İnsan Genomunu Anlamlandırmak Veritabanları ve Biyoinformatik Araçlar

Barış Salman

Gen-Era Diagnostik

Kayseri
20 Şubat, 2019

İnsan Genomu

Genom Haritalama

Genomik İşaretleyiciler

Varyant Önemi

Varyant Yordamlama Araçları

İnsan Genomu

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Gen-Era Diagnostik

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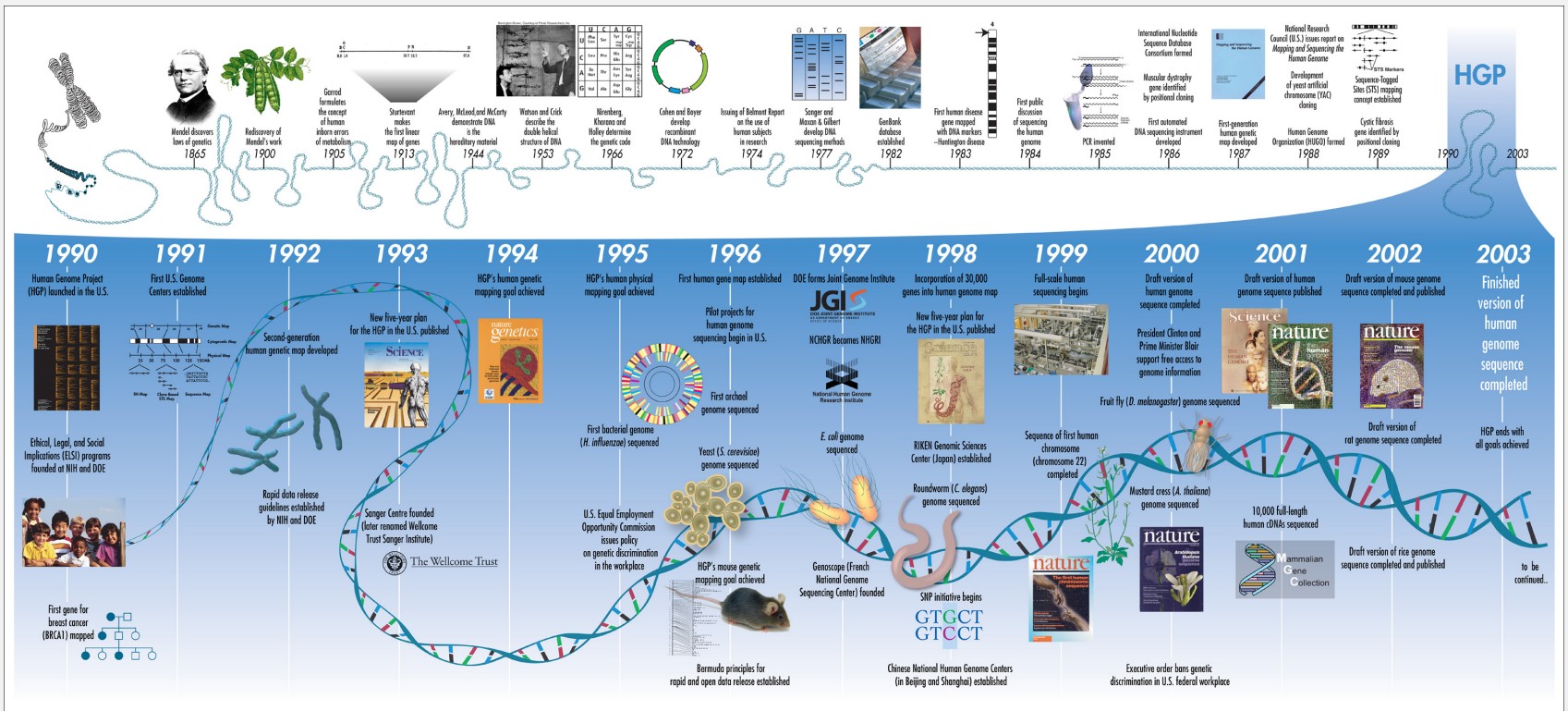

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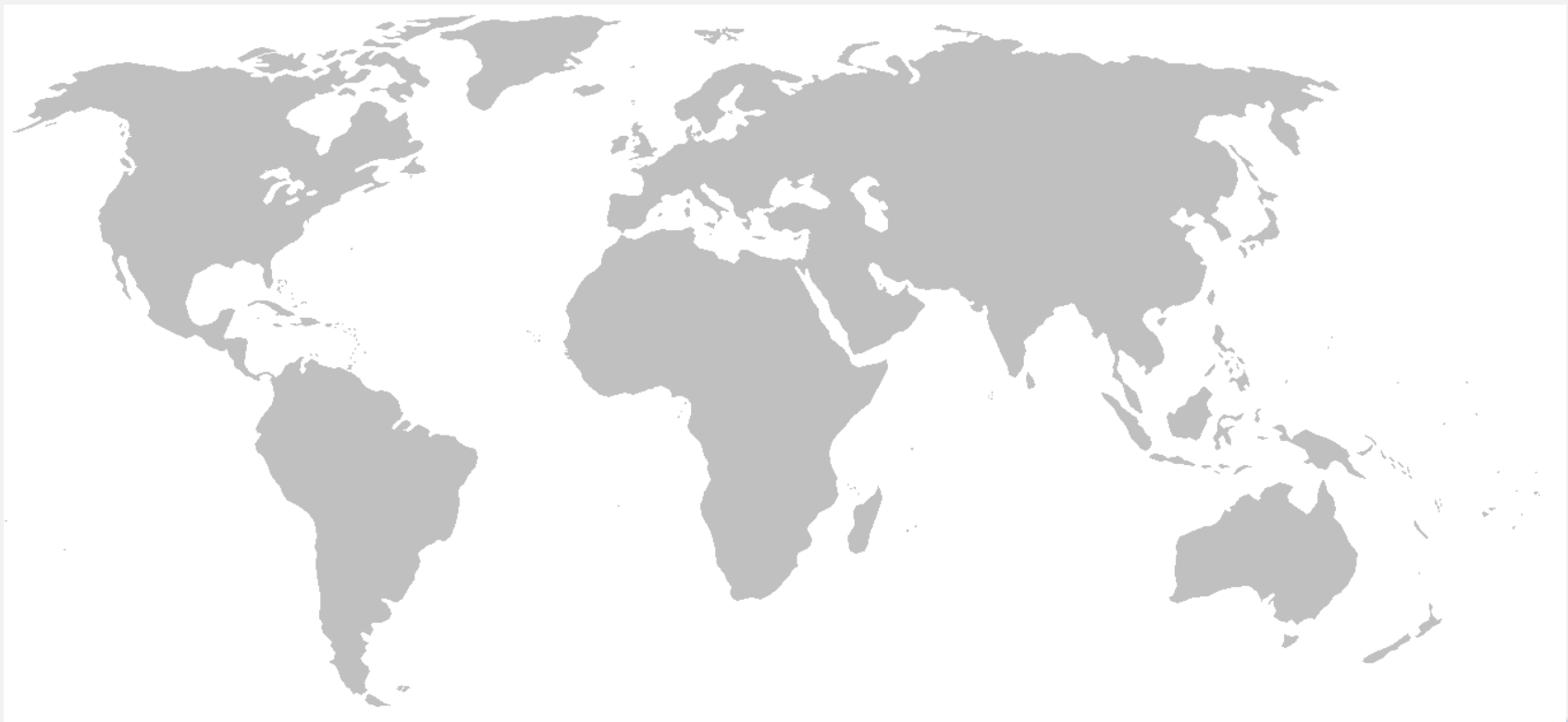


Genom Versiyonları

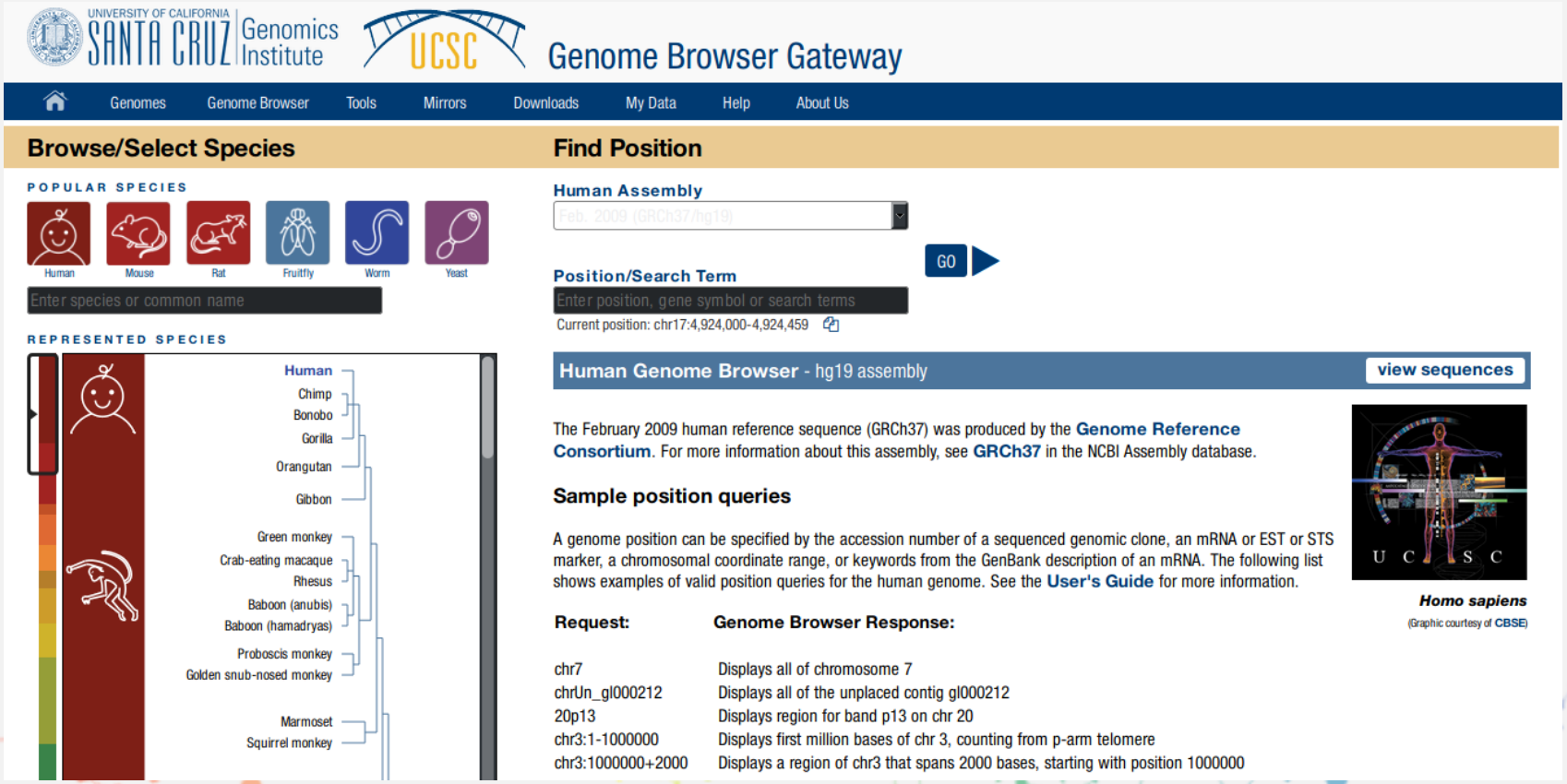
Genome Reference Consortium

- GRC human builds

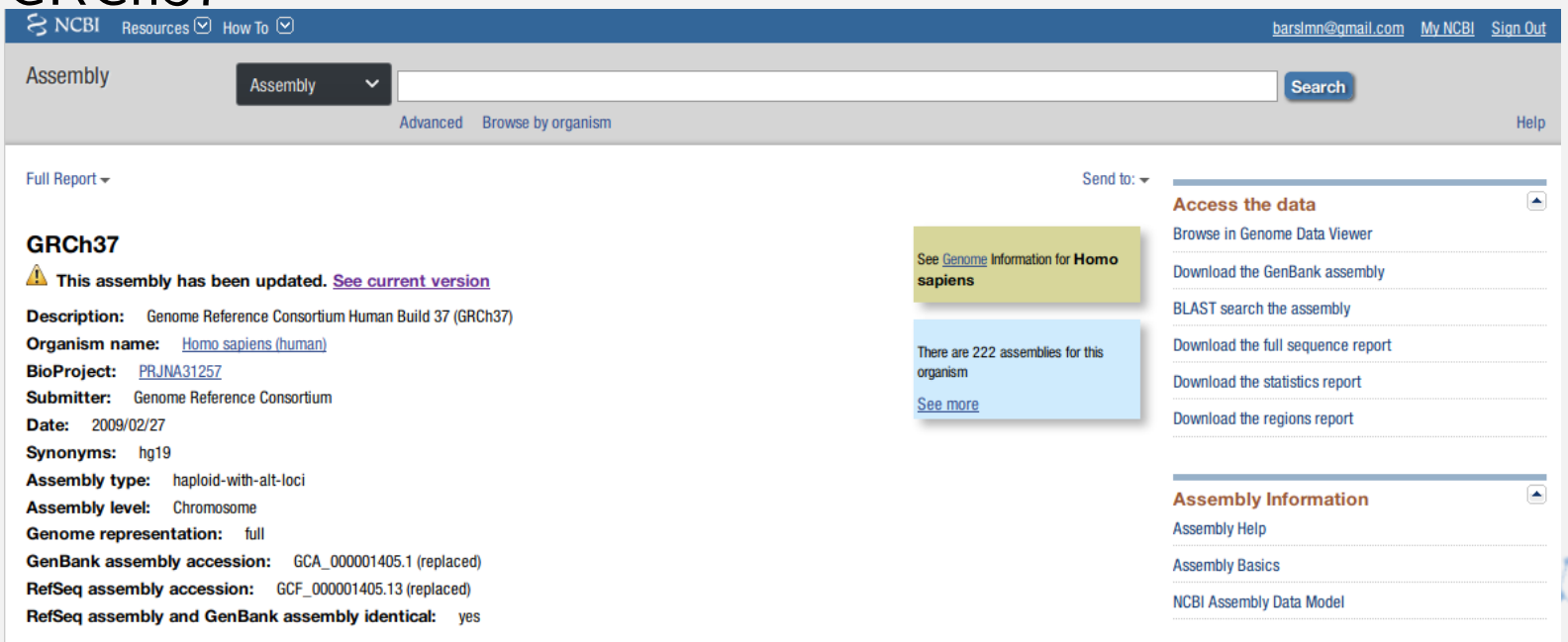
The University of California, Santa Cruz

- human genome (hg)

hg19


UNIVERSITY OF CALIFORNIA SANTA CRUZ Genomics Institute
UCSC
Genome Browser Gateway
Genomes
Genome Browser
Tools
Mirrors
Downloads
My Data
Help
About Us
Browse/Select Species
POPULAR SPECIES
Human
Mouse
Rat
Fruitfly
Worm
Yeast
Enter species or common name
REPRESENTED SPECIES
Human
Chimp
Bonobo
Gorilla
Orangutan
Gibbon
Green monkey
Crab-eating macaque
Rhesus
Baboon (anubis)
Baboon (hamadryas)
Proboscis monkey
Golden snub-nosed monkey
Marmoset
Squirrel monkey
Find Position
Human Assembly
Feb. 2009 (GRCh37/hg19)
GO
Position/Search Term
Enter position, gene symbol or search terms
Current position: chr17:4,924,000-4,924,459
Human Genome Browser - hg19 assembly
view sequences
The February 2009 human reference sequence (GRCh37) was produced by the Genome Reference Consortium. For more information about this assembly, see GRCh37 in the NCBI Assembly database.
Sample position queries
A genome position can be specified by the accession number of a sequenced genomic clone, an mRNA or EST or STS marker, a chromosomal coordinate range, or keywords from the GenBank description of an mRNA. The following list shows examples of valid position queries for the human genome. See the User's Guide for more information.
Request:
Genome Browser Response:
chr7 Displays all of chromosome 7
chrUn_gl000212 Displays all of the unplaced contig gl000212
20p13 Displays region for band p13 on chr 20
chr3:1-1000000 Displays first million bases of chr 3, counting from p-arm telomere
chr3:1000000+2000 Displays a region of chr3 that spans 2000 bases, starting with position 1000000
U C S C
Homo sapiens
(Graphic courtesy of CBSE)

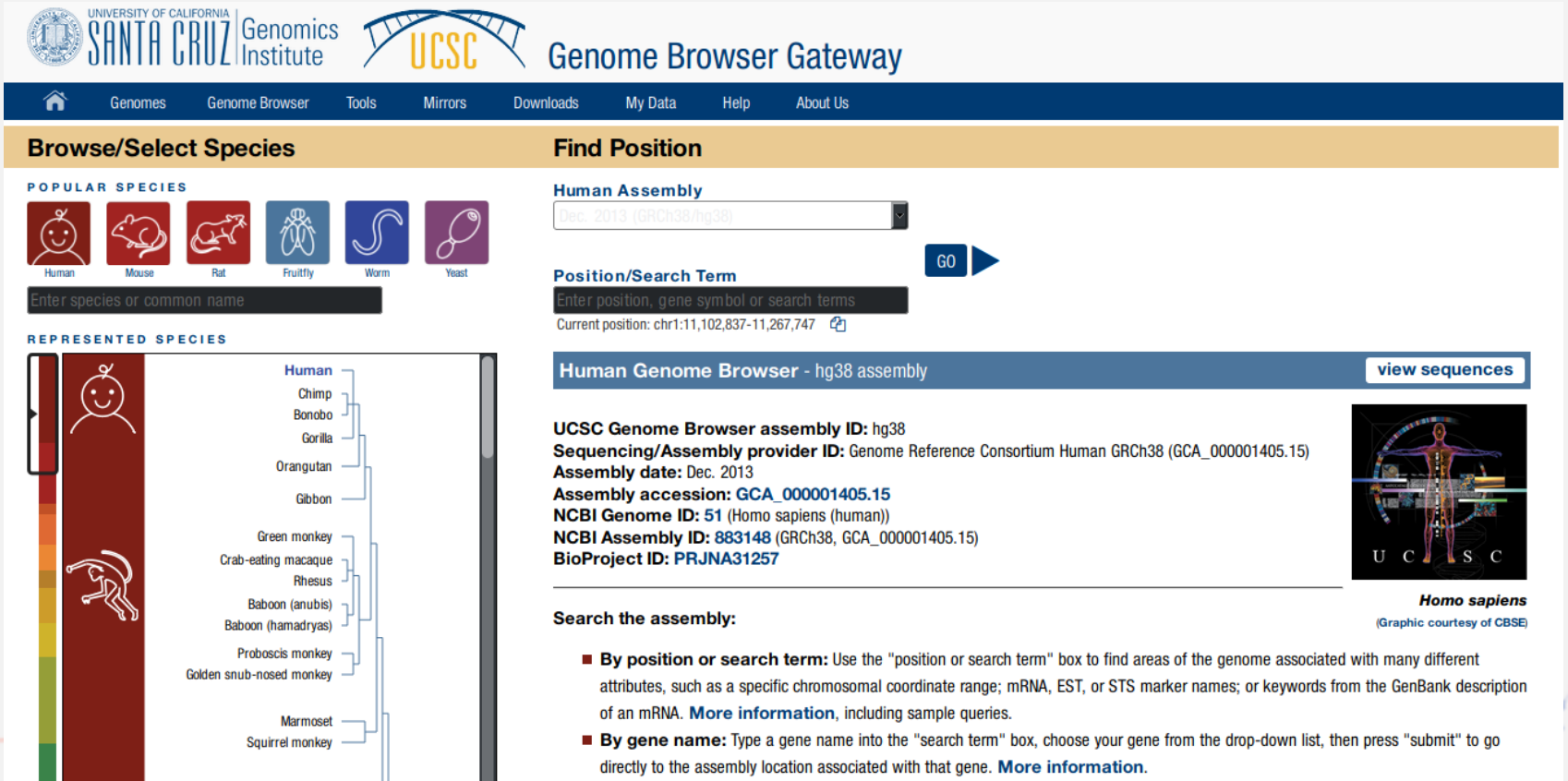
GRCh37


NCBI Resources How To
barslmn@gmail.com My NCBI Sign Out
Assembly
Assembly
Search
Advanced Browse by organism
Help
Full Report
Send to:
GRCh37
This assembly has been updated. See current version
Description: Genome Reference Consortium Human Build 37 (GRCh37)
Organism name: Homo sapiens (human)
BioProject: PRJNA31257
Submitter: Genome Reference Consortium
Date: 2009/02/27
Synonyms: hg19
Assembly type: haploid-with-alt-loci
Assembly level: Chromosome
Genome representation: full
GenBank assembly accession: GCA_000001405.1 (replaced)
RefSeq assembly accession: GCF_000001405.13 (replaced)
RefSeq assembly and GenBank assembly identical: yes
See Genome Information for Homo sapiens
There are 222 assemblies for this organism
See more
Access the data
Browse in Genome Data Viewer
Download the GenBank assembly
BLAST search the assembly
Download the full sequence report
Download the statistics report
Download the regions report
Assembly Information
Assembly Help
Assembly Basics
NCBI Assembly Data Model

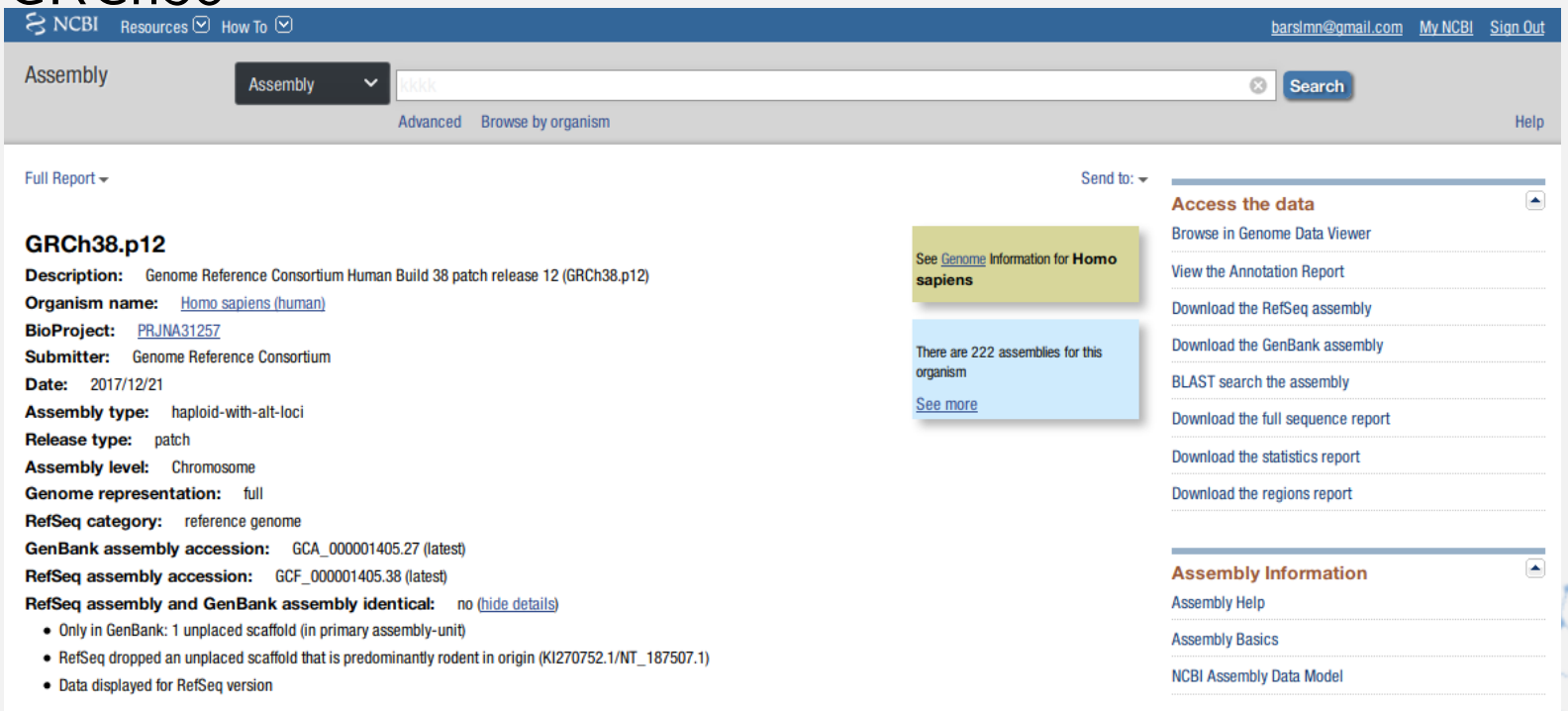
Differences between Google Maps and Yandex Maps (Russian competitor) in Europe

