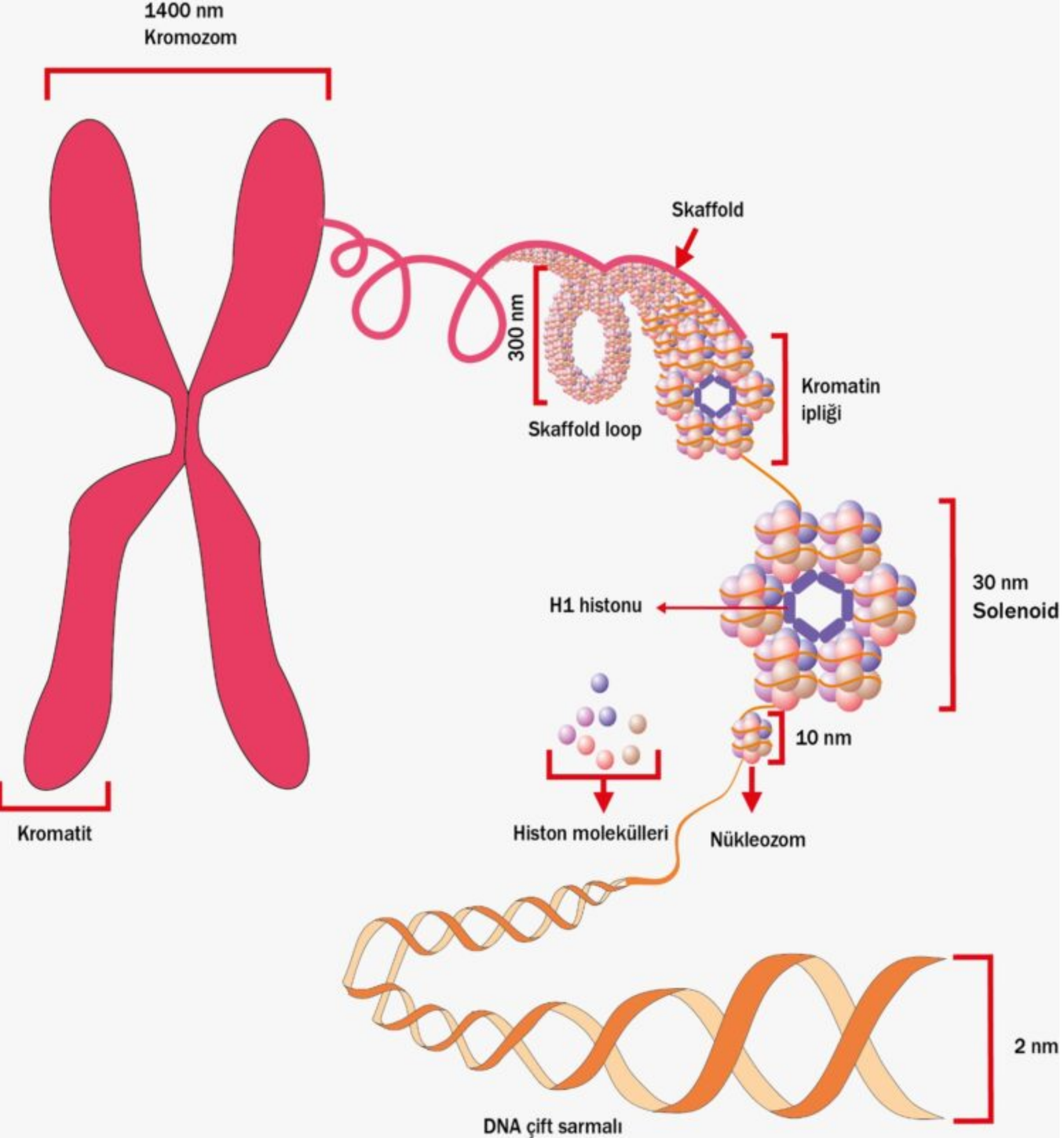
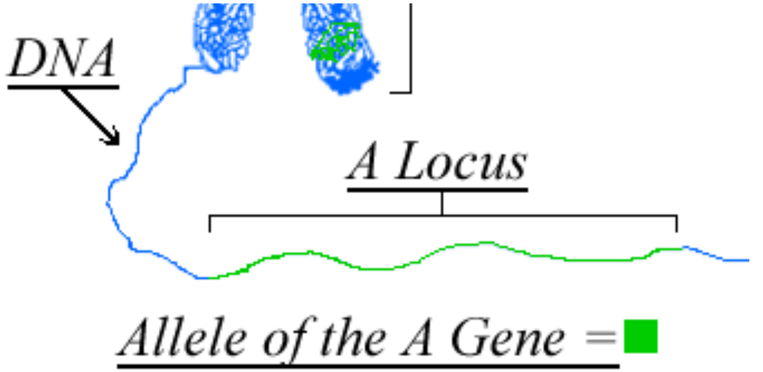
