

**Dr.Yavuz ŞAHİN**

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## **TEMEL GENETİK BİLGİLER**



**Necip Fazıl Şehir Hastanesi  
Kahramanmaraş**

# GENETİK HASTALIKLAR

## Nadir Mi?

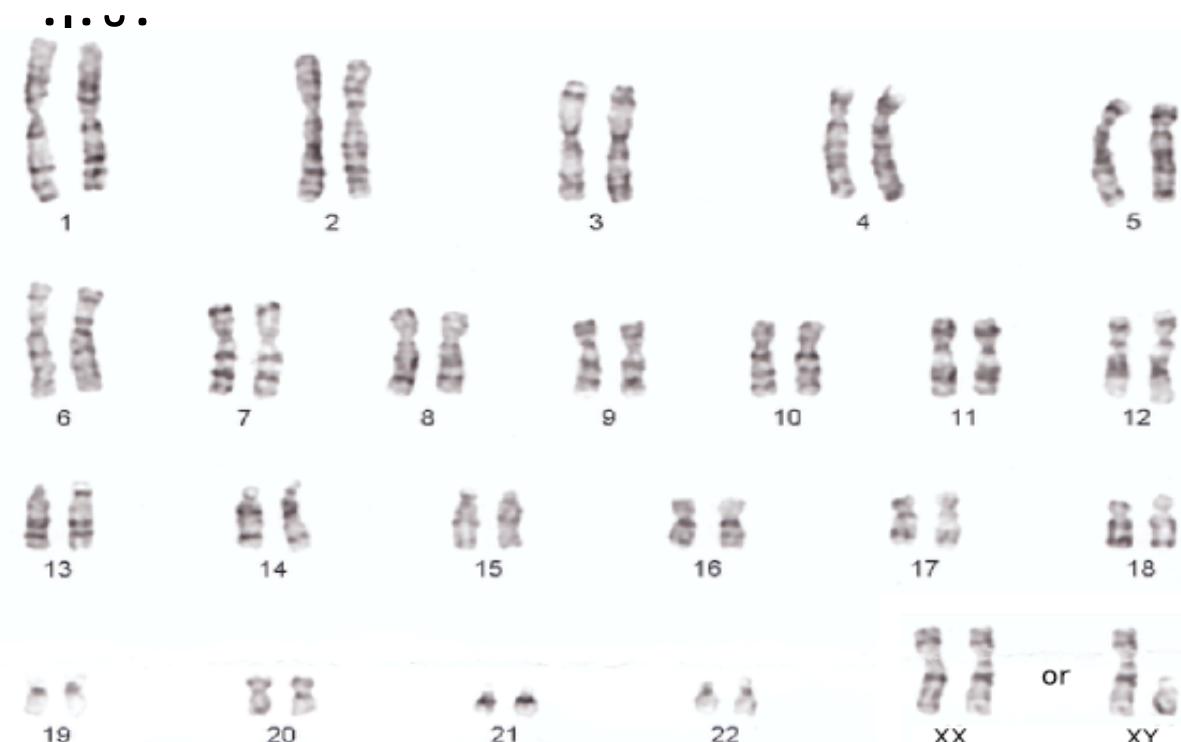


- Dünyada 350 milyon birey
  - %80 Gen
  - %50 Çocukluk Çağı
- Ölümlerin
  - İlk 1 yıl %35'i
  - %30 hasta ilk 5 yıl kaybedilir.
- Hastaların ancak %50'sine tanı konulmaktadır.



# Periferik Kandan Kromozom Analizi

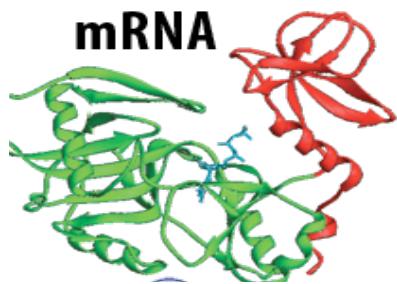
- Zeka Gerilemesi
- Gelişimsel Gerileme
- Atipik Yemek Tüketimi
- Otizm
- Direnç
- Tekrarlı İstekler
- IVF Başarısızlığı
- Tanı Kısıtlığı



DNA



Gen



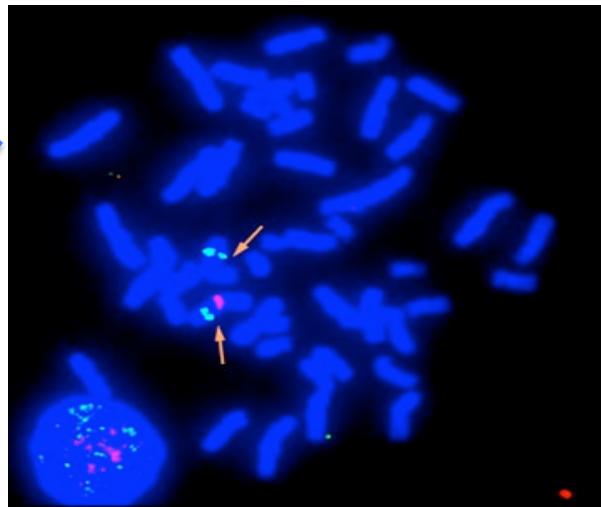
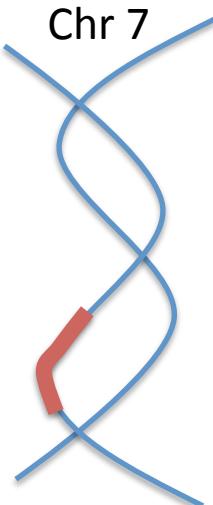
# Mikrodelesyon Sendromları

# Mikrodelesyon Sendromu

- Supravalvuler aort stenozu
- Hafif – orta MR
- Kalın ses
- Periorbital dolgunluk



