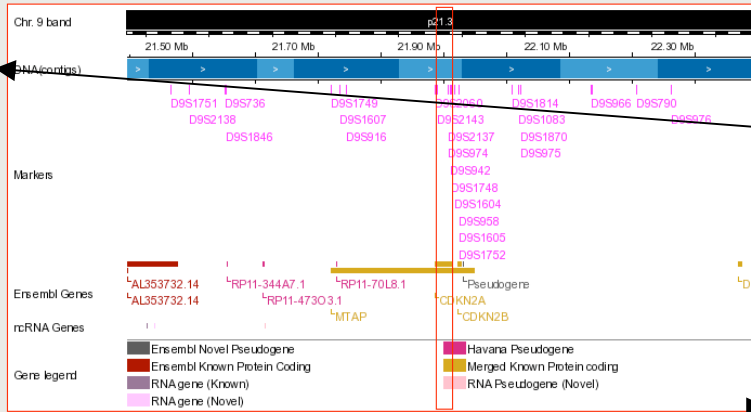
Export from region...



Chromosome 9



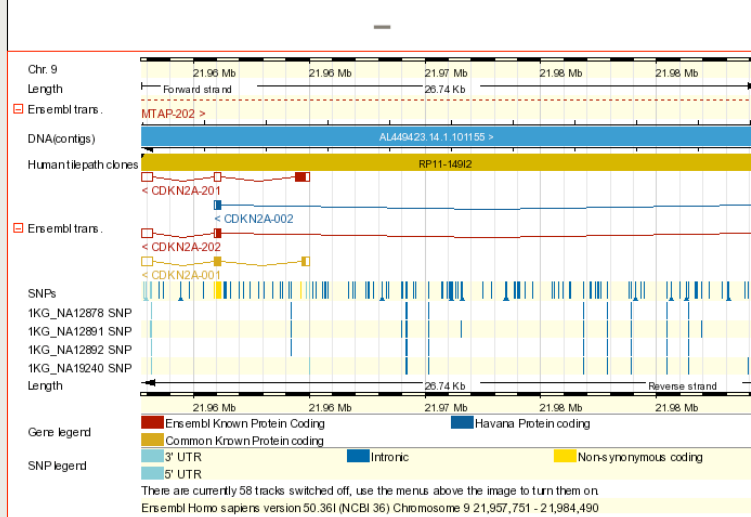
Overview



Detailed view

Features DAS Sources Repeats Decorations Export Image size Help

Jump to region 9 : [21957751] - [21984490] Refresh Band: Refresh

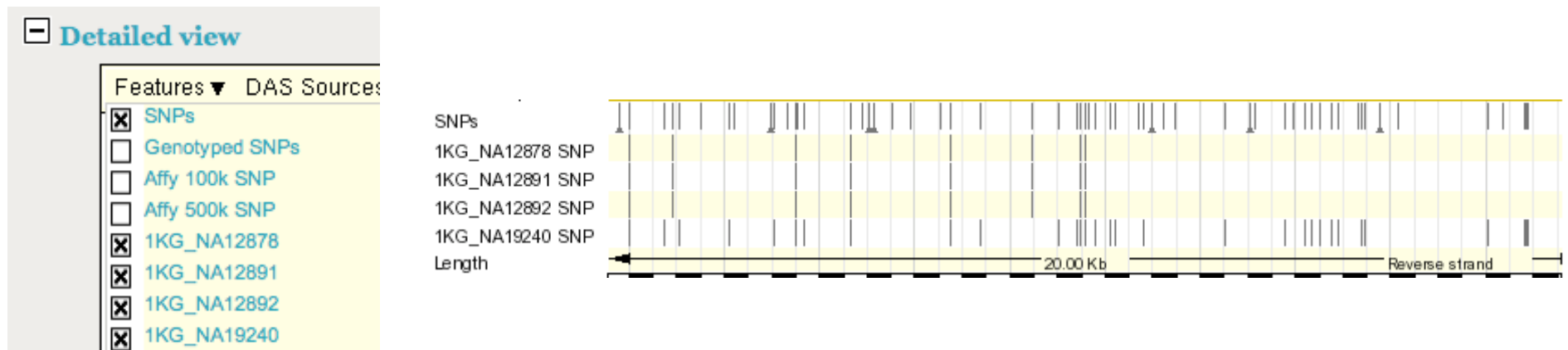


Main view

- Built on Ensembl
- Navigation is on the left hand side
- Options are drop down menus on the tops of the windows
- Includes only human data in current release
 - Comparative genomics information will be available in a future release
 - All appropriate pages have links to current versions of Ensembl and UCSC

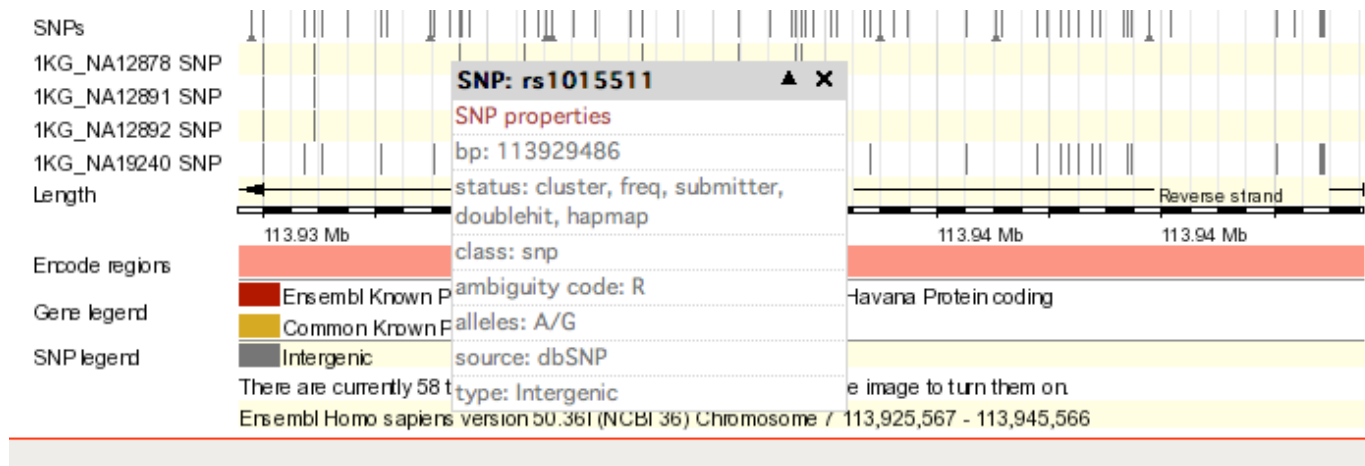
Individual-specific SNPs

- The 1KG individuals can be viewed on the graphical view (contig view) pages
- These are selected from the “Features” menu and appear as tracks near the bottom of the display
- Tracks for all SNPs and the SNPs on selected Affy arrays are also available



SNP Information

- SNPs are clickable which brings up a small window with basic information
- The “SNP properties” link leads to a dedicated page for the SNP with detailed information (mostly imported from dbSNP) about population frequencies, identifiers, individual genotypes and other information



1000 Genomes

A Deep Catalog of Human Genetic Variation

Ensembl release 50 - Jul 2008

HOME

dbSNP: rs1015511

SNP Report

- ENSG00000128573 - GeneSNP info
- ENSG00000133612 - GeneSNP info
- rs1015511 - SNP info
- rs1015511 - LD info

Chromosome 7
113,929,486

- View of Chromosome 7
- Graphical view
- Graphical overview
- Export from region...