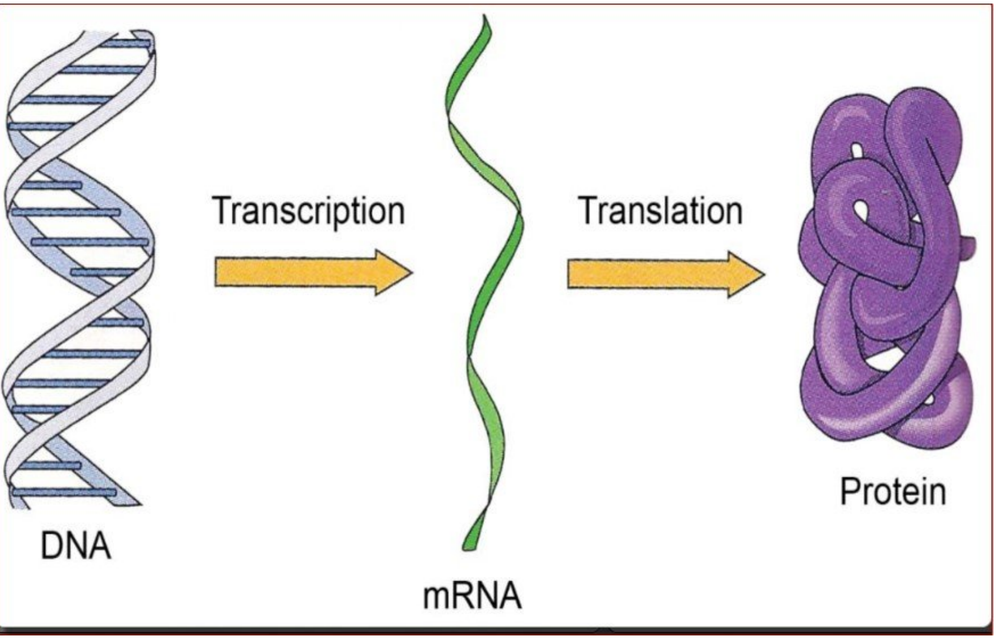
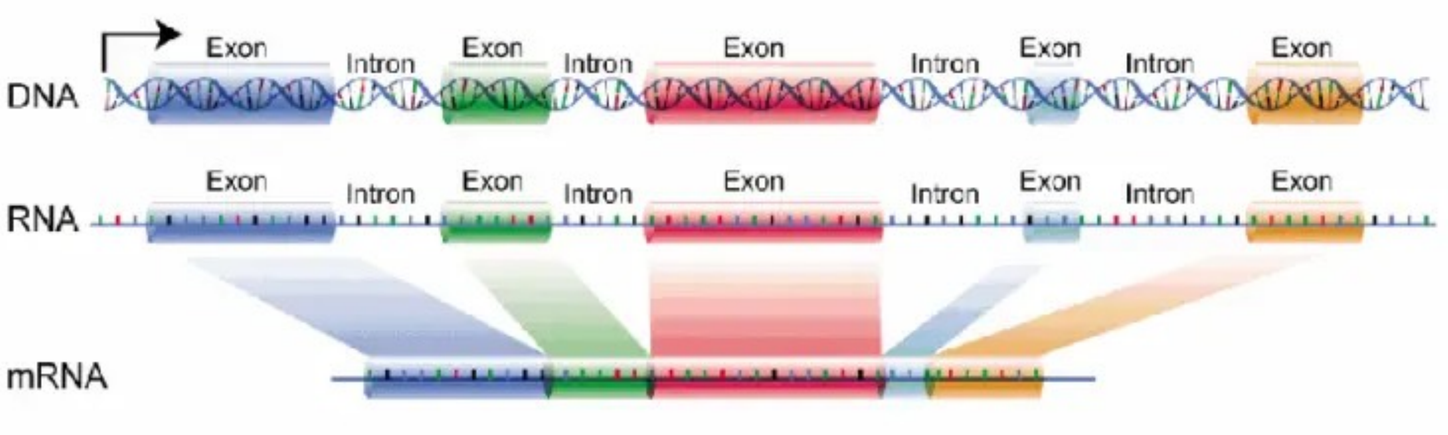
SNP ve GENETİK