# FISH Williams Syndrome



# Mikrodeleseyon Sendromu

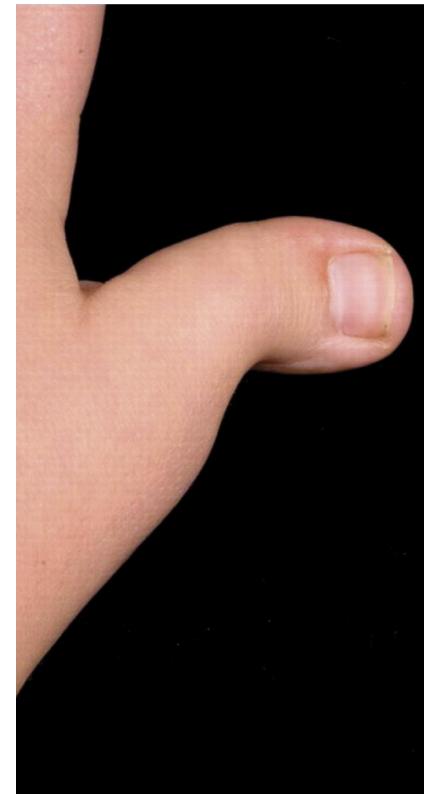


Angelman



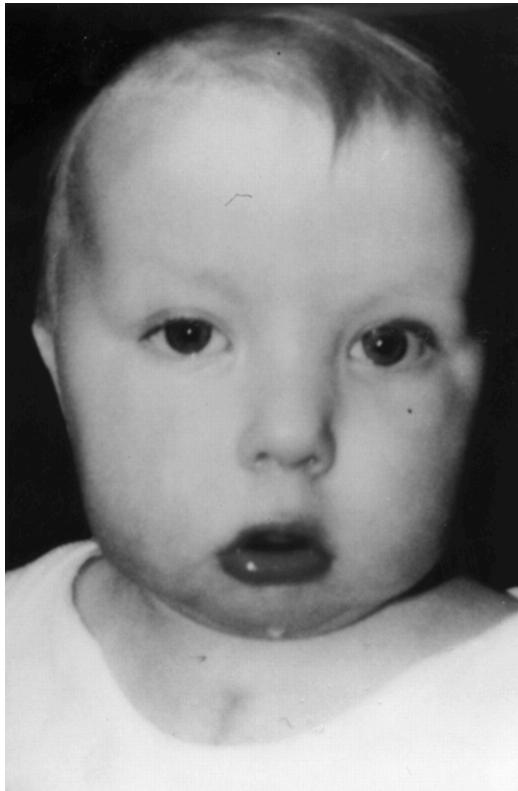
Prader Willi

# Mikrodelesyon Sendromu



Rubinstein Taybi

# Mikrodelesyon Sendromu

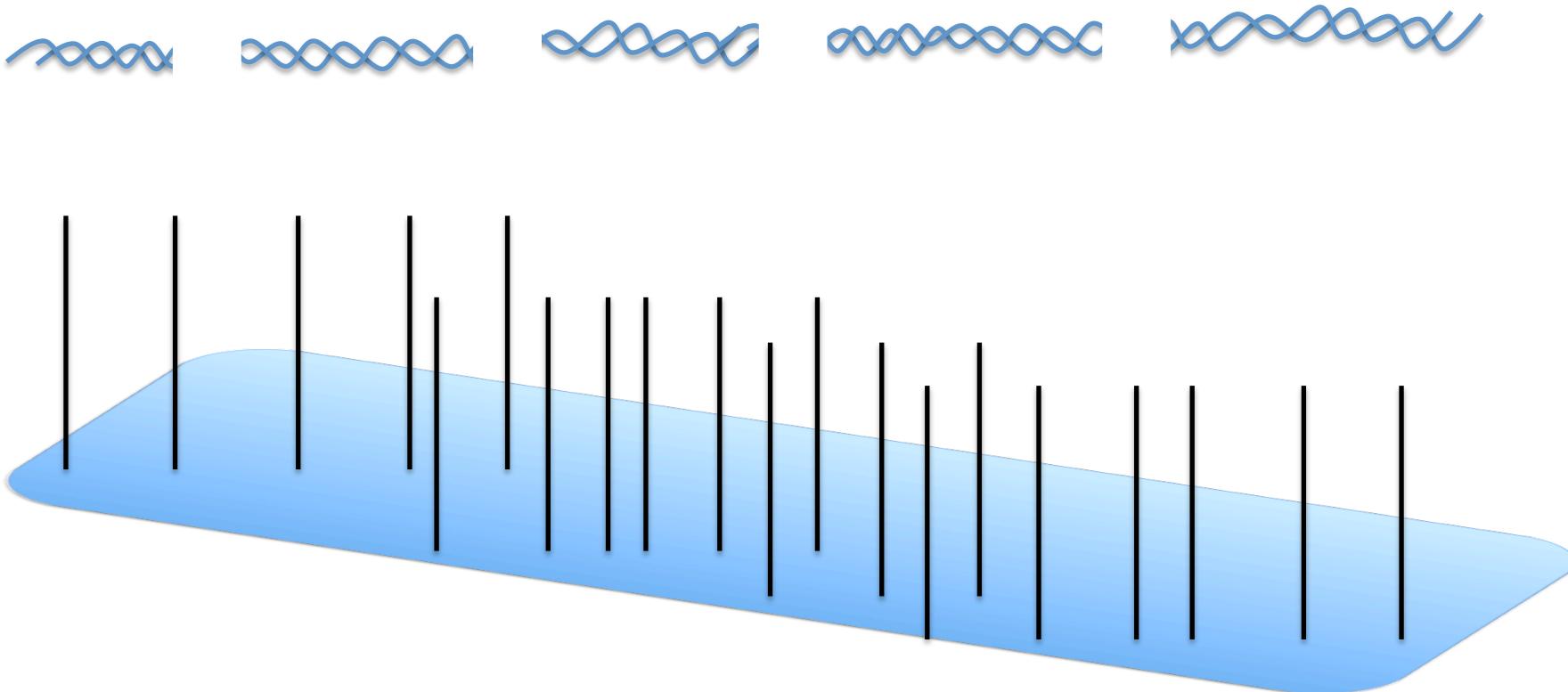


Di George

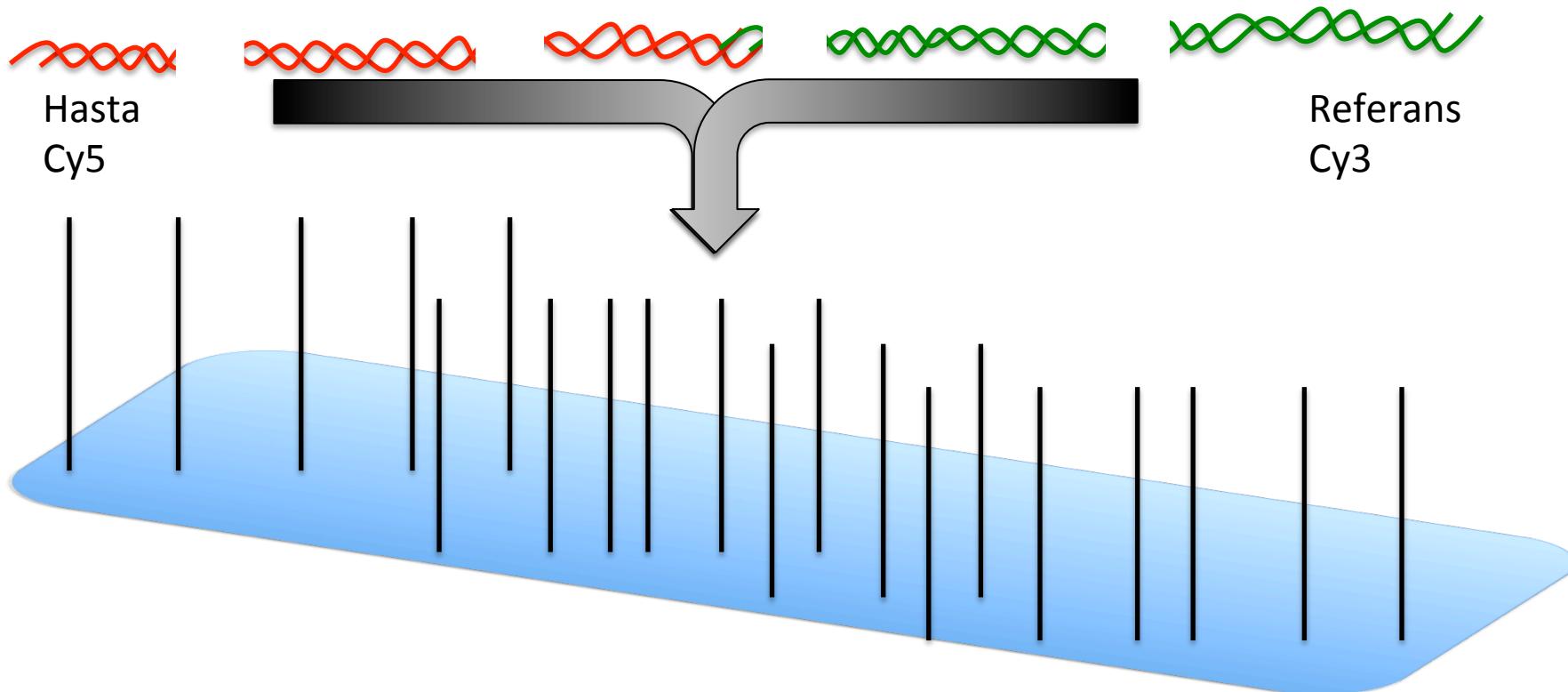


Sotos

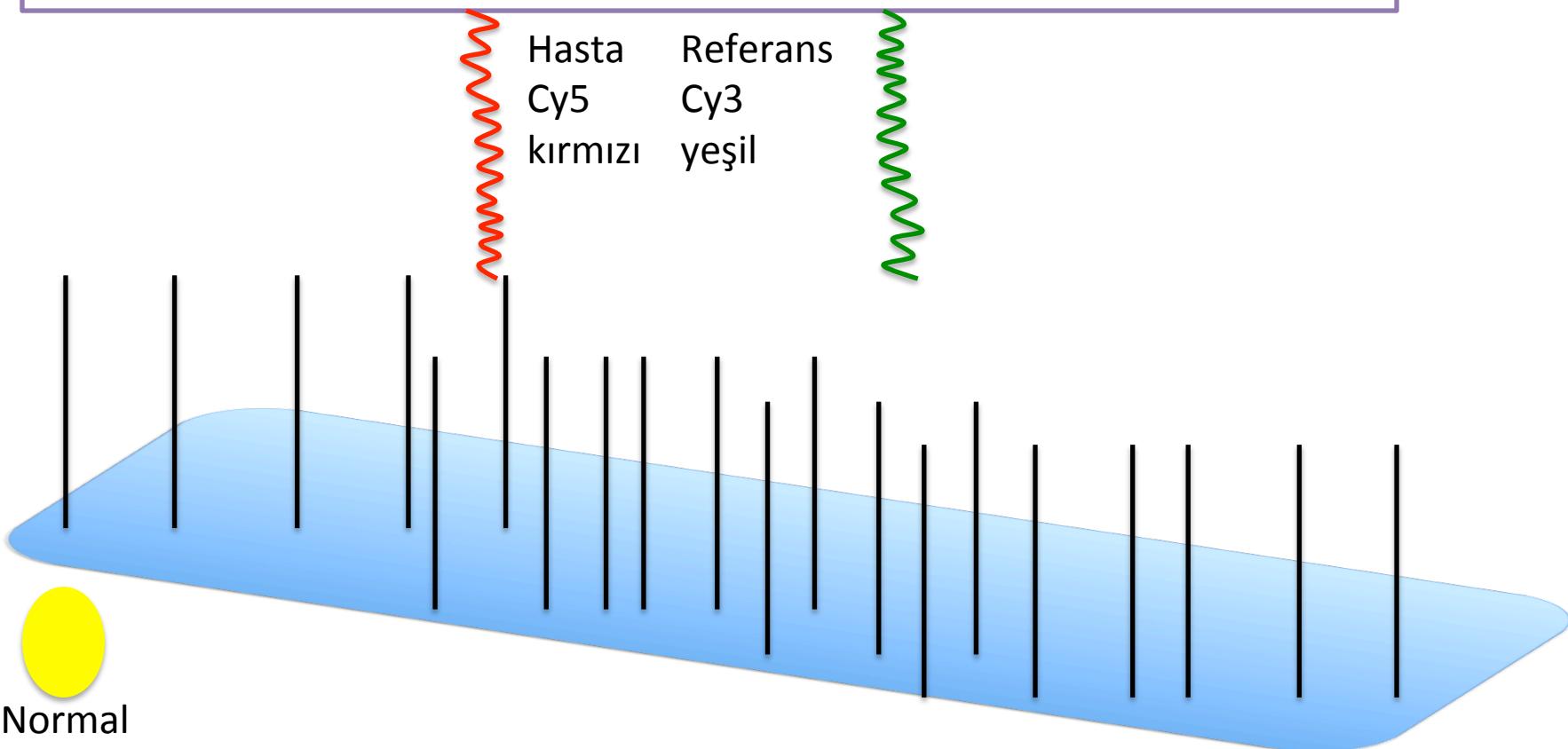
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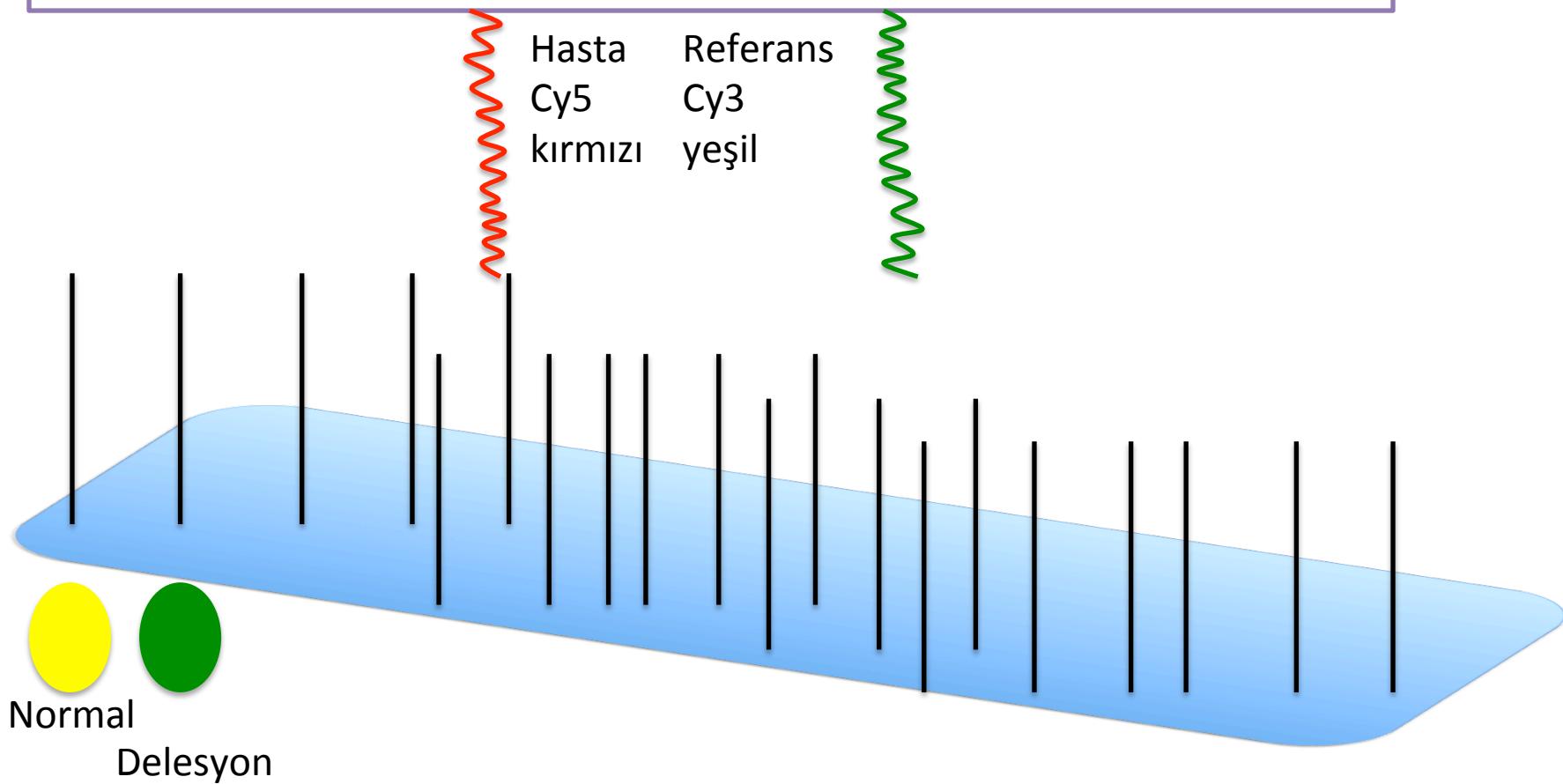
# Array CGH



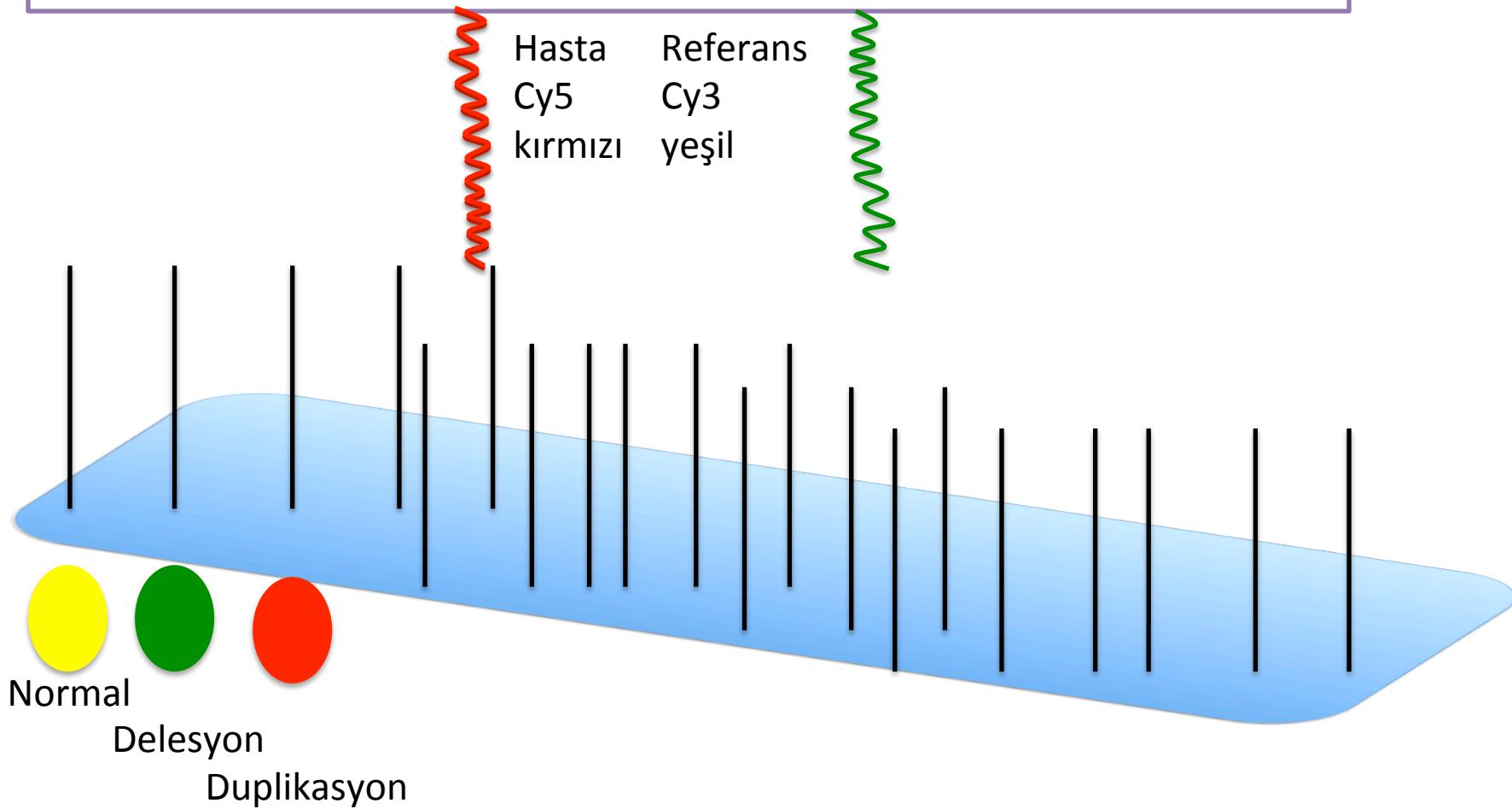
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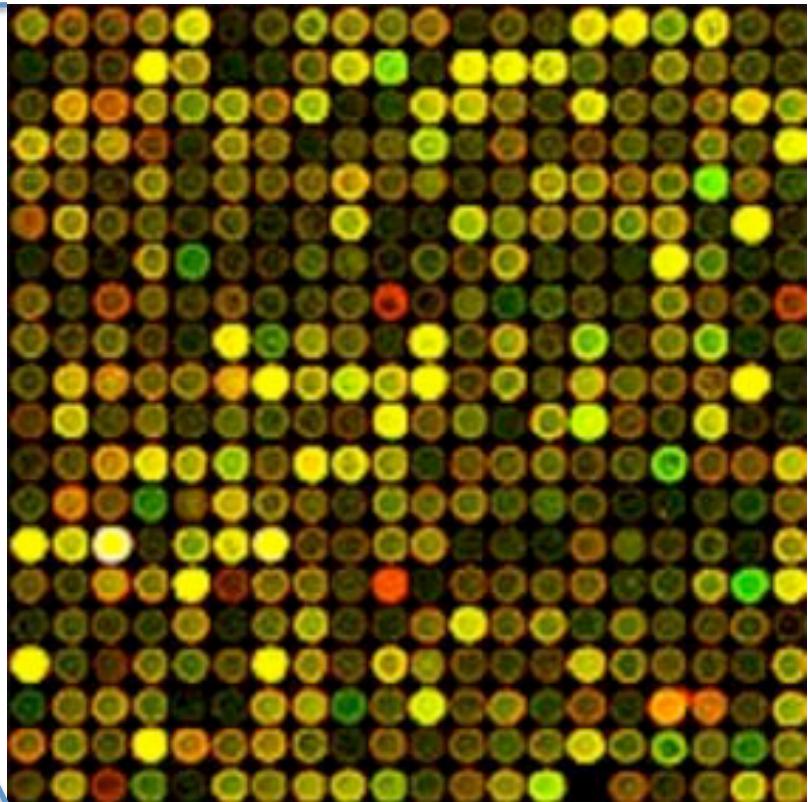
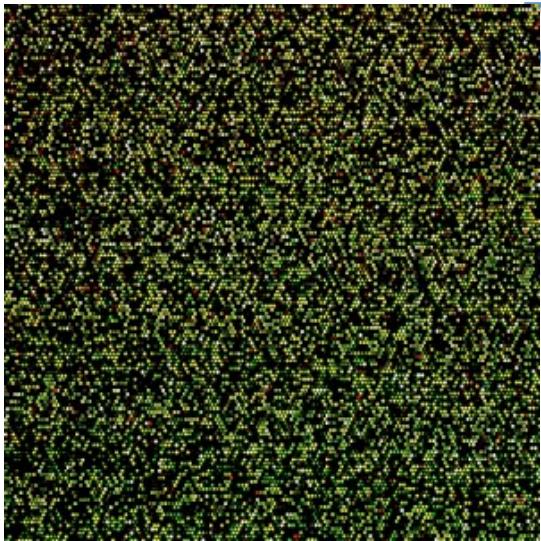
# Array CGH