SNP rs1015511 (dbSNP129)

Synonyms
1KG_NA12892 tmp:7:113929486
ENSEMBL:Watson ENSNP5660229
1KG_NA12891 tmp:7:113929486
Affy GeneChip 100K Array SNP_A-1746777
1KG_NA19240 tmp:7:113929486
1KG_NA12878 tmp:7:113929486
TSC TSC0097629

Alleles
A/G (ambiguity code: **R**)
Ancestral allele: A

Validation status
Proven by cluster, frequency, submitter, doublehit (SNP tested and validated by a non-computational method).
HapMap SNP

Linkage disequilibrium data
Links to LDview per population:
CSHL_HAPMAP:HapMap-YRI (Tag SNP) PERLEGEN:AFD_AFR_PANEL (Tag SNP)

Flanking sequence
ATCACAACTCACTATAGGAGGTGACTGAGTGAAGCTTTACAGTGAATAATTTTCTTTT
GGAGAAATACATCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
GTCTTAATTCAGTCTTCTTCAATGAGAAAATGTGAATTAACCCACAGACTACAG
ATGCTGCTCAGAGACTAAGGTACACTAAATGTGATCTTAAGAACTTCACTTA
AACATCATCAATTTAGAAATAGATTTAAGCAGGTTAATAGGTTAGTTACGCCAA
GCTGTGATATCACTTTCAAAATGCTACTTACTTACTTCACTTCACTTCACTTCACTT
GATACATCTTATCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
TTTCTTAATCAAACTCACTGAGTGAAGTGAAGTGAAGTGAAGTGAAGTGAAGTGAAGTGAAGT
GCTTGAAGAAATACAGAAAATCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
ATAACTGTATCTTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
CAGACACTTGGAGTAAAGTGTATCATGAAAAGTGTGACACTTCACTCACTCACTCACTCACTCACT
TGAATTCGAAAAGTCTTCTTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
GTAAAGTAAATAAATATCTTTATCTAACTATATTTCCAAATTTTACTAATAC
AGAACTCTGCTTCTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
CTTTTAACTAAATCTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
AATACACTTATATCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
TAATTAATAAACTGGAAAAGTCTTGTGCTTCTTCACTCACTCACTCACTCACTCACTCACTCACT
AACATCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
TTCACTTTCAGGCCATCCGGATATTTCTAAAGTGTGGGTTTAAACTGCATTACTCA
AATCTTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
TAAATTTTGTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT
TTAAAAGGAAATTTGCTTCTTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACTCACT

SNP rs1015511 is located in the following transcripts
There is no transcript mapping for this SNP.

Population genotypes and allele frequencies

Population	Alleles		Genotypes		Genotypes		Description
	A	G	A/A	A/G	G/G		
AFFY:AFam	0.167	0.833		0.333	0.667	African-American	
AFFY:Asian	0.833	0.167	0.667	0.333		Asian	
AFFY:CEPH	0.600	0.400	0.200	0.800		Caucasian	
AFFY:Caucasian	0.333	0.667	0.167	0.333	0.500	Caucasian	
AFFY:PDpanel	0.310	0.690	0.095	0.429	0.476	Mixed	
CSHL_HAPMAP:HapMap-CEU	0.450	0.550	0.217	0.467	0.317	30 mother-father-child trios from the CEPH collection (Utah residents with ancestry from northern and western Europe), representing one of the populations studied in the International HapMap project...	

Individual genotypes for SNP rs1015511

SNP Context - 7 113929486

Features: Source, SNP class, SNP type, Decorations, Export, Image size, Help

Length: 20.00 Kb

DNA (contigs): Forward strand, Reverse strand

Length: 20.00 Kb

Ensembl trans, rRNA, tRNA

SNPs: 34 of the 100 variations in this region have been filtered out by the Source, Class and Type menus.