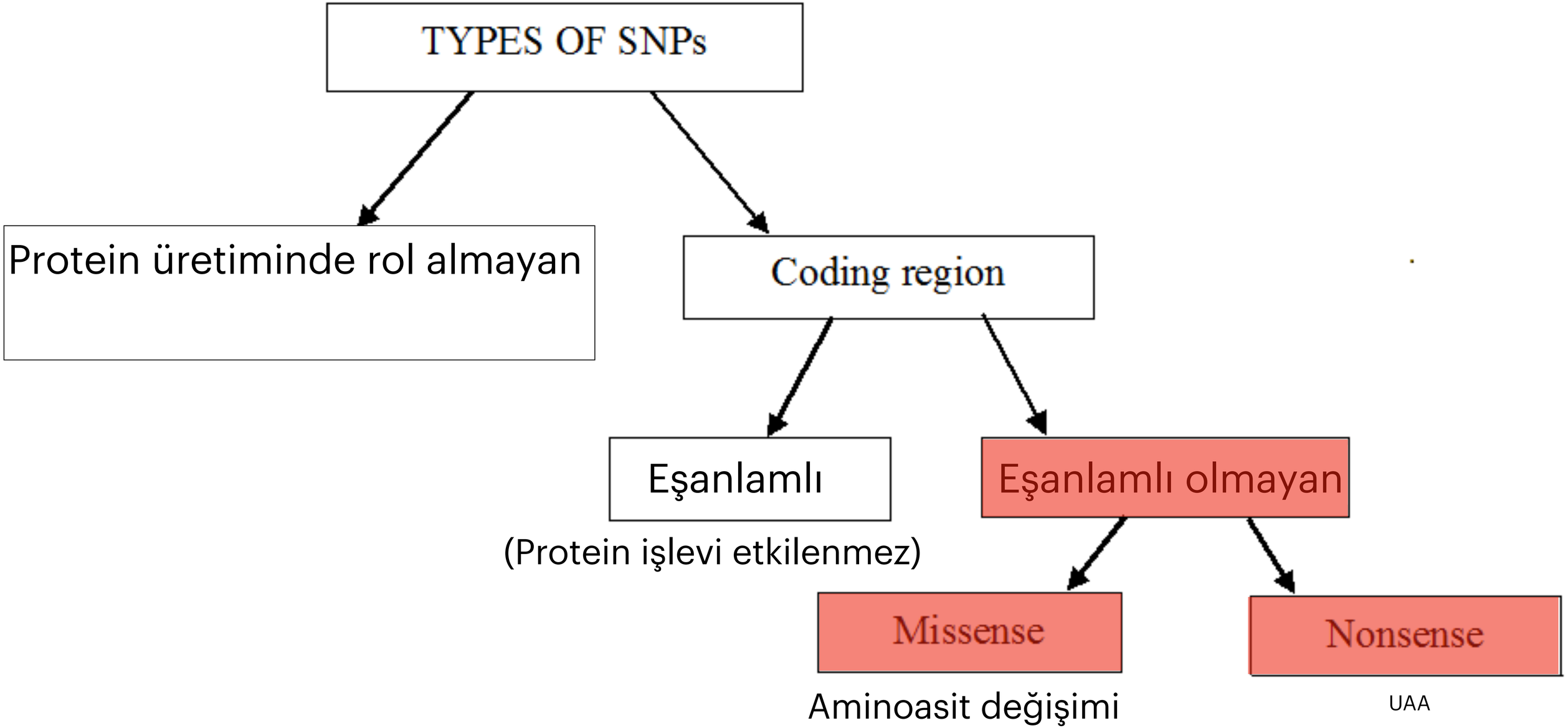
SNP’lerin Oluşumu ve Genetikteki Yeri


1400 nm
Kromozom
Skaffold
300 nm
Skaffold loop
Kromatin
ipliği
H1 histonu
30 nm
Solenoid
10 nm
Kromatit
Histon molekülleri
Nükleozom
2 nm
DNA çift sarmalı