# Array CGH



# Array CGH

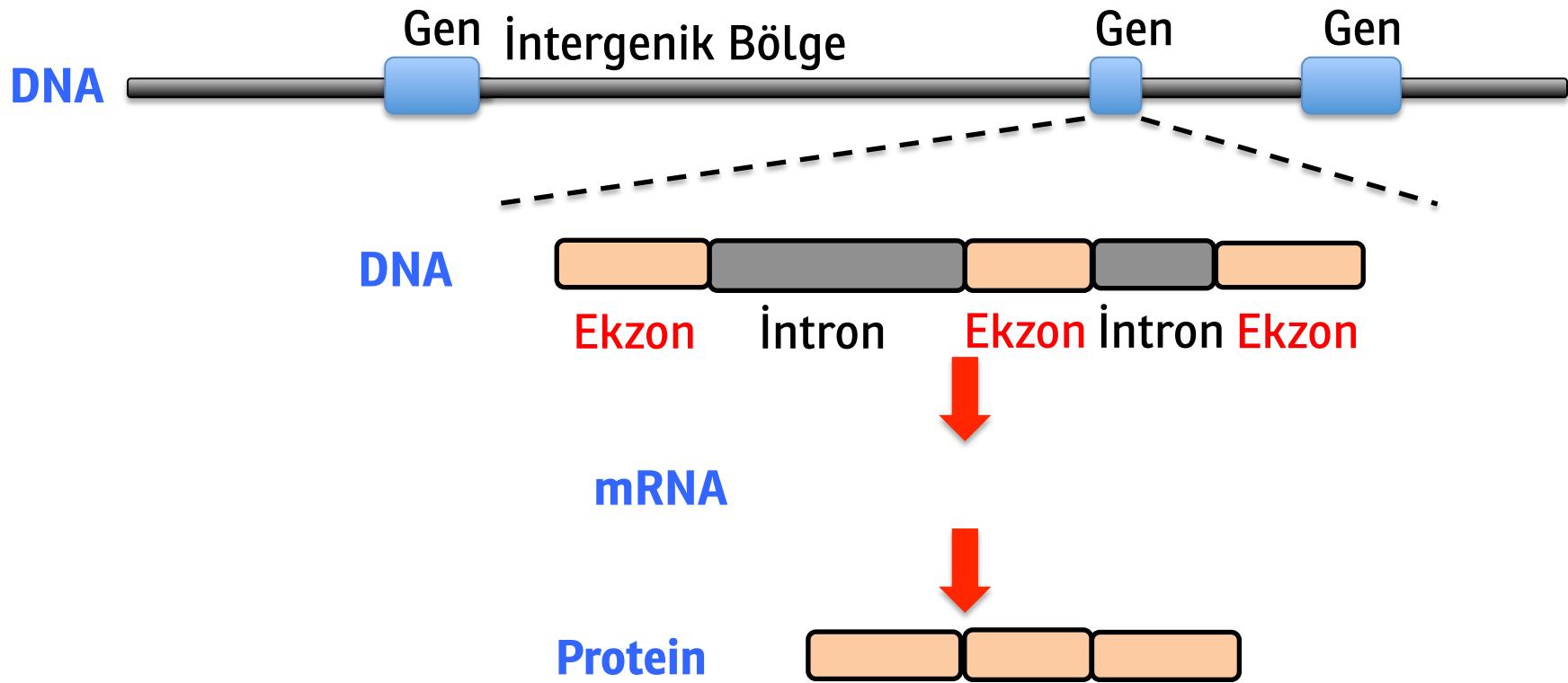


# Array

- Zeka Geriliği
- Gelişme Geriliği
- Atipik Yüz Görünümü
- Dirençli Epilepsi
- Tanı Konulamayan Vakalar