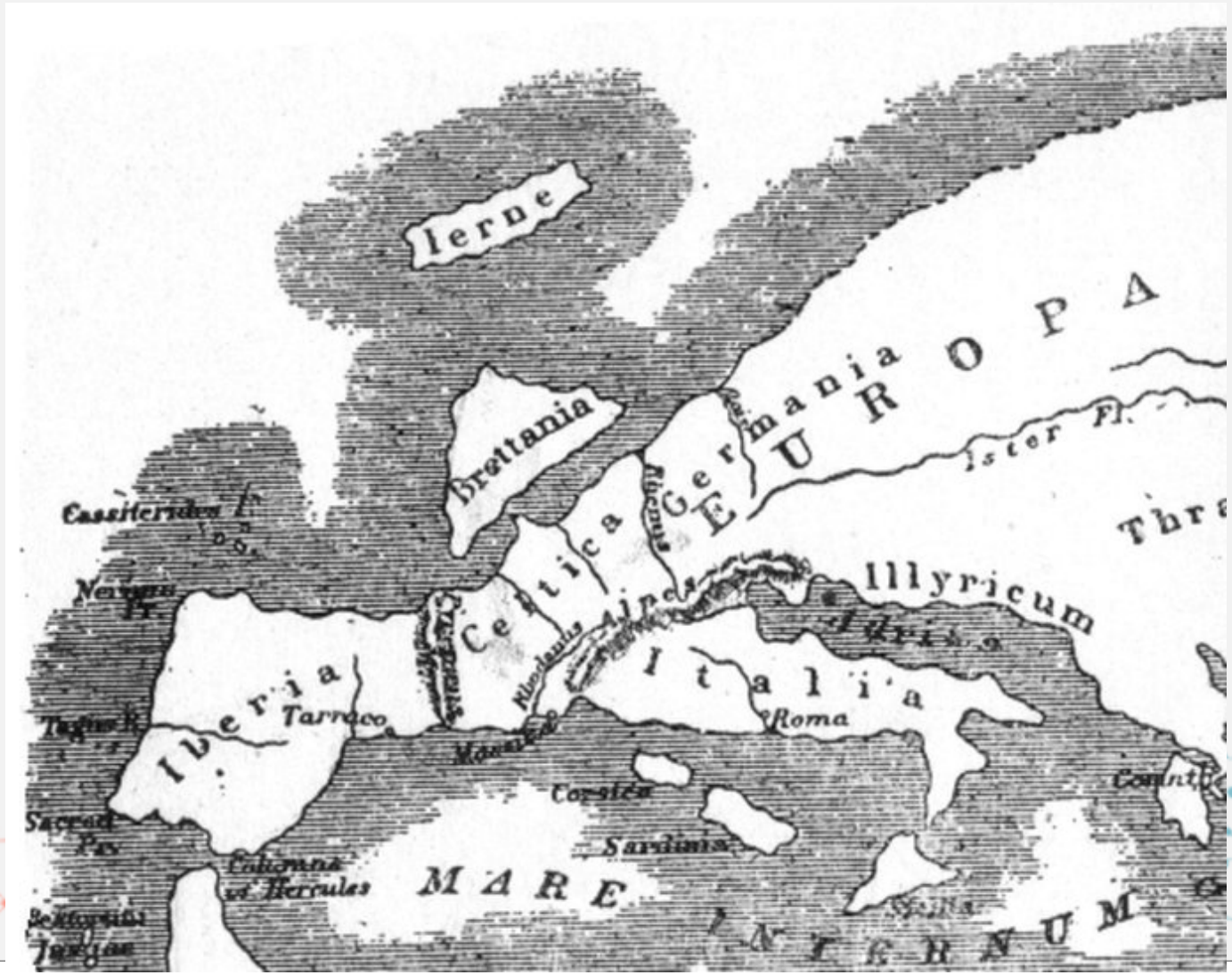
Çıkış Tarihi	UCSC	GRC
Şubat 2009	hg19	GRCh37
Aralık 2013	hg38	GRCh38

hg38


UNIVERSITY OF CALIFORNIA SANTA CRUZ Genomics Institute
UCSC
Genome Browser Gateway
Genomes
Genome Browser
Tools
Mirrors
Downloads
My Data
Help
About Us
Browse/Select Species
POPULAR SPECIES
Human
Mouse
Rat
Fruitfly
Worm
Yeast
Enter species or common name
REPRESENTED SPECIES
Human
Chimp
Bonobo
Gorilla
Orangutan
Gibbon
Green monkey
Crab-eating macaque
Rhesus
Baboon (anubis)
Baboon (hamadryas)
Proboscis monkey
Golden snub-nosed monkey
Marmoset
Squirrel monkey
Find Position
Human Assembly
Dec. 2013 (GRCh38/hg38)
GO
Position/Search Term
Enter position, gene symbol or search terms
Current position: chr1:11,102,837-11,267,747
Human Genome Browser - hg38 assembly
view sequences
UCSC Genome Browser assembly ID: hg38
Sequencing/Assembly provider ID: Genome Reference Consortium Human GRCh38 (GCA_000001405.15)
Assembly date: Dec. 2013
Assembly accession: GCA_000001405.15
NCBI Genome ID: 51 (Homo sapiens (human))
NCBI Assembly ID: 883148 (GRCh38, GCA_000001405.15)
BioProject ID: PRJNA31257
Homo sapiens
(Graphic courtesy of CBSE)
Search the assembly:
By position or search term: Use the "position or search term" box to find areas of the genome associated with many different attributes, such as a specific chromosomal coordinate range; mRNA, EST, or STS marker names; or keywords from the GenBank description of an mRNA. More information, including sample queries.
By gene name: Type a gene name into the "search term" box, choose your gene from the drop-down list, then press "submit" to go directly to the assembly location associated with that gene. More information.

GRCh38


NCBI Resources How To
barslmn@gmail.com My NCBI Sign Out
Assembly
Assembly
Search
Advanced Browse by organism
Help
Full Report
Send to:
GRCh38.p12
Description: Genome Reference Consortium Human Build 38 patch release 12 (GRCh38.p12)
Organism name: Homo sapiens (human)
BioProject: PRJNA31257
Submitter: Genome Reference Consortium
Date: 2017/12/21
Assembly type: haploid-with-alt-loci
Release type: patch
Assembly level: Chromosome
Genome representation: full
RefSeq category: reference genome
GenBank assembly accession: GCA_000001405.27 (latest)
RefSeq assembly accession: GCF_000001405.38 (latest)
RefSeq assembly and GenBank assembly identical: no (hide details)
Only in GenBank: 1 unplaced scaffold (in primary assembly-unit)
RefSeq dropped an unplaced scaffold that is predominantly rodent in origin (KI270752.1/NT_187507.1)
Data displayed for RefSeq version
See Genome Information for Homo sapiens
There are 222 assemblies for this organism
See more
Access the data
Browse in Genome Data Viewer
View the Annotation Report
Download the RefSeq assembly
Download the GenBank assembly
BLAST search the assembly
Download the full sequence report
Download the statistics report
Download the regions report
Assembly Information
Assembly Help
Assembly Basics
NCBI Assembly Data Model

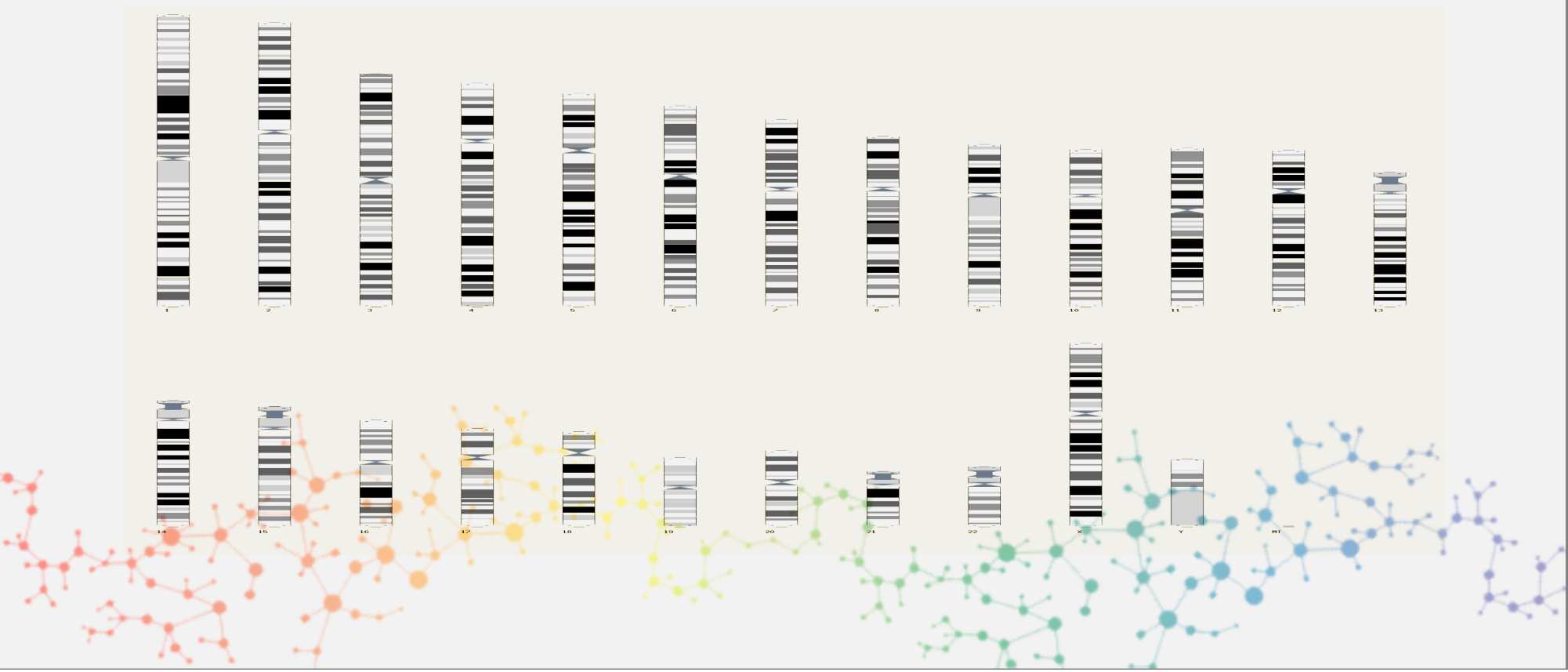

Ierne
Brettania
Germania
EUROPA
Ister F.
Thra
Illyricum
Iberia
Tarraco
Roma
Corsica
Sardinia
MARE
TYRRENUM

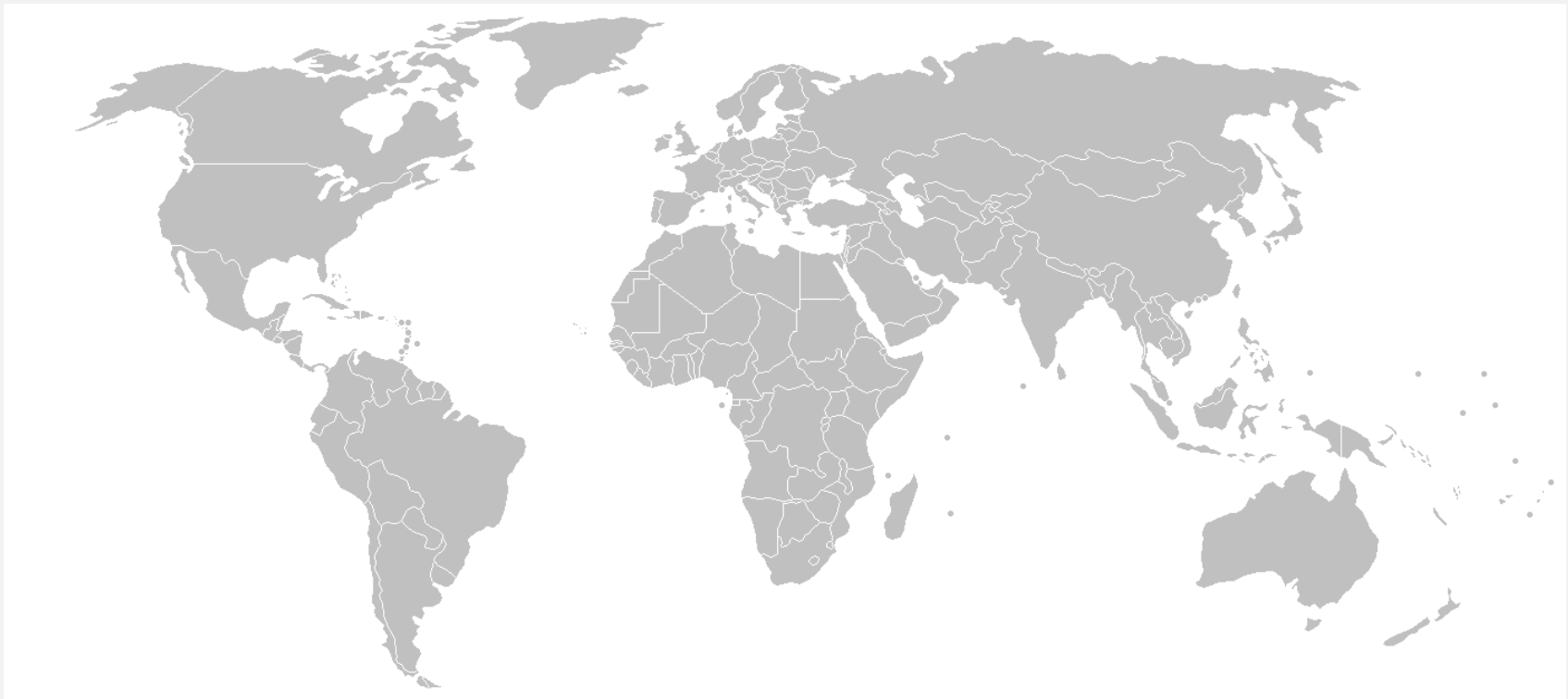
Genom Haritalama

Barış Salman

Gen-Era Diagnostik

Fiziksel Haritalama





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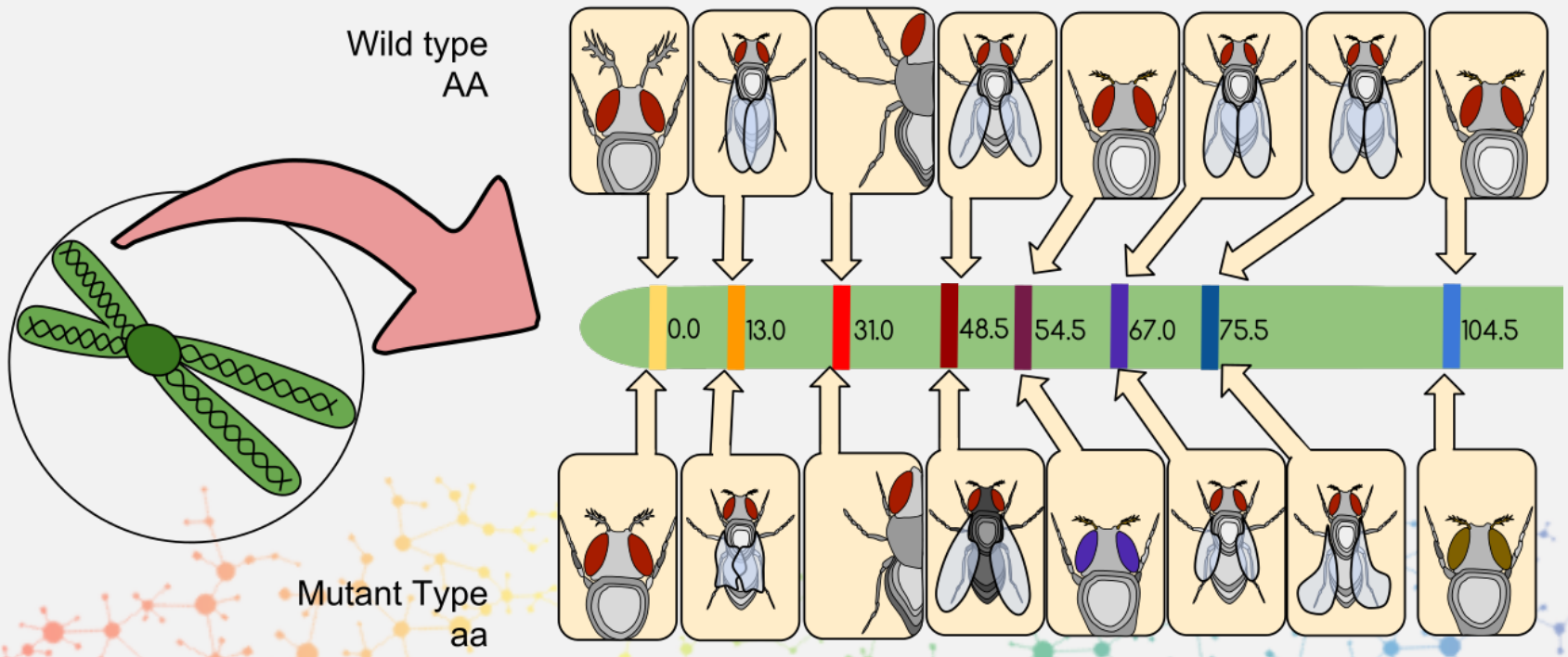
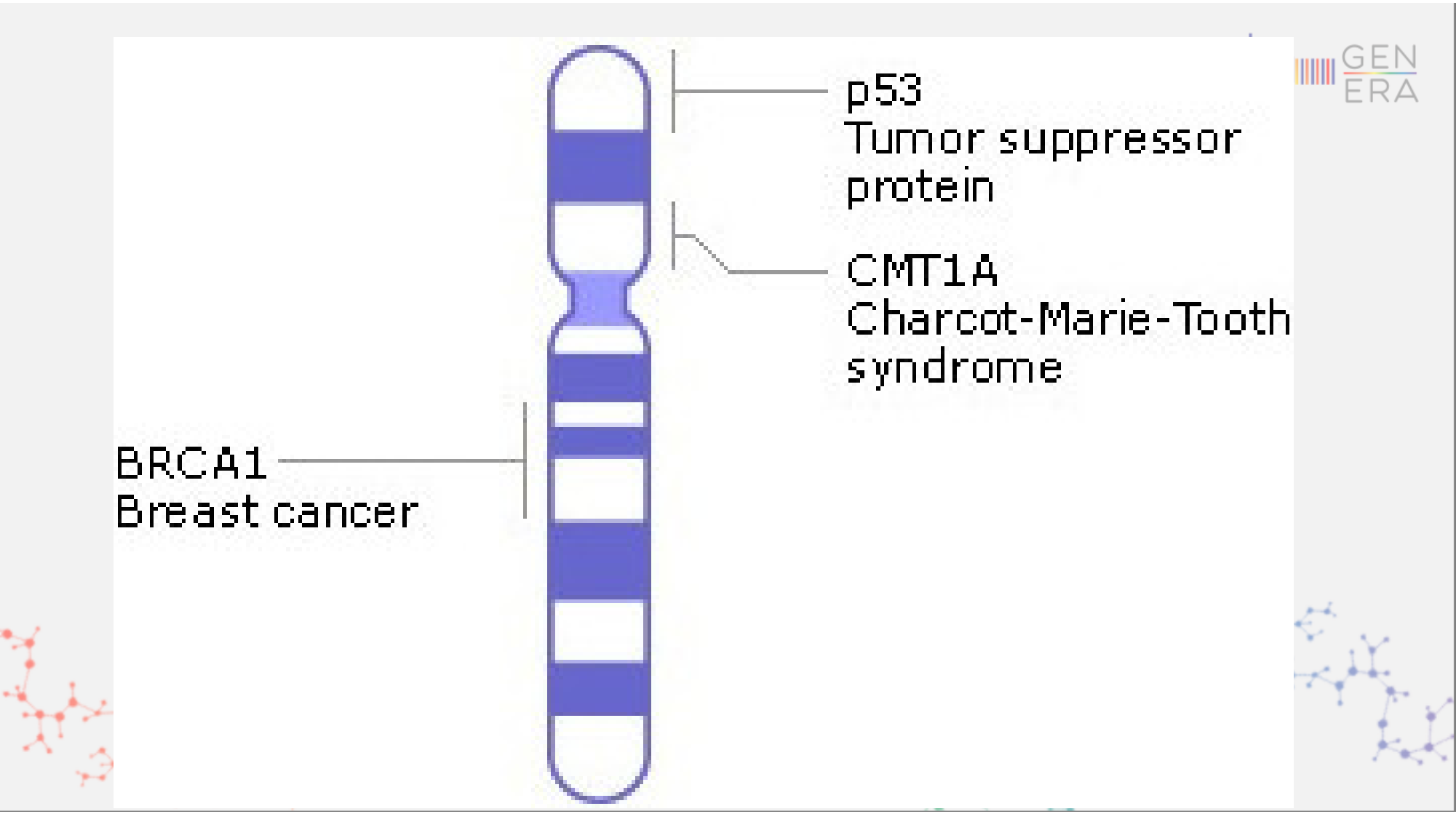
Genetik Haritalama




p53
Tumor suppressor protein
CMT1A
Charcot-Marie-Tooth syndrome
BRCA1
Breast cancer
GEN ERA

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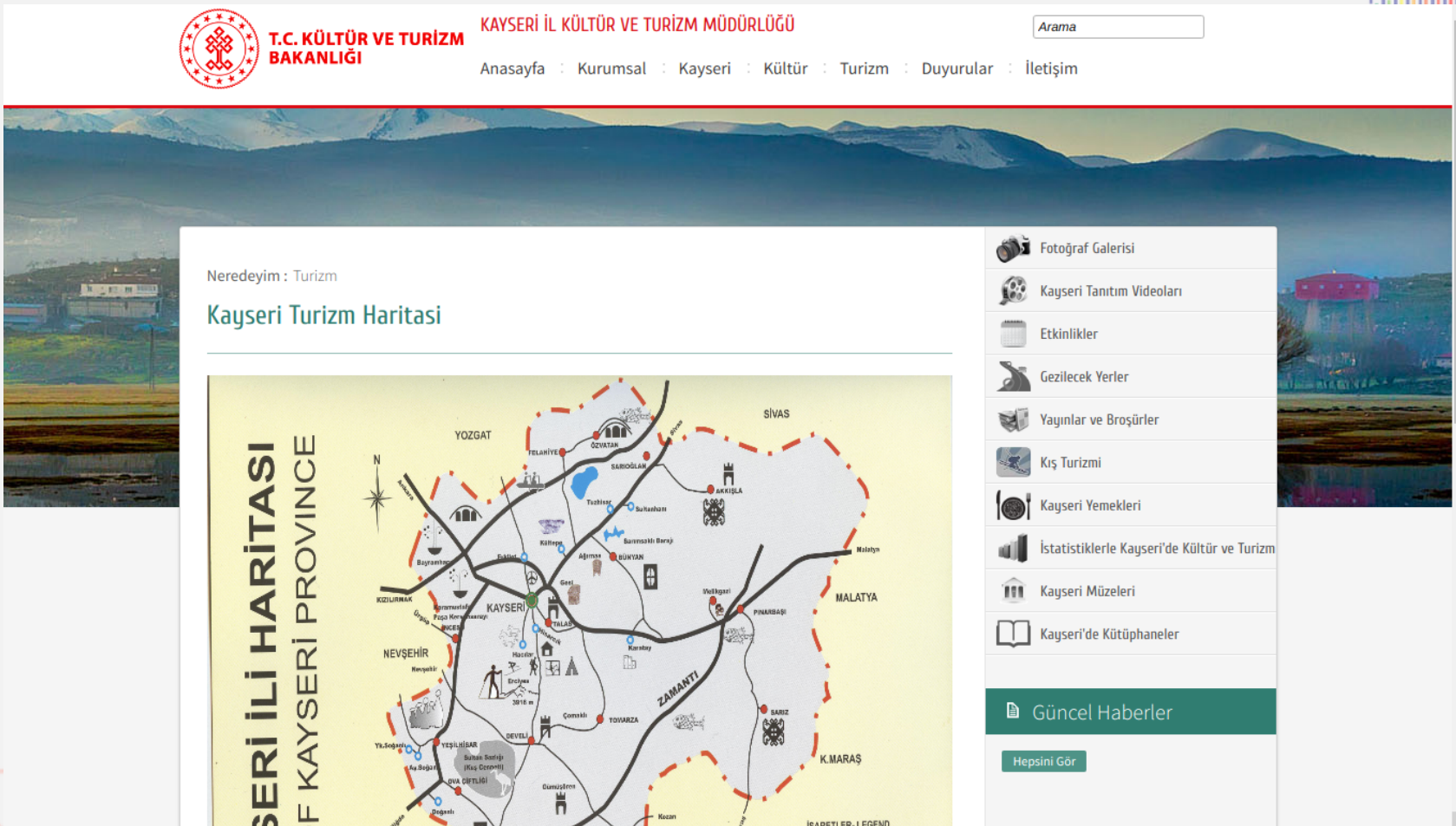