SNPView

- Temporary rsName
- Identifiers
- Flanking sequence
- Population frequency
- Individual genotypes
- Change display options (individuals, SNP type, etc.)
- Location information

Resequencing alignment

- View any region of the genome in alignment with reference, 4 1KG individuals, Watson, Venter
- Assumption made that if there is sequence coverage and not a SNP called, the base is the same as the reference
- Use “Resequencing alignment” link on the left side of pages to access view

The image shows two overlapping web interface panels. The left panel is titled 'Chromosome 9 21,944,015 - 21,964,014' and lists several options: 'View of Chromosome 9', 'Graphical view', 'Graphical overview', 'Resequencing alignment', 'View region at UCSC', and 'View region at EnsEMBL'. Below these is an 'Export data' section with 'Export from region...'. At the bottom are logos for NCBI and EMBL-EBI. The right panel is titled 'ENSG00000147889' and lists a comprehensive set of options: 'Gene information', 'Genomic sequence', 'Genomic sequence alignment', 'Resequencing alignment', 'Gene splice site image', 'Gene tree info.', 'Gene variation info.', 'LD info', 'ID history', 'Compare SNPs in transcript', 'Transcript information', 'Exon information', 'Protein information', and 'Export gene data'. Below this is another 'Chromosome 9 21,957,751 - 21,984,490' section with options: 'View of Chromosome 9', 'Graphical view', 'Graphical overview', and 'Export from region...'. The labels 'ContigView Page' and 'GeneView Page' are positioned below their respective panels.

Chromosome 9
21,944,015 - 21,964,014

- View of Chromosome 9
- Graphical view
- Graphical overview
- Resequencing alignment
- View region at UCSC
- View region at EnsEMBL

Export data

- Export from region...

NCBI EMBL-EBI

ContigView Page

ENSG00000147889

- Gene information
- Genomic sequence
- Genomic sequence alignment
- Resequencing alignment
- Gene splice site image
- Gene tree info.
- Gene variation info.
- LD info
- ID history
- Compare SNPs in transcript
- Transcript information
- Exon information
- Protein information
- Export gene data

Chromosome 9
21,957,751 - 21,984,490

- View of Chromosome 9
- Graphical view
- Graphical overview
- Export from region...

GeneView Page

Resequencing alignment options

Sequence Alignment for chromosome:NCBI36:9:21944015:21964014:1

Genomic Location and Markup options

Chromosome Name *

Start *

End *

Strand ▾

Exons to highlight ▾

Highlight variations ▾

Line numbering ▾

Alignment width *Number of bp per line in alignments

Matching basepairs ▾

Codons ▾ Displayed only for the highlighted exons

Title display ▾ On mouse over displays exon IDs, length of insertions and SNP's allele

Reference individual: NCBI36

Resequenced Human individuals 1KG_NA12878 1KG_NA12891
 1KG_NA12892 1KG_NA19240
 Venter Watson