OD

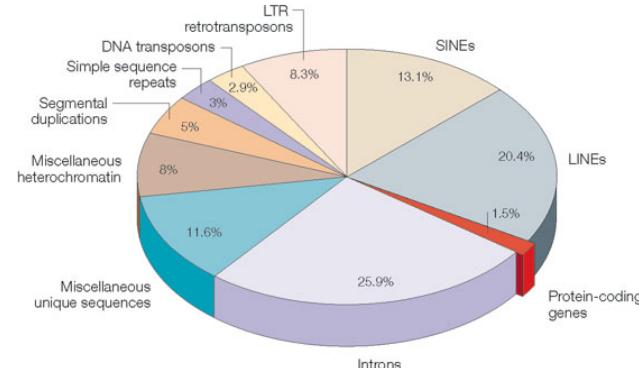


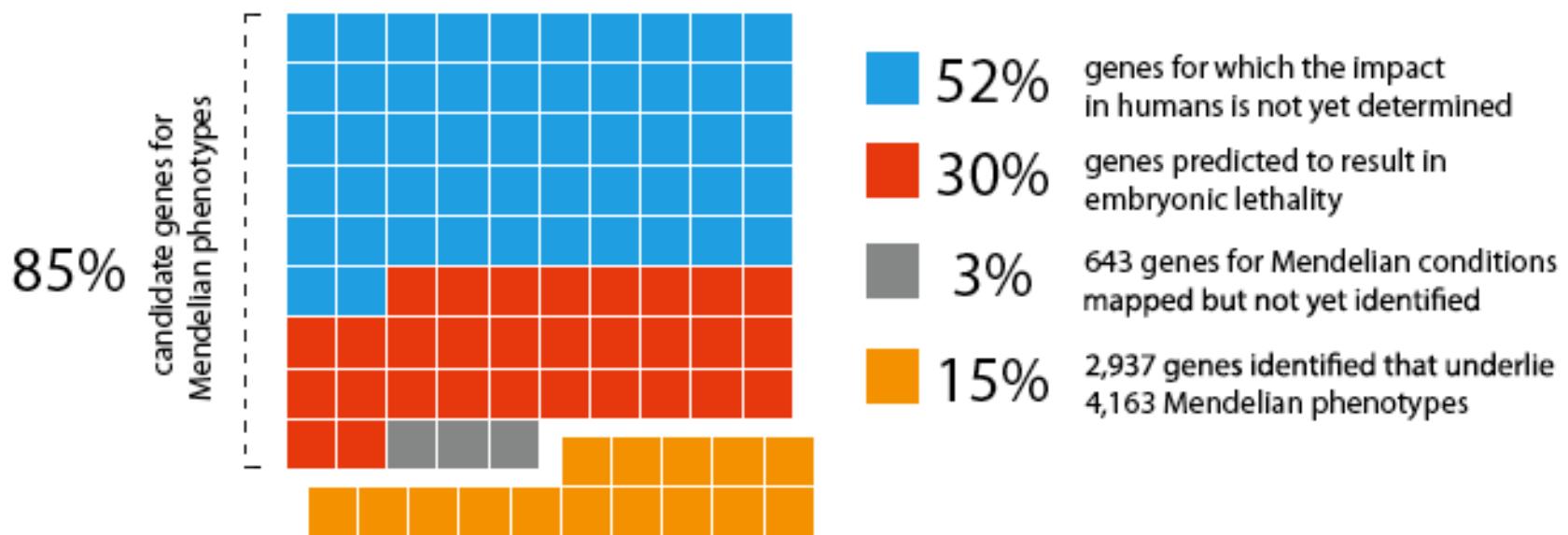


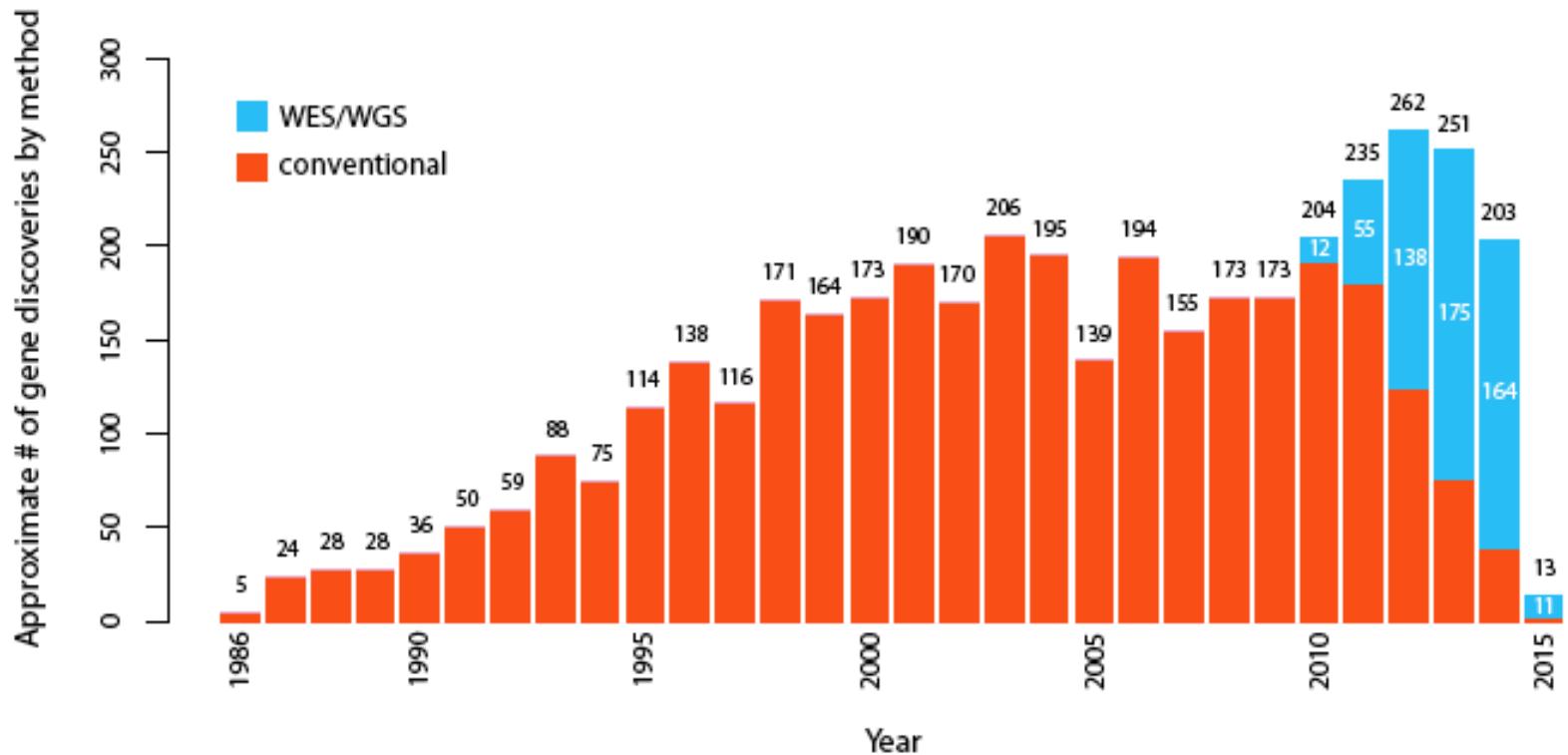
# 20. 000

20.000 gen  
 200.000 ekzon  
 %1.5 coding  
 %98.5 non coding

% 26 intron  
 % 8 -20 regulator

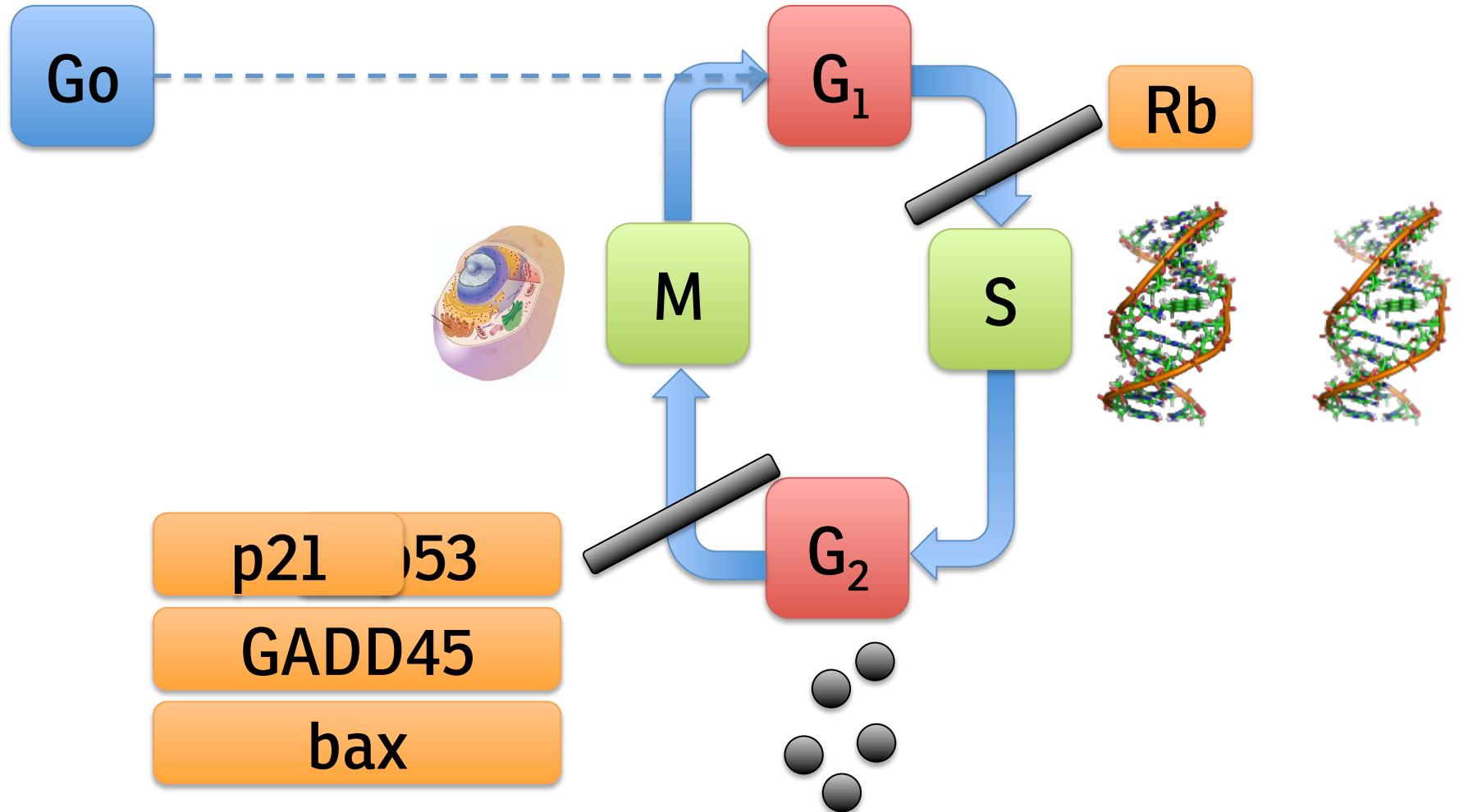


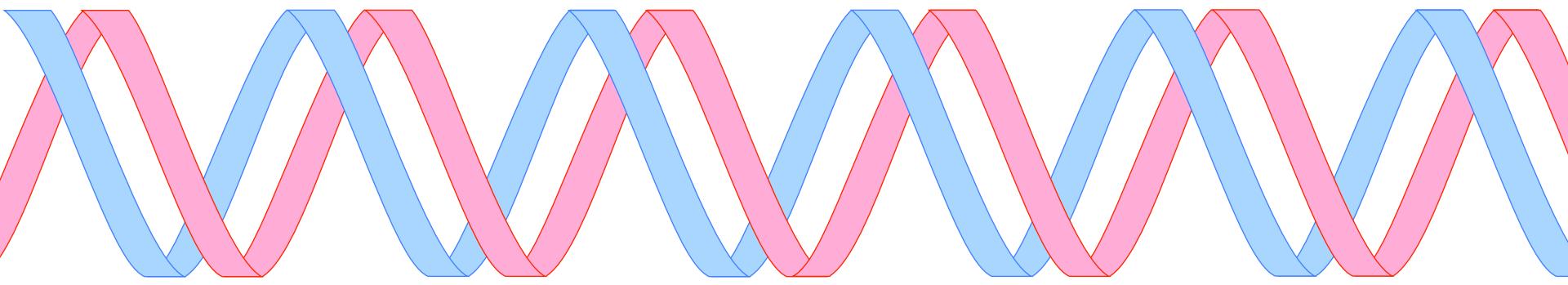


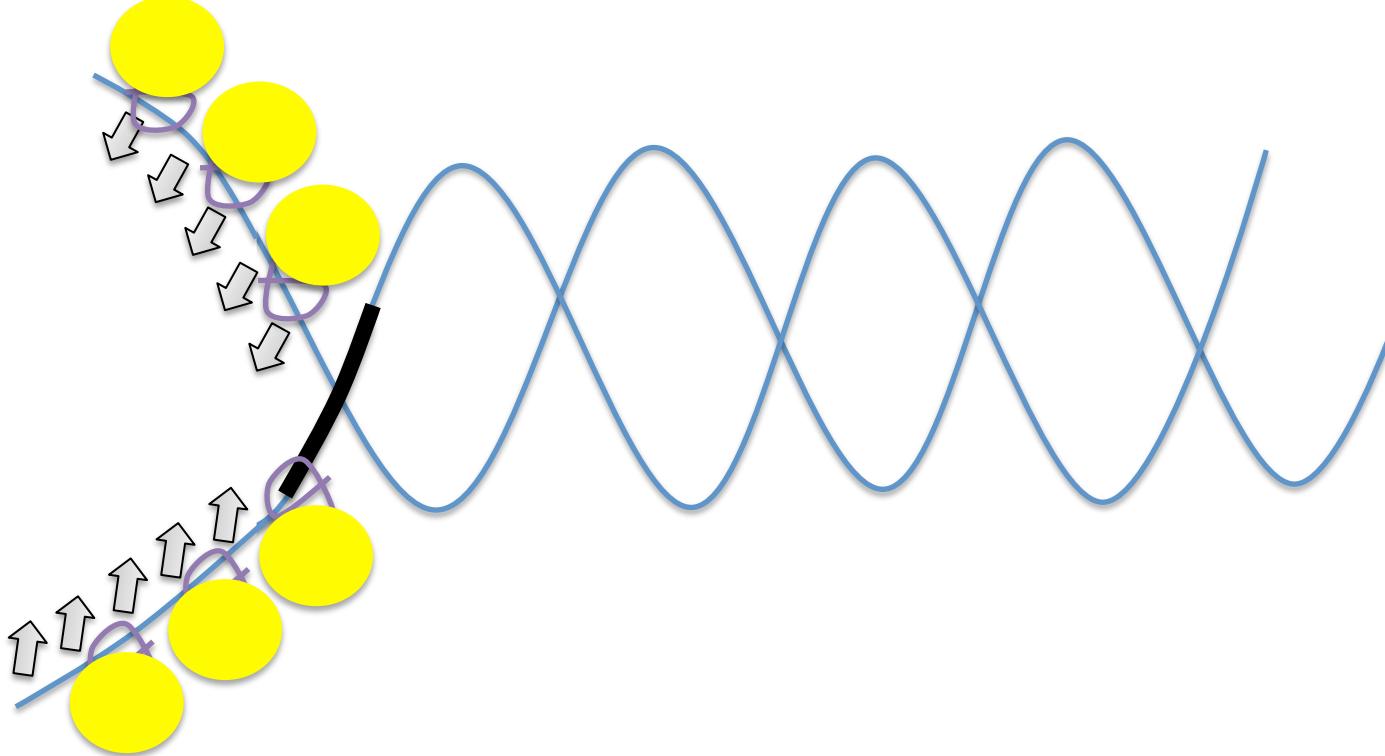


# TEMEL GENETİK MEKANİZMALAR

# **REPLİKASYON**







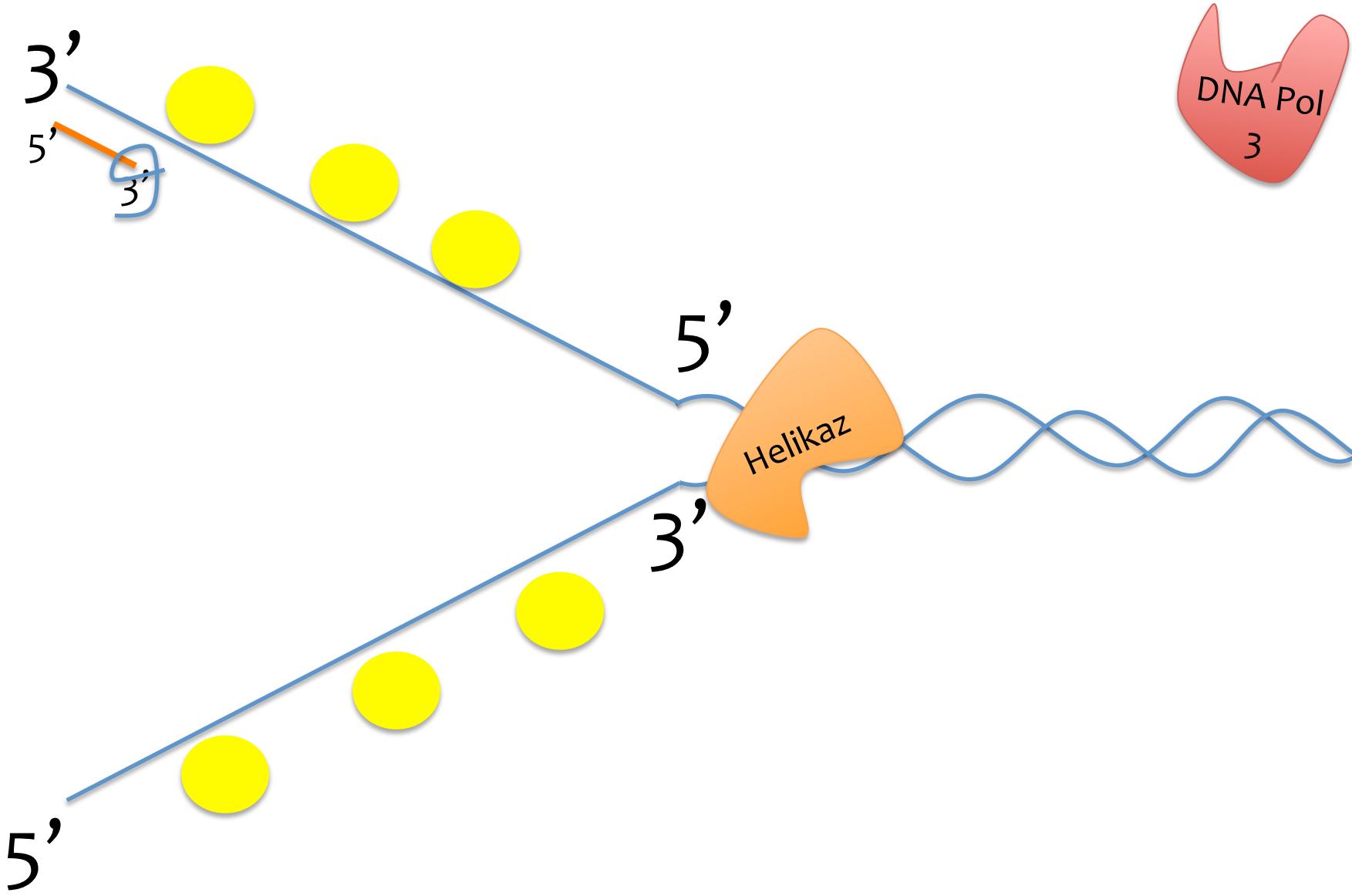
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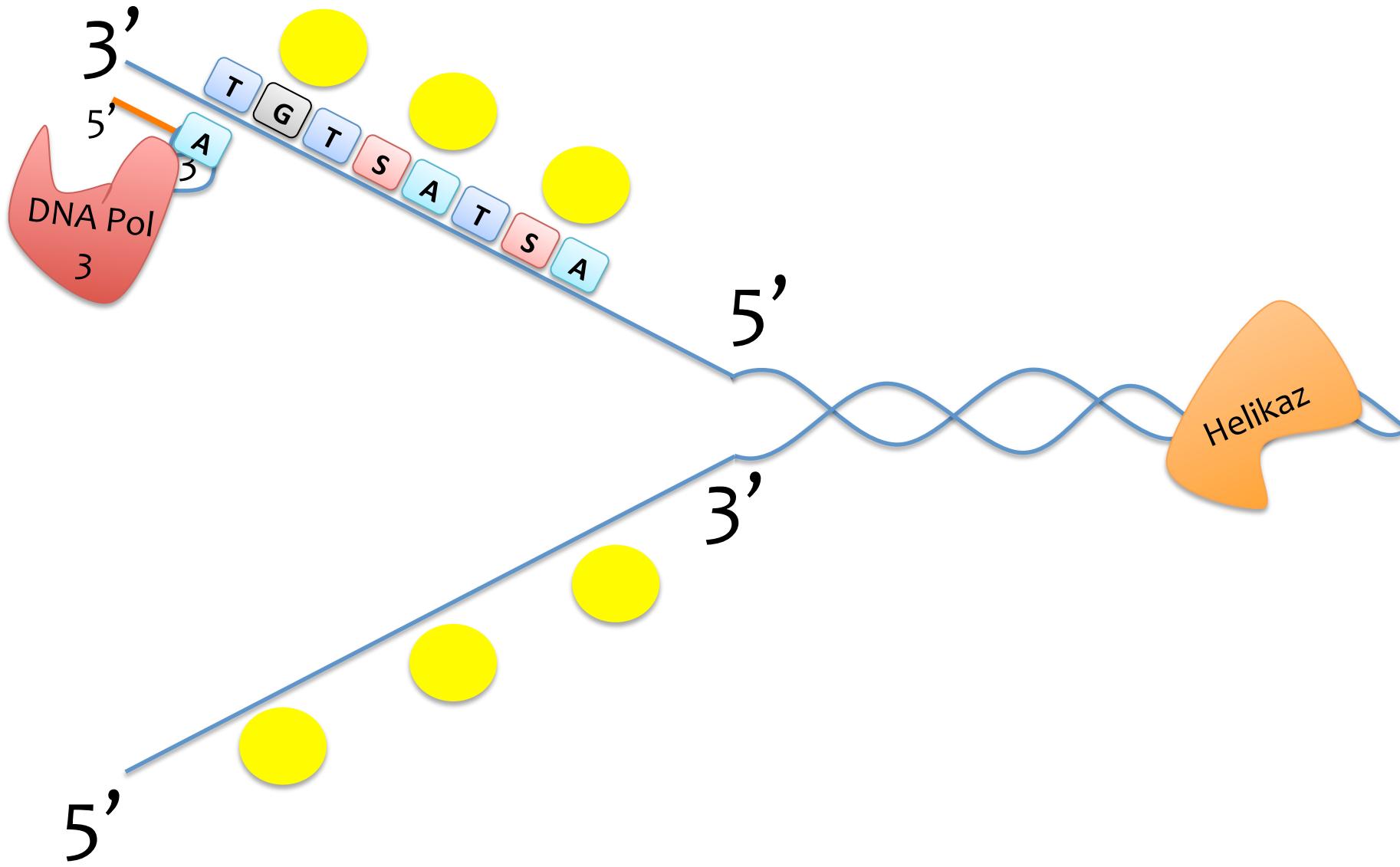
DnaA