Barış Salman
Gen-Era Diagnostik

NCBI Gene

WNT10B Wnt family member 10B [*Homo sapiens* (human)]

Gene ID: 7480, updated on 13-Feb-2019

Summary

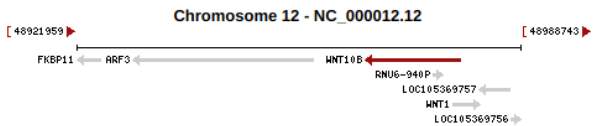
Official Symbol	WNT10B provided by HGNC
Official Full Name	Wnt family member 10B provided by HGNC
Primary source	HGNC:HGNC:12775
See related	Ensembl:ENSG00000169884 MIM:601906
Gene type	protein coding
RefSeq status	REVIEWED
Organism	Homo sapiens
Lineage	Eukaryota; Metazoa; Chordata; Craniata; Vertebrata; Euteleostomi; Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini; Catarrhini; Hominidae; Homo
Also known as	SHFM6; STHAG8; WNT-12
Summary	The WNT gene family consists of structurally related genes which encode secreted signaling proteins. These proteins have been implicated in oncogenesis and in several developmental processes, including regulation of cell fate and patterning during embryogenesis. This gene is a member of the WNT gene family. It may be involved in breast cancer, and its protein signaling is likely a molecular switch that governs adipogenesis. This protein is 96% identical to the mouse Wnt10b protein at the amino acid level. This gene is clustered with another family member, WNT1, in the chromosome 12q13 region. [provided by RefSeq, Jul 2008]
Expression	Broad expression in brain (RPKM 3.0), skin (RPKM 0.8) and 15 other tissues See more
Orthologs	mouse all

Genomic context

Location: 12q13.12 — See WNT10B in Genome Data Viewer

Exon count: 8

Annotation release	Status	Assembly	Chr	Location
109	current	GRCh38.p12 (GCF_000001405.38)	12	NC_000012.12 (48965340..48979587, complement)
105	previous assembly	GRCh37.p13 (GCF_000001405.25)	12	NC_000012.11 (49359123..49365641, complement)



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- Order cDNA clone

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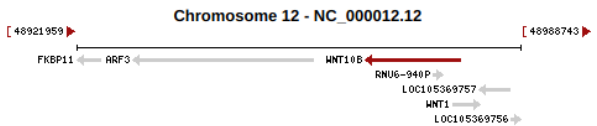

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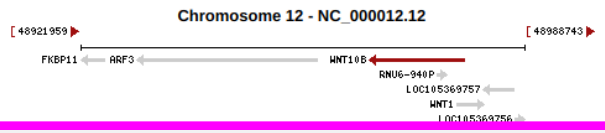

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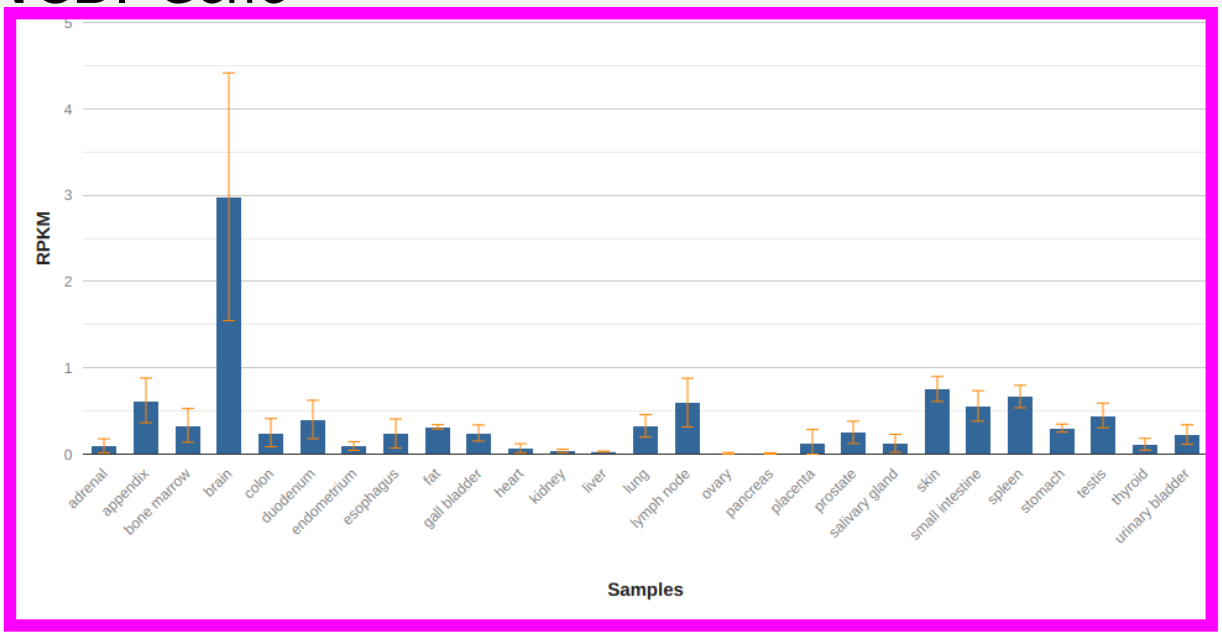

Related articles in PubMed

1. Split hand-foot malformation and a novel WNT10B mutation.
   Kantaputra PN, *et al.* Eur J Med Genet, 2018 Jul. PMID 29427788
2. Relevance of Wnt10b and activation of β-catenin/GCMa/syncytin-1 pathway in BeWo cell fusion.
   Malhotra SS, *et al.* Am J Reprod Immunol, 2017 Oct. PMID 28370659
3. CD8+ T cells mediate the regenerative PTH effect in hPDL cells via Wnt10b signaling.
   Wolf M, *et al.* Innate Immun, 2016 Nov. PMID 28071181
4. Prolonged overexpression of Wnt10b induces epidermal keratinocyte transformation through activating EGF pathway.
   Lei M, *et al.* Histochem Cell Biol, 2015 Sep. PMID 25995040, **Free PMC Article**
5. Novel homozygous mutations in the WNT10B gene underlying autosomal recessive split hand/foot malformation in three consanguineous families.

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UCSC

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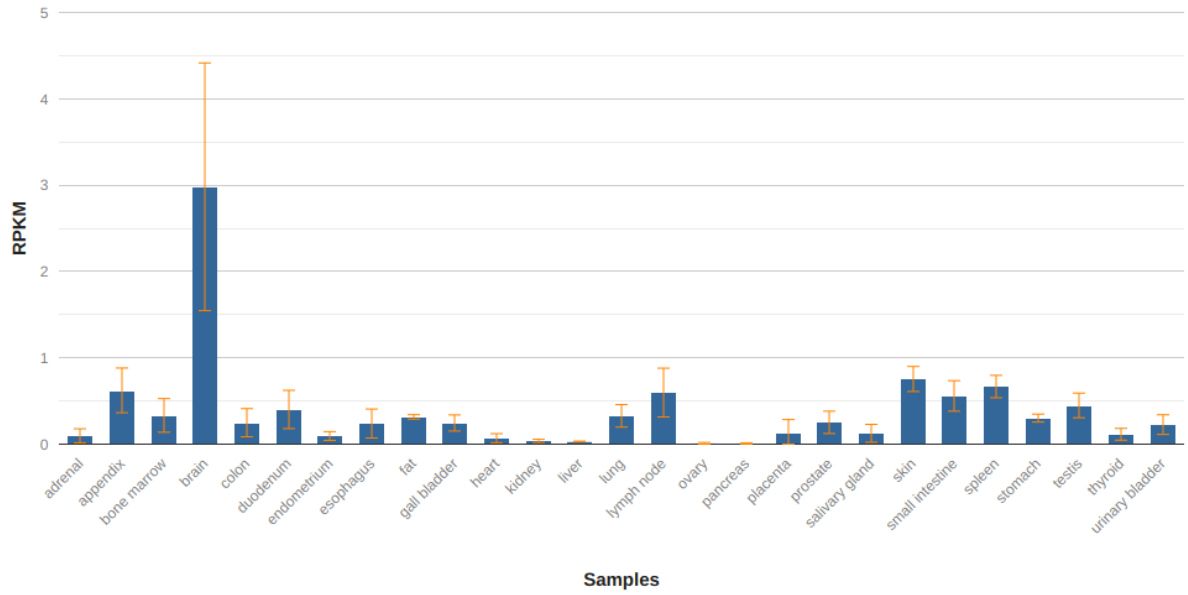
BioAssay by Target (List)
BioAssay by Target (Summary)
BioAssay, by Gene target
BioAssays, RNAi Target, Active

NCBI Gene



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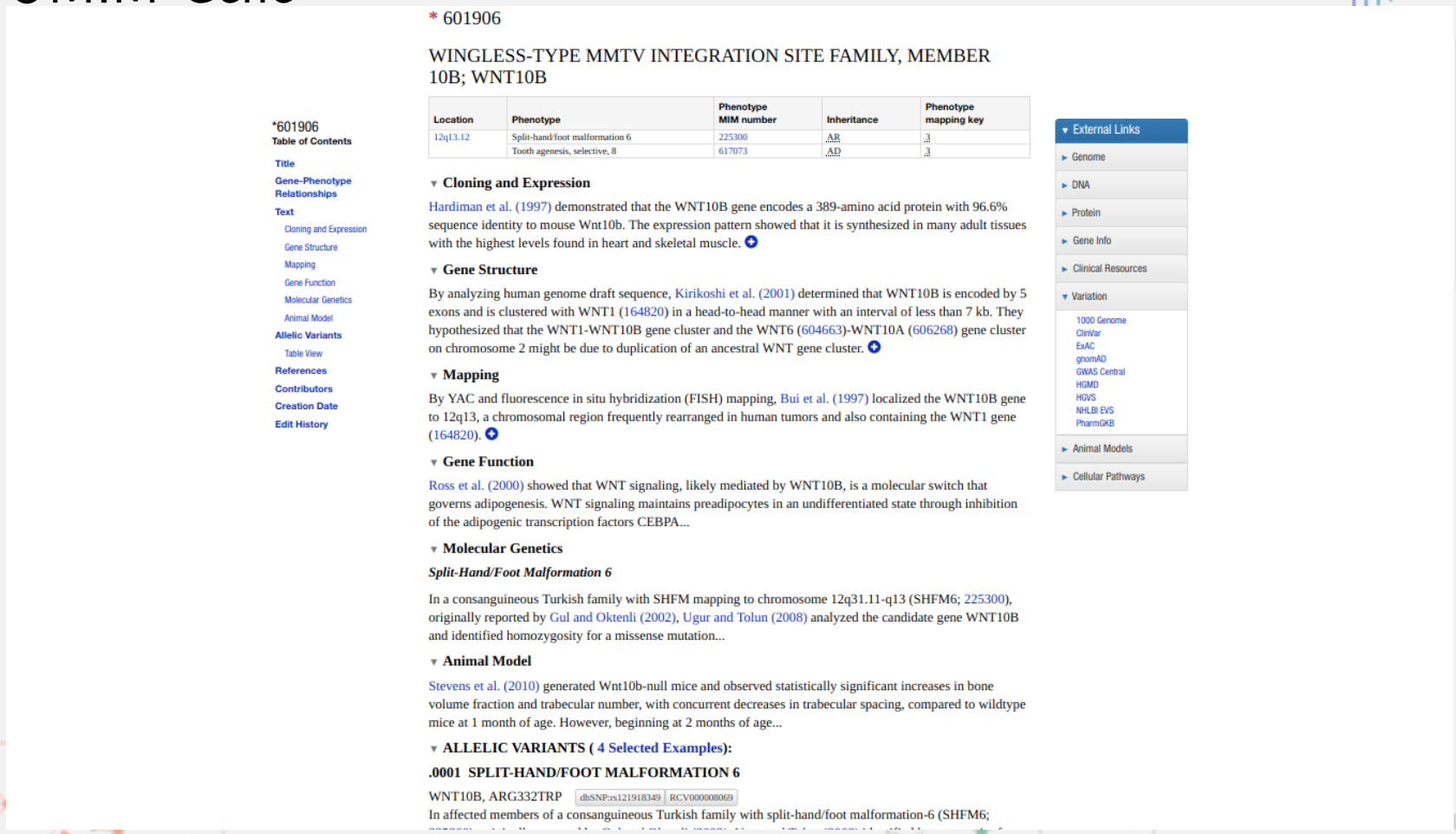
BioAssay by Target (List)
BioAssay by Target (Summary)
BioAssay, by Gene target
BioAssays, RNAi Target, Active

OMIM Gene



* 601906

WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 10B; WNT10B

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key
12q13.12	Split-hand/foot malformation 6	225300	AR	3
	Tooth agenesis, selective, 8	617073	AD	3

*601906
Table of Contents
Title
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Table View
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Contributors
Creation Date
Edit History

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DNA
Protein
Gene Info
Clinical Resources
Variation
1000 Genome
ClinVar
ExAC
gnomAD
GWAS Central
HGMD
HGVS
NHLBI EVS
PharmGKB
Animal Models
Cellular Pathways

Cloning and Expression

Hardiman et al. (1997) demonstrated that the WNT10B gene encodes a 389-amino acid protein with 96.6% sequence identity to mouse Wnt10b. The expression pattern showed that it is synthesized in many adult tissues with the highest levels found in heart and skeletal muscle.

Gene Structure

By analyzing human genome draft sequence, Kirikoshi et al. (2001) determined that WNT10B is encoded by 5 exons and is clustered with WNT1 (164820) in a head-to-head manner with an interval of less than 7 kb. They hypothesized that the WNT1-WNT10B gene cluster and the WNT6 (604663)-WNT10A (606268) gene cluster on chromosome 2 might be due to duplication of an ancestral WNT gene cluster.

Mapping

By YAC and fluorescence in situ hybridization (FISH) mapping, Bui et al. (1997) localized the WNT10B gene to 12q13, a chromosomal region frequently rearranged in human tumors and also containing the WNT1 gene (164820).

Gene Function

Ross et al. (2000) showed that WNT signaling, likely mediated by WNT10B, is a molecular switch that governs adipogenesis. WNT signaling maintains preadipocytes in an undifferentiated state through inhibition of the adipogenic transcription factors CEBPA...

Molecular Genetics

Split-Hand/Foot Malformation 6

In a consanguineous Turkish family with SHFM mapping to chromosome 12q31.11-q13 (SHFM6; 225300), originally reported by Gul and Oktenli (2002), Ugur and Tolun (2008) analyzed the candidate gene WNT10B and identified homozygosity for a missense mutation...

Animal Model

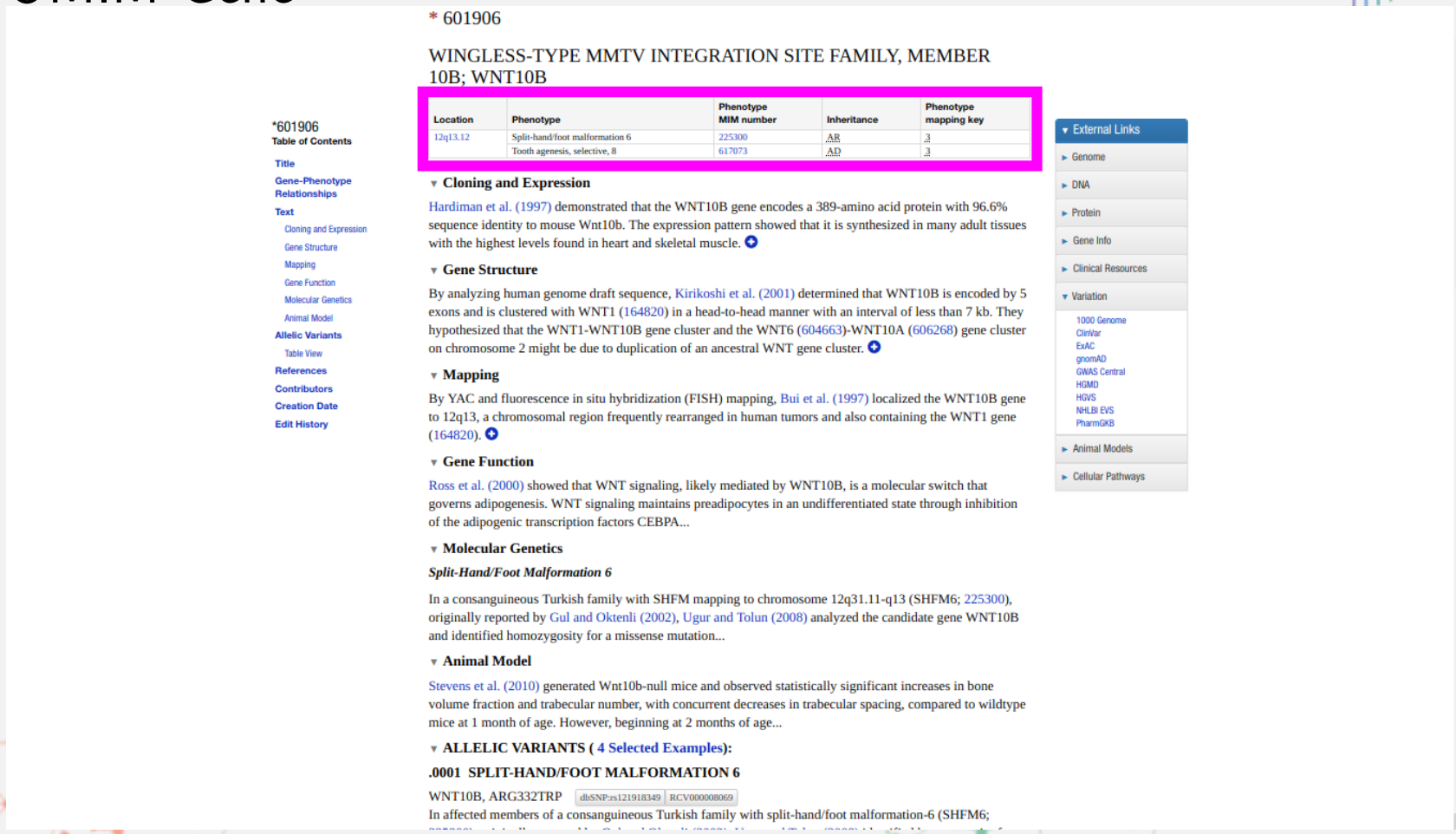
Stevens et al. (2010) generated Wnt10b-null mice and observed statistically significant increases in bone volume fraction and trabecular number, with concurrent decreases in trabecular spacing, compared to wildtype mice at 1 month of age. However, beginning at 2 months of age...

ALLELIC VARIANTS (4 Selected Examples):

.0001 SPLIT-HAND/FOOT MALFORMATION 6

WNT10B, ARG332TRP dbSNP:rs121918349 RCV000008069
In affected members of a consanguineous Turkish family with split-hand/foot malformation-6 (SHFM6;

OMIM Gene



* 601906

WINGLESS-TYPE MMTV INTEGRATION SITE FAMILY, MEMBER 10B; WNT10B

Location	Phenotype	Phenotype MIM number	Inheritance	Phenotype mapping key
12q13.12	Split-hand/foot malformation 6	225300	AR	3
	Tooth agenesis, selective, 8	617073	AD	3

*601906
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Cloning and Expression

Hardiman et al. (1997) demonstrated that the WNT10B gene encodes a 389-amino acid protein with 96.6% sequence identity to mouse Wnt10b. The expression pattern showed that it is synthesized in many adult tissues with the highest levels found in heart and skeletal muscle.

Gene Structure

By analyzing human genome draft sequence, Kirikoshi et al. (2001) determined that WNT10B is encoded by 5 exons and is clustered with WNT1 (164820) in a head-to-head manner with an interval of less than 7 kb. They hypothesized that the WNT1-WNT10B gene cluster and the WNT6 (604663)-WNT10A (606268) gene cluster on chromosome 2 might be due to duplication of an ancestral WNT gene cluster.

Mapping

By YAC and fluorescence in situ hybridization (FISH) mapping, Bui et al. (1997) localized the WNT10B gene to 12q13, a chromosomal region frequently rearranged in human tumors and also containing the WNT1 gene (164820).

Gene Function

Ross et al. (2000) showed that WNT signaling, likely mediated by WNT10B, is a molecular switch that governs adipogenesis. WNT signaling maintains preadipocytes in an undifferentiated state through inhibition of the adipogenic transcription factors CEBPA...

