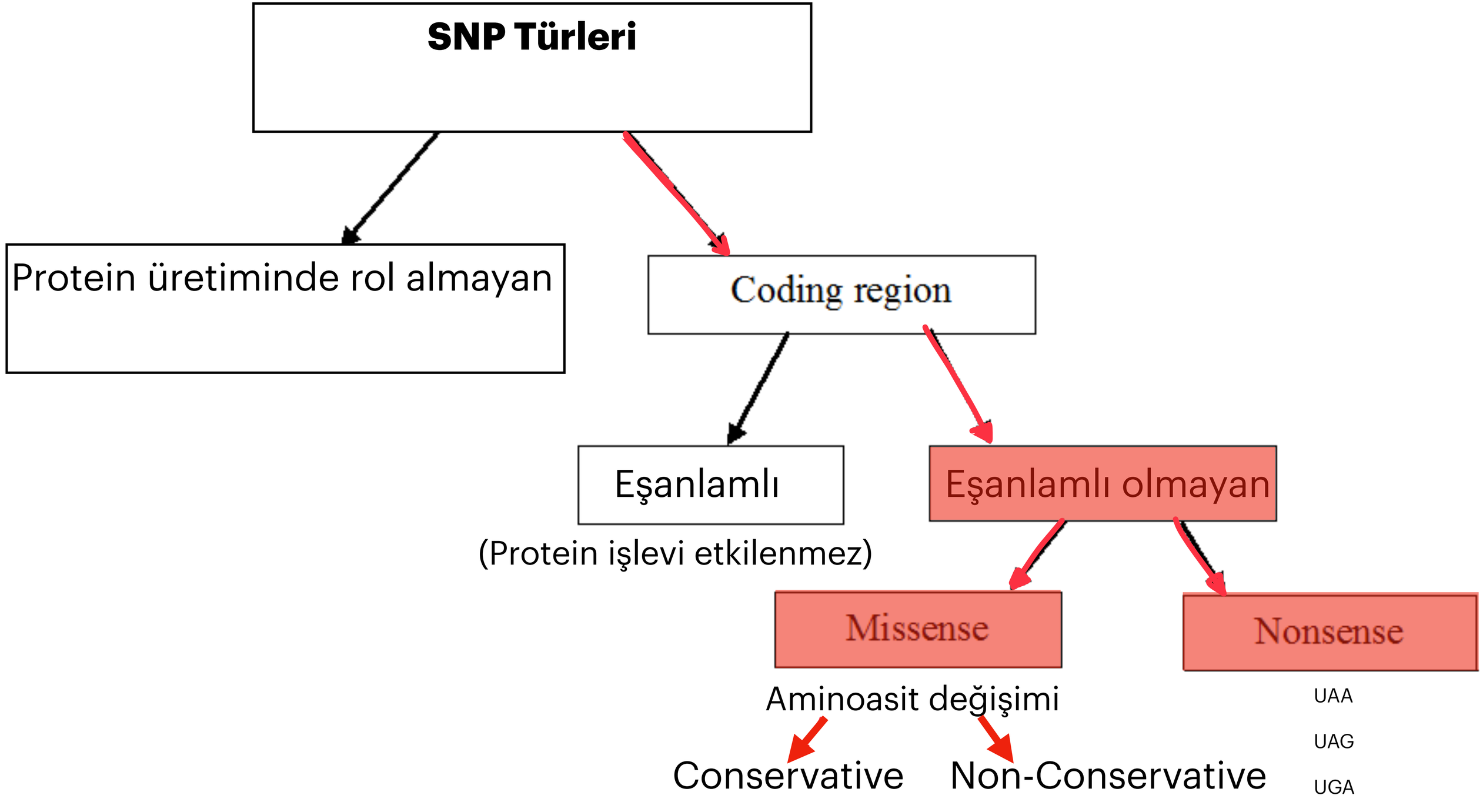
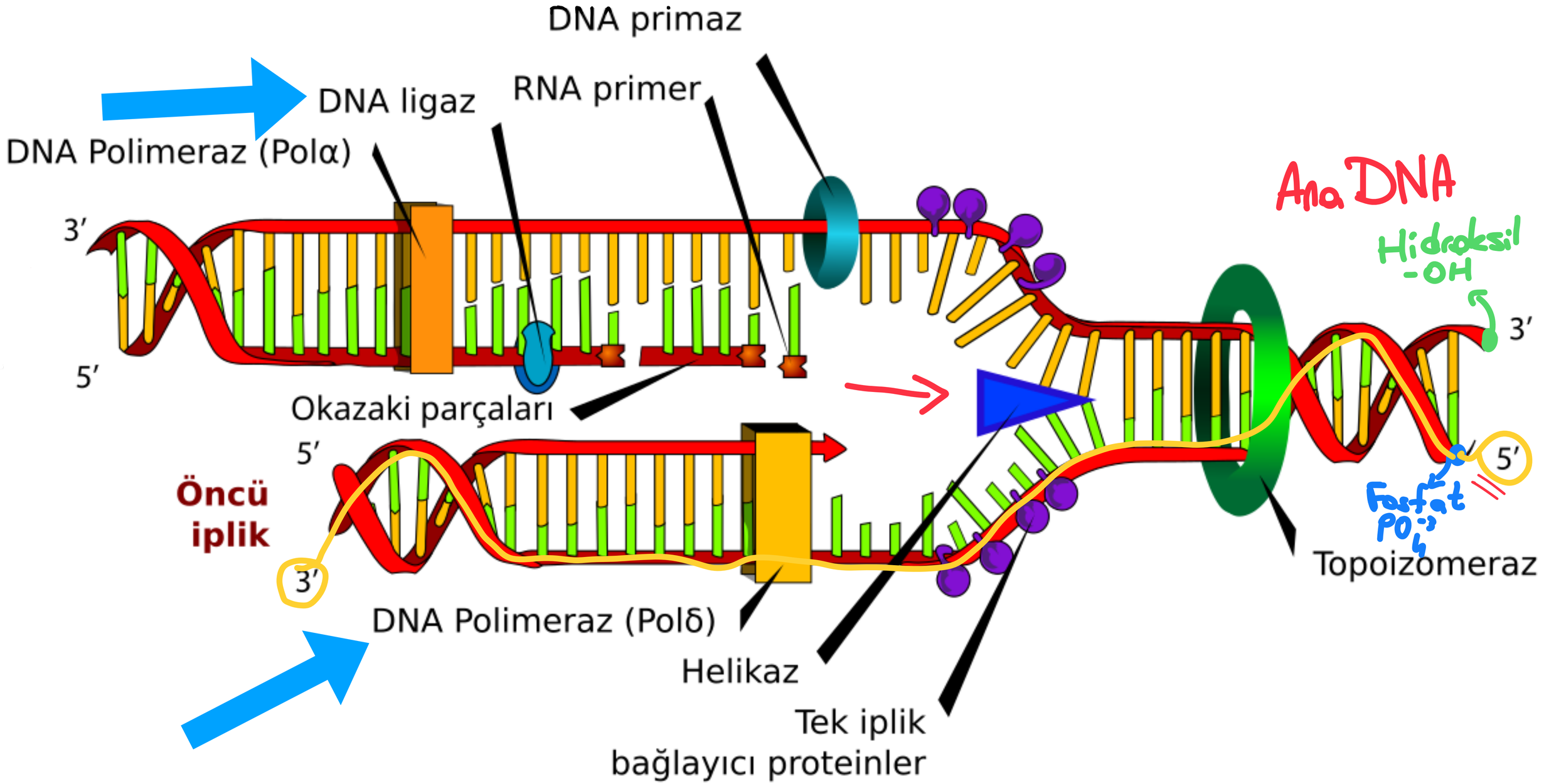
Missense ve **Nonsense** Mutasyonlar

Protein Yapısı ve FASTA format

Doğukan **SAHİL**

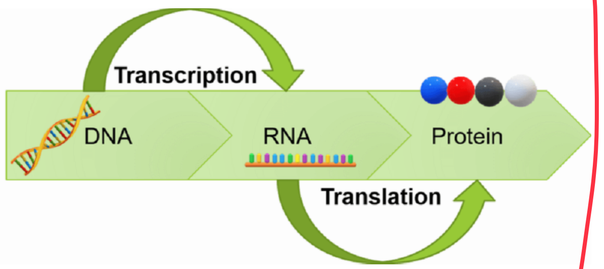

SNP Türleri
Protein üretiminde rol almayan
Coding region
Eşanlamlı
(Protein işlevi etkilenmez)
Eşanlamlı olmayan
Missense
Nonsense
Aminoasit değişimi
Conservative
Non-Conservative
UAA
UAG
UGA

DNA REPLİKASYONU



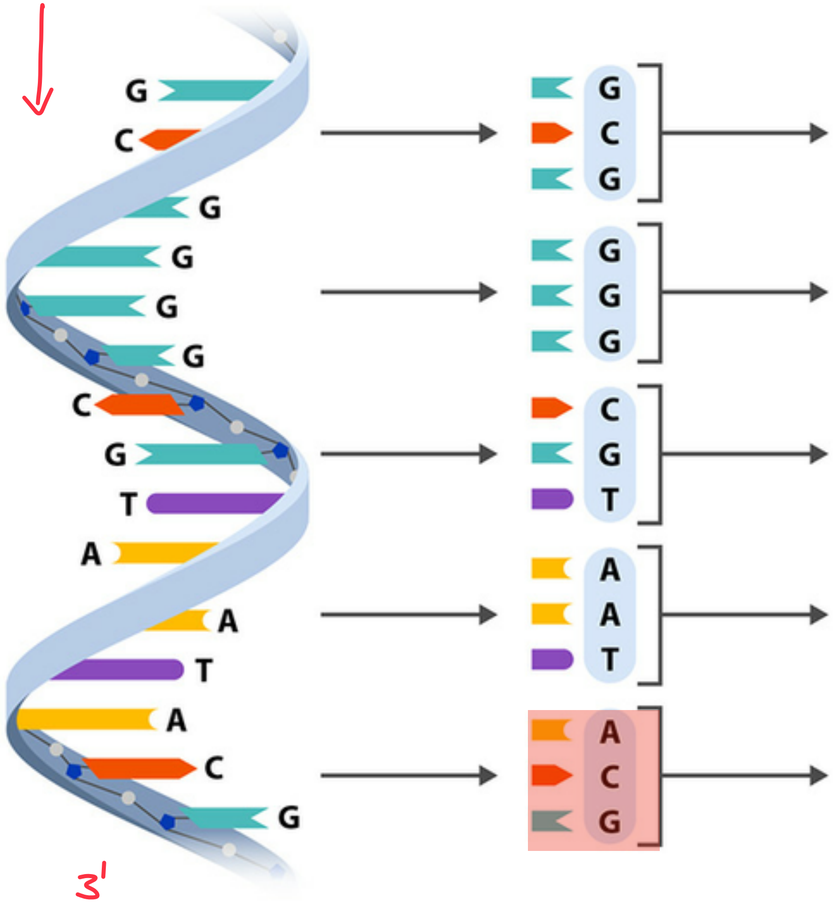
https://pmc.ncbi.nlm.nih.gov/articles/PMC8636028/