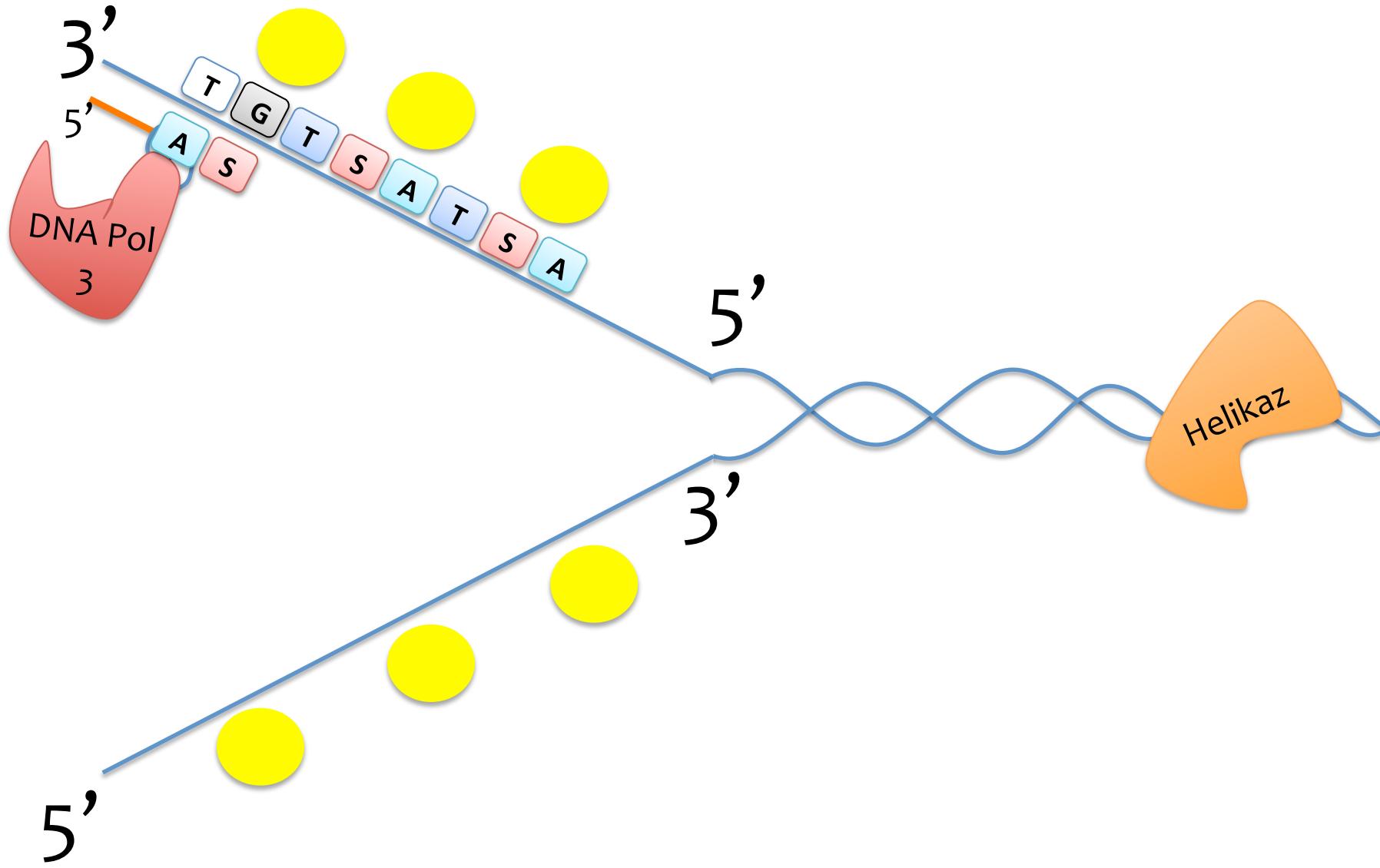
Helikaz

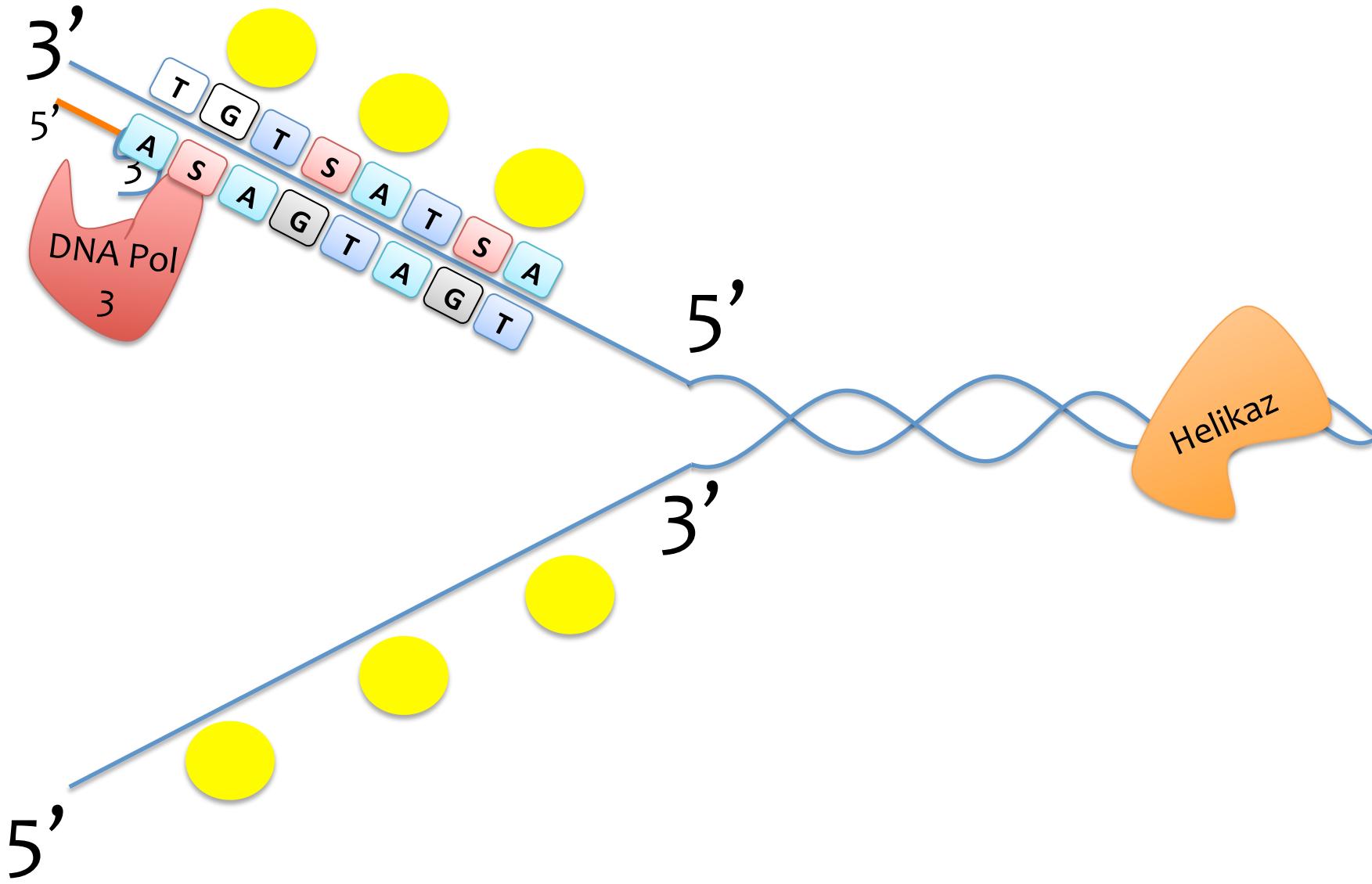
SSBPs

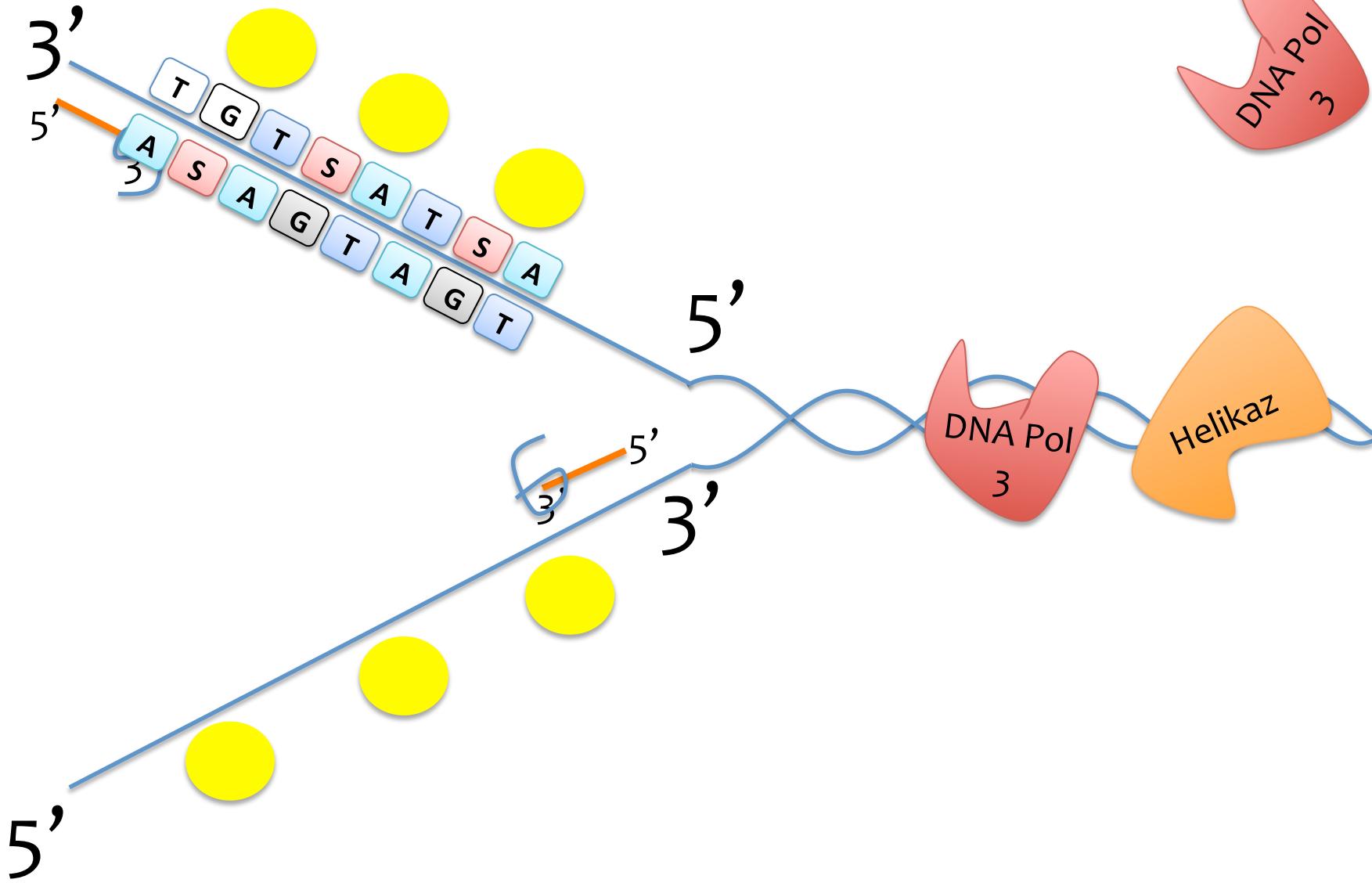
Helikaz

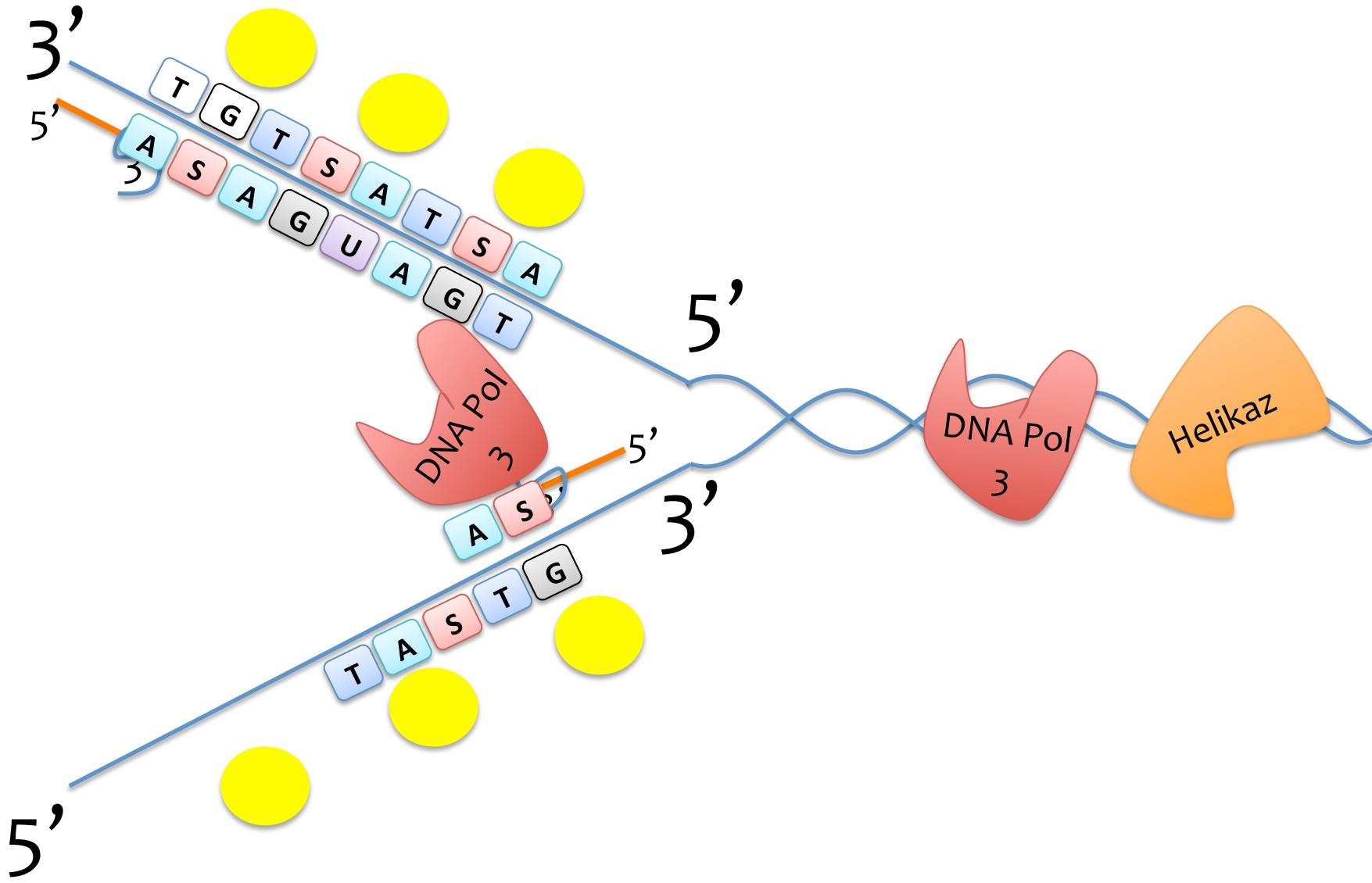


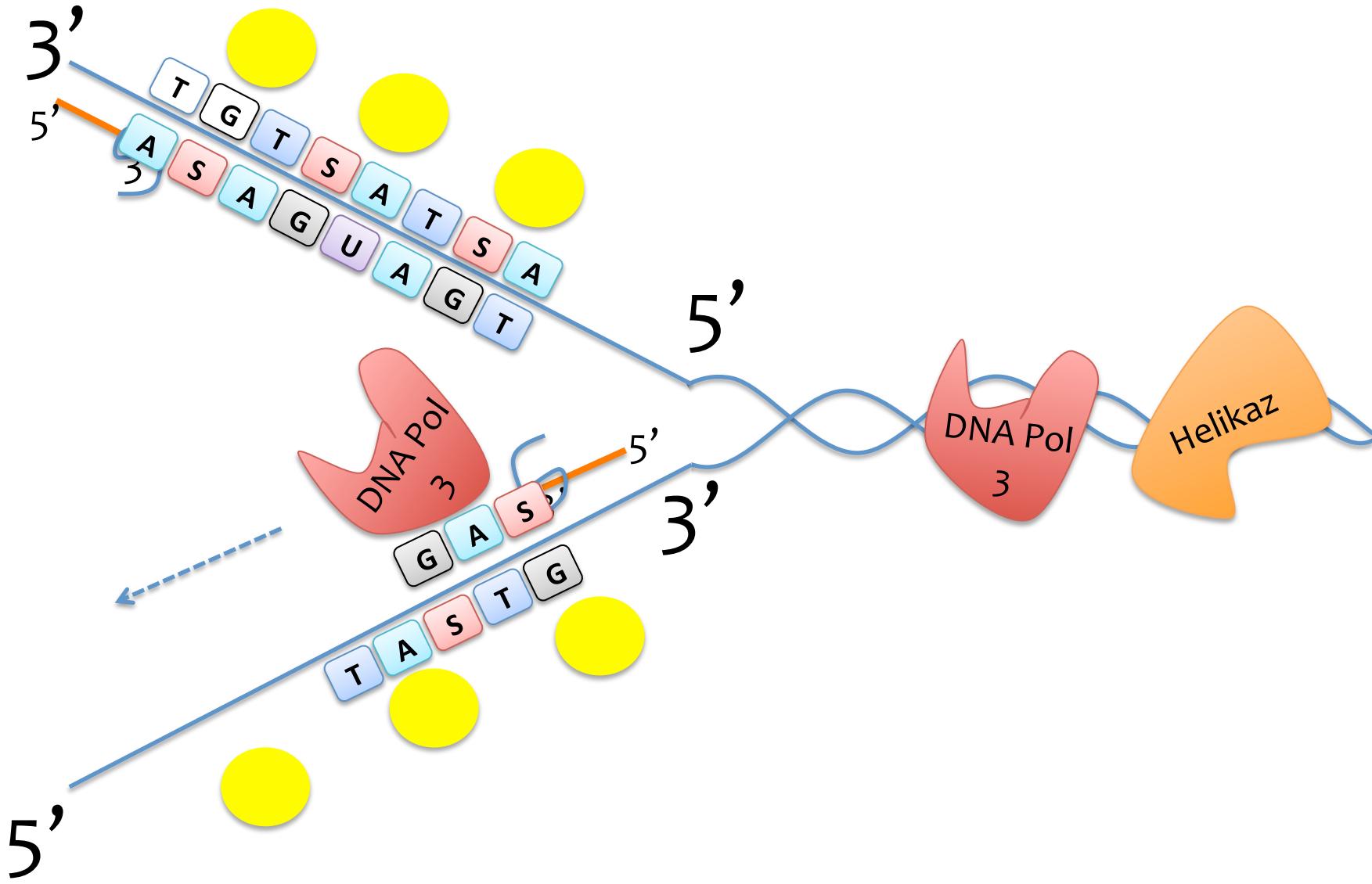