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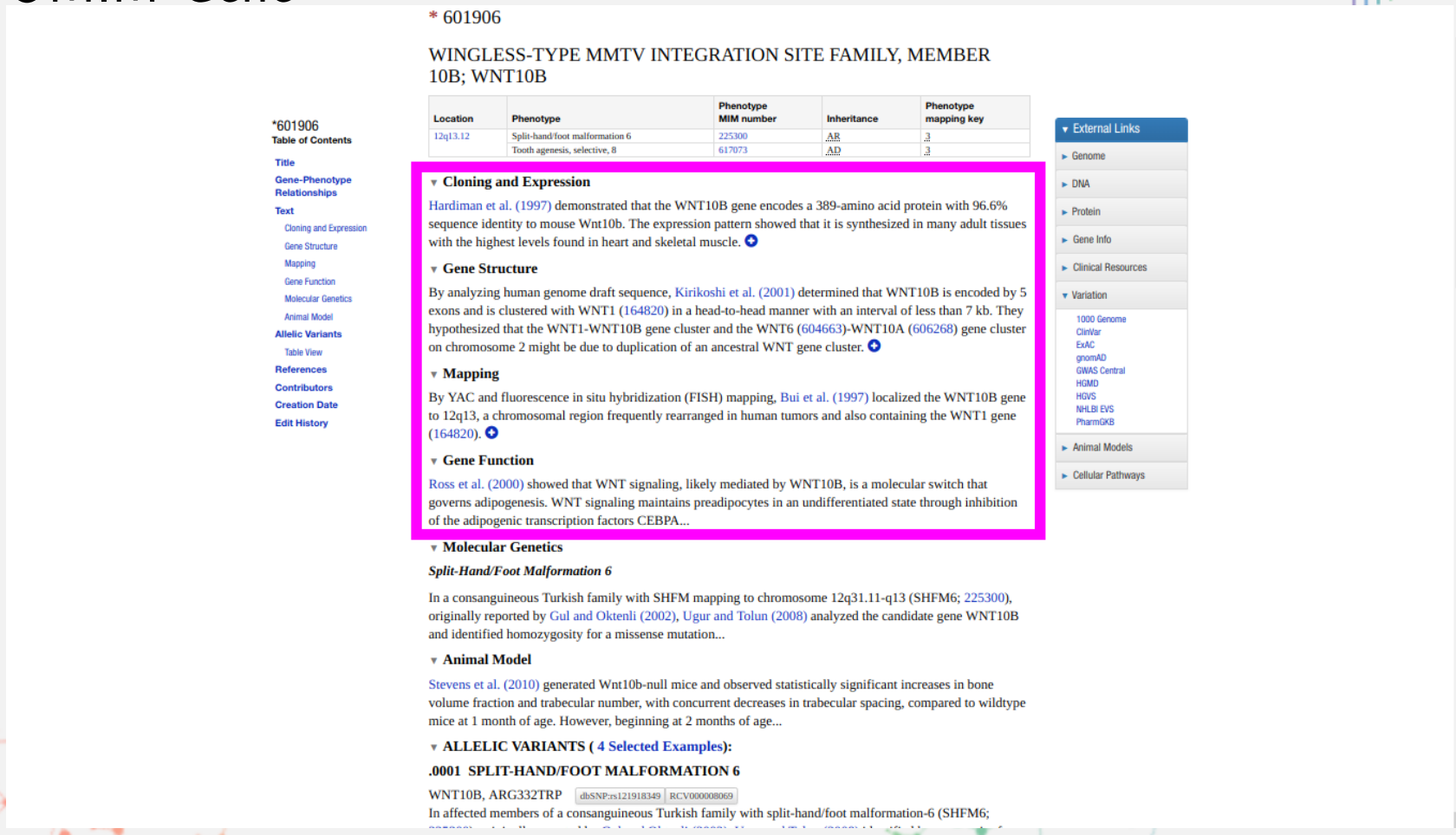

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Genomik İşaretleyiciler

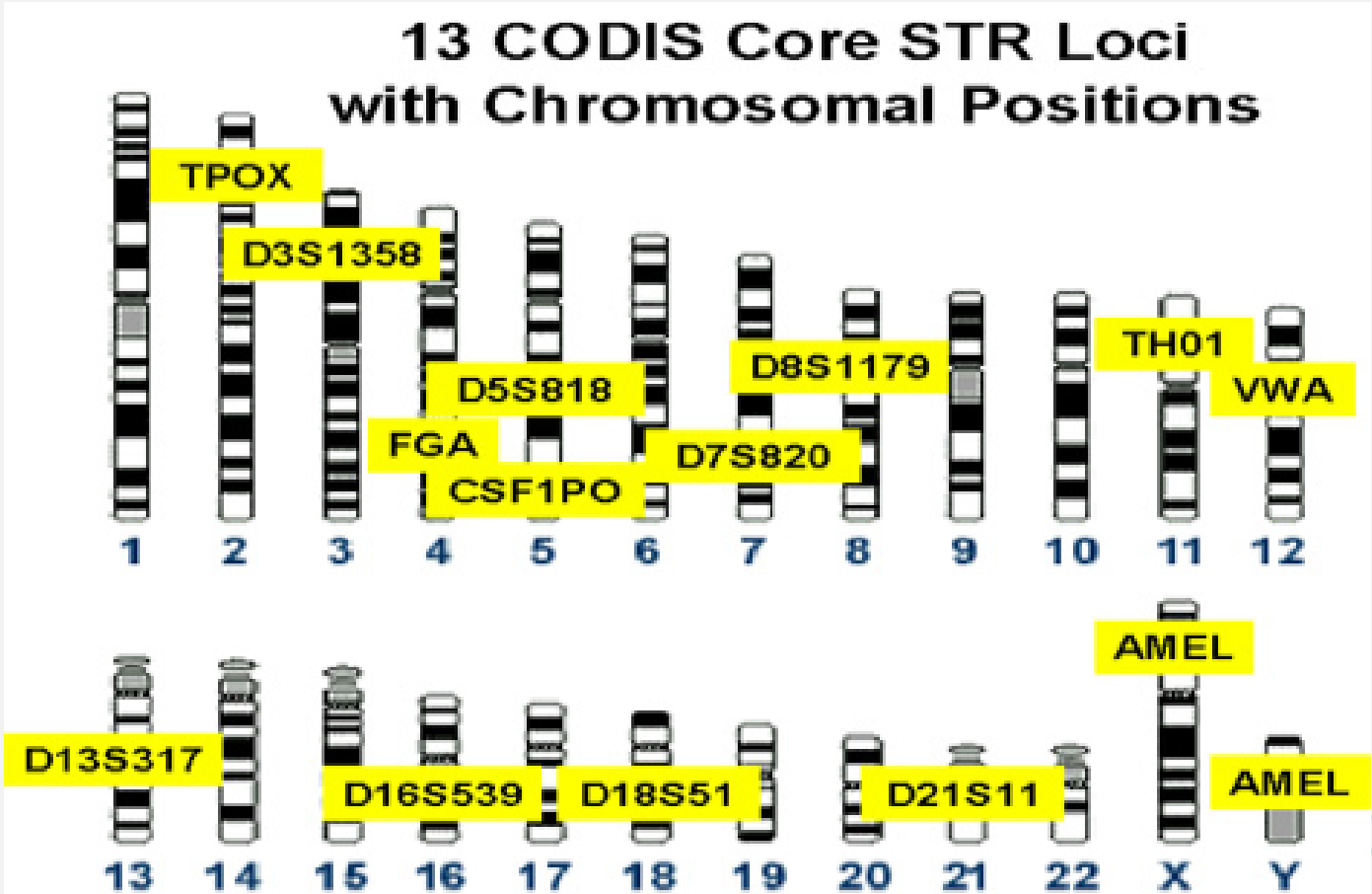
Barış Salman

Gen-Era Diagnostik

Barış Salman

Gen-Era Diagnostik

STR ~ 700k


13 CODIS Core STR Loci
with Chromosomal Positions
TPOX
D3S1358
D5S818
FGA
CSF1PO
D7S820
D8S1179
TH01
VWA
AMEL
AMEL
D13S317
D16S539
D18S51
D21S11
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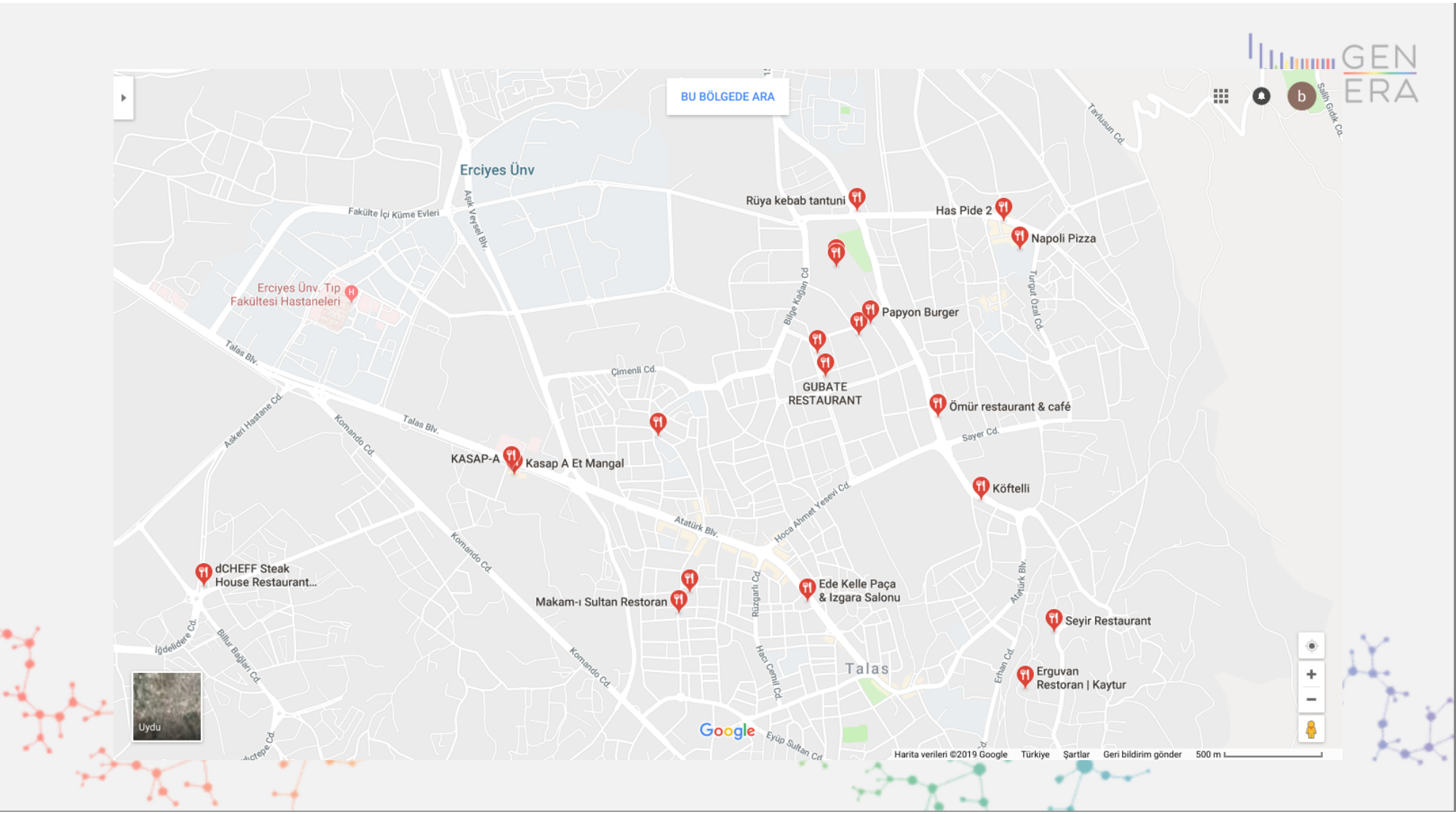
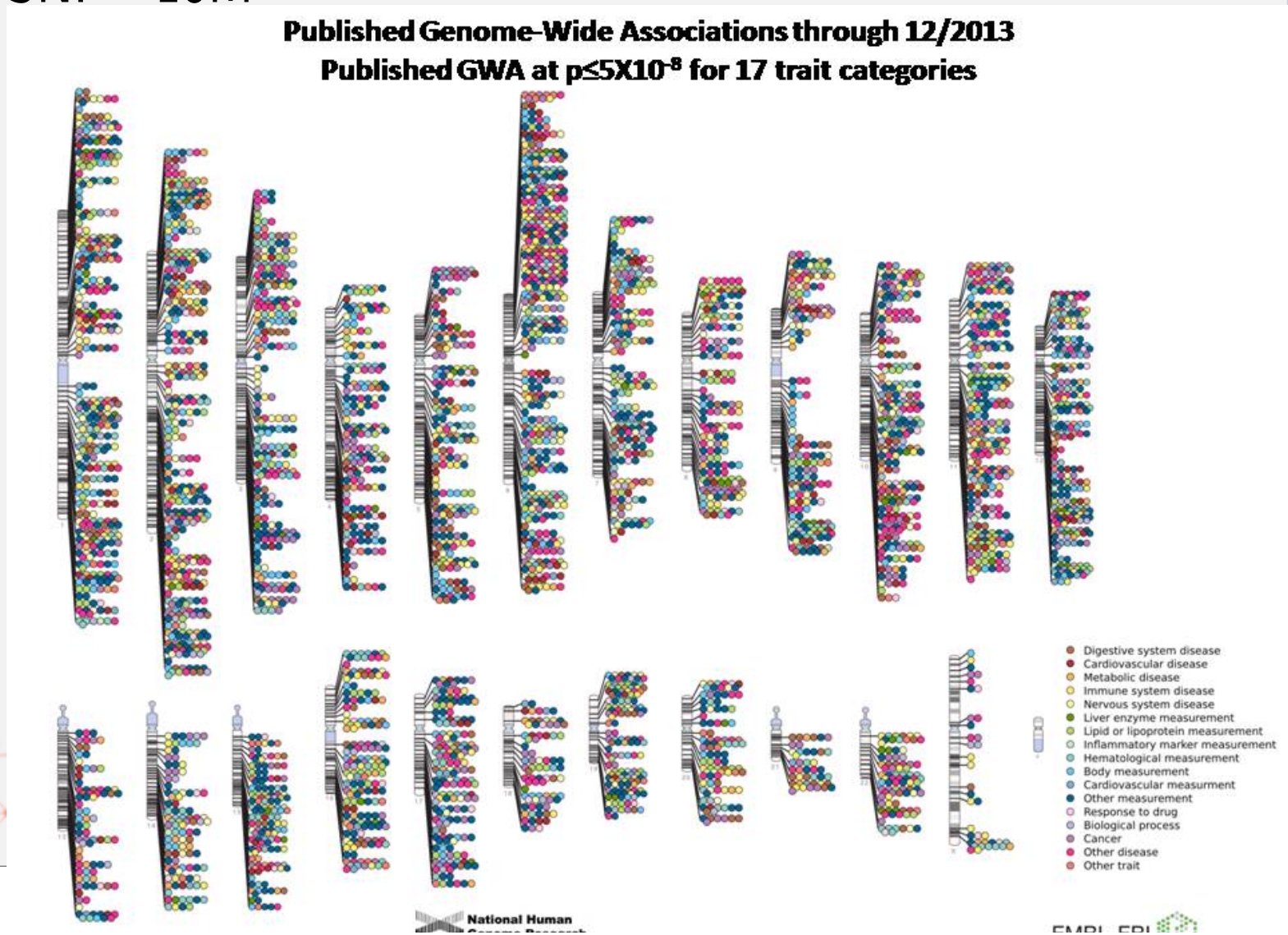
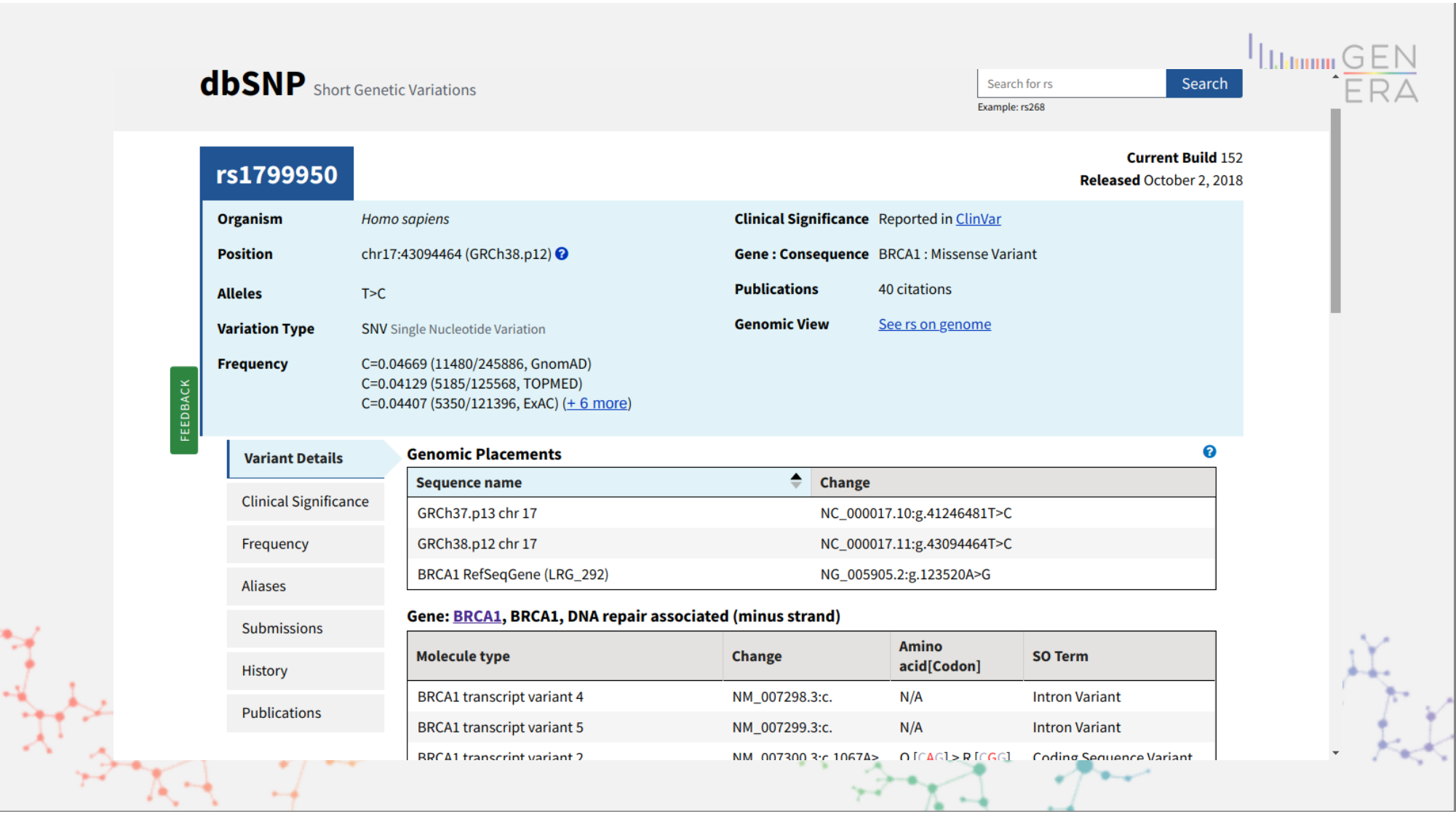
13 14 15 16 17 18 19 20 21 22 X Y


BU BÖLGEDE ARA
Erciyes Ünv
Fakülte İçi Küme Evleri
Aşık Veysel Blv.
Erciyes Ünv. Tıp Fakültesi Hastaneleri
Talas Blv.
Askeri Hastane Cd.
Komando Cd.
Çimenli Cd.
Bilge Kağan Cd
Turgut Özal Cd.
Rüya kebab tantuni
Has Pide 2
Napoli Pizza
Papyon Burger
GUBATE RESTAURANT
Ömür restaurant & café
Sayer Cd.
Köftelli
KASAP-A
Kasap A Et Mangal
Atatürk Blv.
Hoca Ahmet Yesevi Cd.
dCHEFF Steak House Restaurant...
Makam-ı Sultan Restoran
Rüzgarlı Cd.
Ede Kelle Paça & Izgara Salonu
Seyir Restaurant
Erguvan Restoran | Kaytur
Talas
Google
Harita verileri ©2019 Google
Türkiye
Şartlar
Geri bildirim gönder
500 m
Uydu

SNP ~10M


Published Genome-Wide Associations through 12/2013
Published GWA at p≤5X10-8 for 17 trait categories
Digestive system disease
Cardiovascular disease
Metabolic disease
Immune system disease
Nervous system disease
Liver enzyme measurement
Lipid or lipoprotein measurement
Inflammatory marker measurement
Hematological measurement
Body measurement
Cardiovascular measurment
Other measurement
Response to drug
Biological process
Cancer
Other disease
Other trait
National Human


dbSNP Short Genetic Variations
Search for rs
Search
Example: rs268
GEN ERA
rs1799950
Current Build 152
Released October 2, 2018
Organism Homo sapiens
Position chr17:43094464 (GRCh38.p12)
Alleles T>C
Variation Type SNV Single Nucleotide Variation
Frequency C=0.04669 (11480/245886, GnomAD)
C=0.04129 (5185/125568, TOPMED)
C=0.04407 (5350/121396, ExAC) (+ 6 more)
Clinical Significance Reported in ClinVar
Gene : Consequence BRCA1 : Missense Variant
Publications 40 citations
Genomic View See rs on genome
FEEDBACK
Variant Details
Clinical Significance
Frequency
Aliases
Submissions
History
Publications
Genomic Placements
Sequence name
Change
GRCh37.p13 chr 17
NC_000017.10:g.41246481T>C
GRCh38.p12 chr 17
NC_000017.11:g.43094464T>C
BRCA1 RefSeqGene (LRG_292)
NG_005905.2:g.123520A>G
Gene: BRCA1, BRCA1, DNA repair associated (minus strand)
Molecule type
Change
Amino acid[Codon]
SO Term
BRCA1 transcript variant 4
NM_007298.3:c.
N/A
Intron Variant
BRCA1 transcript variant 5
NM_007299.3:c.
N/A
Intron Variant

The Golden State Killer

The Golden State Killer is a rapist and a serial killer who operated between the late 1970s and the mid 1980s. The investigation was at a stalemate for decades, but in the early 1990s, an investigator named Paul Holes decided to utilize the emerging DNA testing technology to solve the case.

It is now commonplace for people to obtain a glimpse of their own genetic profiles using direct-to-consumer (DTC) genetic testing services, like 23andMe and AncestryDNA. Many people use this information to find unknown relatives by comparing their genetic profiles to those contained in large online databases, like GEDmatch. Brilliantly, Detective Holes, had the idea to create a profile using the Golden State Killer's DNA and search for the killer's relatives in the GEDmatch database, where he actually found a distant relative who shared a great-great-great-grandparent with the serial killer (Figure 1).

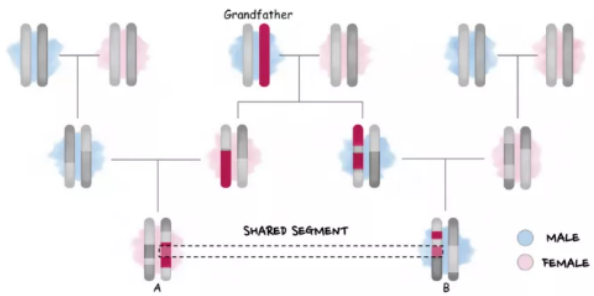
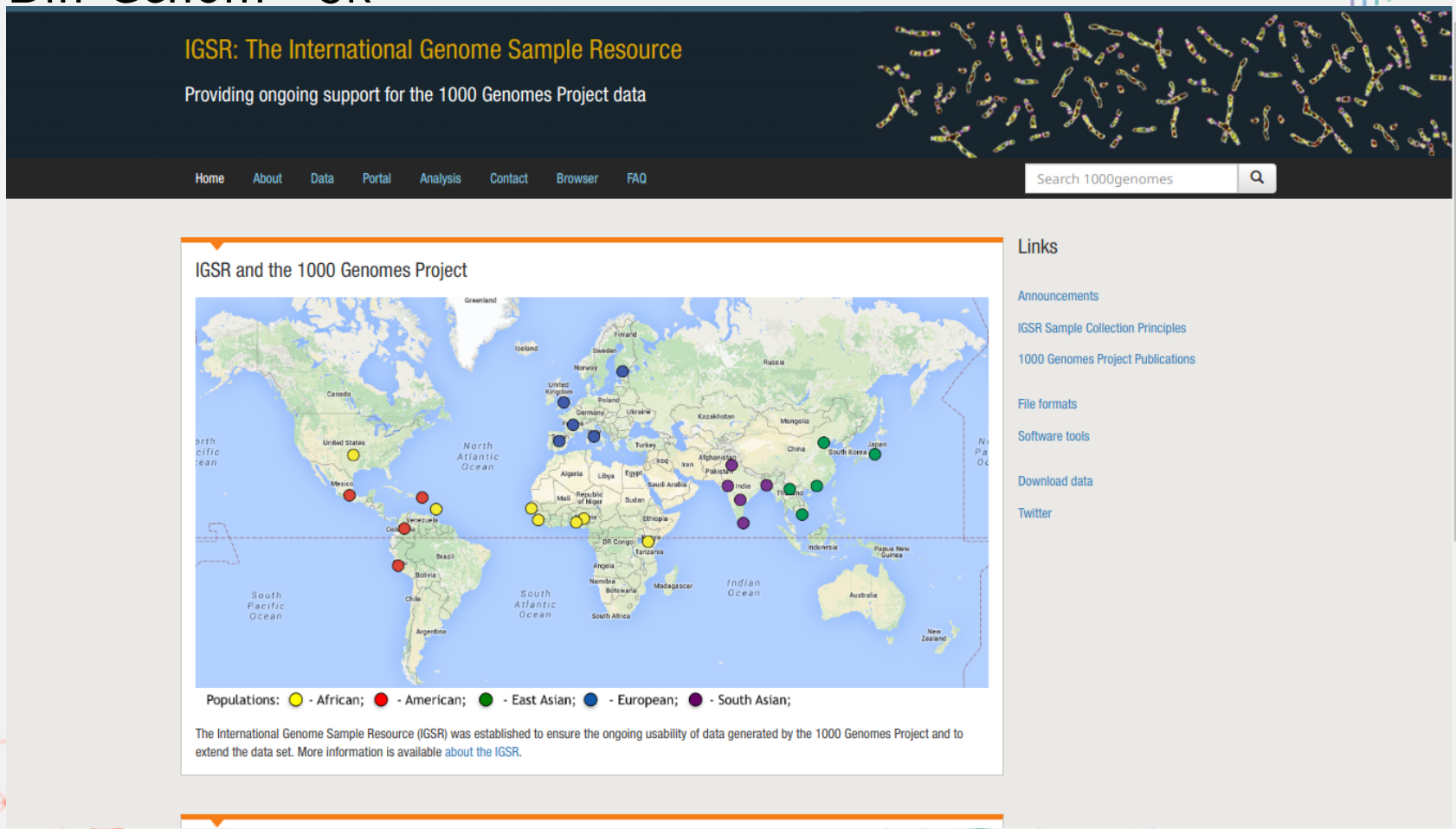


***Figure 1.* The genetic basis of genealogy analyses.** This is an example of a family tree where each individual has two gray bars representing one pair out of the twenty-three pairs of the chromosome. The horizontal lines represent marriage, and the vertical ones, offspring. During one generation, each pair of chromosomes is scrambled before one from each parent is passed down to the offspring. An example is portrayed as the red segments, which are portions of the grandfather's chromosome that were passed down to his children and grandchildren. Tools like GEDmatch are able to find long-lost relatives by computing the degree of shared genetic code between users' profile.