Fields marked with * are required

Resequencing alignment output

```
Marked up sequence

~ No resequencing coverage at this position
THIS STYLE: Location of SNPs
THIS STYLE: Location of deletions

Homo_sapiens > chromosome:NCBI36:9:21944015:21964014:1

NCBI36      AAAAAATTATCCCTGTCTAATAATGACAAAGACATCTAACAAATCCCAAAAGATAGATA
LKG_NA12878 AAAAAATTATCCCTGTCTAATAATGACAAAGACATCTAACAAATCCCAAAAGATAGATA
LKG_NA12891 AAAAAATTATCCCTGTCTAATAATGACAAAGACATCTAACAAATCCCAAAAGATAGATA
LKG_NA12892 AAAAAATTATCCCTGTCTAATAATGACAAAGACATCTAACAAATCCCAAAAGATAGATA
LKG_NA19240 AAAAAATTATCCCTGTCTAATAATGACAAAGACATCTAACAAATCCCAAAAGATAGATA
Ventur      AAAAAATTATCCCTGTCTAATAATGACAAAGACATCTAACAAATCCCAAAAGATAGATA
Watson      AAAAAATTATCCCTGTCTAATAATGACAAAGACATCTAACAAATCCCAAAAGATAGATA

NCBI36      TTTAAAAATACCTGACCCATTCTTCTCAAAGTGTCAAGATGACCAAAGCAAGGAACAAAC
LKG_NA12878 TTTAAAAATACCTGACCCATTCTTCTCAAAGTGTCAAGATGACCAAAGCAAGGAACAAAC
LKG_NA12891 TTTAAAAATACCTGACCCATTCTTCTCAAAGTGTCAAGATGACCAAAGCAAGGAACAAAC
LKG_NA12892 TTTAAAAATACCTGACCCATTCTTCTCAAAGTGTCAAGATGACCAAAGCAAGGAACAAAC
LKG_NA19240 TTTAAAAATACCTGACCCATTCTTCTCAAAGTGTCAAGATGACCAAAGCAAGGAACAAAC
Ventur      TTTAAAAATACCTGACCCATTCTTCTCAAAGTGTCAAGATGACCAAAGCAAGGAACAAAC
Watson      TTTAAAAATACCTGACCCATTCTTCTCAAAGTGTCAAGATGACCAAAGCAAGGAACAAAC

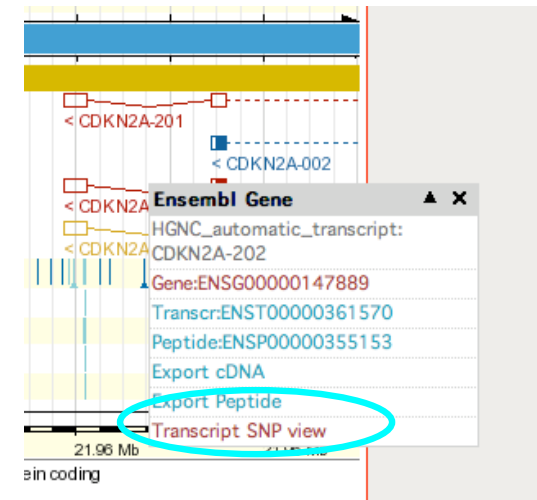
NCBI36      TGAAAAGCTGTCATAACCAAGAGGGGCTGAGAAAGACATGATGATGAAATGTTATATGAT
LKG_NA12878 TGAAAAGCTGTCATAACCAAGAGGGGCTGAGAAAGACATGATGATGAAATGTTATATGAT
LKG_NA12891 TGAAAAGCTGTCATAACCAAGAGGGGCTGAGAAAGACATGATGATGAAATGTTATATGAT
LKG_NA12892 TGAAAAGCTGTCATAACCAAGAGGGGCTGAGAAAGACATGATGATGAAATGTTATATGAT
LKG_NA19240 TGAAAAGCTGTCATAACCAAGAGGGGCTGAGAAAGACATGATGATGAAATGTTATATGAT
Ventur      TGAAAAGCTGTCATAACCAAGAGGGGCTGAGAAAGACATGATGATGAAATGTTATATGAT
Watson      *****CCTGAGAAAGACATGATGATGAAATGTTATATGAT

NCBI36      ATTCTGGATGGGTTCTTGGTTTGCAAAAGGGAATTTATGCAAAAATCAAGGAAATTTGAT
LKG_NA12878 ATTCTGGATGGGTTCTTGGTTTGCAAAAGGGAATTTATGCAAAAATCAAGGAAATTTGAT
LKG_NA12891 ATTCTGGATGGGTTCTTGGTTTGCAAAAGGGAATTTATGCAAAAATCAAGGAAATTTGAT
LKG_NA12892 ATTCTGGATGGGTTCTTGGTTTGCAAAAGGGAATTTATGCAAAAATCAAGGAAATTTGAT
LKG_NA19240 ATTCTGGATGGGTTCTTGGTTTGCAAAAGGGAATTTATGCAAAAATCAAGGAAATTTGAT
Ventur      ATTCTGGATGGGTTCTTGGTTTGCAAAAGGGAATTTATGCAAAAATCAAGGAAATTTGAT
Watson      A*****CTTGGTTTGCAAAAGGGAATTTATGCAAAAATCAAGGAAATTTGAT
```