PROTEİN SENTEZİ

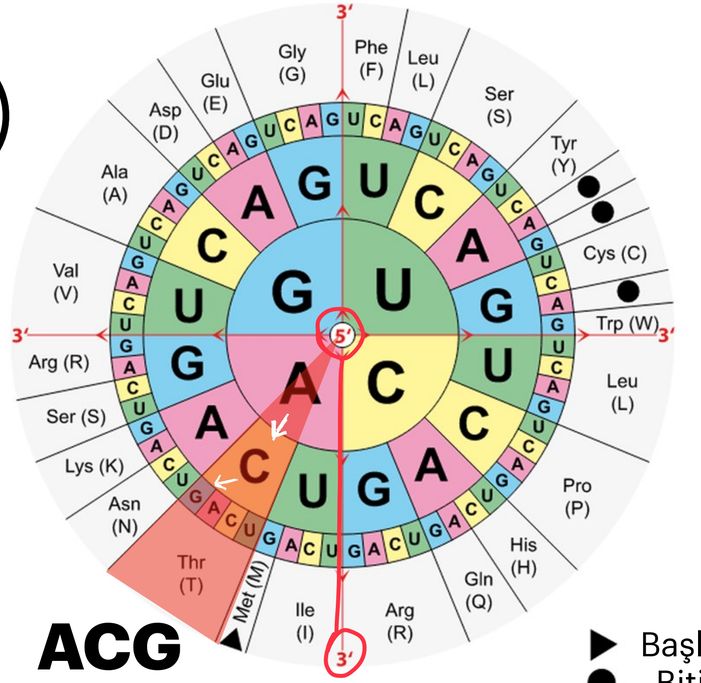


1

DNA 5'



2

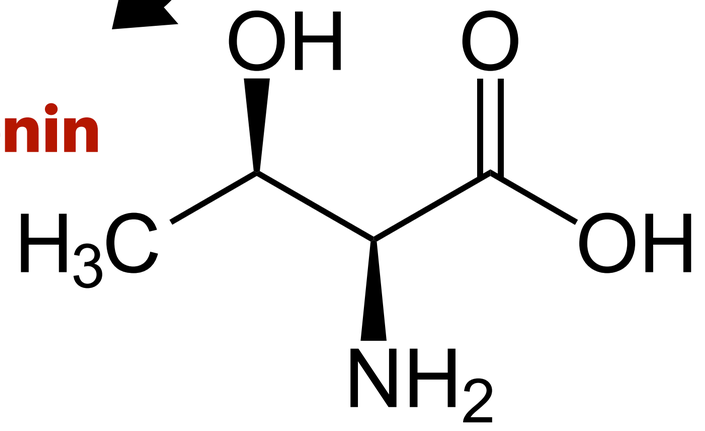


ACG

▶ Başlat
● Bitir

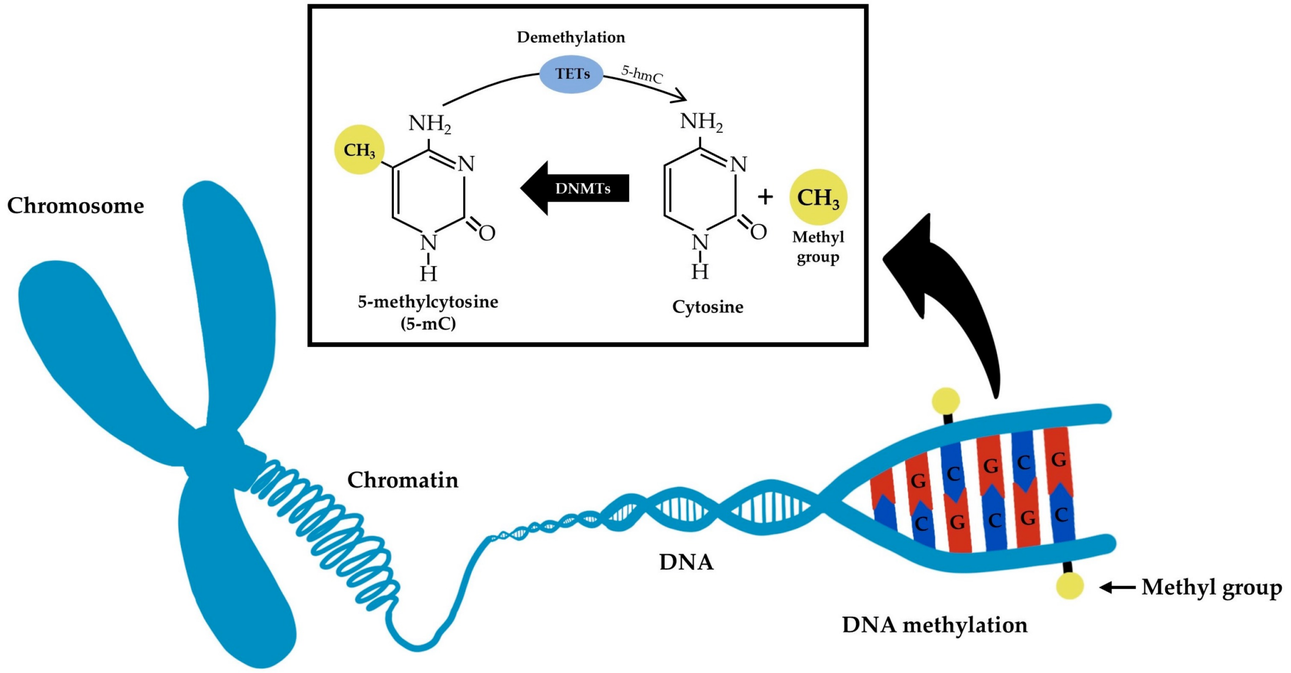
3

Treonin



DNA Metilasyonu ve Mutasyon

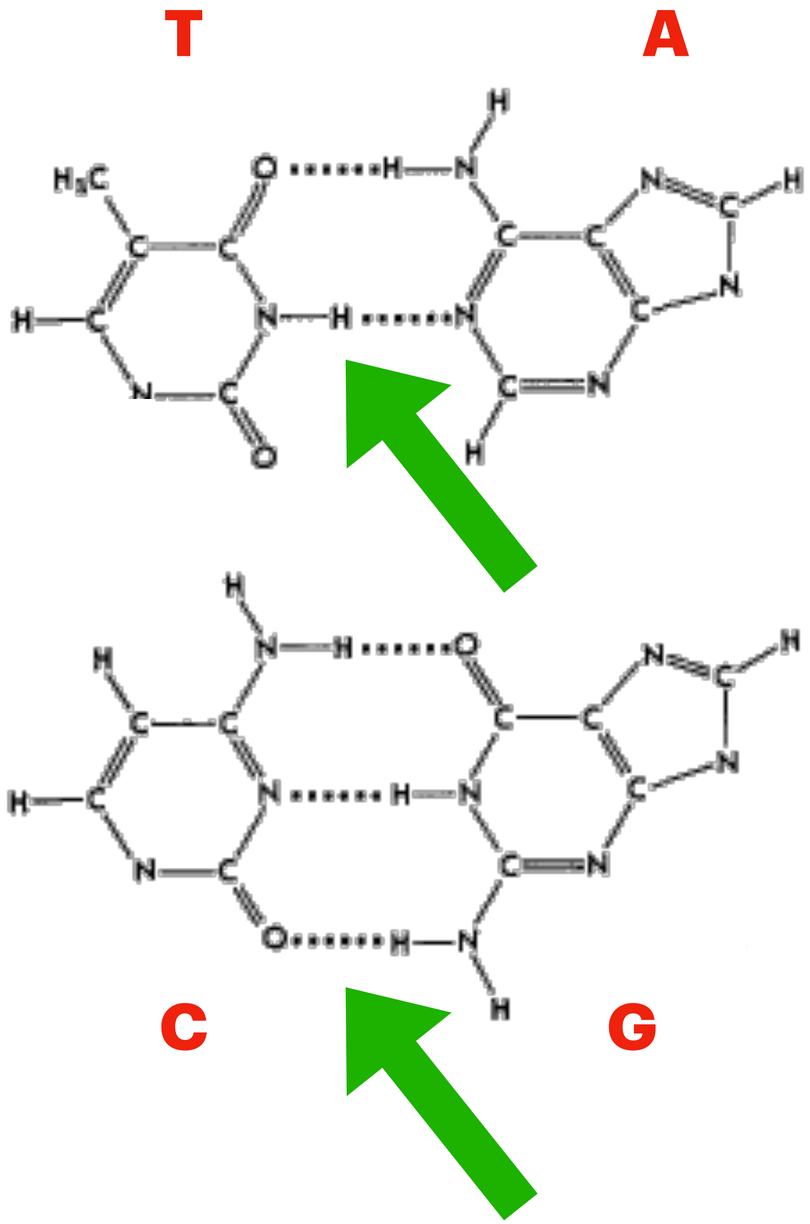
- DNA'ya **Metil** grubu eklenmesi
- Gen susturulur veya ifadesi azalır
- Genellikle **C ve G** arasında görülür