DNA
A Locus
Allele of the A Gene =


Transcription
Translation
DNA
mRNA
Protein


DNA
RNA
mRNA
Exon
Intron
Exon
Intron
Exon
Intron
Exon
Intron
Exon


TYPES OF SNPs
Protein üretiminde rol almayan
Coding region
Eşanlamlı
(Protein işlevi etkilenmez)
Eşanlamlı olmayan
Missense
Aminoasit değişimi
Nonsense
UAA
UAG
UGA

TP53 tumor protein p53 [*Homo sapiens* (human)]

Gene ID: 7157, updated on 12-Nov-2024

Download Datasets

Summary

Official Symbol TP53 provided by HGNC
Official Full Name tumor protein p53 provided by HGNC
Primary source HGNC:HGNC:11998
See related Ensembl:ENSG00000141510 MIM:191170; AllianceGenome:HGNC:11998
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 P53; BCC7; LFS1; BMFS5; TRP53
Summary This gene encodes a tumor suppressor protein containing transcriptional activation, DNA binding, and oligomerization domains. The encoded protein responds to diverse cellular stresses to regulate expression of target genes, thereby inducing cell cycle arrest, apoptosis, senescence, DNA repair, or changes in metabolism. Mutations in this gene are associated with a variety of human cancers, including hereditary cancers such as Li-Fraumeni syndrome. Alternative splicing of this gene and the use of alternate promoters result in multiple transcript variants and isoforms. Additional isoforms have also been shown to result from the use of alternate translation initiation codons from identical transcript variants (PMIDs: 12032546, 20937277). [provided by RefSeq, Dec 2016]
Expression Ubiquitous expression in spleen (RPKM 13.2), lymph node (RPKM 13.1) and 25 other tissues See more
Orthologs mouse all

NEW **Try the new Gene table**
Try the new Transcript table

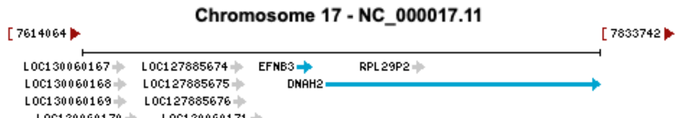
Genomic context

Location: 17p13.1

Exon count: 13

See TP53 in Genome Data Viewer

Annotation release	Status	Assembly	Chr	Location
RS_2024_08	current	GRCh38.p14 (GCF_000001405.40)	17	NC_000017.11 (7668421..7687490, complement)
RS_2024_08	current	T2T-CHM13v2.0 (GCF_009914755.1)	17	NC_060941.1 (7572544..7591594, complement)
RS_2024_09	previous assembly	GRCh37.p13 (GCF_000001405.25)	17	NC_000017.10 (7571739..7590808, complement)