- Hets
- SNPs
- No coverage

Individual SNP consequences

- TranscriptSNPView
 - Linked off all Ensembl Genes
 - From transcript pages in left hand menu
 - Color-coded display of how SNPs affect transcripts



ENST00000304494											
<ul style="list-style-type: none">Gene informationGene splice site imageGenomic sequenceGene variation info.ID historyCompare transcript SNPsResequencing alignmentTranscript informationExon informationProtein informationExport transcript data	<h3>Ensembl Transcript Report</h3> <table border="1"><tbody><tr><td>Transcript</td><td>CDKN2A-001 (HGNC (curated)) To view all Ensembl genes linked to the name click here. This transcript is a member of the Human CCDS set: CCDS6510 Havana transcript having same CDS: OTTHUMT00000051915</td></tr><tr><td>Ensembl Transcript ID</td><td>ENST00000304494</td></tr><tr><td>Transcript information</td><td>Exons: 3 Transcript length: 1,160 bps Translation length: 156 residues This transcript is a product of gene: ENSG00000147889</td></tr><tr><td>Genomic Location</td><td>This transcript can be found on Chromosome 9 at location 21,957,751-21,965,038. The start of this transcript is located in Contig AL449423.14.1.101155.</td></tr><tr><td>Description</td><td>Cyclin-dependent kinase inhibitor 2A, isoform 4 (p14ARF) (p19ARF). Source: Uniprot/SWISSPROT Q8N726</td></tr></tbody></table>	Transcript	CDKN2A-001 (HGNC (curated)) To view all Ensembl genes linked to the name click here . This transcript is a member of the Human CCDS set: CCDS6510 Havana transcript having same CDS: OTTHUMT00000051915	Ensembl Transcript ID	ENST00000304494	Transcript information	Exons: 3 Transcript length: 1,160 bps Translation length: 156 residues This transcript is a product of gene: ENSG00000147889	Genomic Location	This transcript can be found on Chromosome 9 at location 21,957,751-21,965,038 . The start of this transcript is located in Contig AL449423.14.1.101155 .	Description	Cyclin-dependent kinase inhibitor 2A, isoform 4 (p14ARF) (p19ARF). Source: Uniprot/SWISSPROT Q8N726
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Description	Cyclin-dependent kinase inhibitor 2A, isoform 4 (p14ARF) (p19ARF). Source: Uniprot/SWISSPROT Q8N726										

Main display

Summary table

ENST00000361570

[Gene information](#)
[Gene splice site image](#)
[Genomic sequence](#)
[Gene variation info.](#)
[ID history](#)
[Compare transcript](#)
[SNPs](#)
[Resequencing alignment](#)
[Transcript information](#)
[Exon information](#)
[Protein information](#)
[Export transcript data](#)

Chromosome 9
21,957,751 - 21,984,490

[View of Chromosome 9](#)
[Graphical view](#)
[Graphical overview](#)
[Export from region...](#)

NCBI EMBL-EBI

Ensembl Transcript Variation Report for ENST00000361570

Transcript **CDKN2A** (HGNC (automatic)) **Synonyms:** ARF, CDK4I, CDKN2, CMM2, INK4, INK4a, MLM, MTS1, p14, p16, p16INK4a, p19
To view all Ensembl genes linked to the name [click here](#).
This transcript is a member of the Human CCDS set: [CCDS6511](#)

Genomic Location This transcript can be found on Chromosome 9 at location [21,957,751-21,984,490](#).
The start of this transcript is located in [Contig AL449423.14.1.101155](#).

Description Cyclin-dependent kinase inhibitor 2A, isoform 4 (p14ARF) (p19ARF). [Source: Uniprot/SWISSPROT Q8N7Z6](#)

SNPs and variations in region of transcript ENST00000361570

Where there is resequencing coverage, SNPs have been called using a computational method. Here we display the SNP calls observed by transcript from these sources: dbSNP, HGVbase, ENSEMBL:celera, TSC, Affy GeneChip 100K Array, Affy GeneChip 500K Array, Affy GenomeWideSNP_6.0, ENSEMBL:Venter, ENSEMBL:Watson, EGA, 1KG_NA12892, 1KG_NA12878, 1KG_NA12891, 1KG_NA19240.