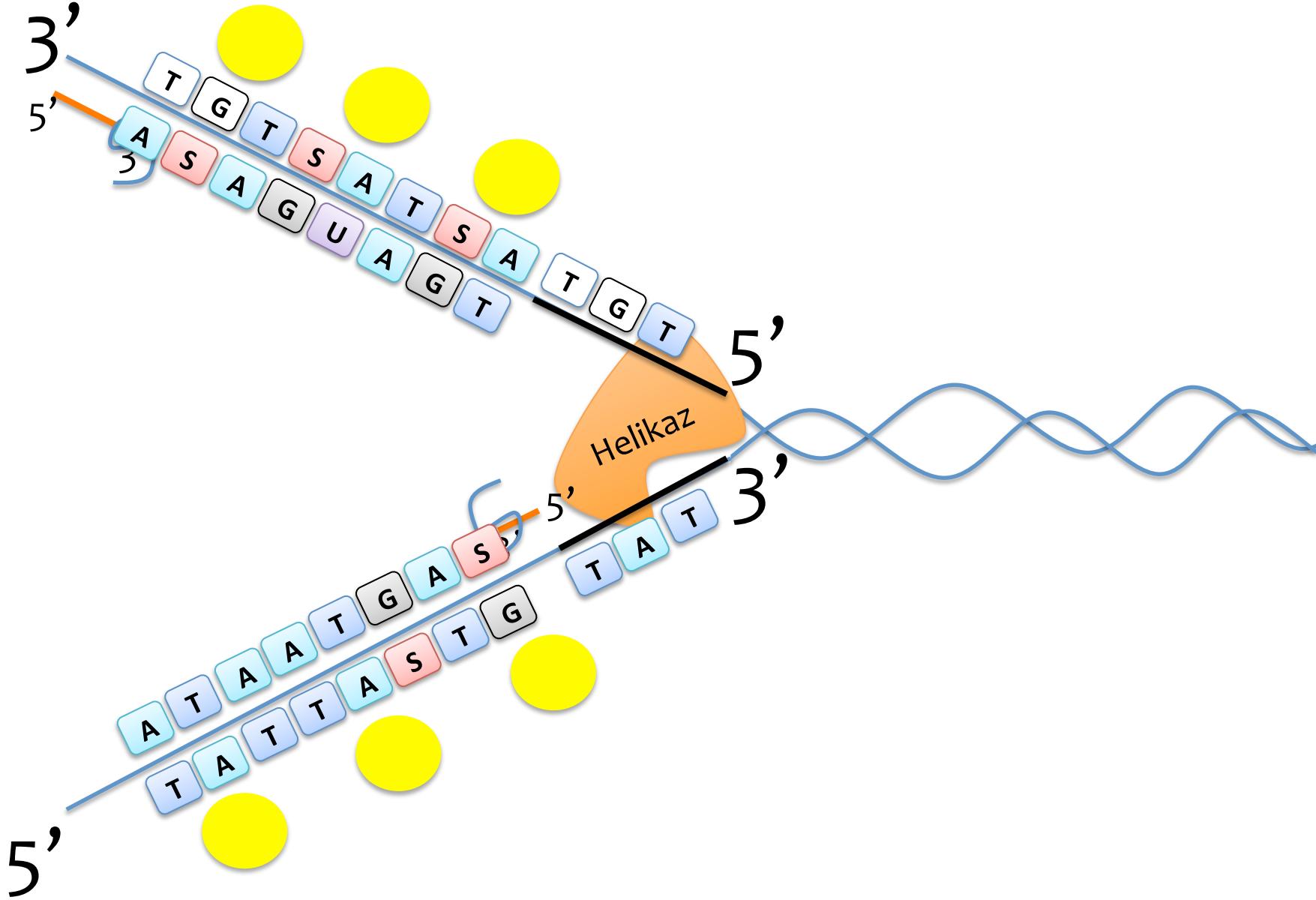


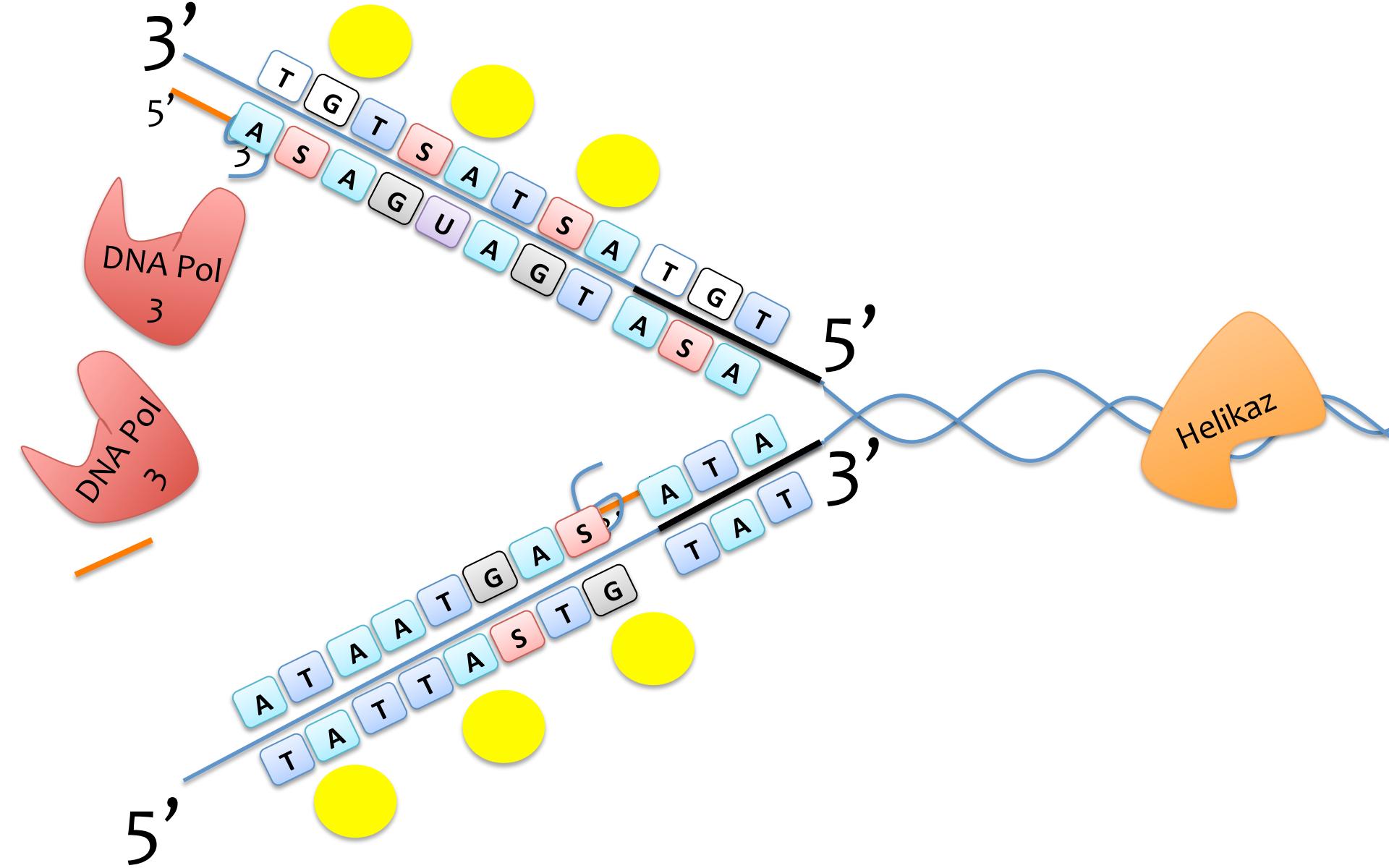


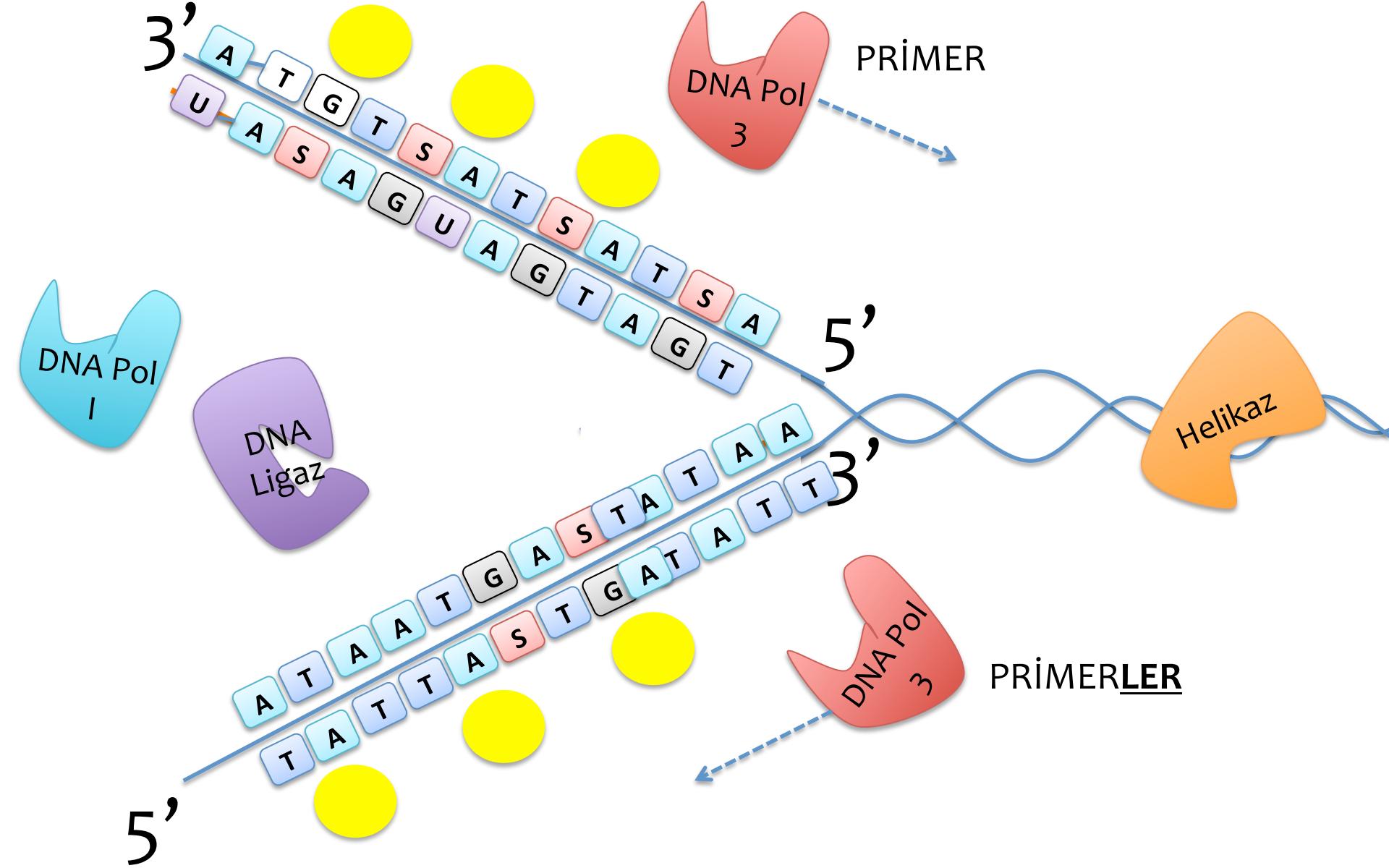


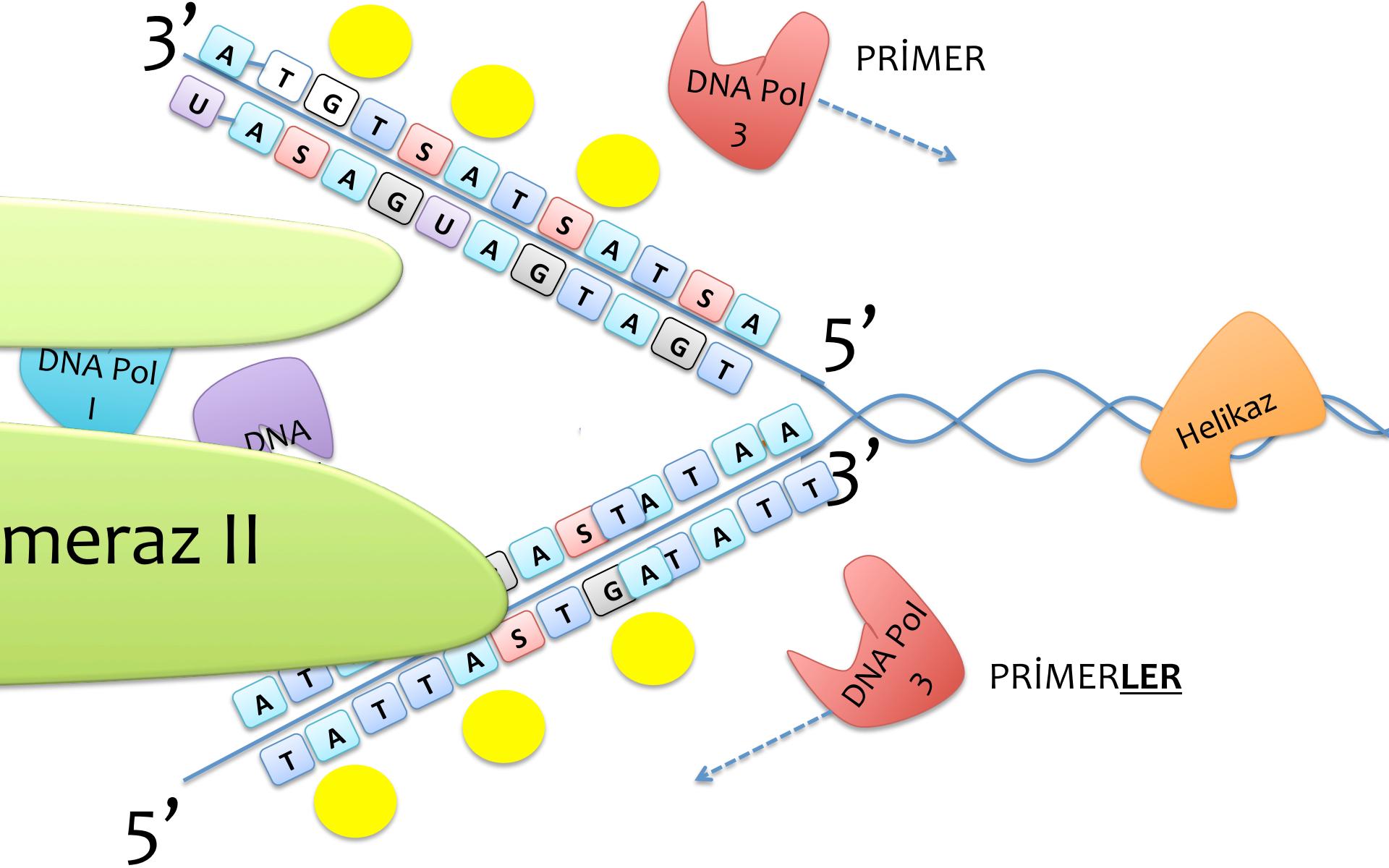


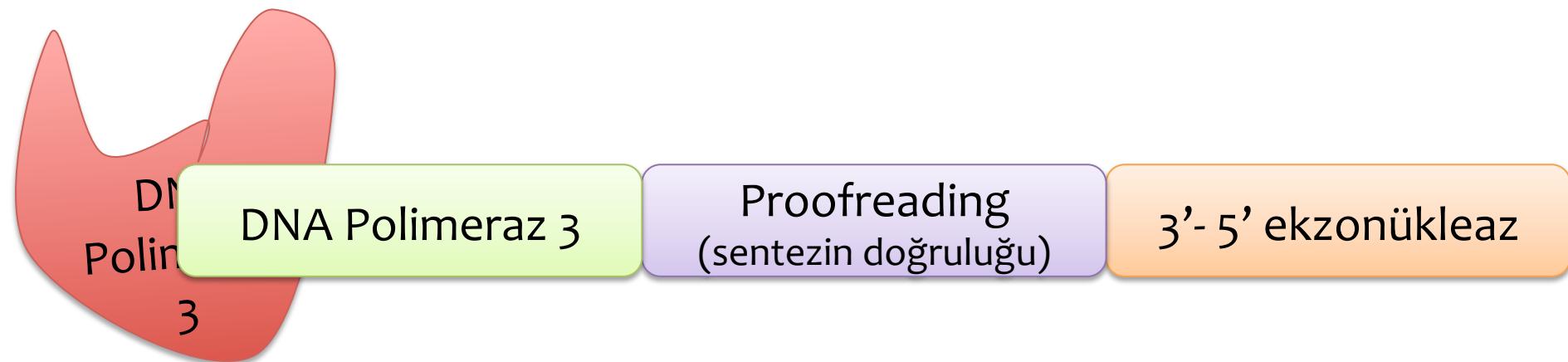