SNV

Single Nucleotide Variation

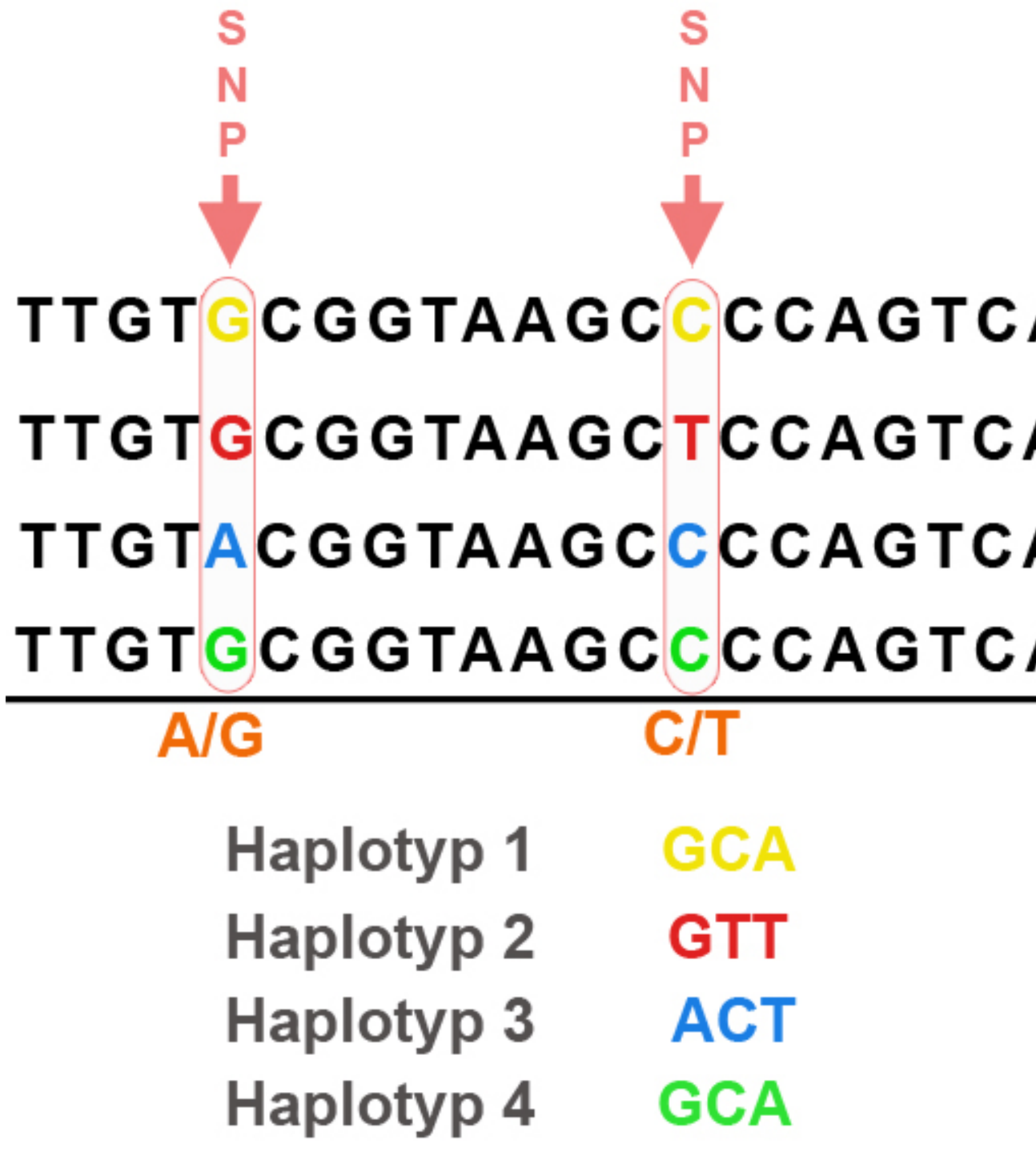
- <%1
- Nadir görülür
- Aile veya belirli bireylerle sınırlıdır

Single Nucleotide Variation

SNP

Tek Nükleotid Polimorfizmi

- %1>
- İki insan arasında benzerlik %99,9 olmasına rağmen SNP 30 - 300 milyon vardır.
- Her **SNP** ayrıca **SNV**'dir.



$2^{10,000,000}$

Evrendeki atom sayısı 2^{66}

Genetik Varyasyon

- Genetik çeşitlilik
- Kişiselleştirilmiş Tıp

- **Karmaşık**
- **SNP - SNP etkileşimi**

Kocaeli Üniversitesi Sağlık Bilir

ARAŞTIRMA MAKALESİ

TR EN

PDF BibTex RIS Kaynak Gö

Meme Kanserinde TP53 (RS1042522) Polimorfizmi

Yıl 2016, Cilt: 2 Sayı: 1, 28 - 31, 31.01.2016

Nihal Üren, Tuğcan Korak *, Duygu Altınok, Emel [illegible]gül, Bahadır Güllüoğlu, Turgay Şimşe
, Zafer Cantürk, Zafer Utkan, Ali Sazcı