https://www.mdpi.com/2079-4991/13/12/1880

Esnek**, az stabil**

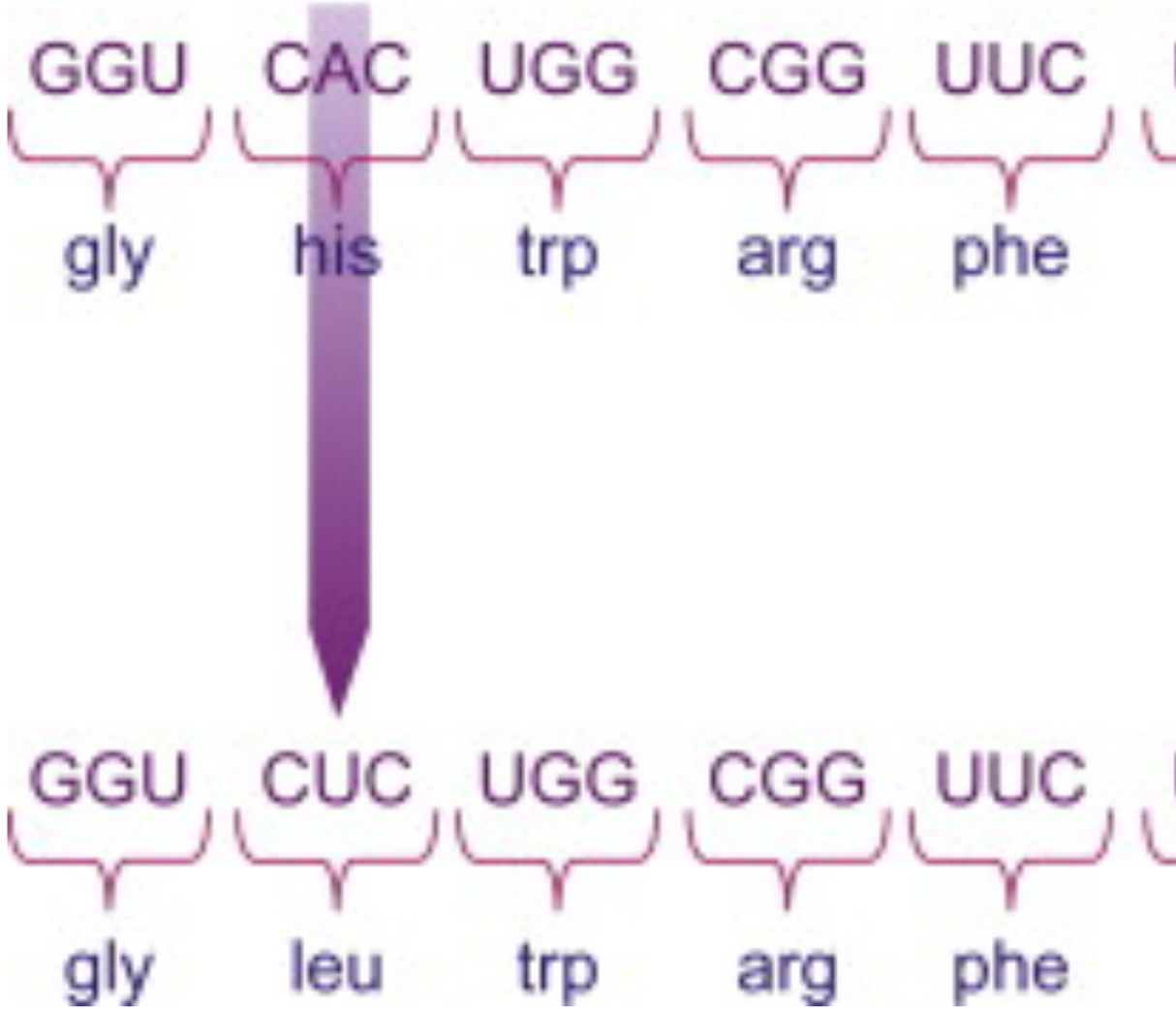
Sert, stabil



Missense Mutation

Yanlış Anlamlı Mutasyon

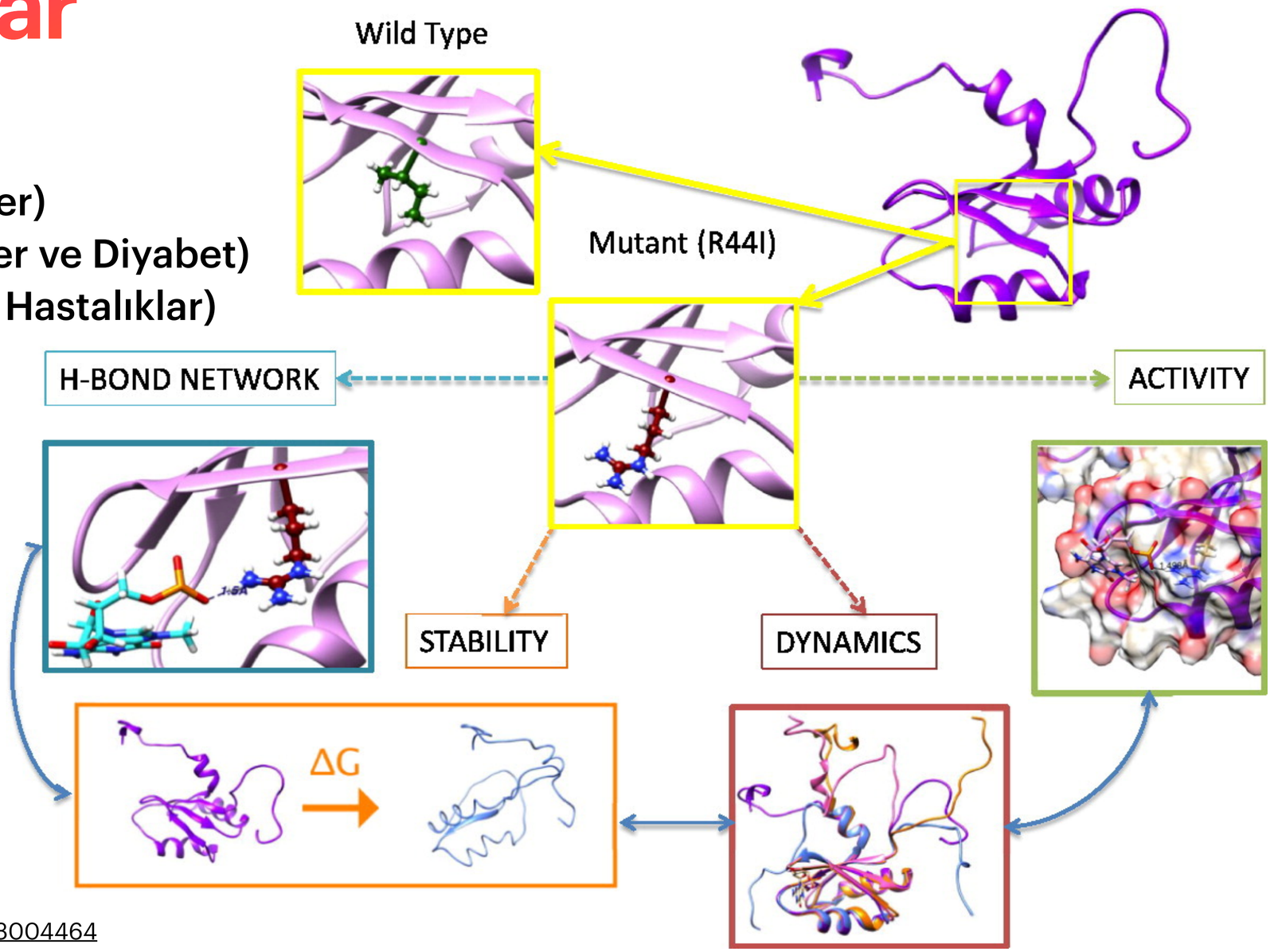
- Aminoasit değişmesine sebep olur.
- Proteinin yapısı, stabilitesi veya fonksiyonunu da değiştirebilir.
- Hastalıklarla doğrudan ilişkili.



https://pmc.ncbi.nlm.nih.gov/articles/PMC3346971/

Missense İlişkili Hastalıklar

A. Protein **stabilitesi** (KAS)
B. **Hidrojen bağı** değişikliği (Alzheimer)
C. Protein **esnekliği** değişikliği (Kanser ve Diyabet)
D. Protein aktivitesi değişikliği (Nadir Hastalıklar)