SNP legend

- Green: same as reference
- Purple: different
- Hatched: heterozygous

Comparison to NCBI 936 alleles

Sample	C	T	G	A	C	G	C	C	C	G	A	C	C	C	G	G	G	G	C
1KG_NA12878	C	T	G	-	C	G	C	C	C	G	A	C	C	C	G	G	G	G	C
1KG_NA12891	C	T	G	-	C	G	C	C	C	G	A	C	C	C	G	G	G	G	C
1KG_NA12892	C	T	G	-	C	G	C	C	C	G	A	C	C	C	G	G	G	G	C
1KG_NA19240	C	T	G	-	C	G	C	C	C	G	A	C	C	C	G	G	G	G	C

Haplotype legend

- Green: Same allele
- Purple: Different allele
- Hatched: Heterozygous
- White: Missing data

- Individual
- Coverage
- SNPs
- Legend
- SNP summary
 - Green
 - same as reference
 - Purple:
 - different
 - Hatched
 - heterozygous

Variations in 1KG_NA12878

ID	Type	Chr: bp	Ref. allele	Individual allele genotype	Ambiguity	Transcript codon	CDS coord.	AA	AA change	Class	Source	Validation
rs11515	3PRIME_UTR	9:21958199	C	GIG	S	-	-	-	-	SNP	ENSEMBL:Watson, 1KG_NA12892, 1KG_NA12891, ENSEMBL:celera, 1KG_NA12878, 1KG_NA19240, dbSNP	cluster, frequency, doublehit, hapmap

Variations in 1KG_NA12891

ID	Type	Chr: bp	Ref. allele	Individual allele genotype	Ambiguity	Transcript codon	CDS coord.	AA	AA change	Class	Source	Validation
rs308844Q	3PRIME_UTR	9:21958159	G	AIG	R	-	-	-	-	SNP	1KG_NA12891, HGVbase, dbSNP	cluster, frequency, hapmap
rs11515	3PRIME_UTR	9:21958199	C	GIG	S	-	-	-	-	SNP	ENSEMBL:Watson, 1KG_NA12892, 1KG_NA12891, ENSEMBL:celera, 1KG_NA12878, 1KG_NA19240, dbSNP	cluster, frequency, doublehit, hapmap

Variations in 1KG_NA12892

ID	Type	Chr: bp	Ref. allele	Individual allele genotype	Ambiguity	Transcript codon	CDS coord.	AA	AA change	Class	Source	Validation
rs11515	3PRIME_UTR	9:21958199	C	GIG	S	-	-	-	-	SNP	ENSEMBL:Watson, 1KG_NA12892, 1KG_NA12891, ENSEMBL:celera, 1KG_NA12878, 1KG_NA19240, dbSNP	cluster, frequency, doublehit, hapmap

Variations in 1KG_NA19240

ID	Type	Chr: bp	Ref. allele	Individual allele genotype	Ambiguity	Transcript codon	CDS coord.	AA	AA change	Class	Source	Validation
rs11515	3PRIME_UTR	9:21958199	C	GIG	S	-	-	-	-	SNP	ENSEMBL:Watson, 1KG_NA12892, 1KG_NA12891, ENSEMBL:celera, 1KG_NA12878, 1KG_NA19240, dbSNP	cluster, frequency, doublehit, hapmap

Dump data

Dump of SNP data per individual (SNPs in rows, individuals in columns). For more advanced data queries use [BioMart](#).