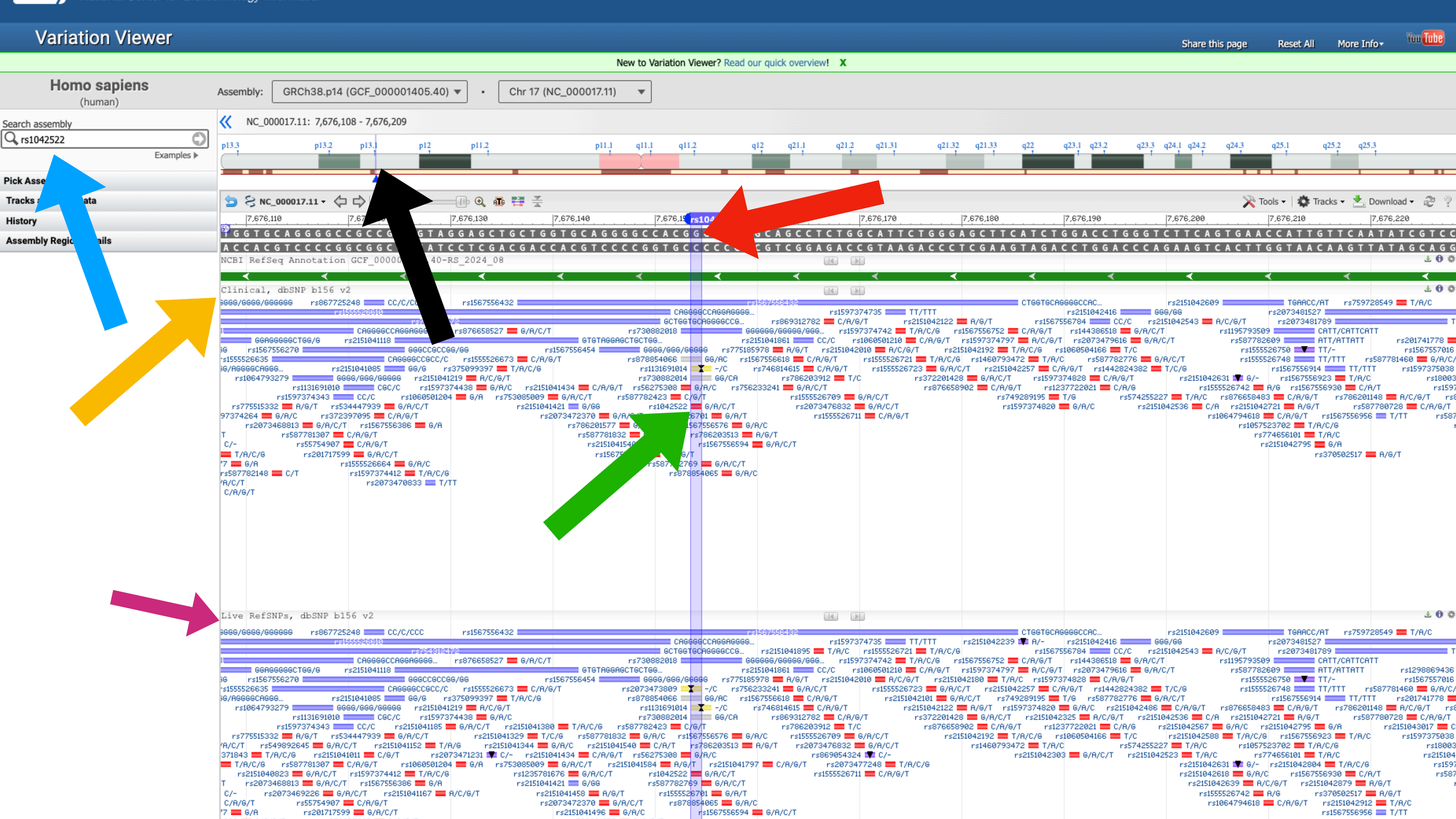
https://doi.org/10.30934/kusbed.358489

Cited By: 1

Öz

Amaç: TP53 geni temel olarak DNA tamiri, apoptozis, hücre yaşlanması ve hücre döngü kontrolünde görev alan en önemli tümör baskılayıcı genlerden biridir. TP53 rs10425 (Arg72Pro) polimorfizmi tümör baskılama sırasında P53 protein yapısında değişikliğe ned olan bir polimorfizmdir. Bu verilere dayanarak, bu çalışmanın amacı TP53 rs10425 polimorfizmi ve meme kanseri riski arasındaki ilişkiyi araştırmaktır.