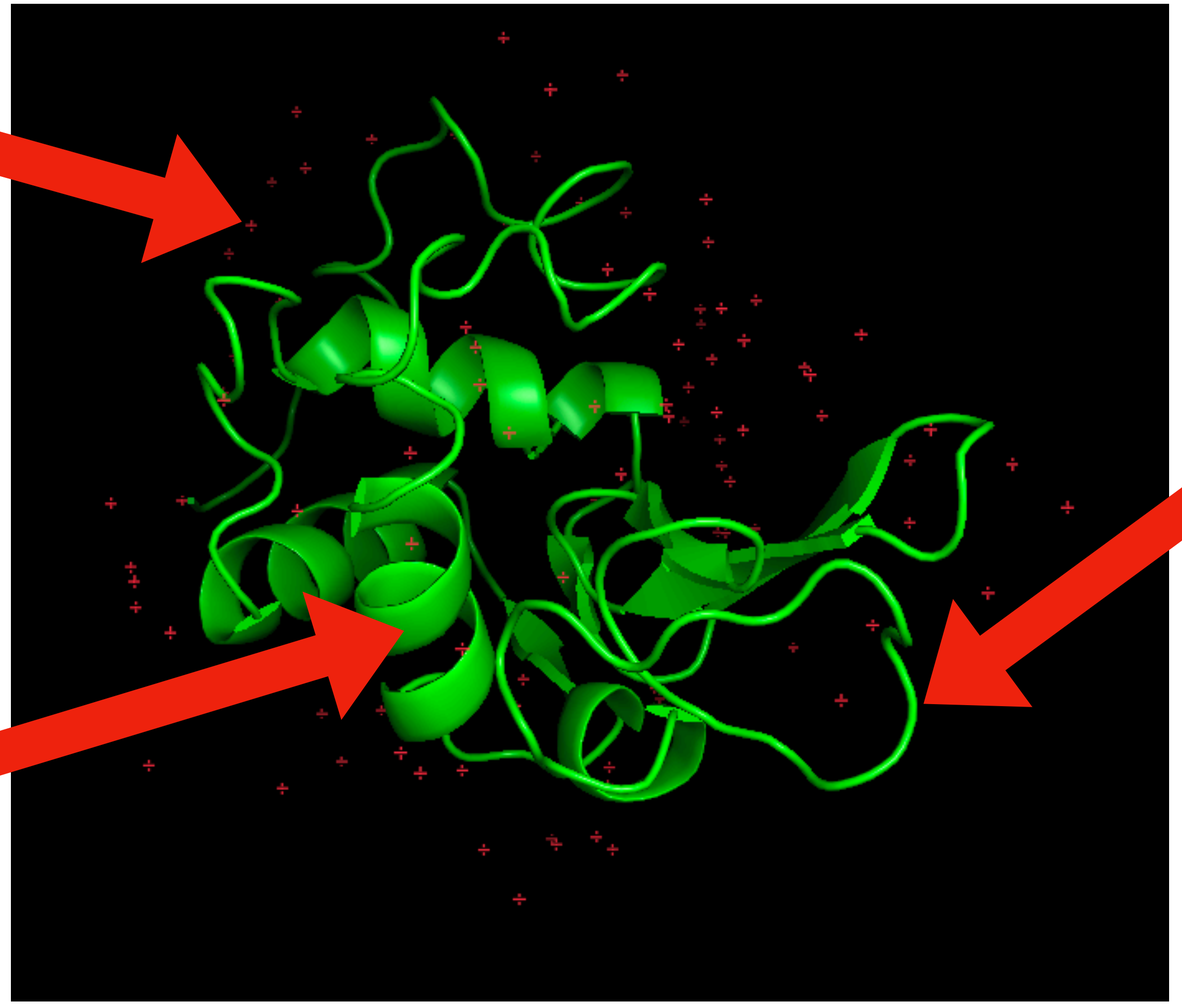


https://pmc.ncbi.nlm.nih.gov/articles/PMC3796015/
https://www.sciencedirect.com/science/article/abs/pii/S0022283613004464

Missense Mutasyonlar

Protein Yapısı

Hidrojen Bağı Alıcıları



Loops (Bağlantı Çizgileri)

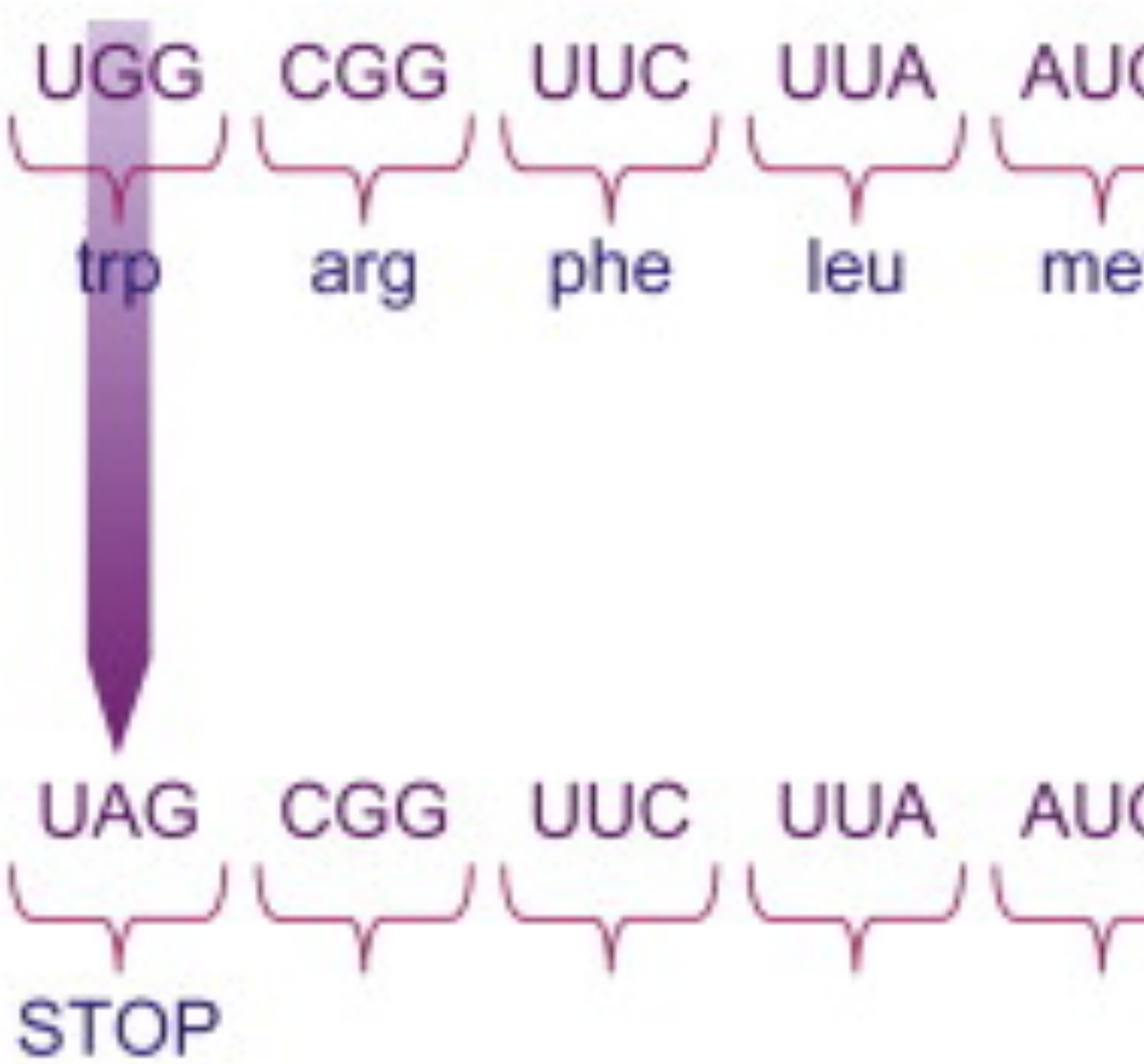
Alfa Heliks

1LYZ proteini https://www.rcsb.org/3d-view/jsmol/1lyz/0

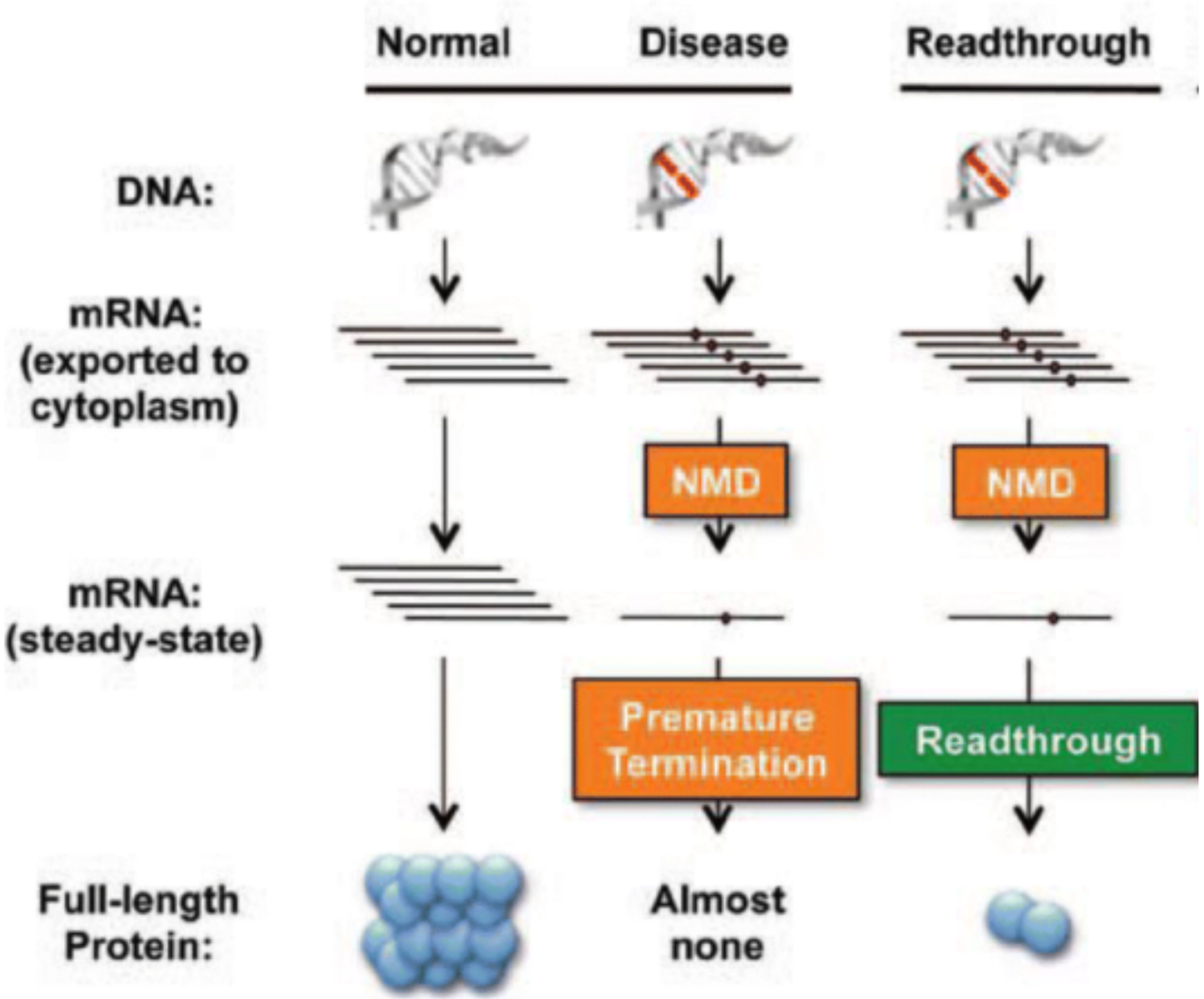
https://www.sciencedirect.com/science/article/abs/pii/S0022283613004464