Bin Genom ~3k


IGSR: The International Genome Sample Resource
Providing ongoing support for the 1000 Genomes Project data
Home
About
Data
Portal
Analysis
Contact
Browser
FAQ
Search 1000genomes
IGSR and the 1000 Genomes Project
Populations: - African; - American; - East Asian; - European; - South Asian;
The International Genome Sample Resource (IGSR) was established to ensure the ongoing usability of data generated by the 1000 Genomes Project and to extend the data set. More information is available about the IGSR.
Links
Announcements
IGSR Sample Collection Principles
1000 Genomes Project Publications
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Software tools
Download data
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gnomAD Broad Enstitüsü

Variant: 12-49360054-G-C

Current Dataset: gnomAD v2.1

Annotations

This variant falls on 6 transcript(s) in 1 gene(s).

missense
- WNT10B
  - ENST00000301061 *
    HGVSp: p.Arg332Gly
    Polyphen: probably_damaging
    SIFT: deleterious

3' UTR
- WNT10B
  - ENST00000403957
  - ENST00000407467

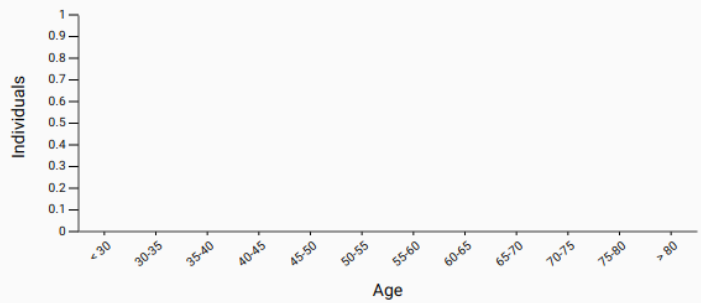
downstream gene
- WNT10B
  - ENST00000413630
  - ENST00000420388
  - ENST00000475740

Population Frequencies

Population	Allele Count	Allele Number	Number of Homozygotes	Allele Frequency
European (non-Finnish)	1	113348	0	0.000008822
African	0	16130	0	0.000
Latino	0	34584	0	0.000
Ashkenazi Jewish	0	10056	0	0.000
East Asian	0	18378	0	0.000
European (Finnish)	0	21602	0	0.000
Other	0	6130	0	0.000
South Asian	0	30614	0	0.000
Total	**1**	**250842**	**0**	**0.000003987**

Include: Exomes Genomes

Age Distribution

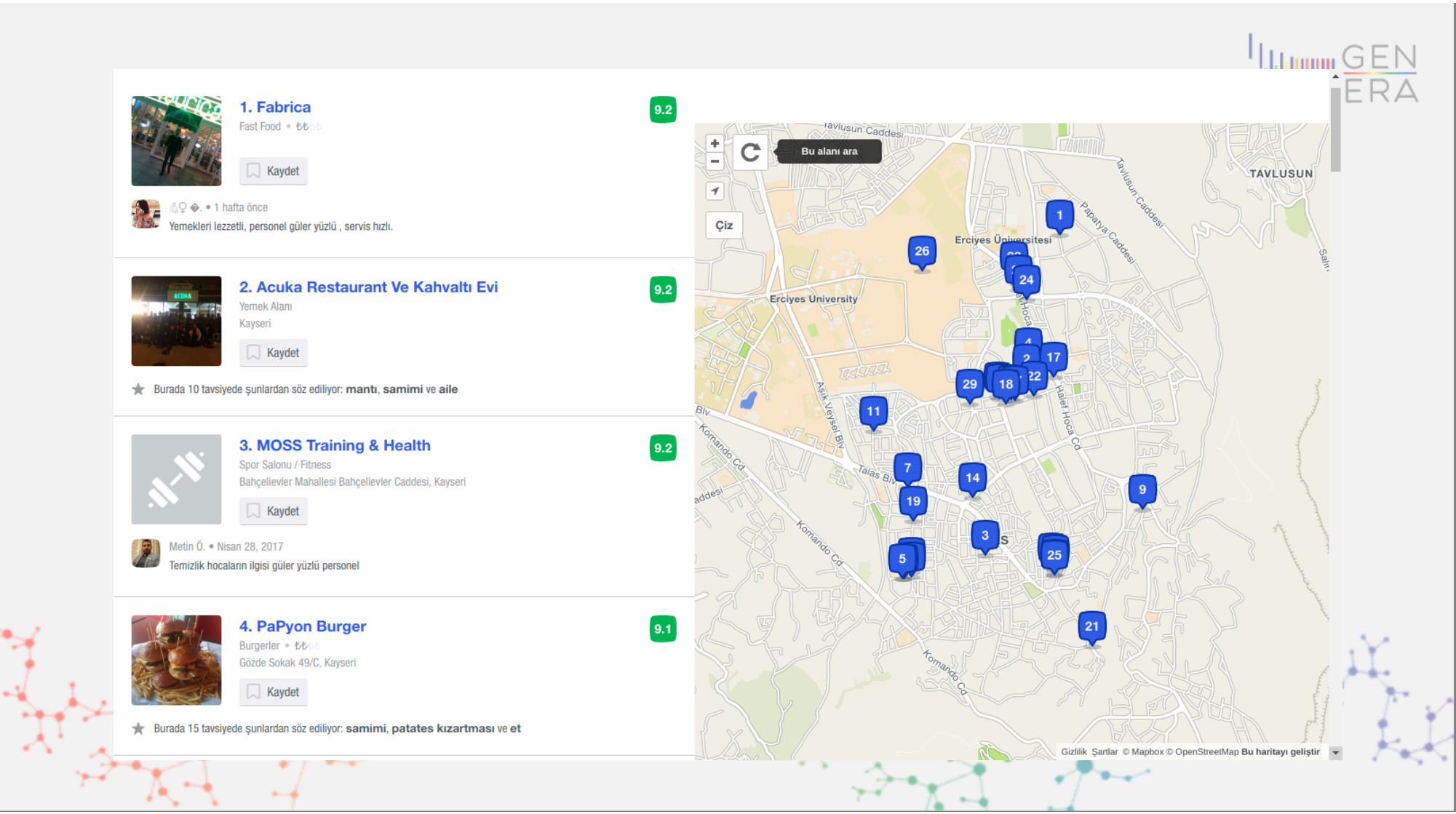


Heterozygotes | Homozygotes

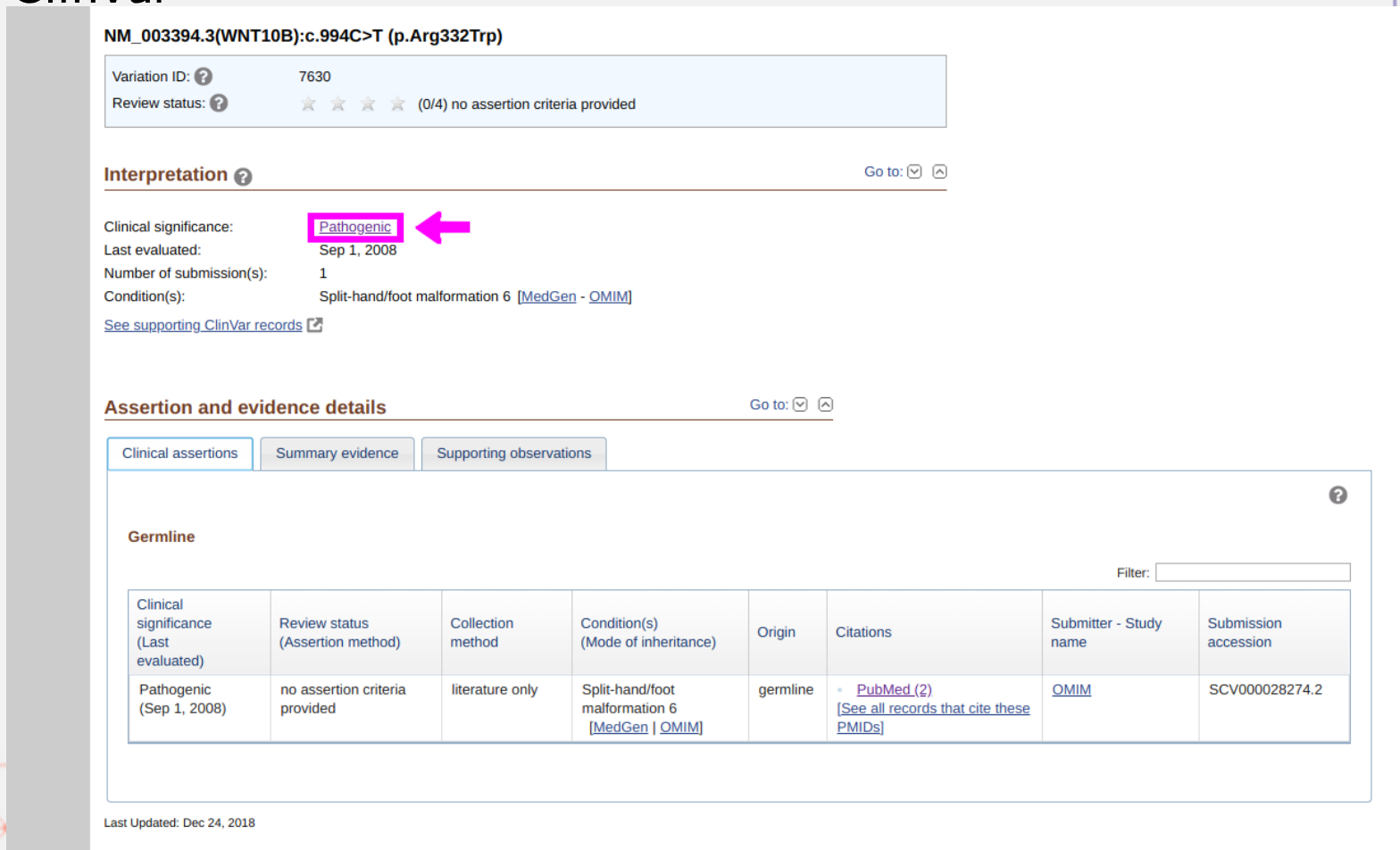
Varyant Önemi

Barış Salman

Gen-Era Diagnostik


1. Fabrica
Fast Food
9.2
Kaydet
1 hafta önce
Yemekleri lezzetli, personel güler yüzlü , servis hızlı.
2. Acuka Restaurant Ve Kahvaltı Evi
Yemek Alanı
Kayseri
9.2
Kaydet
Burada 10 tavsiyede şunlardan söz ediliyor: mantı, samimi ve aile
3. MOSS Training & Health
Spor Salonu / Fitness
Bahçelievler Mahallesi Bahçelievler Caddesi, Kayseri
9.2
Kaydet
Metin Ö. • Nisan 28, 2017
Temizlik hocaların ilgisi güler yüzlü personel
4. PaPyon Burger
Burgerler
Gözde Sokak 49/C, Kayseri
9.1
Kaydet
Burada 15 tavsiyede şunlardan söz ediliyor: samimi, patates kızartması ve et
Bu alanı ara
Çiz
TAVLUSUN
Erciyes Üniversitesi
Erciyes University
Tavlusun Caddesi
Papatya Caddesi
Komando Cd
Talas Blv
Gizlilik Şartlar © Mapbox © OpenStreetMap Bu haritayı geliştir

Clinvar



NM_003394.3(WNT10B):c.994C>T (p.Arg332Trp)

Variation ID: 7630
Review status: ☆ ☆ ☆ ☆ (0/4) no assertion criteria provided

Interpretation Go to:

Clinical significance: Pathogenic
Last evaluated: Sep 1, 2008
Number of submission(s): 1
Condition(s): Split-hand/foot malformation 6 [MedGen - OMIM]

See supporting ClinVar records

Assertion and evidence details Go to:

Clinical assertions | Summary evidence | Supporting observations

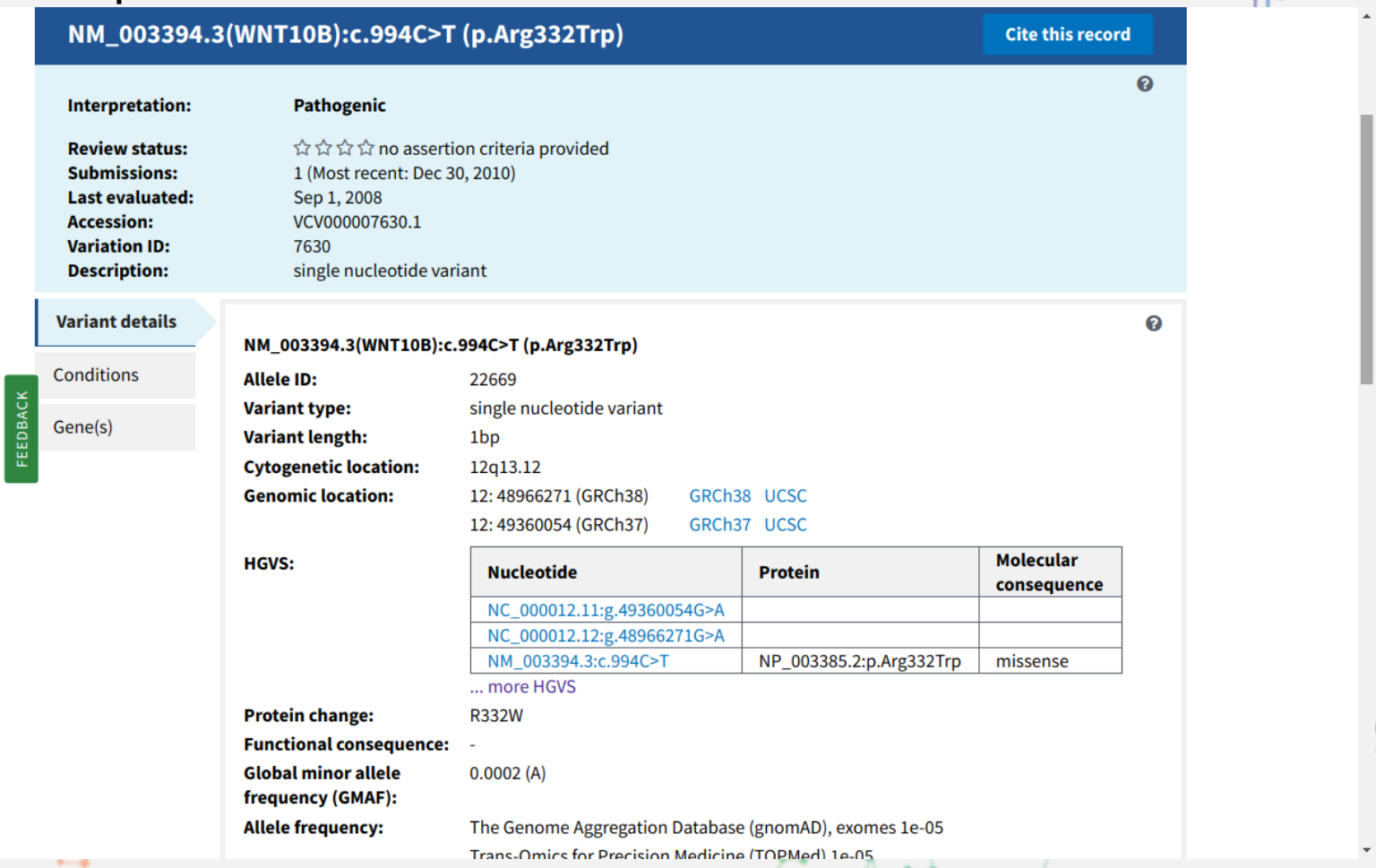
Germline

Filter:

Clinical significance (Last evaluated)	Review status (Assertion method)	Collection method	Condition(s) (Mode of inheritance)	Origin	Citations	Submitter - Study name	Submission accession
Pathogenic (Sep 1, 2008)	no assertion criteria provided	literature only	Split-hand/foot malformation 6 [MedGen | OMIM]	germline	PubMed (2) [See all records that cite these PMIDs]	OMIM	SCV000028274.2

Last Updated: Dec 24, 2018

Clinvar Rapor



NM_003394.3(WNT10B):c.994C>T (p.Arg332Trp) — Cite this record

Interpretation:	**Pathogenic**
Review status:	☆☆☆☆ no assertion criteria provided
Submissions:	1 (Most recent: Dec 30, 2010)
Last evaluated:	Sep 1, 2008
Accession:	VCV000007630.1
Variation ID:	7630
Description:	single nucleotide variant

Variant details | Conditions | Gene(s) — FEEDBACK

NM_003394.3(WNT10B):c.994C>T (p.Arg332Trp)

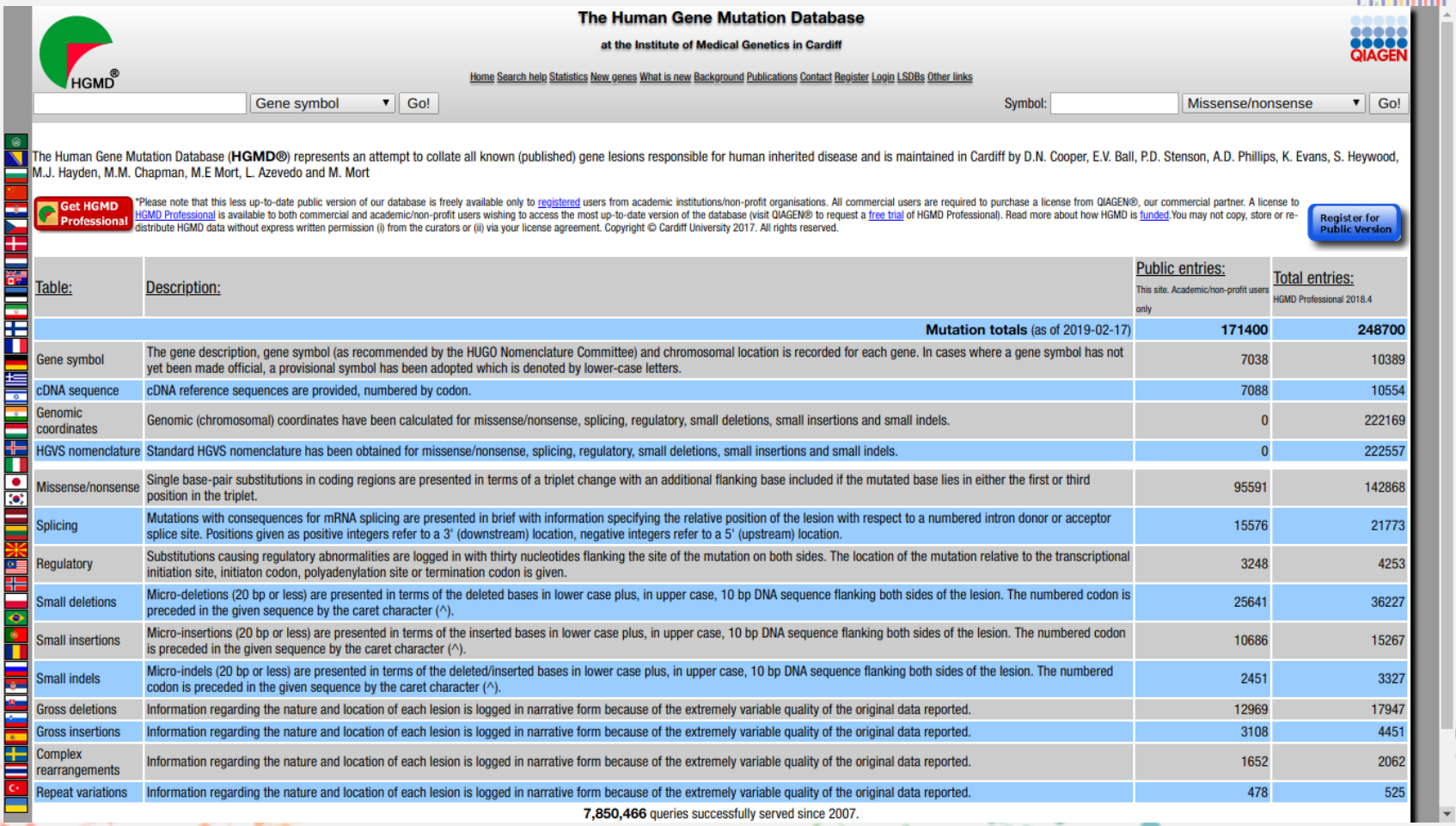
Allele ID:	22669
Variant type:	single nucleotide variant
Variant length:	1bp
Cytogenetic location:	12q13.12
Genomic location:	12: 48966271 (GRCh38) GRCh38 UCSC 12: 49360054 (GRCh37) GRCh37 UCSC

HGVS:

Nucleotide	Protein	Molecular consequence
NC_000012.11:g.49360054G>A		
NC_000012.12:g.48966271G>A		
NM_003394.3:c.994C>T	NP_003385.2:p.Arg332Trp	missense

... more HGVS

Protein change:	R332W
Functional consequence:	-
Global minor allele frequency (GMAF):	0.0002 (A)
Allele frequency:	The Genome Aggregation Database (gnomAD), exomes 1e-05
	Trans-Omics for Precision Medicine (TOPMed) 1e-05



The Human Gene Mutation Database

at the Institute of Medical Genetics in Cardiff

HGMD®

Home Search help Statistics New genes What is new Background Publications Contact Register Login LSDBs Other links

Gene symbol Go! Symbol: Missense/nonsense Go!

The Human Gene Mutation Database (**HGMD®**) represents an attempt to collate all known (published) gene lesions responsible for human inherited disease and is maintained in Cardiff by D.N. Cooper, E.V. Ball, P.D. Stenson, A.D. Phillips, K. Evans, S. Heywood, M.J. Hayden, M.M. Chapman, M.E Mort, L. Azevedo and M. Mort

Get HGMD Professional

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Register for Public Version

Table:	Description:	Public entries: This site. Academic/non-profit users only	Total entries: HGMD Professional 2018.4
	Mutation totals (as of 2019-02-17)	**171400**	**248700**
Gene symbol	The gene description, gene symbol (as recommended by the HUGO Nomenclature Committee) and chromosomal location is recorded for each gene. In cases where a gene symbol has not yet been made official, a provisional symbol has been adopted which is denoted by lower-case letters.	7038	10389
cDNA sequence	cDNA reference sequences are provided, numbered by codon.	7088	10554
Genomic coordinates	Genomic (chromosomal) coordinates have been calculated for missense/nonsense, splicing, regulatory, small deletions, small insertions and small indels.	0	222169
HGVS nomenclature	Standard HGVS nomenclature has been obtained for missense/nonsense, splicing, regulatory, small deletions, small insertions and small indels.	0	222557
Missense/nonsense	Single base-pair substitutions in coding regions are presented in terms of a triplet change with an additional flanking base included if the mutated base lies in either the first or third position in the triplet.	95591	142868
Splicing	Mutations with consequences for mRNA splicing are presented in brief with information specifying the relative position of the lesion with respect to a numbered intron donor or acceptor splice site. Positions given as positive integers refer to a 3' (downstream) location, negative integers refer to a 5' (upstream) location.	15576	21773
Regulatory	Substitutions causing regulatory abnormalities are logged in with thirty nucleotides flanking the site of the mutation on both sides. The location of the mutation relative to the transcriptional initiation site, initiaton codon, polyadenylation site or termination codon is given.	3248	4253
Small deletions	Micro-deletions (20 bp or less) are presented in terms of the deleted bases in lower case plus, in upper case, 10 bp DNA sequence flanking both sides of the lesion. The numbered codon is preceded in the given sequence by the caret character (^).	25641	36227
Small insertions	Micro-insertions (20 bp or less) are presented in terms of the inserted bases in lower case plus, in upper case, 10 bp DNA sequence flanking both sides of the lesion. The numbered codon is preceded in the given sequence by the caret character (^).	10686	15267
Small indels	Micro-indels (20 bp or less) are presented in terms of the deleted/inserted bases in lower case plus, in upper case, 10 bp DNA sequence flanking both sides of the lesion. The numbered codon is preceded in the given sequence by the caret character (^).	2451	3327
Gross deletions	Information regarding the nature and location of each lesion is logged in narrative form because of the extremely variable quality of the original data reported.	12969	17947
Gross insertions	Information regarding the nature and location of each lesion is logged in narrative form because of the extremely variable quality of the original data reported.	3108	4451
Complex rearrangements	Information regarding the nature and location of each lesion is logged in narrative form because of the extremely variable quality of the original data reported.	1652	2062
Repeat variations	Information regarding the nature and location of each lesion is logged in narrative form because of the extremely variable quality of the original data reported.	478	525

7,850,466 queries successfully served since 2007.