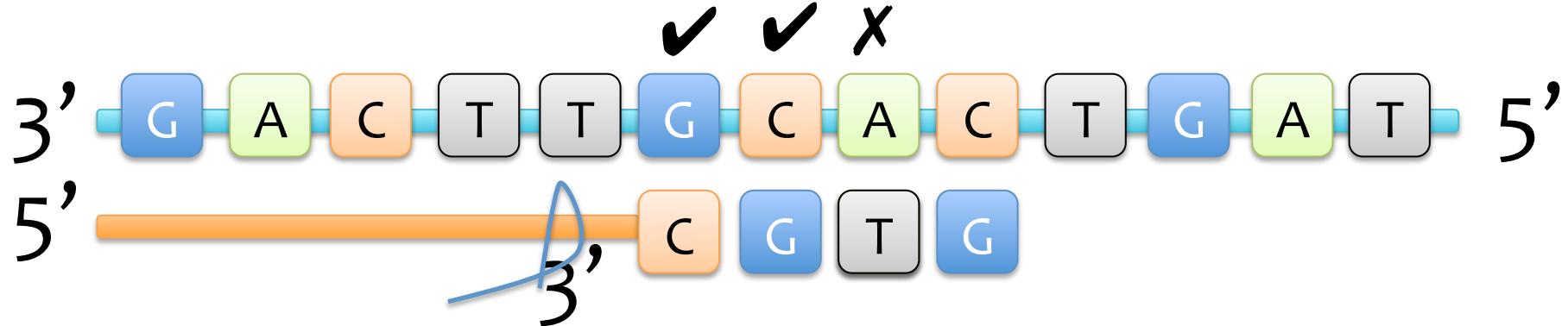








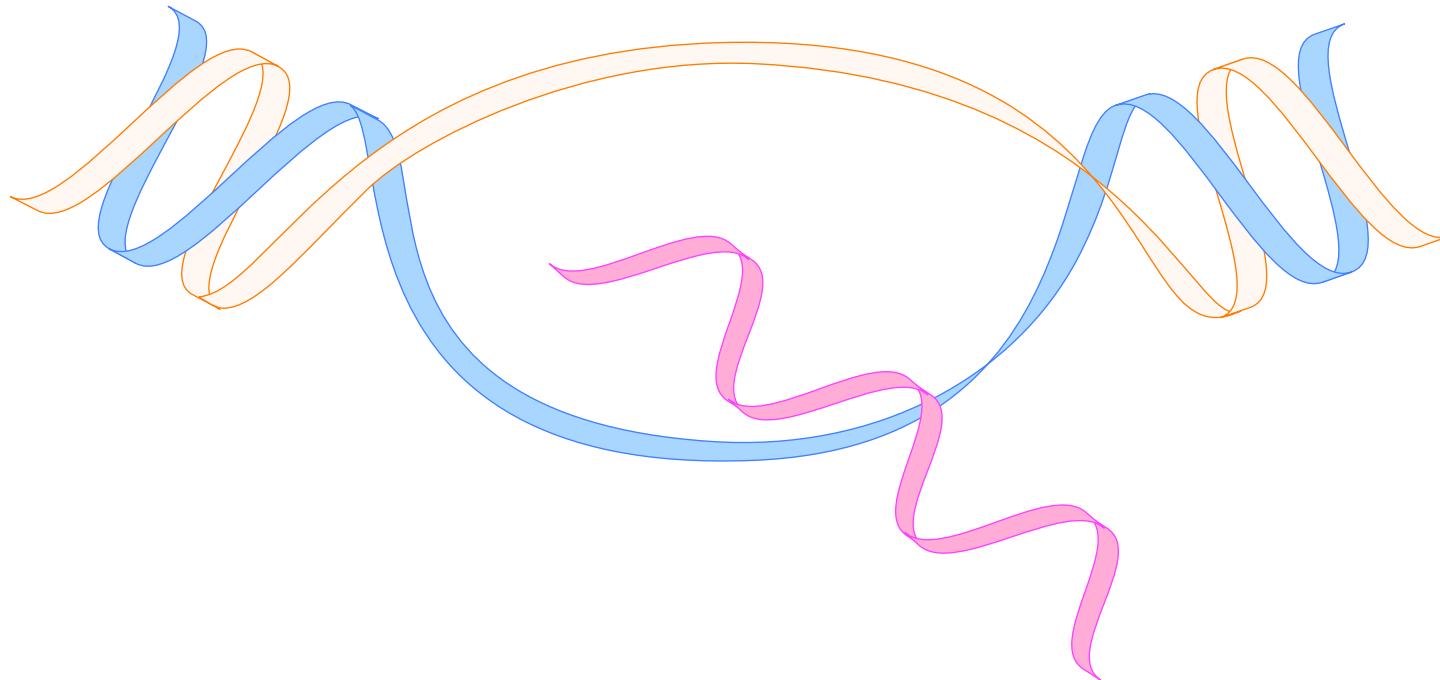




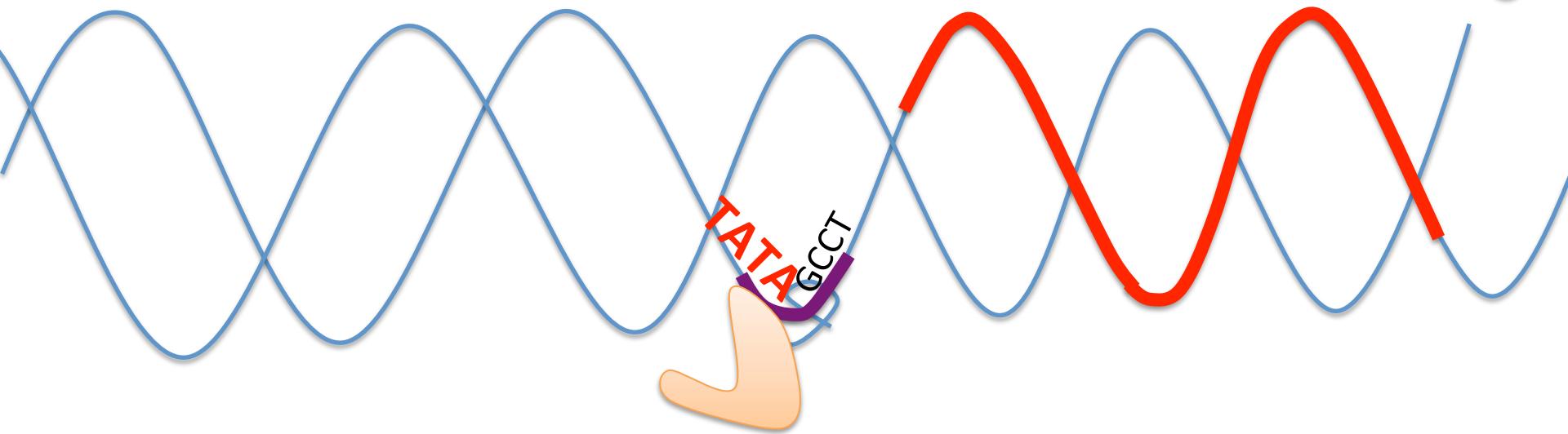
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# **TRANSKRİPSİYON**