Nonsense Mutation

- Durdurma
- İşlevsiz veya eksik protein
- **Readthrough**



https://pmc.ncbi.nlm.nih.gov/articles/PMC7437896/

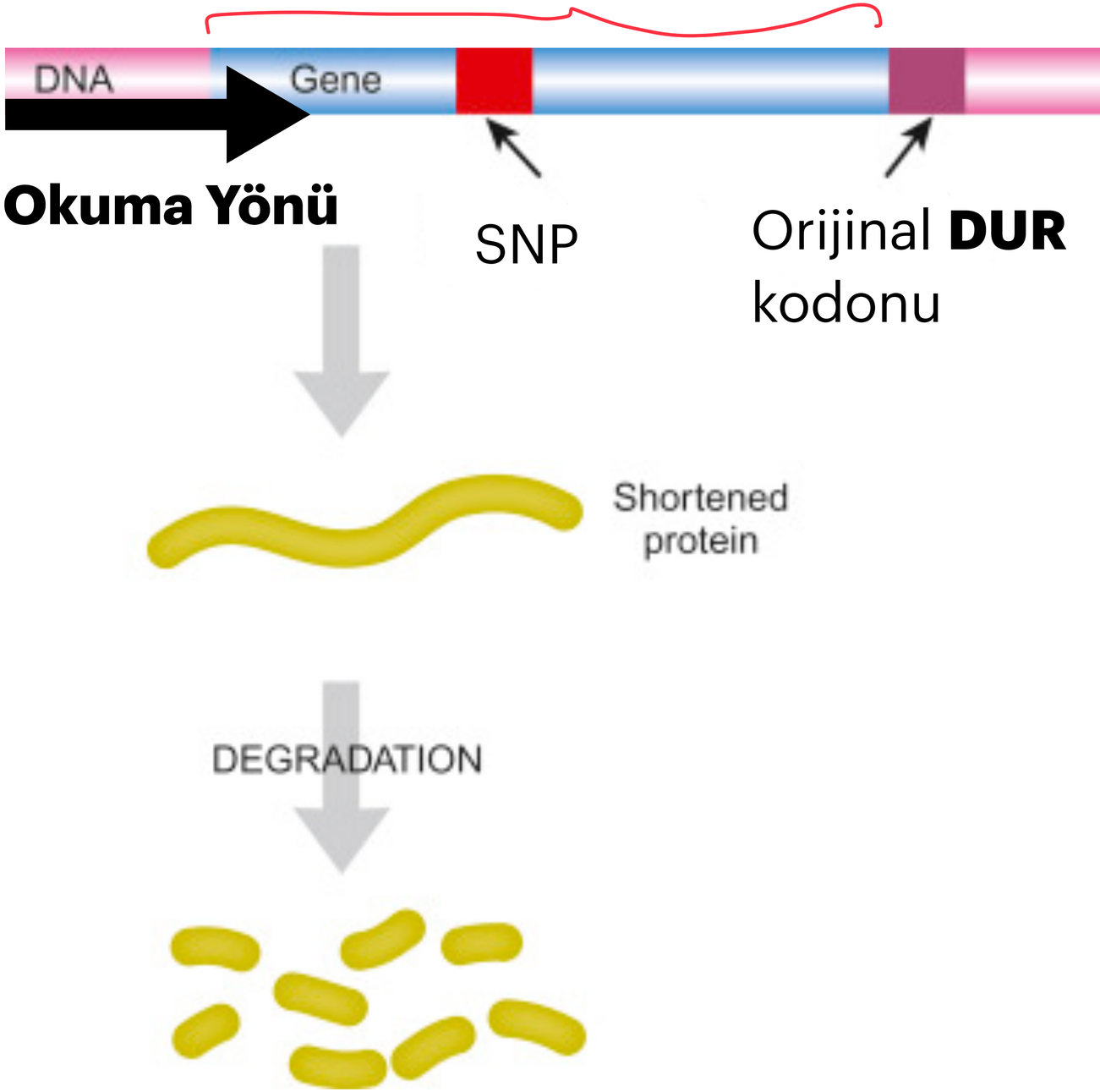


+200.000 £

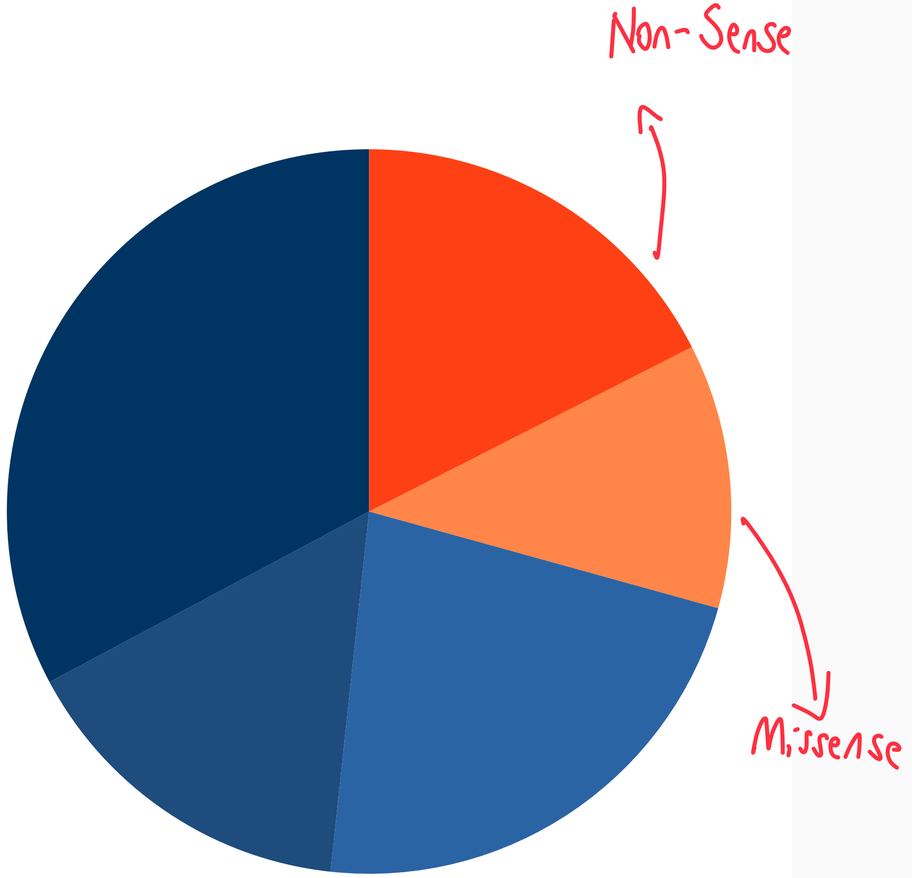
https://www.bbc.com/news/health-36056759

Nonsense İlişkili Hastalıklar

Kas hastalıkları, Görme kayıpları, kistik fibrozis ve kanserler



https://pubmed.ncbi.nlm.nih.gov/18454449/
https://pmc.ncbi.nlm.nih.gov/articles/PMC7352488/



Acta Oncologica Turcica 2009; 42: 13-16

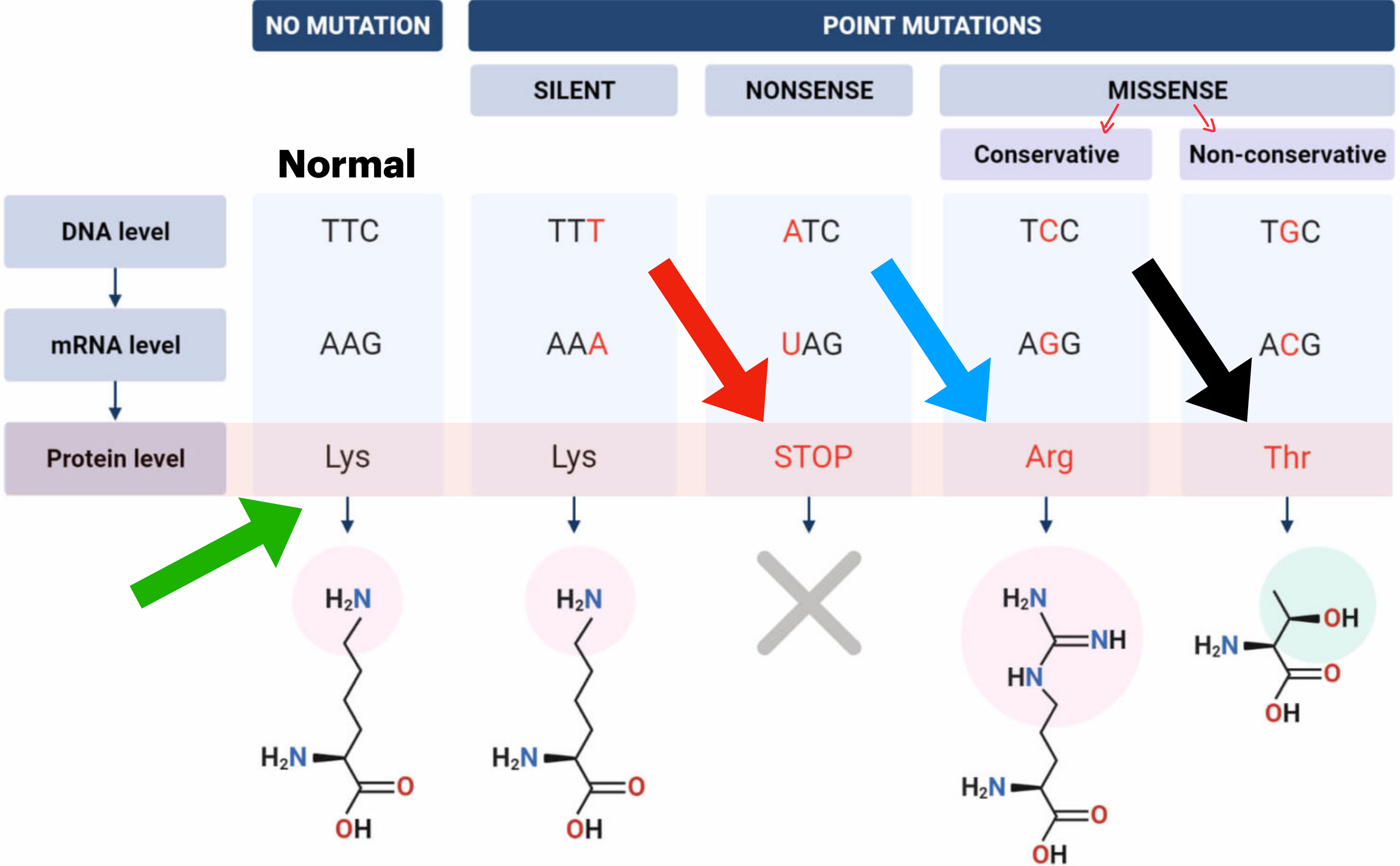
Türk Toplumundaki Nörofibromatozis Tip 1'li Hastalarda Gen Mutasyonlarının Araştırılması*