Ensembl SNP


Human (GRCh38.p12)
Location: 12:48,965,340-48,971,763
Gene: WNT10B
Transcript: WNT10B-201
Variant: rs766021478
Jobs
Variant displays
Explore this variant
Genomic context
Genes and regulation
Flanking sequence
Population genetics
Phenotype data
Sample genotypes
Linkage disequilibrium
Phylogenetic context
Citations
3D Protein model
Configure this page
Custom tracks
Export data
Share this page
Bookmark this page
rs766021478 SNP
Most severe consequence
stop gained | See all predicted consequences
Alleles
C/T | Ancestral: C | Highest population MAF: < 0.01
Location
Chromosome 12:48966479 (forward strand) | VCF: 12 48966479 rs766021478 C T
Evidence status
Clinical significance
HGVS names
This variant has 7 HGVS names - Show
Synonyms
This variant has 3 synonyms - Show
Original source
Variants (including SNPs and indels) imported from dbSNP (release 151) | View in dbSNP
About this variant
This variant overlaps 3 transcripts, 1 regulatory feature, is associated with 1 phenotype and is mentioned in 1 citation.
Description from SNPedia
Description not available [More information from SNPedia]
Explore this variant
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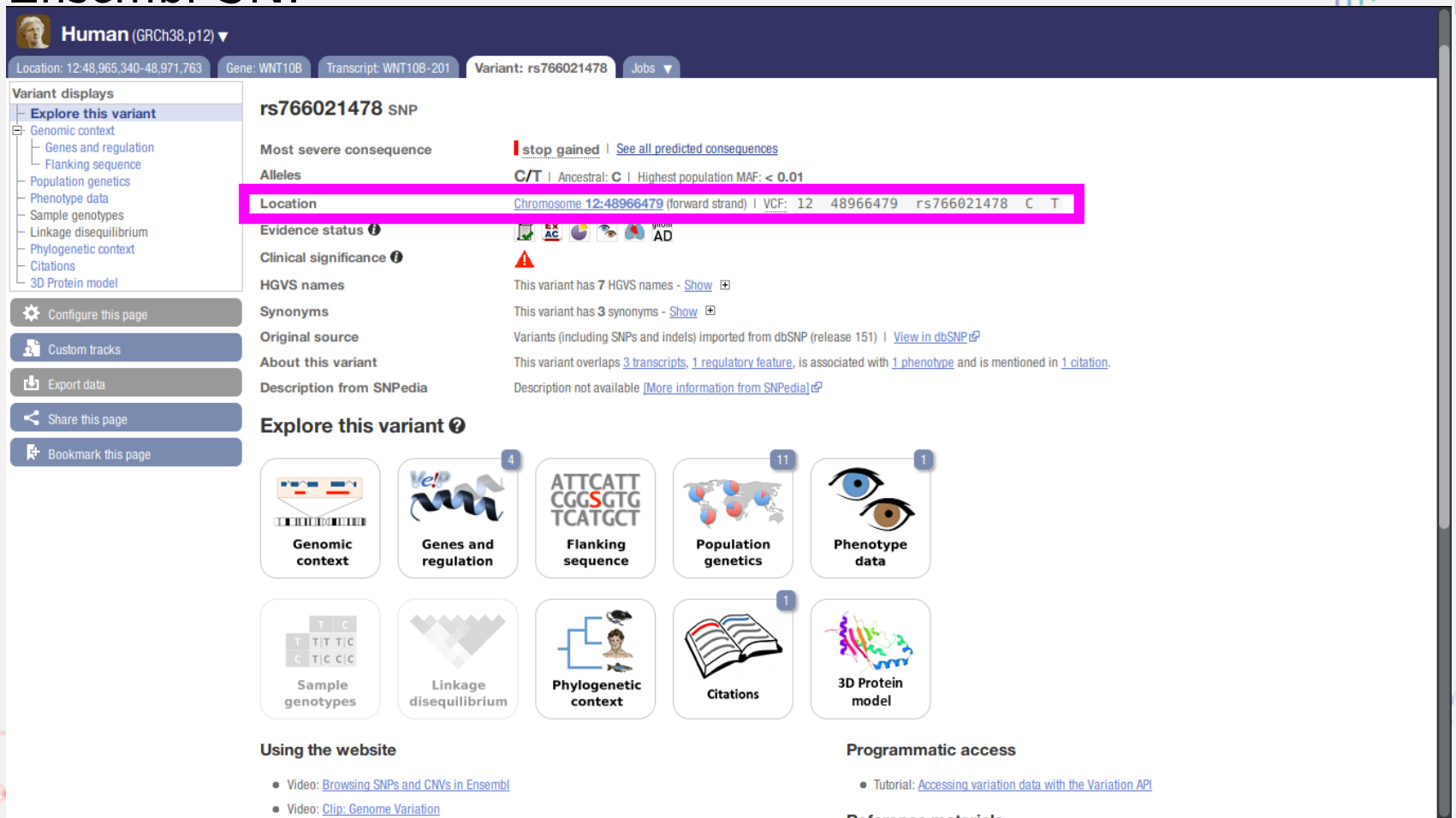
Ensembl SNP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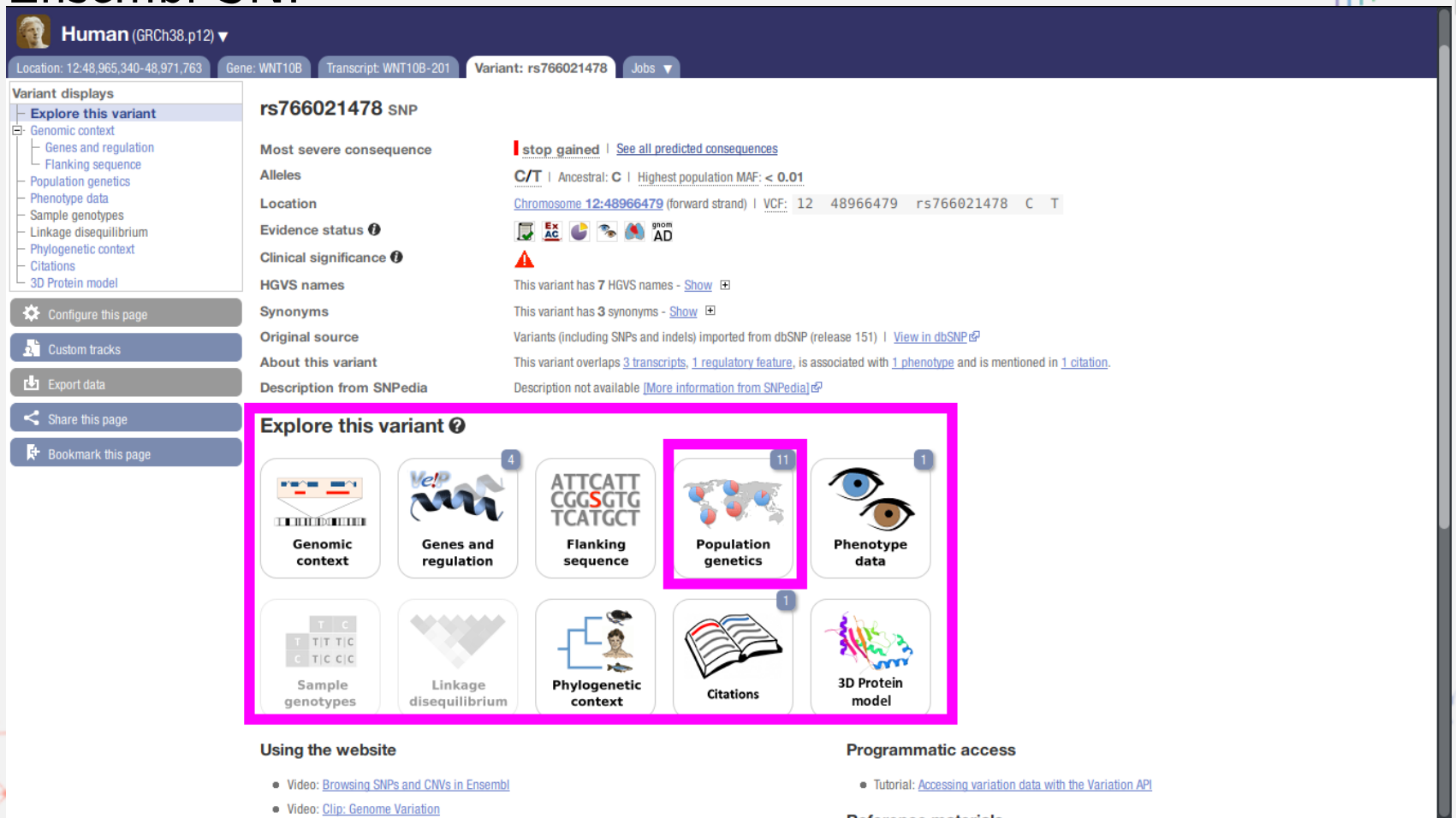
Human (GRCh38.p12)
Location: 12:48,965,340-48,971,763
Gene: WNT10B
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Share this page
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rs766021478 SNP
Most severe consequence
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Alleles
C/T | Ancestral: C | Highest population MAF: < 0.01
Location
Chromosome 12:48966479 (forward strand) | VCF: 12 48966479 rs766021478 C T
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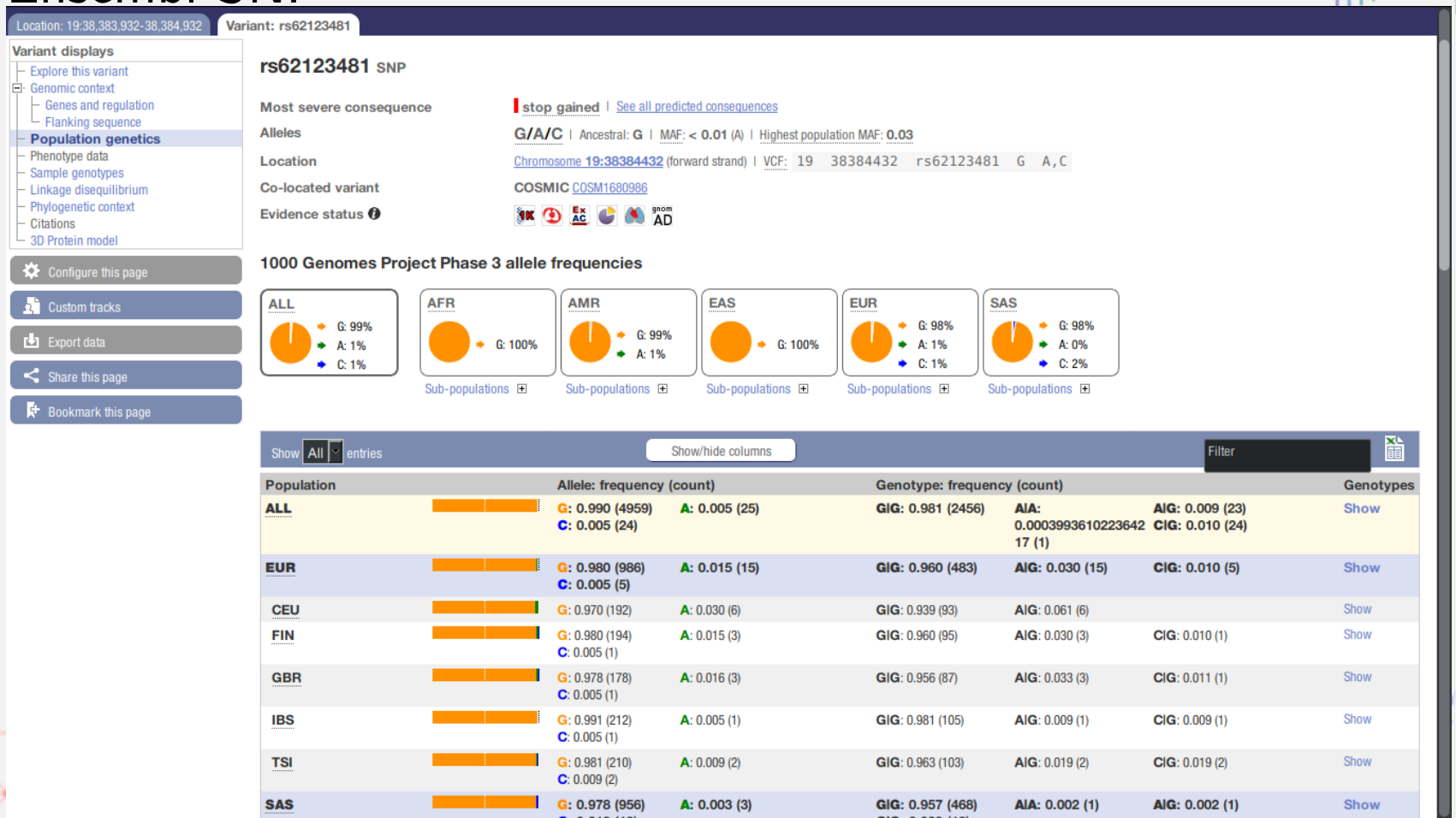

Location: 19:38,383,932-38,384,932
Variant: rs62123481
Variant displays
Explore this variant
Genomic context
Genes and regulation
Flanking sequence
Population genetics
Phenotype data
Sample genotypes
Linkage disequilibrium
Phylogenetic context
Citations
3D Protein model
Configure this page
Custom tracks
Export data
Share this page
Bookmark this page
rs62123481 SNP
Most severe consequence
stop gained | See all predicted consequences
Alleles
G/A/C | Ancestral: G | MAF: < 0.01 (A) | Highest population MAF: 0.03
Location
Chromosome 19:38384432 (forward strand) | VCF: 19 38384432 rs62123481 G A,C
Co-located variant
COSMIC COSM1680986
Evidence status
1000 Genomes Project Phase 3 allele frequencies
ALL G: 99% A: 1% C: 1%
AFR G: 100%
AMR G: 99% A: 1%
EAS G: 100%
EUR G: 98% A: 1% C: 1%
SAS G: 98% A: 0% C: 2%
Sub-populations
Show All entries
Show/hide columns
Filter
Population
Allele: frequency (count)
Genotype: frequency (count)
Genotypes
ALL G: 0.990 (4959) A: 0.005 (25) C: 0.005 (24) GIG: 0.981 (2456) AIA: 0.0003993610223642 17 (1) AIG: 0.009 (23) CIG: 0.010 (24) Show
EUR G: 0.980 (986) A: 0.015 (15) C: 0.005 (5) GIG: 0.960 (483) AIG: 0.030 (15) CIG: 0.010 (5) Show
CEU G: 0.970 (192) A: 0.030 (6) GIG: 0.939 (93) AIG: 0.061 (6) Show
FIN G: 0.980 (194) A: 0.015 (3) C: 0.005 (1) GIG: 0.960 (95) AIG: 0.030 (3) CIG: 0.010 (1) Show
GBR G: 0.978 (178) A: 0.016 (3) C: 0.005 (1) GIG: 0.956 (87) AIG: 0.033 (3) CIG: 0.011 (1) Show
IBS G: 0.991 (212) A: 0.005 (1) C: 0.005 (1) GIG: 0.981 (105) AIG: 0.009 (1) CIG: 0.009 (1) Show
TSI G: 0.981 (210) A: 0.009 (2) C: 0.009 (2) GIG: 0.963 (103) AIG: 0.019 (2) CIG: 0.019 (2) Show
SAS G: 0.978 (956) A: 0.003 (3) GIG: 0.957 (468) AIA: 0.002 (1) AIG: 0.002 (1) Show

Varyant Yordamlama Araçları

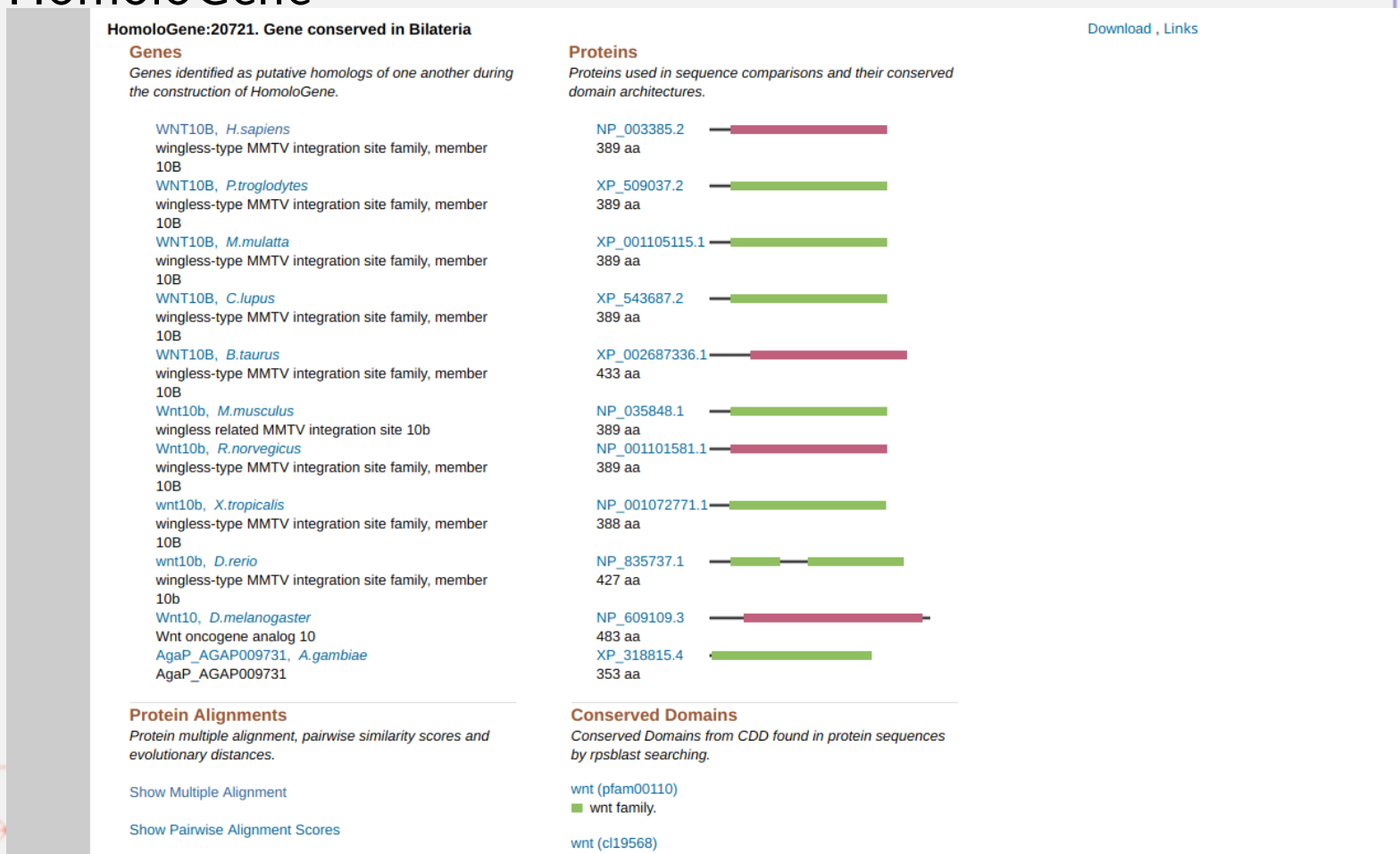
Barış Salman

Gen-Era Diagnostik

İnsan Genomunu Anlamlandırmak Veritabanları ve Biyoinformatik Araçlar

- Evrimsel korunmuşluk
- Aminoasit değişimi
- Kırpılma mekanizması üzerindeki etkiler

HomoloGene



HomoloGene:20721. Gene conserved in Bilateria

Download , Links

Genes

Genes identified as putative homologs of one another during the construction of HomoloGene.

WNT10B, *H.sapiens*
wingless-type MMTV integration site family, member 10B
WNT10B, *P.troglodytes*
wingless-type MMTV integration site family, member 10B
WNT10B, *M.mulatta*
wingless-type MMTV integration site family, member 10B
WNT10B, *C.lupus*
wingless-type MMTV integration site family, member 10B
WNT10B, *B.taurus*
wingless-type MMTV integration site family, member 10B
Wnt10b, *M.musculus*
wingless related MMTV integration site 10b
Wnt10b, *R.norvegicus*
wingless-type MMTV integration site family, member 10B
wnt10b, *X.tropicalis*
wingless-type MMTV integration site family, member 10B
wnt10b, *D.rerio*
wingless-type MMTV integration site family, member 10b
Wnt10, *D.melanogaster*
Wnt oncogene analog 10
AgaP_AGAP009731, *A.gambiae*
AgaP_AGAP009731

Proteins

Proteins used in sequence comparisons and their conserved domain architectures.

NP_003385.2
389 aa
XP_509037.2
389 aa
XP_001105115.1
389 aa
XP_543687.2
389 aa
XP_002687336.1
433 aa
NP_035848.1
389 aa
NP_001101581.1
389 aa
NP_001072771.1
388 aa
NP_835737.1
427 aa
NP_609109.3
483 aa
XP_318815.4
353 aa

Protein Alignments

Protein multiple alignment, pairwise similarity scores and evolutionary distances.

Show Multiple Alignment

Show Pairwise Alignment Scores

Conserved Domains

Conserved Domains from CDD found in protein sequences by rpsblast searching.

wnt (pfam00110)
wnt family.