Dump format: Text format HTML format

Dump

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Chromosome 8
19,851,160 - 19,871,160

- View of Chromosome 8
- Graphical view
- Graphical overview
- Resequencing alignment
- View region at UCSC
- View region at Ensembl

Export data

Export from region...

dbSNP: rs299

- GeneSNP info
- rs299 - SNP info
- rs299 - LD info



Linkage disequilibrium report: SNP rs299

Focus: SNP	rs299 (dbSNP 129)
Prediction method	LD values were calculated by a pairwise estimation between SNPs genotyped in the same individuals and within a 100kb window. An established method was used to estimate the maximum likelihood of the proportion that each possible haplotype contributed to the double heterozygote.
Population	Population: CSH_HAPMAP:HapMap-CEU [size: unknown] Description: 30 mother-father-child trios from the CEPH collection (Utah residents with ancestry from northern and western Europe), representing one of the populations studied in the International HapMap project (http://www.hapmap.org) SNP in tagged set for this population: Yes Super-population: EUROPE [size: unknown] Description: Samples from Europe north and west of Caucasus Mountains, Scandinavia, Atlantic Islands.

Context - Chromosome 8 19,851,160



Dump data

Use the yellow drop down menus at the top of the image to configure display and data you wish to dump. If no LD values are displayed, zoom out, choose another population or another region.

- Dump format
- As text
 - In Excel format
 - HTML format
 - For upload into Haploview software (may take a while)

Dump

LDView

- Based on data from HapMap and Perlegen populations
- Populations selectable from drop down tab

More SNPs displays

GeneSeqView

5' Flanking sequence: 600

3' Flanking sequence: 600

Exons to display: Ensembl exons

Exons on strand: Both orientations

Show variations: All Variations

Genomic sequence

GeneView

Ensembl Human GeneView

Essential Gene Report for BRCA2:BRCA2

Gene: BRCA2 (BRCA2) (BRCA2) (BRCA2)

Transcript: BRCA2

Transcript Location: chr13:31,273,811-31,273,811

Transcript Orientation: +

Transcript Length: 35,200 bp

Transcript Description: The protein product of BRCA2 is a tumor suppressor protein that is involved in DNA repair and cell cycle regulation. It is a member of the BRCA2 protein family and is highly conserved across species.

Transcript Structure: 13 Exons

Transcript Features: 13 Exons, 12 Introns

Transcript Coordinates: chr13:31,273,811-31,273,811

Gene variation info.

Protein information

Transcript information

GeneSNPView

GeneSNPView visualization showing multiple sequence alignments with SNPs highlighted in red and green.

TransView
















TransView visualization showing the genomic context of a gene (SNX1) with exons and introns, and protein coding regions.

ProtView

ProtView visualization showing protein structure, SNPs, and other features like low complexity sequences and coiled coils.

SNP types

Non-synonymous	In coding sequence, resulting in an aa change
Synonymous	In coding sequence, not resulting in an aa change
Frameshift	In coding sequence, resulting in a frameshift
Stop lost	In coding sequence, resulting in the loss of a stop codon
Stop gained	In coding sequence, resulting in the gain of a stop codon
Essential splice site	In the first 2 or the last 2 basepairs of an intron
Splice site	1-3 bps into an exon or 3-8 bps into an intron
Upstream	Within 5 kb upstream of the 5'-end of a transcript
Regulatory region	In regulatory region annotated by Ensembl
5' UTR	In 5' UTR
Intronic	In intron
3' UTR	In 3' UTR
Downstream	Within 5 kb downstream of the 3'-end of a transcript
Intergenic	More than 5 kb away from a transcript

 Regulatory region	 UTR	 3' UTR
 5' UTR	 Synonymous coding SNP	 Intronic
 Essential splice site	 Splice site SNP	 Non-synonymous coding SNP
 Intergenic	 Frameshift coding	 Downstream
 Upstream	 Stop lost	 Stop gained

Credits

- Eugene Kulesha, Stephen Keenan
- Yuan Chen, Fiona Cunningham
- Laura Clarke, Zam Iqbal

- 1000 Genome Data providers
- Entire Ensembl Team