Yöntem: TP53 rs1042522 polimorfizmi için 508 meme kanserli kadın hastadan ve 367 sağl kadından alınan periferik kanlardan DNA izole edilerek PCR-RFLP yöntemi ile genotipler


Variation Viewer
Share this page
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More Info
New to Variation Viewer? Read our quick overview!
Homo sapiens
(human)
Assembly:
GRCh38.p14 (GCF_000001405.40)
Chr 17 (NC_000017.11)
Search assembly
rs1042522
Examples
Pick Asse
Tracks
ata
History
Assembly Regio
ails
NC_000017.11: 7,676,108 - 7,676,209
NC_000017.11
Tools
Tracks
Download
NCBI RefSeq Annotation GCF_0000
40-RS_2024_08
Clinical, dbSNP b156 v2
Live RefSNPs, dbSNP b156 v2

Nokta Mutasyonu

%38

Welcome to the Reference SNP (rs) Report
All alleles are reported in the Forward orientation. Click on the Variant Details tab for details on Genomic Placement, Gene, and Amino Acid changes. HGVS names are in the HGVS tab.

Reference SNP (rs) Report

Download

Current Build 156
Released September 21, 2022

rs1042522

Organism	*Homo sapiens*
P	chr17:7676154 (GRCh38.p14)
Alleles	G>A / G>C / G>T
Variation Type	SNV Single Nucleotide Variation
Frequency	G=0.382621 (101276/264690, TOPMED) G=0.331835 (83136/250534, GnomAD_exome) G=0.373457 (52281/139992, GnomAD) (+ 23 more)
Clinical Significa	ported in ClinVar
Gene : Consequence	TP53 : Missense Variant
Publications	390 citations LitVar 2054
Genomic View	See rs on genome

Frequency | Variant Details | Clinical Significance | HGVS | Submissions | History | Publications | Flanks

ALFA Allele Frequency

The ALFA project provide aggregate allele frequency from dbGaP. More information is available on the project page including descriptions, data access, and terms of use.

Release Version: 20231103111315

Search:

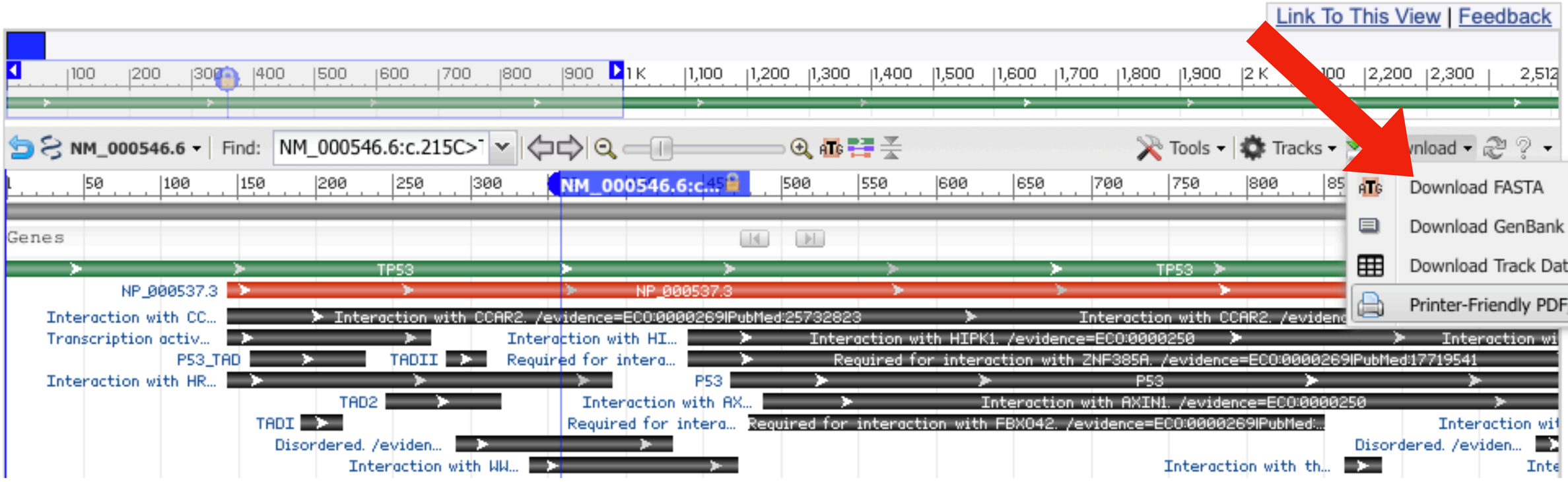
Population	Group	Sample Size	Ref Allele	Alt Allele	Ref HMOZ	Alt HMOZ	HTRZ	HWEP
Total	Global	105516	G=0.285663	C=0.714337, T=0.000000	0.094659	0.523333	0.382008	32
European	Sub	90388	G=0.26336	C=0.73664, T=0.00000	0.073439	0.54671	0.379851	11
African	Sub	2988	G=0.6854	C=0.3146, T=0.0000	0.523427	0.15261	0.323963	32