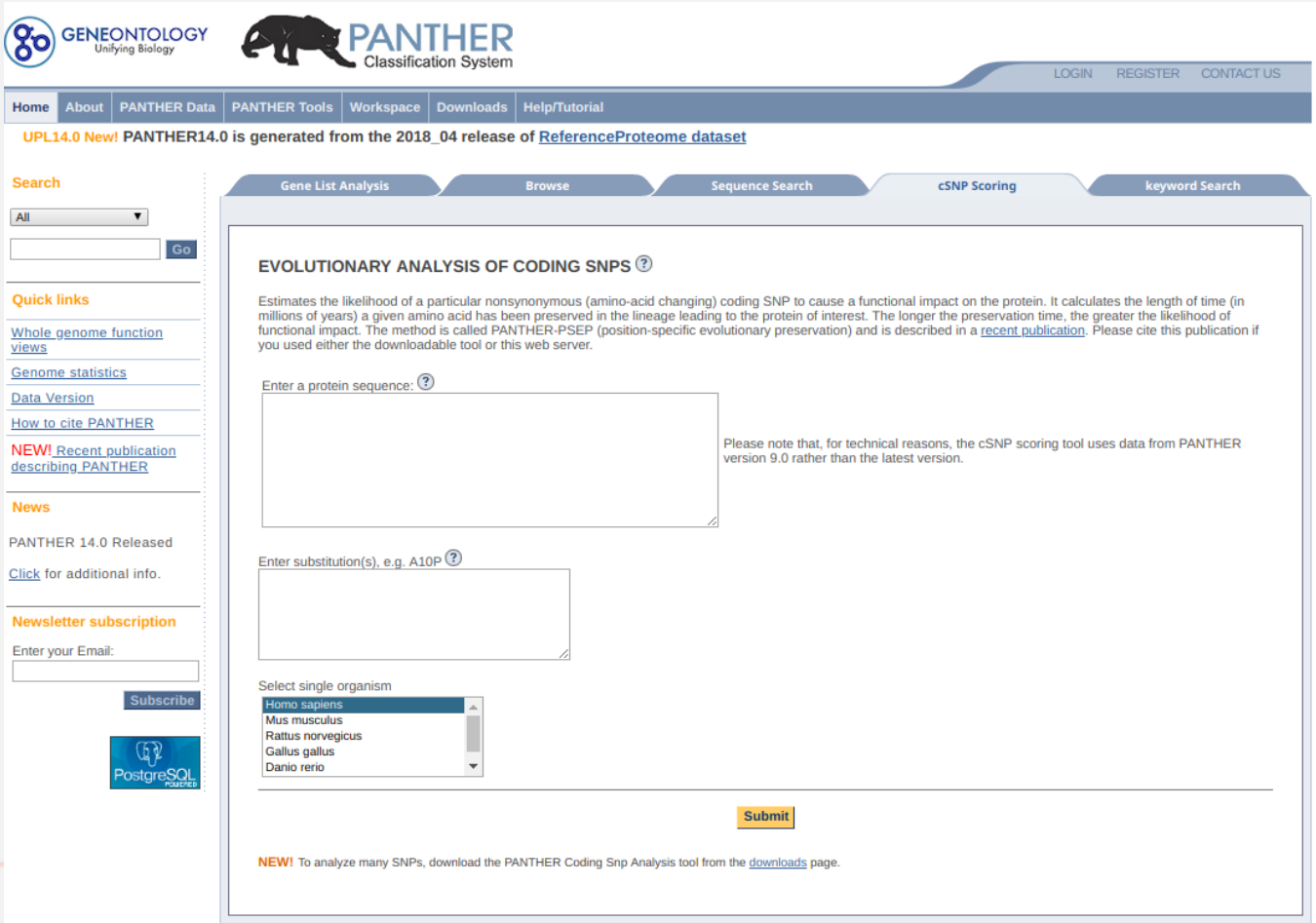
wnt (cl19568)

HomoloGene

NP_003385.2	9	PPPSGLAGLLFLALCSRALSNEILGLKLP--GEPPLTANTVCLTLSGLSK	56
XP_509037.2	9	PPPSGLAGLLFLALCSRALSNEILGLKLP--GEPPLTANTVCLTLSGLSK	56
XP_001105115.1	9	PPPSGLAGLLFLALCSRALSNEILGLKLP--GEPPLTANTVCLTLSGLSK	56
XP_543687.2	9	PPPSGFAGLLLLALCSRALSNEILGLKLP--GEPPLTANTVCLTLSGLSK	56
XP_002687336.1	51	PPPSGLAGLLFLALCSRALGNEIQGLKLPGGGEPPLTANTVCLTLSGLSK	100
NP_035848.1	9	PPPLGLAGLLFLALFSRALSNEILGLKLP--GEPPLTANTVCLTLSGLSK	56
NP_001101581.1	9	PPPLGLAGLLFLALFSRALSNEILGLKLP--GEPPLTANTVCLTLSGLSK	56
NP_835737.1	9	LGRVLIVTAALLSPAFTVLGNDILGLKVA--GEPVLTPNAVCLRLAGLTK	56
NP_609109.3	35	SSNNLVATPATSRHCNLHLIVMIILACCTRWLYGLPDGRATCRSVPGLTK	84
XP_318815.4		--	
NP_001072771.1	9	TWPWILA--IAIWLCSRVLCNDILGLKLP--NDPILTPNTVCLTLPGLSK	54

NP_003385.2	57	RQLGLCLRNPDVTASALQGLHIAVHECQHQLRDQRWNCSALEGGGRLPHH	106
XP_509037.2	57	RQLGLCLRNPDVTASALQGLHIAVHECQHQLRDQRWNCSALEGGGRPAHH	106
XP_001105115.1	57	RQLGLCLRNPDVTASALQGLHIAVHECQHQLRDQRWNCSALEGGGRLPHH	106
XP_543687.2	57	RQLGLCLRSPDVTASALQGLHIAVHECQHQLRDQRWNCSALEGGGRLPHH	106
XP_002687336.1	101	QQLGLCLRSPDVTASALQGLHIAVHECQHQLRDQRWNCSALEGGGRLPHH	150
NP_035848.1	57	RQLGLCLRSPDVTASALQGLHIAVHECQHQLRDQRWNCSALEGGGRLPHH	106
NP_001101581.1	57	RQLGLCLRSPDVTASALQGLHIAVHECQHQLRDQRWNCSALEGGGRLPHH	106
NP_835737.1	57	KQMRLCVRSPDVTASALQGIQVAIHECQHQLRDQRWNCSSLENHGKLPHQ	106
NP_609109.3	85	DQVELCYKASDVTAAALEGLDMAIRECQIQFQWHRWNCSSLSTKSRNPHA	134
XP_318815.4		--	
NP_001072771.1	55	RQMGLCVRNPDVTASALQGIQIAIHECQHQLKGQRWNCSTLETMGKMPHD	104

NP_003385.2	107	SAILKRGFRESAFSFSMLAAGVMHAVATACSLGKLVSCGCG--WKGSGEQ	154
XP_509037.2	107	SVILKARFRESAFSFSMLAAGVMHAVATACSLGKLVSCGCG--WKGSGEQ	154
XP_001105115.1	107	SAILKRGFRESAFSFSMLAAGVMHAVATACSLGKLVSCGCG--WKGSGEQ	154
XP_543687.2	107	SAILKRGFRESAFSFSMLAAGVMHAVATACSLGRLVSCGCG--WKGSGEQ	154
XP_002687336.1	151	SAILKRGFRESAFSFSMLAAGVMHAVATACSLGKLVSCGCG--WKGSGEQ	198
NP_035848.1	107	SAILKRGFRESAFSFSMLAAGVMHAVATACSLGKLVSCGCG--WKGSGEQ	154
NP_001101581.1	107	SAILKRGFRESAFSFSMLAAGVMHAVATACSLGKLVSCGCG--WKGSGEQ	154
NP_835737.1	107	SAILNRGFRESAFSLSLLAAGVVHSVASACSLGKLRGCGCE--AKRRLDD	154
NP_609109.3	135	SSLLKKGYRESAFAFAISAAGVAHSVARACSQGRLMSCGCDPTINRKTLN	184
XP_318815.4	1	MIFISTGYRESAFAYAIAAAGVTHSVARACAQGRLISCGCDPSVNRRGMS	50
NP_001072771.1	105	SAILKRGFRESAFAFSLLAAGVMHSVATACSLGKLQGCGCE--WKRRGTE	152


GENEONTOLOGY
Unifying Biology
PANTHER
Classification System
LOGIN REGISTER CONTACT US
Home About PANTHER Data PANTHER Tools Workspace Downloads Help/Tutorial
UPL14.0 New! PANTHER14.0 is generated from the 2018_04 release of ReferenceProteome dataset
Search
All
Go
Quick links
Whole genome function views
Genome statistics
Data Version
How to cite PANTHER
NEW! Recent publication describing PANTHER
News
PANTHER 14.0 Released
Click for additional info.
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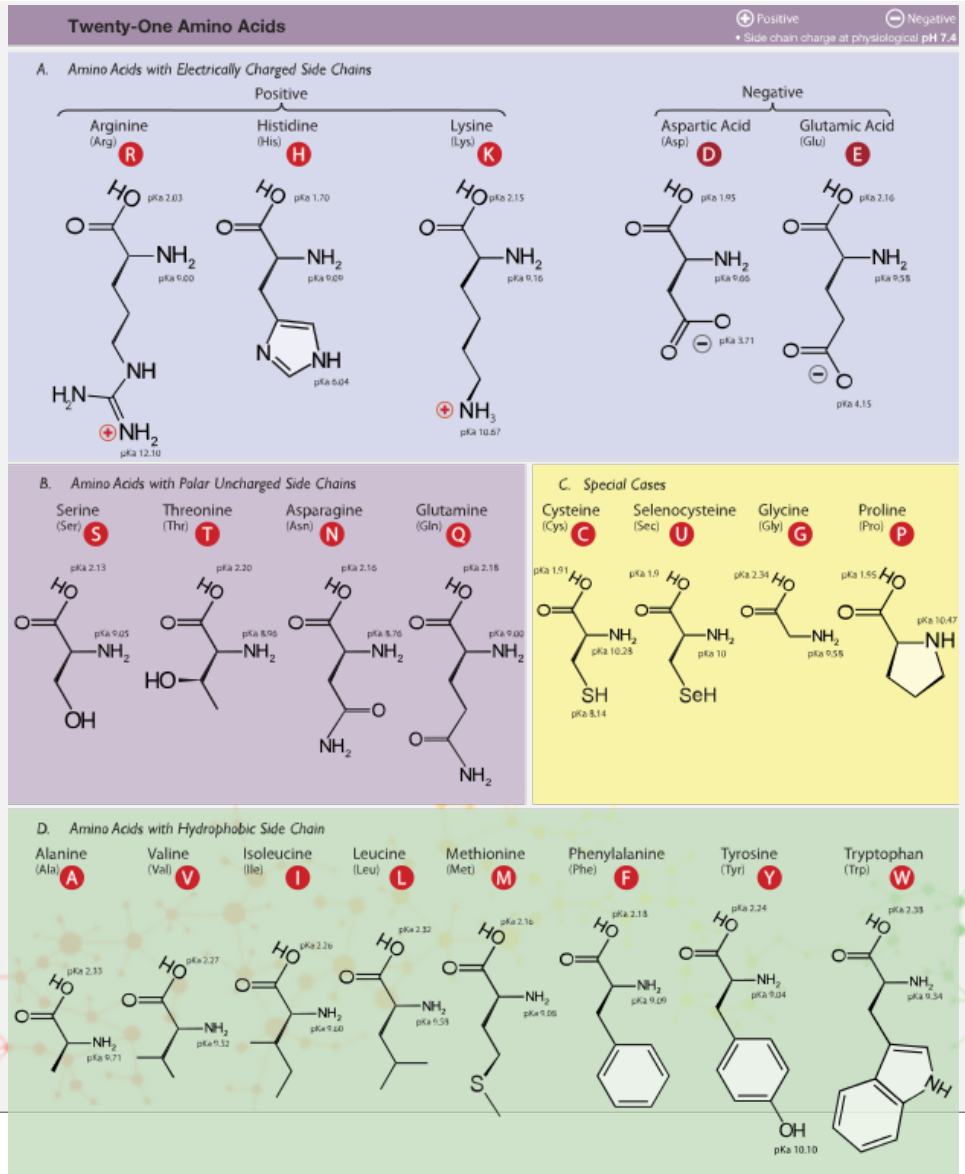
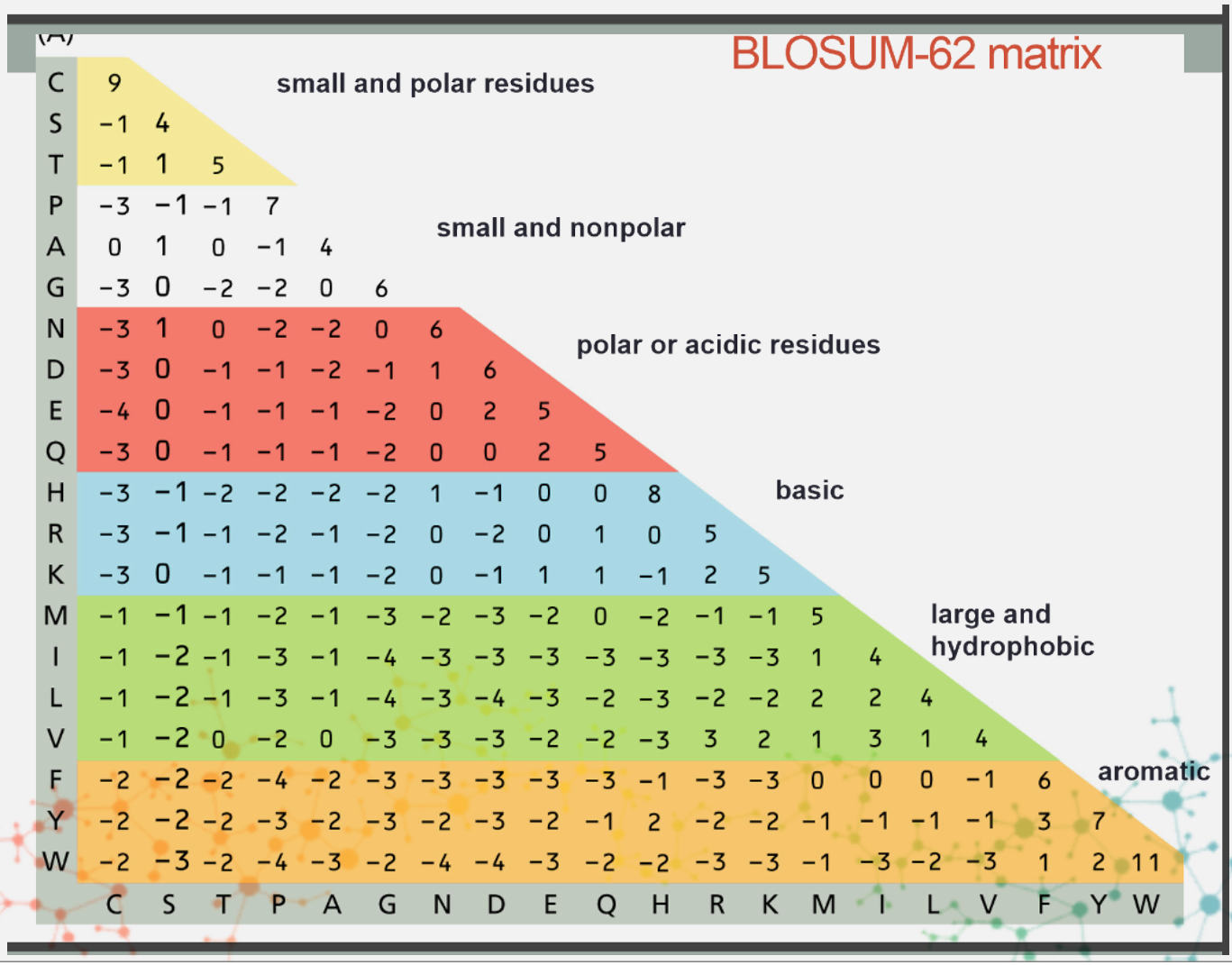
Subscribe
PostgreSQL
Gene List Analysis
Browse
Sequence Search
cSNP Scoring
keyword Search
EVOLUTIONARY ANALYSIS OF CODING SNPS
Estimates the likelihood of a particular nonsynonymous (amino-acid changing) coding SNP to cause a functional impact on the protein. It calculates the length of time (in millions of years) a given amino acid has been preserved in the lineage leading to the protein of interest. The longer the preservation time, the greater the likelihood of functional impact. The method is called PANTHER-PSEP (position-specific evolutionary preservation) and is described in a recent publication. Please cite this publication if you used either the downloadable tool or this web server.
Enter a protein sequence:
Please note that, for technical reasons, the cSNP scoring tool uses data from PANTHER version 9.0 rather than the latest version.
Enter substitution(s), e.g. A10P
Select single organism
Homo sapiens
Mus musculus
Rattus norvegicus
Gallus gallus
Danio rerio
Submit
NEW! To analyze many SNPs, download the PANTHER Coding Snp Analysis tool from the downloads page.

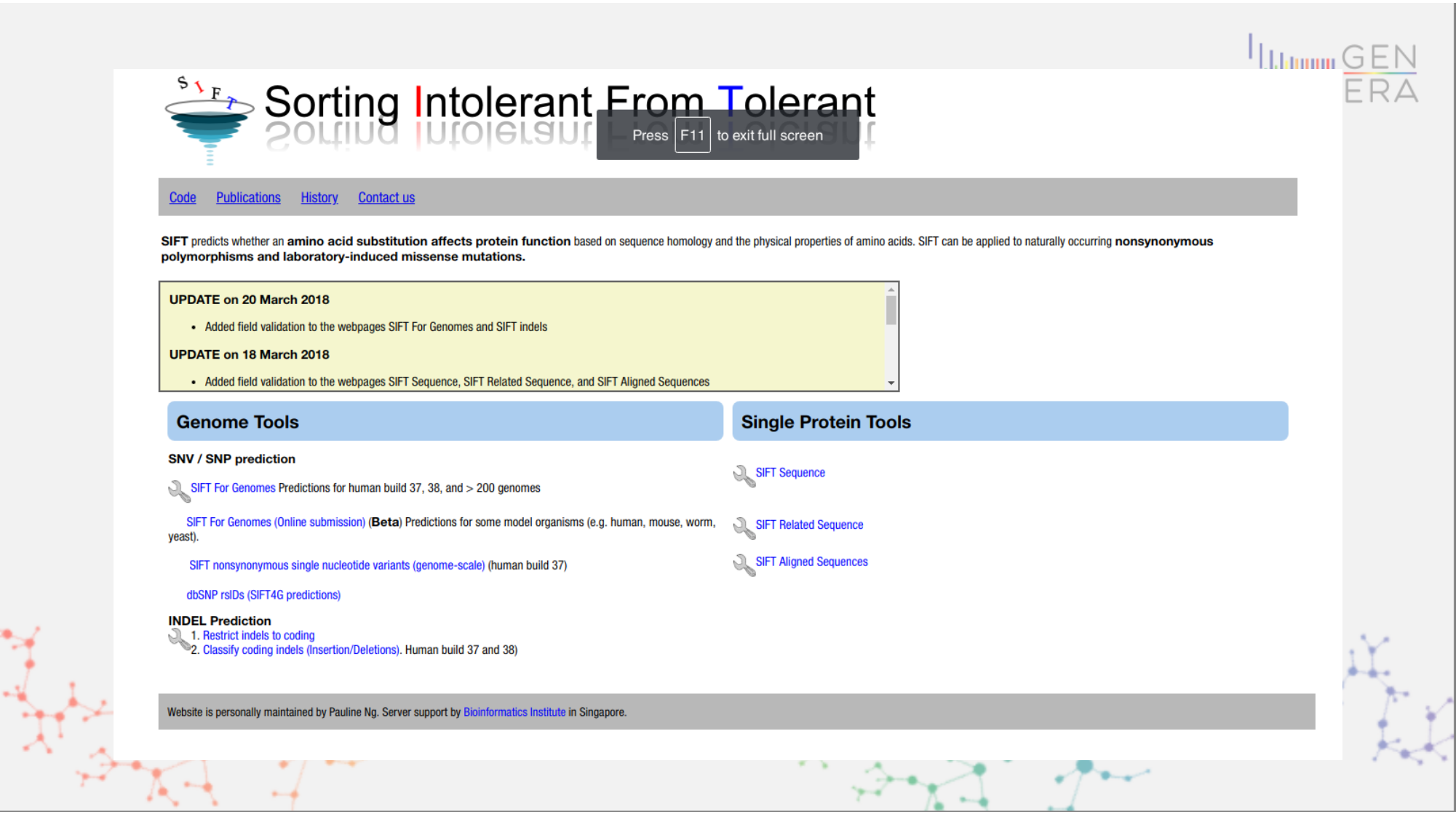


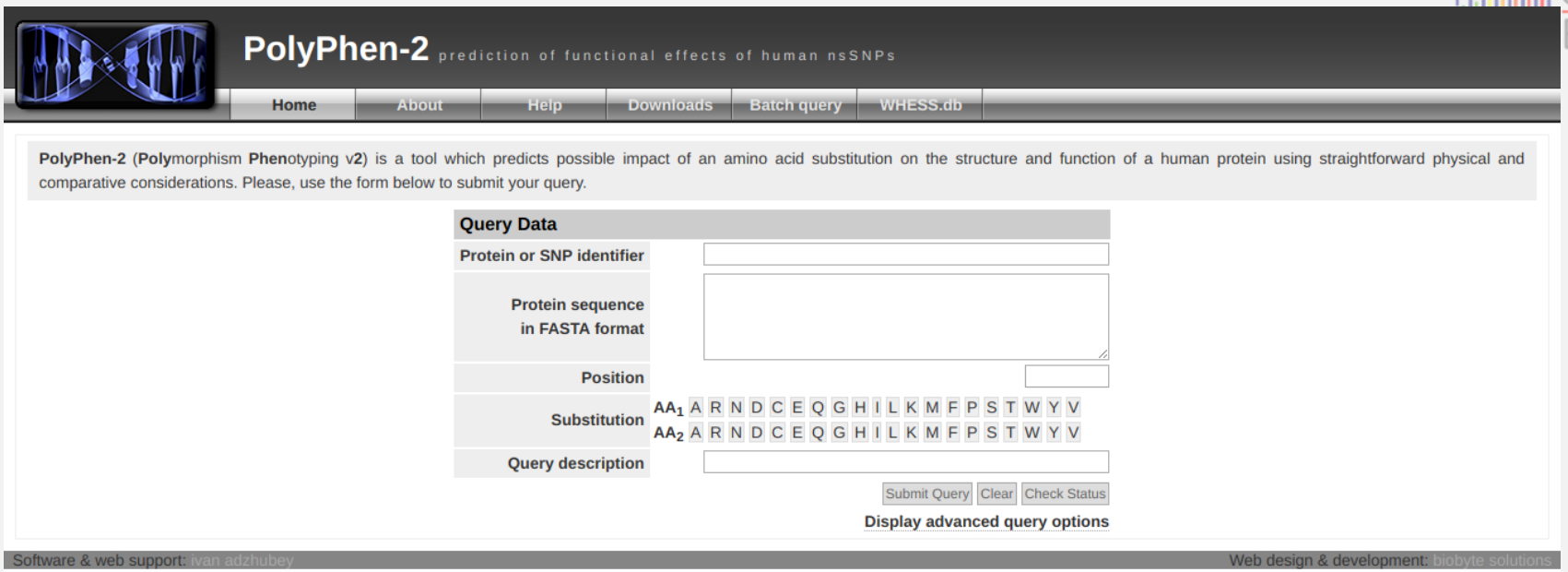
Barış Salman

Gen-Era Diagnostik



	C	S	T	P	A	G	N	D	E	Q	H	R	K	M	I	L	V	F	Y	W
C	9																			
S	−1	4																		
T	−1	1	5																	
P	−3	−1	−1	7																
A	0	1	0	−1	4															
G	−3	0	−2	−2	0	6														
N	−3	1	0	−2	−2	0	6													
D	−3	0	−1	−1	−2	−1	1	6												
E	−4	0	−1	−1	−1	−2	0	2	5											
Q	−3	0	−1	−1	−1	−2	0	0	2	5										
H	−3	−1	−2	−2	−2	−2	1	−1	0	0	8									
R	−3	−1	−1	−2	−1	−2	0	−2	0	1	0	5								
K	−3	0	−1	−1	−1	−2	0	−1	1	1	−1	2	5							
M	−1	−1	−1	−2	−1	−3	−2	−3	−2	0	−2	−1	−1	5						
I	−1	−2	−1	−3	−1	−4	−3	−3	−3	−3	−3	−3	−3	1	4					
L	−1	−2	−1	−3	−1	−4	−3	−4	−3	−2	−3	−2	−2	2	2	4				
V	−1	−2	0	−2	0	−3	−3	−3	−2	−2	−3	3	2	1	3	1	4			
F	−2	−2	−2	−4	−2	−3	−3	−3	−3	−3	−1	−3	−3	0	0	0	−1	6		
Y	−2	−2	−2	−3	−2	−3	−2	−3	−2	−1	2	−2	−2	−1	−1	−1	−1	3	7	
W	−2	−3	−2	−4	−3	−2	−4	−4	−3	−2	−2	−3	−3	−1	−3	−2	−3	1	2	11


GEN ERA
Sorting Intolerant From Tolerant
Press F11 to exit full screen
Code Publications History Contact us
SIFT predicts whether an amino acid substitution affects protein function based on sequence homology and the physical properties of amino acids. SIFT can be applied to naturally occurring nonsynonymous polymorphisms and laboratory-induced missense mutations.
UPDATE on 20 March 2018
Added field validation to the webpages SIFT For Genomes and SIFT indels
UPDATE on 18 March 2018
Added field validation to the webpages SIFT Sequence, SIFT Related Sequence, and SIFT Aligned Sequences
Genome Tools
SNV / SNP prediction
SIFT For Genomes Predictions for human build 37, 38, and > 200 genomes
SIFT For Genomes (Online submission) (Beta) Predictions for some model organisms (e.g. human, mouse, worm, yeast).
SIFT nonsynonymous single nucleotide variants (genome-scale) (human build 37)
dbSNP rsIDs (SIFT4G predictions)
INDEL Prediction
1. Restrict indels to coding
2. Classify coding indels (Insertion/Deletions). Human build 37 and 38)
Single Protein Tools
SIFT Sequence
SIFT Related Sequence
SIFT Aligned Sequences
Website is personally maintained by Pauline Ng. Server support by Bioinformatics Institute in Singapore.