Research of Gene Mutations in Patient Suffering from Neurofibromatozis Type 1 in Turkey

Sacide PEHLİVAN[1,2], Hüseyin ONAY[2], Gülçin ITIRLI[2], Ayşe ERBAY[3], Ahmet KOMAN[4], Duri Şehvar ÖZEL ÜNAL[4], Ferda ÖZKINAY[2]

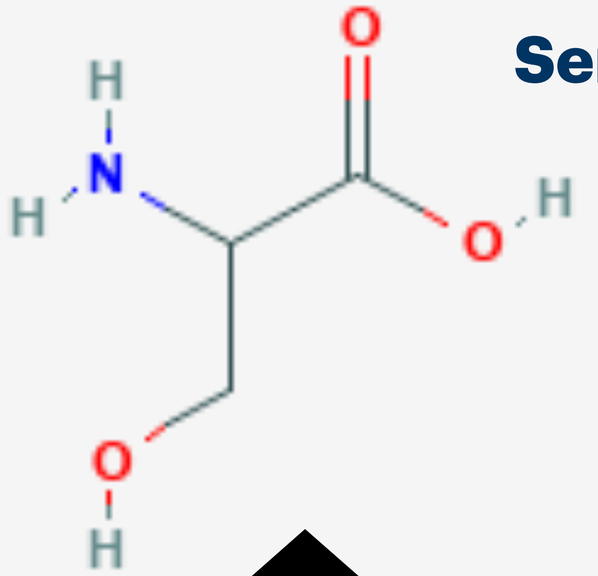
[1] Gaziantep Üniversitesi Tıp Fakültesi, Tıbbi Biyoloji Anabilim Dalı, GAZİANTEP
2 Ege Üniversitesi Tıp Fakültesi, Tıbbi Genetik Anabilim Dalı,
3 Dr. Behçet Uz Çocuk Hastanesi, Onkoloji Birimi, İZMİR
4 Boğaziçi Üniversitesi Fen Fakültesi, Moleküler Biyoloji ve Genetik Anabilim Dalı, İSTANBUL

* Bu çalışma; XII. International Biomedical Science and Technology Symposium, 20-23 September 2005, Izmir-Turkey toplantısında sözlü olarak sunulmuştur.

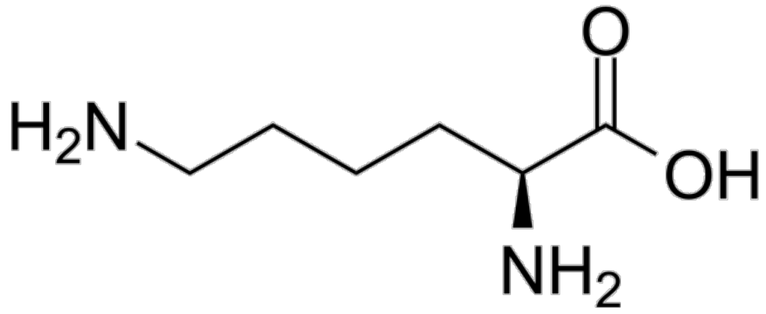

NO MUTATION
POINT MUTATIONS
SILENT
NONSENSE
MISSENSE
Conservative
Non-conservative
Normal
DNA level
mRNA level
Protein level
TTC
AAG
Lys
TTT
AAA
Lys
ATC
UAG
STOP
TCC
AGG
Arg
TGC
ACG
Thr

Tür	Amino Asit Özellikleri	Örnek Değişim	Etkisi	Protein Yapısı ve İşlevine Etkisi	Klinik Örnek
Conservative	- Yeni amino asit, orijinaline benzer özellikler taşır (yük, polarite, hidrofobiklik).	Serin → Treonin	- Protein fonksiyonu büyük oranda korunur.	- Protein yapısı korunur. - Genelde stabil kalır ve katlanma bozulmaz.	**- Genelde klinik sonuçlar yaratmaz.**
Non-Conservative	- Yeni amino asit, orijinaline farklı özellikler taşır (yük, polarite, hidrofobiklik).	Lizin → Prolin	- Protein fonksiyonunu bozabilir.	- Protein yapısı ve katlanması değişir. - Biyolojik aktivite kaybı olabilir.	**- Klinik sonuçlar bekleniyor.**

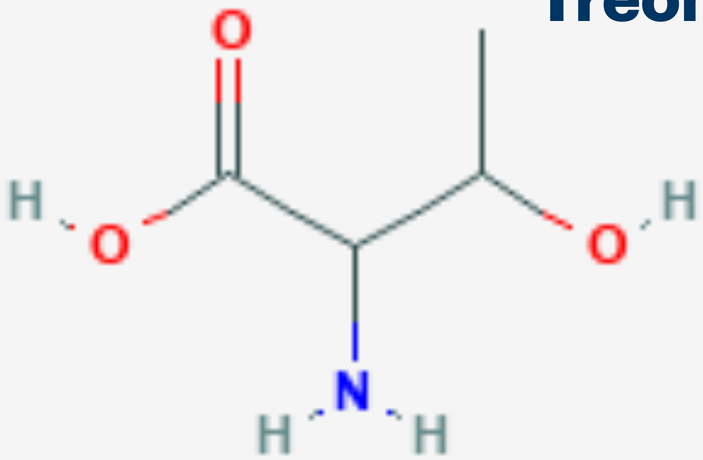
Serin



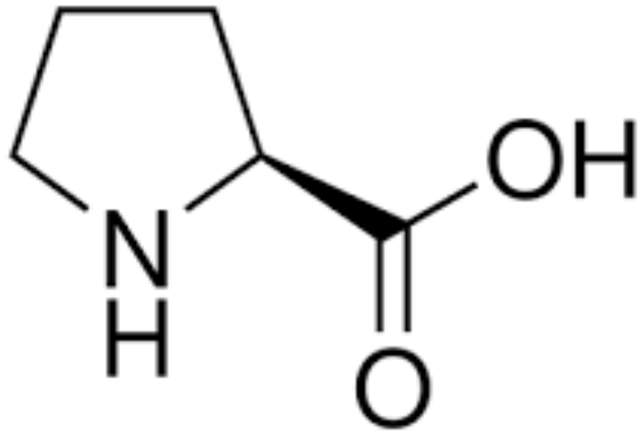
Lizin



Treonin



Prolin



benign
conflicting interpretations of pathogenicity
likely benign
likely pathogenic
pathogenic
pathogenic likely pathogenic
risk factor

Validation Status
by-ALFA
by-cluster
by-frequency

Publication
PubMed Cited
PubMed Linked

Function Class clear
inframe deletion
inframe indel
inframe insertion
initiator codon variant
intron
✓ missense
non coding transcript variant
synonymous

Variation Class
mnv

Search results

Items: 1 to 20 of 7168

Filters activated: missense. Clear all to show 112

The following term was not found in SNP: nonse

rs1129506 *[Homo sapiens]*

1.

Variant type:	SNV
Alleles:	G>A,C [Show Flanl
Chromosome:	17:31319014 (GRCh
	17:29646032 (GRCh
Canonical SPDI:	NC_000017.11:313190
Gene:	EVI2A (Varview), NF1 (
Functional Consequence:	
missense_variant,5_prime_UTR_variant,genic_	
Clinical significance:	benign
Validated:	by frequency,by alfa,
MAF:	G=0.375506/93251 (
	G=0.26087/108 (SG
	G=0.263889/57 (Qat
HGVS:	NC_000017.11:g.3131
	NC_000017.10:g.2964
	NG_009018.1:g.22903

rs2230852 *[Homo sapiens]*

18.

Variant type:	SNV
Alleles:	C>A,G,T [Show Flanks]
Chromosome:	17:31229168 (GRCh38)
	17:29556186 (GRCh37)
Canonical SPDI:	NC_000017.11:31229167:C:A,N
Gene:	NF1 (Varview)
Functional Consequence:	
missense_variant,stop_gained,genic_downstream_transc	
Clinical significance:	pathogenic,benign-likely-benig
Validated:	by frequency,by alfa,by cluste
MAF:	T=0.000653/30 (ALFA)
	T=0.000463/116 (GnomAD_e
	T=0.000522/63 (ExAC)
HGVS:	NC_000017.11:g.31229168C>A,
	NC_000017.11:g.31229168C>T,
	NC_000017.10:g.29556186C>G,