Graphics ▾ Send to: ▾

Homo sapiens tumor protein p53 (TP53), transcript variant 1, mRNA

NCBI Reference Sequence: NM_000546.6

GenBank FASTA



Analyze th
Run BLAST
Pick Primer
Show in Ge
Articles al
The p53 pro
Reference

```
2151085896.fasta dosyası masaüstüne indirildi.
2151085754.fasta dosyası masaüstüne indirildi.
2151085636.fasta dosyası masaüstüne indirildi.
2151085616.fasta dosyası masaüstüne indirildi.
2151085508.fasta dosyası masaüstüne indirildi.
2151085459.fasta dosyası masaüstüne indirildi.
2151085443.fasta dosyası masaüstüne indirildi.
2151085436.fasta dosyası masaüstüne indirildi.
2151085294.fasta dosyası masaüstüne indirildi.
(base) sahil@sahil-3 ~ %
```

a.py

```python
from Bio import Entrez
import os

Entrez.email = "dogukansahil@gmail.
Entrez.api_key = "a6190b141aea42289

desktop_path = os.path.join(os.path.expanduser("~"), "Desktop")

def fetch_snp_fasta(gene_name):
    # SNP'leri bulmak için arama yap
    search_handle = Entrez.esearch(db="snp", term=gene_name, retmax=10)
```

```
ACCCTCTTTTCTTATCATTGACATTTAAACTCTGGGGCAGGTCCTCGCGTAGAACGCGGCT
GCCACTTCCCCTGCCGAGCGGCGGTGAGAAGTGTGGGAACCGGCGCTGCCAGGCTCACCTG
CCTCCGCTCCCAGGTAACCGCCCGGGCTCCGGCCCCGGCCCGGCTCGGGGCCCGCGGGGCC
CCAGCGACTGCTGTCCCCAAATCAAAGCCCGCCCCAAGTGGCCCCGGGGCTTGATTTTTGC
GAGGCATACAAAGATGGAAGCGAGTTACTGAGGGAGGGATAGGAAGGGGGGTGGAGGAGGC
TGCCGAGTGTGCTCTTCTGCAAAAGTAGCAAAATGTTCCACTCCTAAGAGTGGACTTCCAC
GAGCTGGGAGTAGGGGGCGGGAGTCTGCTGCTGCTGTCTGCTAAAGCCACTCGCGACCGCG
GGAGGTGGGGACGCACTTTGCATCCAGACCTCCTCTGCATCGCAGTTCACGACATCCACGC
TCCGTACCCGCGCCTGGAGCGCTTAAAGACACCCTGCCGCGGGTCGGGCGAGGTGCAGCAC
```

- https://www.ncbi.nlm.nih.gov/snp/
- https://pmc.ncbi.nlm.nih.gov/articles/PMC1235544/
- https://pmc.ncbi.nlm.nih.gov/articles/PMC24792/