PolyPhen-2
prediction of functional effects of human nsSNPs
Home
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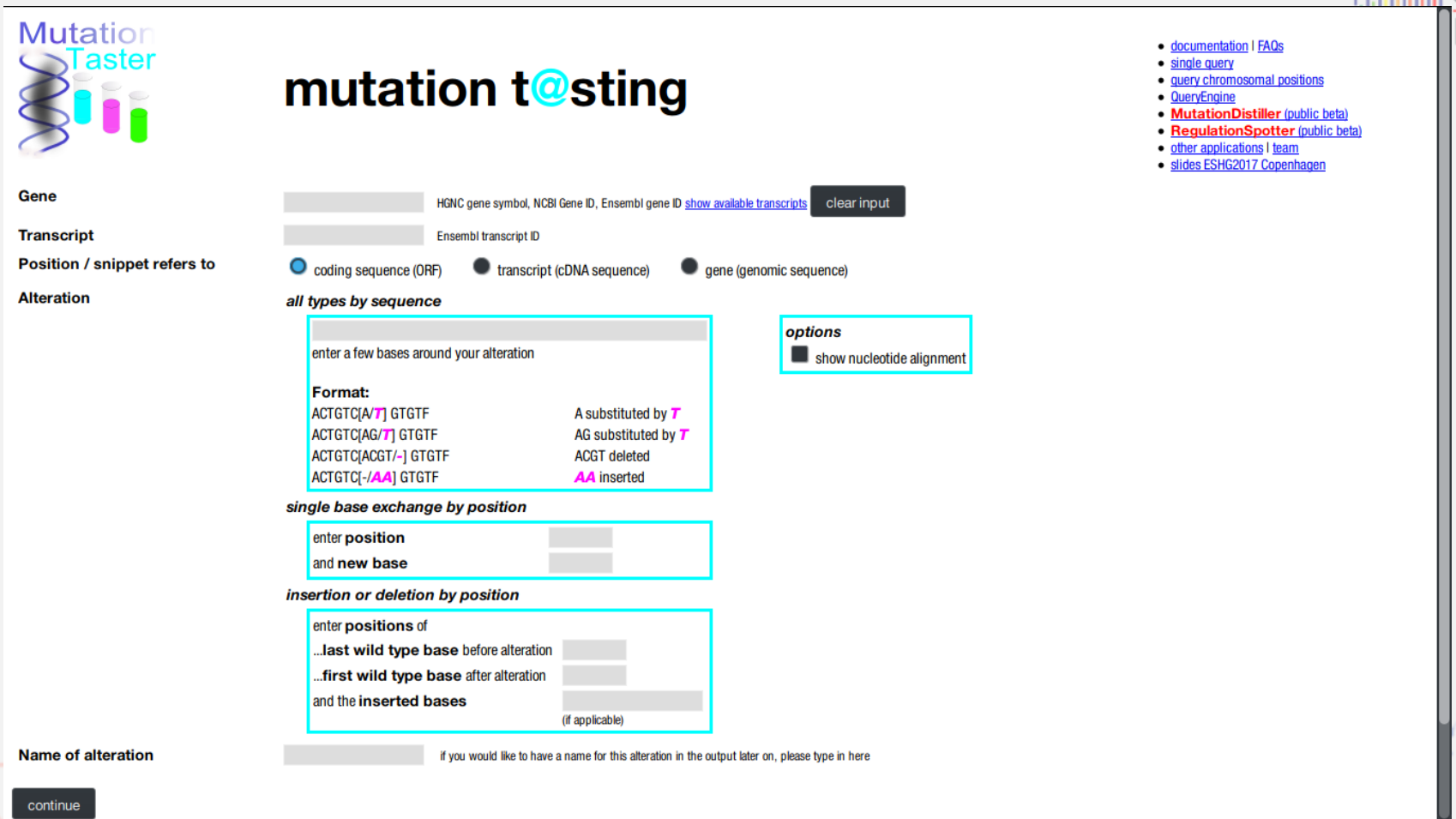
Batch query
WHESS.db
PolyPhen-2 (Polymorphism Phenotyping v2) is a tool which predicts possible impact of an amino acid substitution on the structure and function of a human protein using straightforward physical and comparative considerations. Please, use the form below to submit your query.
Query Data
Protein or SNP identifier
Protein sequence in FASTA format
Position
Substitution
AA1 A R N D C E Q G H I L K M F P S T W Y V
AA2 A R N D C E Q G H I L K M F P S T W Y V
Query description
Submit Query
Clear
Check Status
Display advanced query options
Software & web support: ivan adzhubey
Web design & development: biobyte solutions


MutationTaster
mutation t@sting
documentation | FAQs
single query
query chromosomal positions
QueryEngine
MutationDistiller (public beta)
RegulationSpotter (public beta)
other applications | team
slides ESHG2017 Copenhagen
Gene
HGNC gene symbol, NCBI Gene ID, Ensembl gene ID show available transcripts
clear input
Transcript
Ensembl transcript ID
Position / snippet refers to
coding sequence (ORF)
transcript (cDNA sequence)
gene (genomic sequence)
Alteration
all types by sequence
enter a few bases around your alteration
Format:
ACTGTC[A/T] GTGTF
A substituted by T
ACTGTC[AG/T] GTGTF
AG substituted by T
ACTGTC[ACGT/-] GTGTF
ACGT deleted
ACTGTC[-/AA] GTGTF
AA inserted
options
show nucleotide alignment
single base exchange by position
enter position
and new base
insertion or deletion by position
enter positions of
...last wild type base before alteration
...first wild type base after alteration
and the inserted bases
(if applicable)
Name of alteration
if you would like to have a name for this alteration in the output later on, please type in here
continue


Human Splicing Finder
Aix*Marseille université
Inserm
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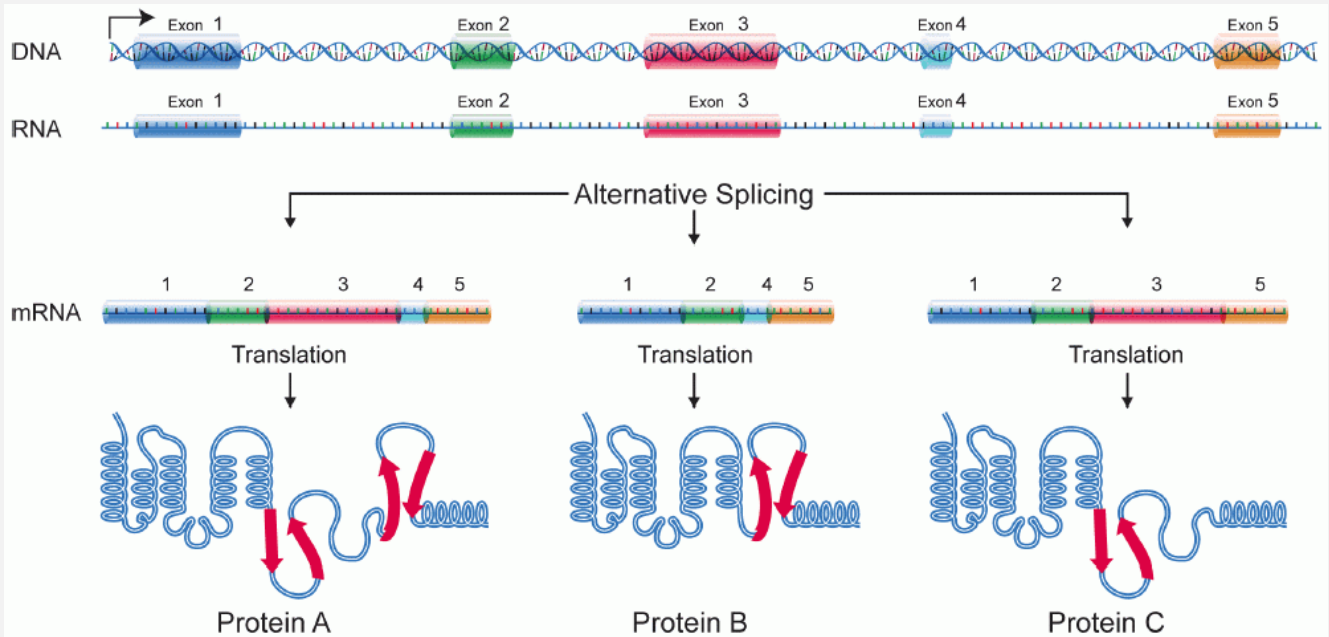
Description
With the completion of the Human Genome Project our vision of human genetic diseases has changed. Thousands of mutations are identified in diagnostic and research laboratories yearly. The knowledge of these mutations associated with clinical and biological data is essential for clinicians, geneticists and researchers.
In order to better understand intronic and exonic mutations leading to splicing defects, we decided to create the Human Splicing Finder website. This tool is aimed to help studying the pre-mRNA splicing [more about splicing background].
To calculate the consensus values of potential splice sites and search for branch points, new algorithms were developed. Furthermore, we have integrated all available matrices to identify exonic and intronic motifs, as well as new matrices to identify hnRNP A1, Tra2-β and 9G8.
We hope that this tool will be useful for your research. In order to improve it, please send us comments and new matrices to identify specific sequences involved in splicing.
HSF (Human Splicing Finder) is freely available for non-commercial users. Nevertheless it is not allowed to copy all or part of the database content without specific authorisation from us. If you are a commercial user please contact us to obtain a dedicated license.
For more information please contact Prof. Christophe Béroud or Dr. David Salgado
Other Splicing Tools
MaxEntScan
SROOGLE: Splicing RegulatiOn Online GraphicaL Engine
RegRNA: A Regulatory RNA Motifs and Elements Finder
EBI Splice Signal Analysis
GeneSplicer
Splice Predictor (DK)
MIT splice predictor
ASPic
Get Started
Start an Analysis with
HSF 3.1
Fundings
Inserm
Institut national de la santé et de la recherche médicale
Aix*Marseille université
GEN2PHEN
RD-Connect
Marseille Medical Genetics (MMG) - UMR 1251
Director: Nicolas LEVY
Bioinformatics & Genetics Team
Director: Christophe BEROUD



Barış Salman
Gen-Era Diagnostik


UniProtKB - O00744 (WN10B_HUMAN)
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Display
BLAST
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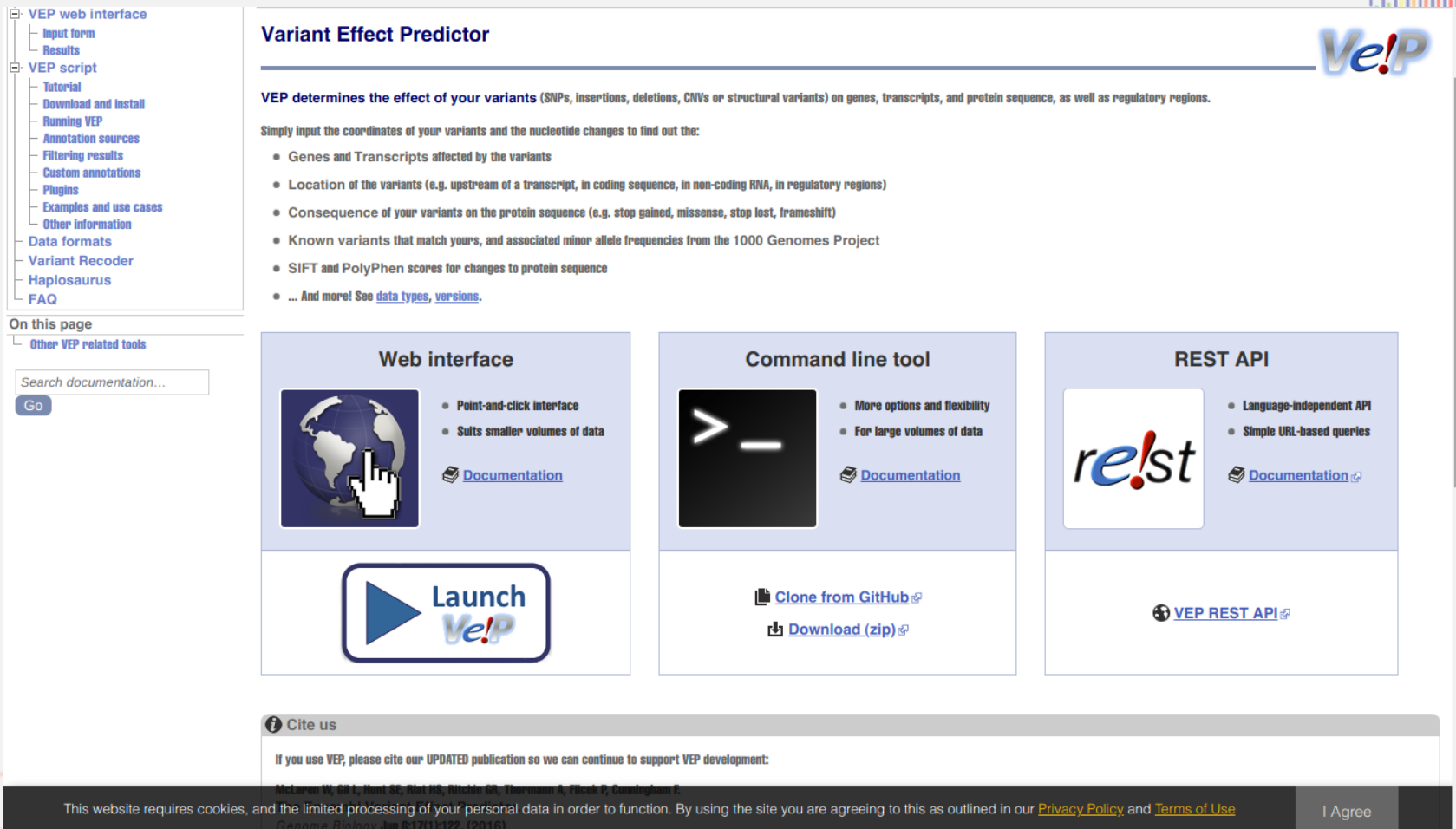
Add to basket
History
Other tutorials and videos
Help video
Feedback
Entry
Publications
Feature viewer
Feature table
None
Function
Names & Taxonomy
Subcellular location
Pathology & Biotech
PTM / Processing
Expression
Interaction
Structure
Family & Domains
Sequences (2+)
Similar proteins
Cross-references
Entry information
Miscellaneous
Top
Protein Protein Wnt-10b
Gene WNT10B
Organism Homo sapiens (Human)
Status Reviewed - Annotation score: - Experimental evidence at protein level
Function
Member of the Wnt ligand gene family that encodes for secreted proteins, which activate the Wnt signaling cascade. Specifically activates canonical Wnt/beta-catenin signaling and thus triggers beta-catenin/LEF/TCF-mediated transcriptional programs. Involved in signaling networks controlling stemness, pluripotency and cell fate decisions. Acts in the immune system, mammary gland, adipose tissue, bone and skin. 3 Publications
GO - Molecular function
frizzled binding Source: GO_Central
receptor ligand activity Source: BHF-UCL
View the complete GO annotation on QuickGO ...
GO - Biological process
bone trabecula formation Source: Ensembl
canonical Wnt signaling pathway Source: BHF-UCL
cell cycle arrest Source: Ensembl
cell fate commitment Source: GO_Central
cellular response to cAMP Source: Ensembl
cellular response to parathyroid hormone stimulus Source: Ensembl
cellular response to retinoic acid Source: UniProtKB
chondrocyte differentiation Source: UniProtKB
fungiform papilla development Source: Ensembl
G2/M transition of mitotic cell cycle Source: Ensembl
hematopoietic stem cell proliferation Source: BHF-UCL
lipid metabolic process Source: Ensembl


VEP web interface
Input form
Results
VEP script
Tutorial
Download and install
Running VEP
Annotation sources
Filtering results
Custom annotations
Plugins
Examples and use cases
Other information
Data formats
Variant Recoder
Haplosaurus
FAQ
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Variant Effect Predictor
VEP determines the effect of your variants (SNPs, insertions, deletions, CNVs or structural variants) on genes, transcripts, and protein sequence, as well as regulatory regions.
Simply input the coordinates of your variants and the nucleotide changes to find out the:
Genes and Transcripts affected by the variants
Location of the variants (e.g. upstream of a transcript, in coding sequence, in non-coding RNA, in regulatory regions)
Consequence of your variants on the protein sequence (e.g. stop gained, missense, stop lost, frameshift)
Known variants that match yours, and associated minor allele frequencies from the 1000 Genomes Project
SIFT and PolyPhen scores for changes to protein sequence
... And more! See data types, versions.
Web interface
Point-and-click interface
Suits smaller volumes of data
Documentation
Command line tool
More options and flexibility
For large volumes of data
Documentation
REST API
Language-independent API
Simple URL-based queries
Documentation
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Clone from GitHub
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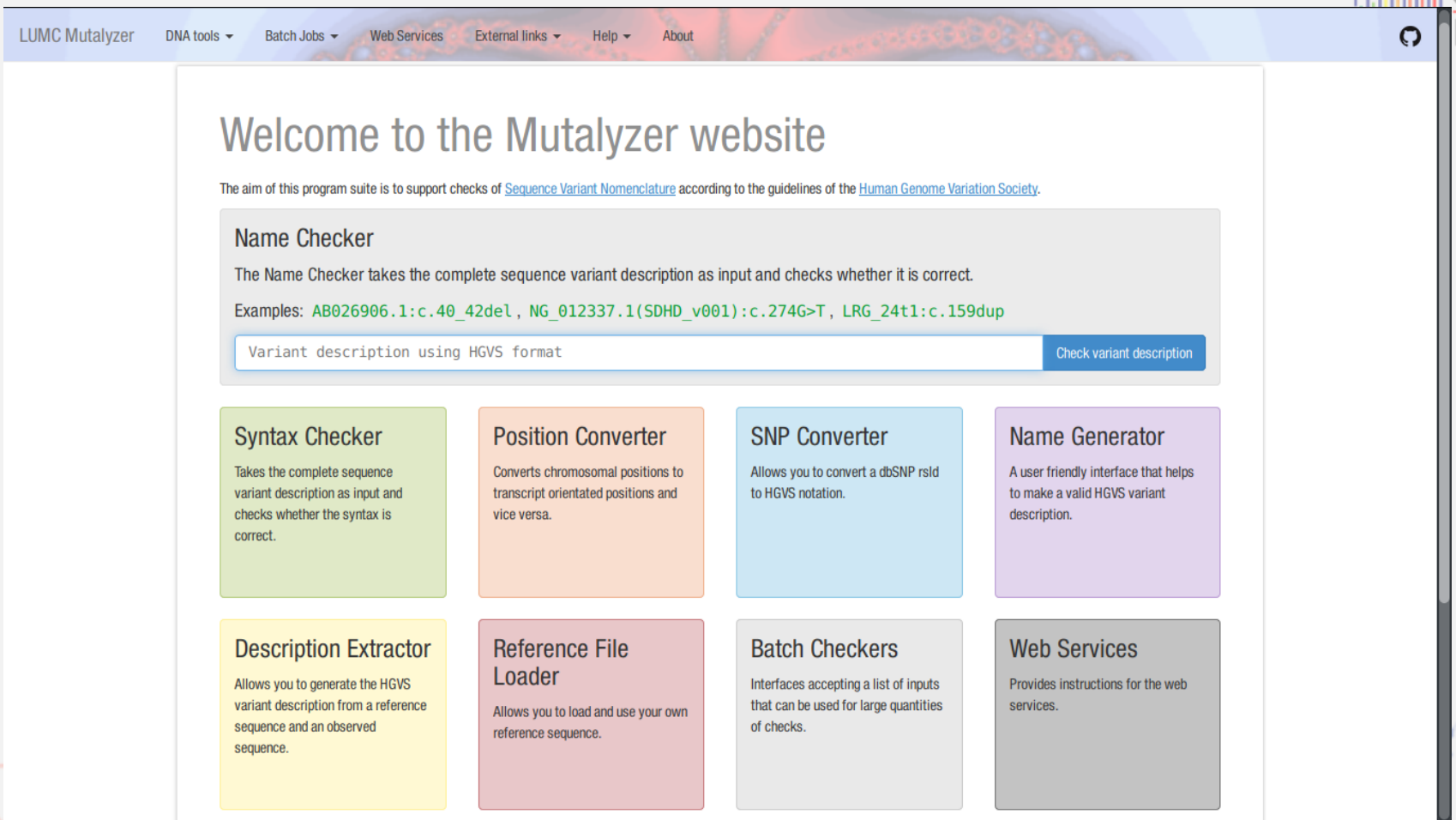
Web Tools
Web Tools
BLAST/BLAT
Variant Effect Predictor
VEP analysis of pasted data
Linkage Disequilibrium Calculator
File Chameleon
Assembly Converter
ID History Converter
VCF to PED Converter
Data Slicer
Configure this page
Custom tracks
Export data
Share this page
Bookmark this page
Variant Effect Predictor results
Category Count
Variants processed 1
Variants filtered out 0
Novel / existing variants 0 (0.0) / 1 (100.0)
Overlapped genes 4
Overlapped transcripts 15
Overlapped regulatory features 1
Consequences (all)
downstream_gene_variant: 59%
upstream_gene_variant: 12%
regulatory_region_variant: 6%
non_coding_transcript_exon_variant: 6%
NMD_transcript_variant: 6%
3_prime_UTR_variant: 6%
missense_variant: 3%
stop_gained: 3%
Coding consequences
missense_variant: 50%
stop_gained: 50%
Uploaded variant Location Allele Consequence Impact Symbol Gene Feature type Feature Biotype Exon
rs62123481 19:38384432-38384432 A downstream_gene_variant MODIFIER PSMD8 ENSG00000099341 Transcript ENST00000215071 protein_coding - -
rs62123481 19:38384432-38384432 C downstream_gene_variant MODIFIER PSMD8 ENSG00000099341 Transcript ENST00000215071 protein_coding - -
rs62123481 19:38384432-38384432 A stop_gained HIGH GGN ENSG00000179168 Transcript ENST00000334928 protein_coding 4/4 20
rs62123481 19:38384432-38384432 C missense_variant MODERATE GGN ENSG00000179168 Transcript ENST00000334928 protein_coding 4/4 20
rs62123481 19:38384432-38384432 A upstream_gene_variant MODIFIER AC005789.1 ENSG00000267090 Transcript ENST00000585411 antisense - -
rs62123481 19:38384432-38384432 C upstream_gene_variant MODIFIER AC005789.1 ENSG00000267090 Transcript ENST00000585411 antisense - -
rs62123481 19:38384432-38384432 A downstream_gene_variant MODIFIER PSMD8 ENSG00000099341 Transcript ENST00000585598 protein_coding - -
rs62123481 19:38384432-38384432 C downstream_gene_variant MODIFIER PSMD8 ENSG00000099341 Transcript ENST00000585598 protein_coding - -
rs62123481 19:38384432-38384432 A 3_prime_UTR_variant, NMD_transcript_variant MODIFIER GGN ENSG00000179168 Transcript ENST00000585737 nonsense_mediated_decay 5/5 16
rs62123481 19:38384432-38384432 C 3_prime_UTR_variant, NMD_transcript_variant MODIFIER GGN ENSG00000179168 Transcript ENST00000585737 nonsense_mediated_decay 5/5 16
rs62123481 19:38384432-38384432 A downstream_gene_variant MODIFIER GGN ENSG00000179168 Transcript ENST00000586599 protein_coding - -


LUMC Mutalyzer
DNA tools
Batch Jobs
Web Services
External links
Help
About
Welcome to the Mutalyzer website
The aim of this program suite is to support checks of Sequence Variant Nomenclature according to the guidelines of the Human Genome Variation Society.
Name Checker
The Name Checker takes the complete sequence variant description as input and checks whether it is correct.
Examples: AB026906.1:c.40_42del, NG_012337.1(SDHD_v001):c.274G>T, LRG_24t1:c.159dup
Variant description using HGVS format
Check variant description
Syntax Checker
Takes the complete sequence variant description as input and checks whether the syntax is correct.
Position Converter
Converts chromosomal positions to transcript orientated positions and vice versa.
SNP Converter
Allows you to convert a dbSNP rsId to HGVS notation.
Name Generator
A user friendly interface that helps to make a valid HGVS variant description.
Description Extractor
Allows you to generate the HGVS variant description from a reference sequence and an observed sequence.
Reference File Loader
Allows you to load and use your own reference sequence.
Batch Checkers
Interfaces accepting a list of inputs that can be used for large quantities of checks.
Web Services
Provides instructions for the web services.

Teşekkürler

Barış Salman

Gen-Era Diagnostik