Download

Released S

Clinical Significanc Reported in ClinVar

Gene : Consequence : Missense Variant
NF1 : Intron Variant

Publications 1 citation

LitVar 2 39

Genomic View See rs on genome

Submissions | History | Publications

e Variant Details tab for details on Genomic Placement, Gene, and

Download

Released Se

Clinical Significance orted in ClinVar

Gene : Consequence NF1 : Stop Gained

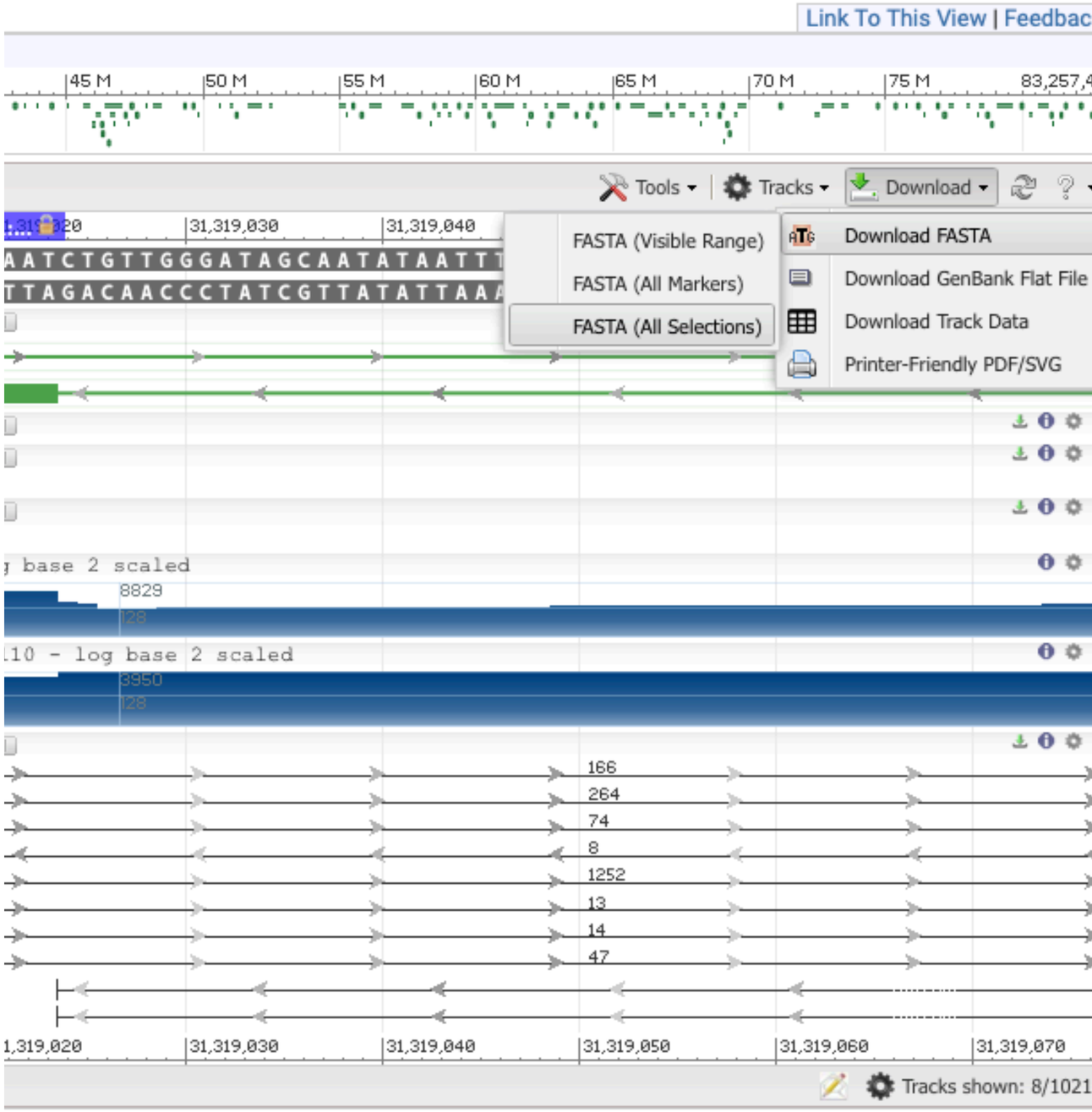
Publications 2 citations

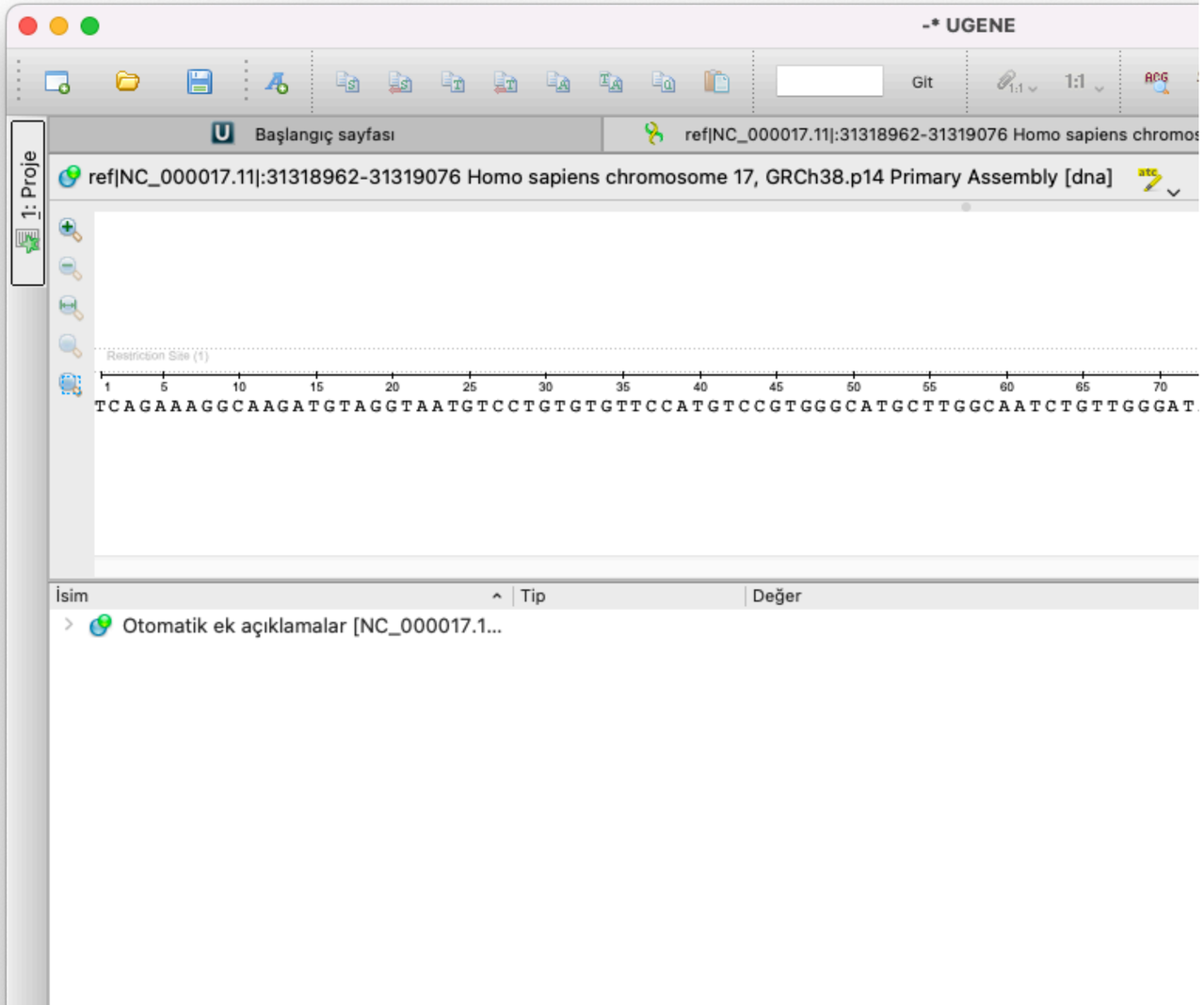
LitVar 2 3

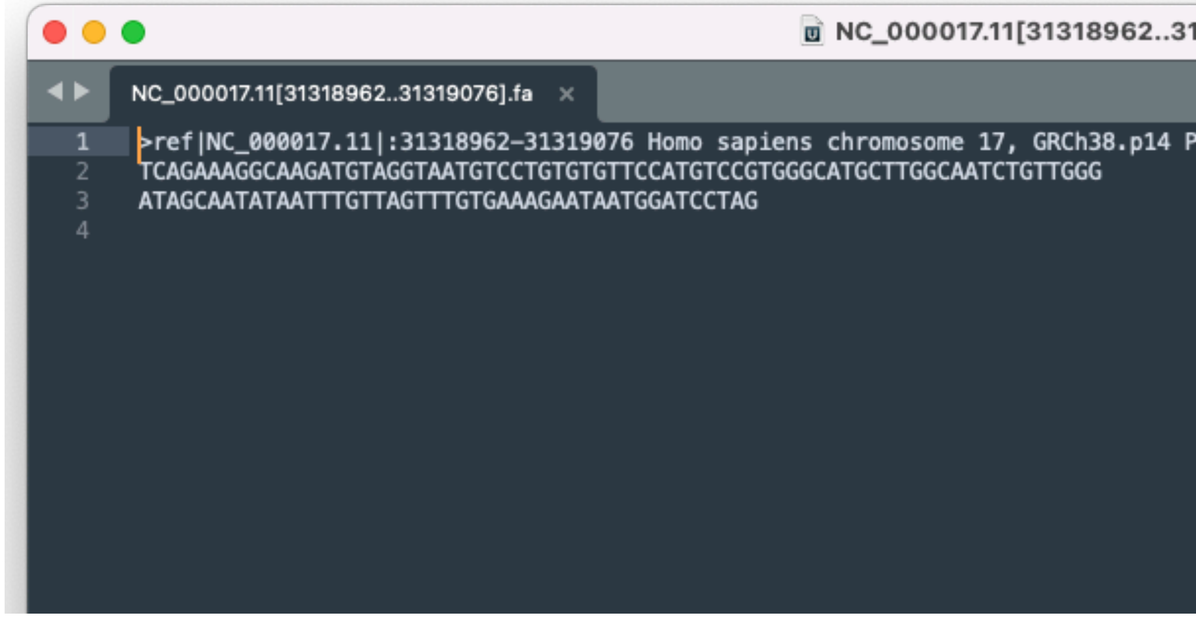
Genomic View See rs on genome

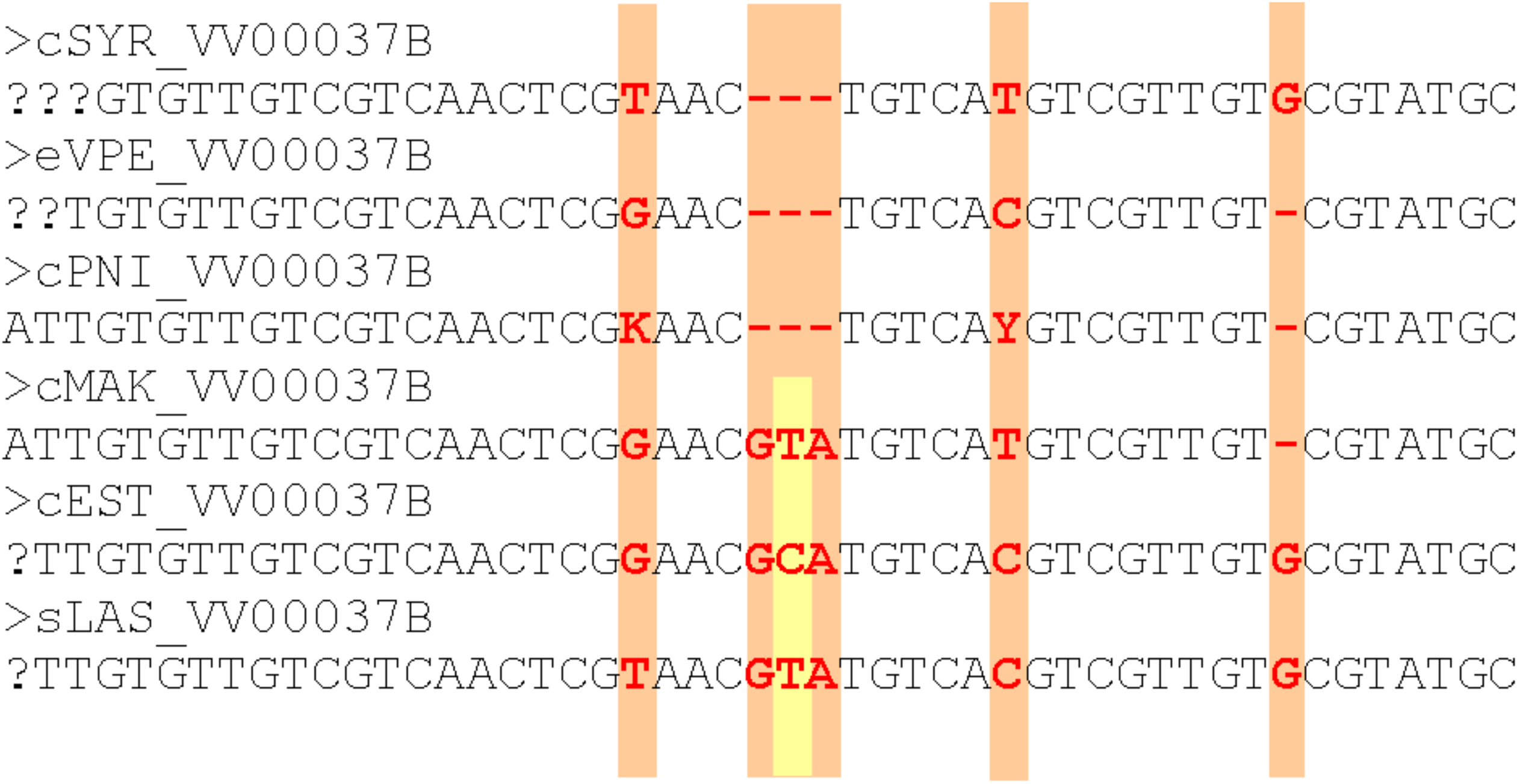
FASTA

- Metin belgesi
- A(U),T,G,C'den oluşur
- DNA/RNA veya Protein




-* UGENE
Git
1:1
Başlangıç sayfası
ref|NC_000017.11|:31318962-31319076 Homo sapiens chromos
ref|NC_000017.11|:31318962-31319076 Homo sapiens chromosome 17, GRCh38.p14 Primary Assembly [dna]
1: Proje
Restriction Site (1)
1 5 10 15 20 25 30 35 40 45 50 55 60 65 70
TCAGAAAGGCAAGATGTAGGTAATGTCCTGTGTGTTCCATGTCCGTGGGCATGCTTGGCAATCTGTTGGGAT
İsim
Tip
Değer
Otomatik ek açıklamalar [NC_000017.1...


NC_000017.11[31318962..31
NC_000017.11[31318962..31319076].fa
1 >ref|NC_000017.11|:31318962-31319076 Homo sapiens chromosome 17, GRCh38.p14 P
2 TCAGAAAGGCAAGATGTAGGTAATGTCCTGTGTGTTCCATGTCCGTGGGCATGCTTGGCAATCTGTTGGG
3 ATAGCAATATAATTTGTTAGTTTGTGAAAGAATAATGGATCCTAG
4


>cSYR_VV00037B
???GTGTTGTCGTCAACTCGTAAC---TGTCATGTCGTTGTGCGTATGC
>eVPE_VV00037B
??TGTGTTGTCGTCAACTCGGAAC---TGTCACGTCGTTGT-CGTATGC
>cPNI_VV00037B
ATTGTGTTGTCGTCAACTCGKAAC---TGTCAYGTCGTTGT-CGTATGC
>cMAK_VV00037B
ATTGTGTTGTCGTCAACTCGGAACGTATGTCATGTCGTTGT-CGTATGC
>cEST_VV00037B
?TTGTGTTGTCGTCAACTCGGAACGCATGTCACGTCGTTGTGCGTATGC
>sLAS_VV00037B
?TTGTGTTGTCGTCAACTCGTAACGTATGTCACGTCGTTGTGCGTATGC

snp.py — UNREGISTERED

```python
from Bio import Entrez
import os
import time

Entrez.email = "dogukansahil@gmail.co
Entrez.api_key = "a6190b141aea42289dfc308"

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

if not os.path.exists(snp_folder):
    os.makedirs(snp_folder)

def fetch_snp_fasta_by_variant(variant_id):
    try:
        fasta_handle = Entrez.efetch(db="snp", id=variant_id, rettype="fasta", retmode="text")
        fasta_data = fasta_handle.read()
        fasta_handle.close()

        file_path = os.path.join(snp_folder, f"{variant_id}.fasta")

        with open(file_path, "w") as file:
            file.write(fasta_data)
            print(f"{variant_id}.fasta dosyası 'snp' klasörüne indirildi.")

    except Exception as e:
        print(f"Hata oluştu: {e}")

fetch_snp_fasta_by_variant("rs1042522")

def fetch_snp_fasta_by_clinical_significance(gene_name, clinical_significance):
    term = f"{gene_name}[Gene Name] AND {clinical_significance}[Clinical Significance] AND human[Organism]"
    search_handle = Entrez.esearch(db="snp", term=term, retmax=1000)
    search_results = Entrez.read(search_handle)
    search_handle.close()
    snp_ids = search_results.get("IdList", [])

    print(f"Toplam {len(snp_ids)} SNP bulundu. İndiriliyor...")

    for snp_id in snp_ids:
        try:
            fasta_handle = Entrez.efetch(db="snp", id=snp_id, rettype="fasta", retmode="text")
            fasta_data = fasta_handle.read()
            fasta_handle.close()

            file_path = os.path.join(snp_folder, f"{snp_id}.fasta")

            with open(file_path, "w") as file:
                file.write(fasta_data)
                print(f"{snp_id}.fasta dosyası 'snp' klasörüne indirildi.")

            time.sleep(0.3)
        except Exception as e:
            print(f"{snp_id} indirilemedi. Hata: {e}")

    print("İndirme işlemi tamamlandı.")

fetch_snp_fasta_by_clinical_significance("TP53", "pathogenic")
```

Line 10, Column 1 — Spaces: 4 — Python

sahil — python3 ~/Desktop/snp.py — 90×26

```
Last login: Tue Dec  3 10:53:01 on ttys000
^[[A
(base) sahil@sahil-3 ~ % python3 /Users/sahil/Desktop/snp.py

rs1042522.fasta dosyası 'snp' klasörüne indirildi.
Toplam 500 SNP bulundu. İndiriliyor...
2151047224.fasta dosyası 'snp' klasörüne indirildi.
2151045484.fasta dosyası 'snp' klasörüne indirildi.
2151043421.fasta dosyası 'snp' klasörüne indirildi.
2151043336.fasta dosyası 'snp' klasörüne indirildi.
2151043202.fasta dosyası 'snp' klasörüne indirildi.
2151042795.fasta dosyası 'snp' klasörüne indirildi.
2151042631.fasta dosyası 'snp' klasörüne indirildi.
2151042609.fasta dosyası 'snp' klasörüne indirildi.
2151042416.fasta dosyası 'snp' klasörüne indirildi.
2151041861.fasta dosyası 'snp' klasörüne indirildi.
2151041421.fasta dosyası 'snp' klasörüne indirildi.
2151041118 indirilemedi. Hata: HTTP Error 400: Bad Request
```

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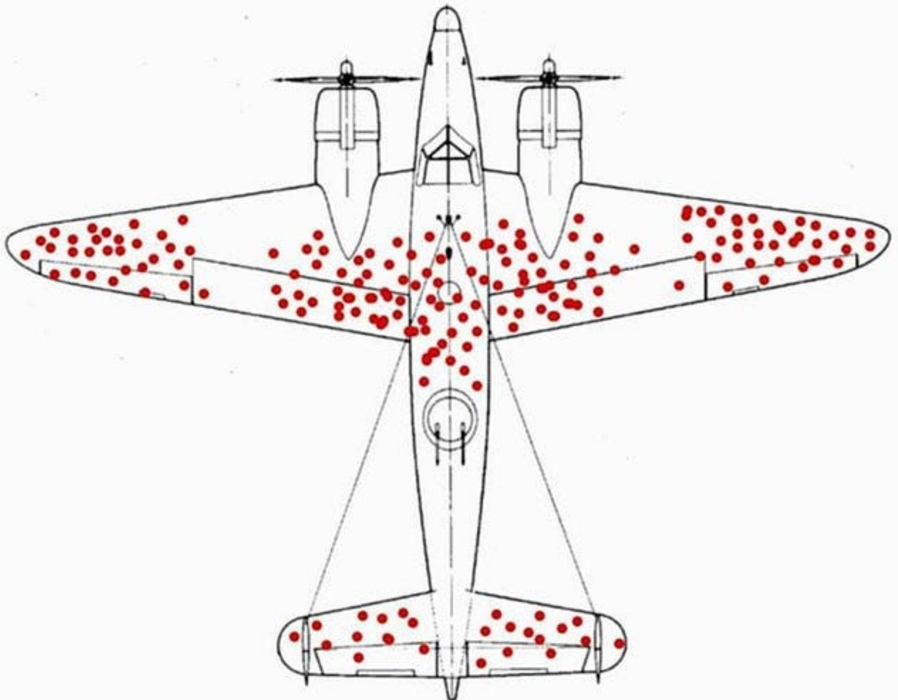
Abraham **Wald**

- **Sağkalım yanlılığı**



Kaynaklar

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