mRNA



# Belirli Bir Bölgemin (Gen) Sentezi

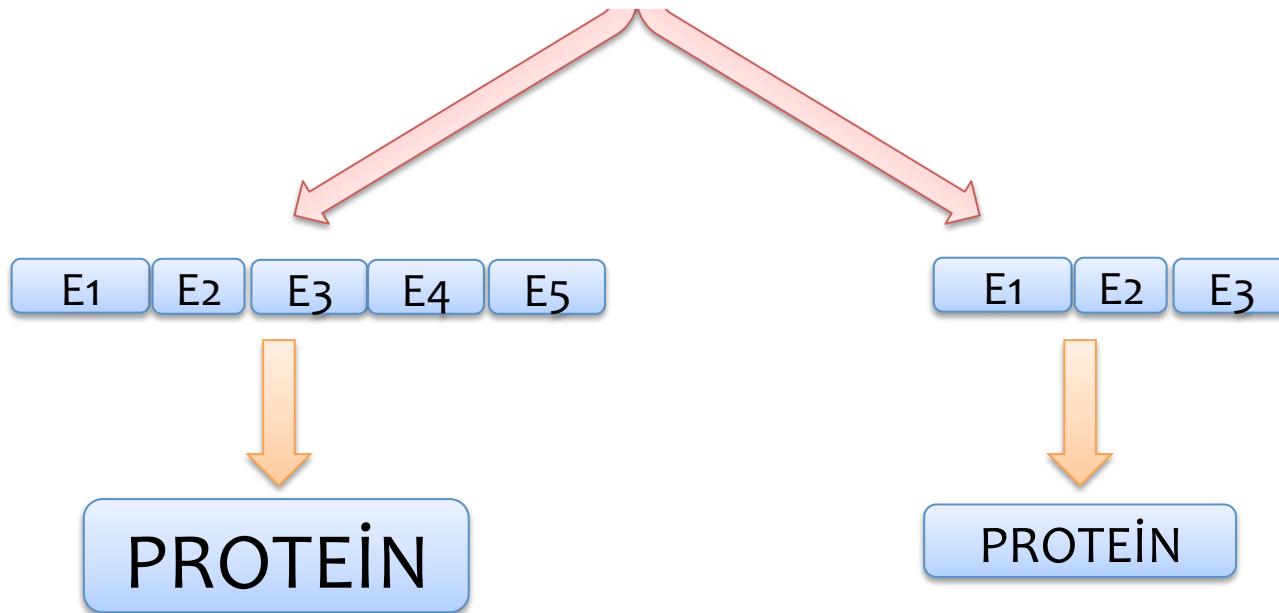
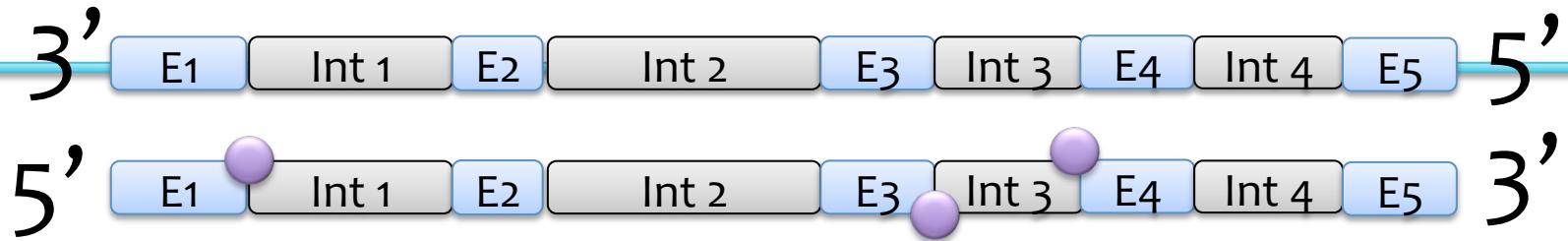


PROMOTOR (TATA, GACA)

Sigma ( $\sigma$ ) alt birimi

mRNA  
RNA Polimeraz

# Alternatif Splicing



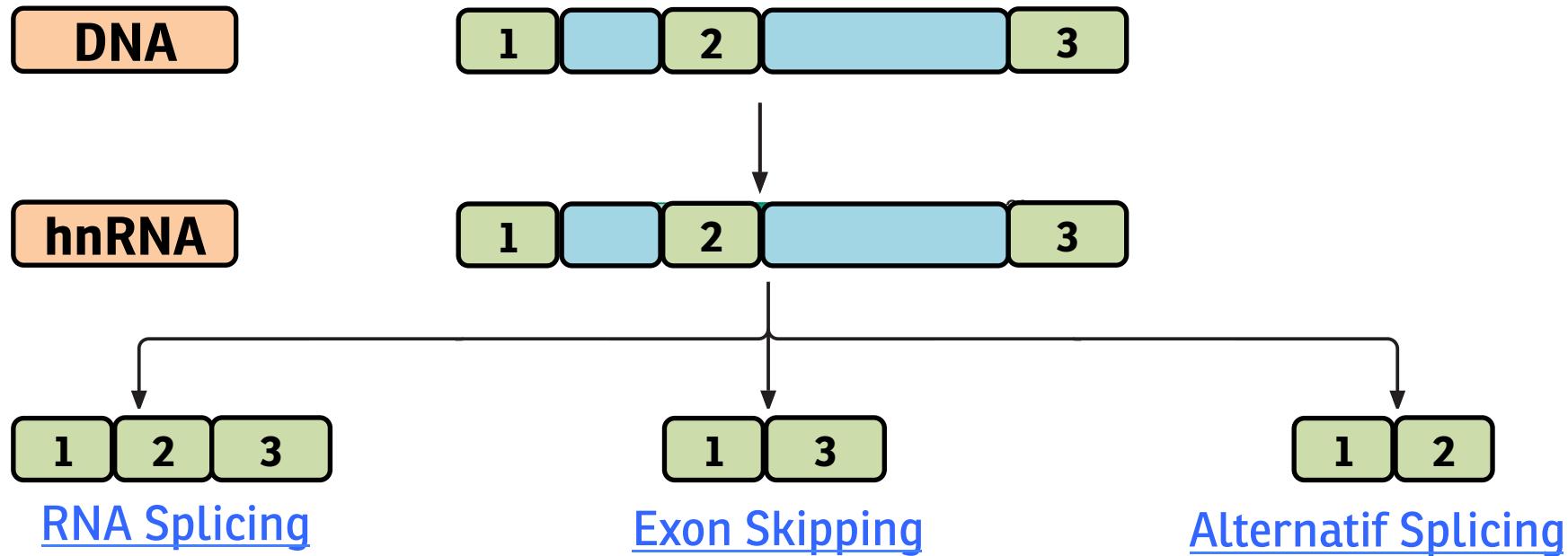
AYNI GEN'den

ALTERNATİF

PROTEİN ELDE ETMEK

SPLICİNG

# RNA Splicing



# PCR

AGCACGTTATGGAGACGCTGG**ACAATGAGTCCTTTACAGCAAAGC**GTCTGCAGGCAAATCGGTTCAAC



# PCR

AGCACGTTATGGAGACGCTGGACAATGAGTCCTTTACAGCAAAGCGTCTGCAGGCAAATCGGTTCAAC

AGCACGTTATGGAGACGCTGGACAATGAGTCCTTTACAGCAAAGCGTCTGCAGGCAAATCGGTTCAAC



# PCR

TATGGAGACGCTGG**ACAATGAGTCCTTTACAGCAAAGC**GTCTGCAGGCAAATCGGTTCAAC

AGCACGTTATGGAGACGCTGG**ACAATGAGTCCTTTACAGCAAAGC**GTCTGCAGGCAAAC



# PCR\_400-1000bp

TATGGAGACGCTGGACAATGAGTCCTTTACAGCAAAGC GTCTGCAGGCAAA

TATGGAGACGCTGGACAATGAGTCCTTTACAGCAAAGC GTCTGCAGGCAAA

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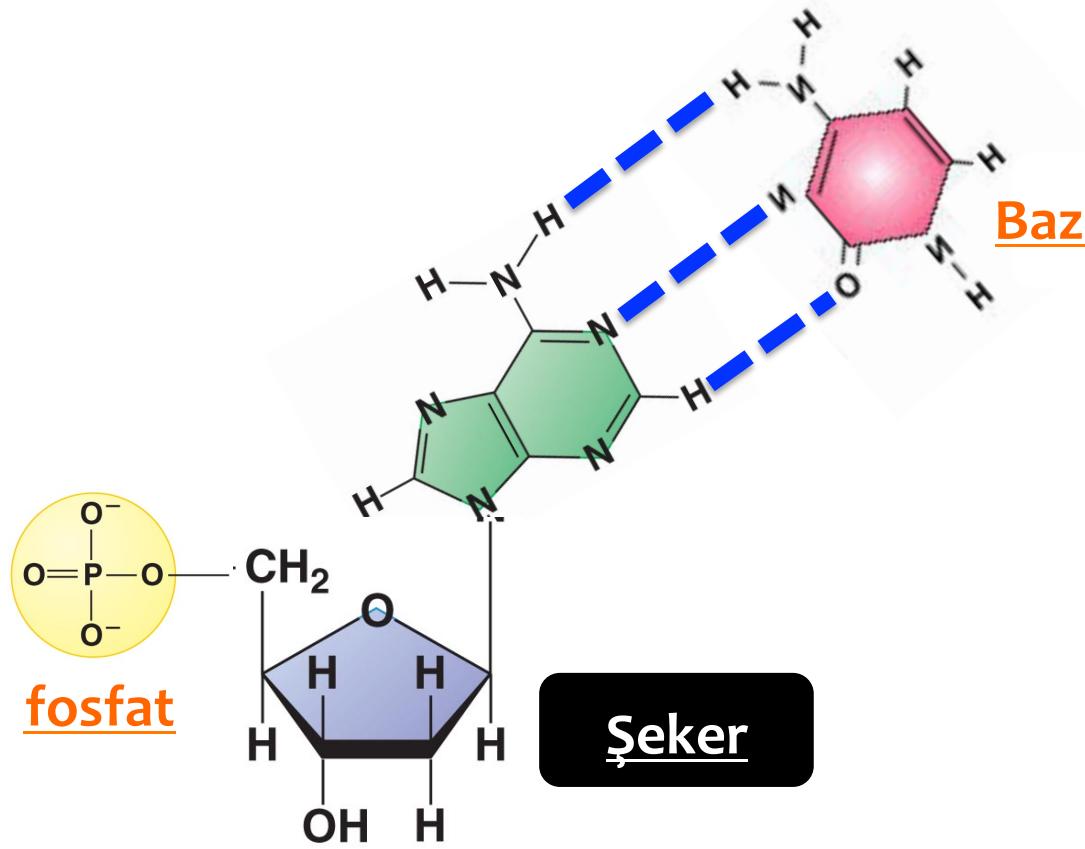
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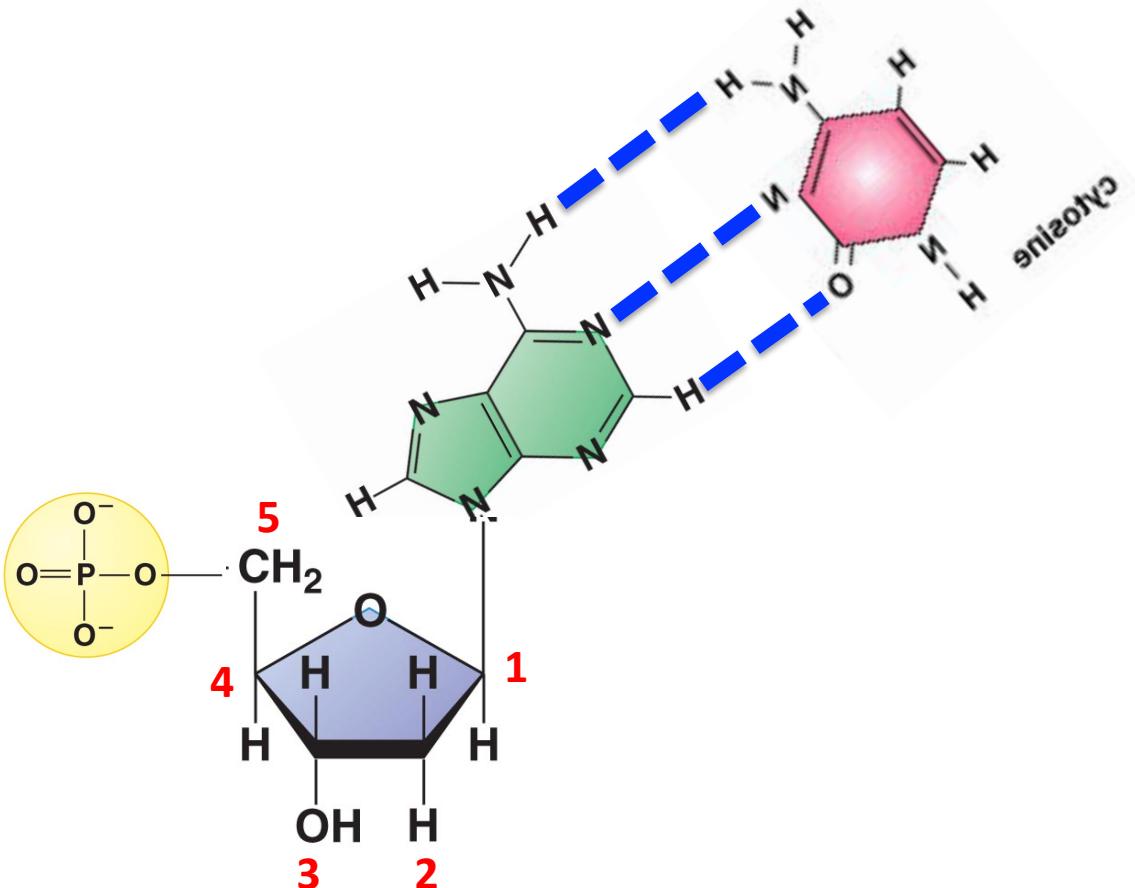
# Sanger Sekans

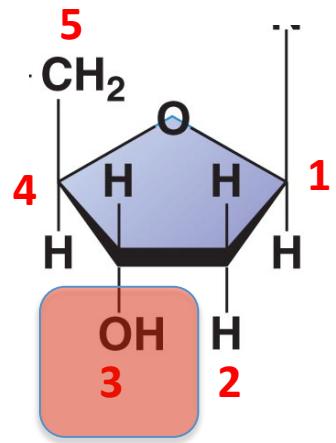


fosfat

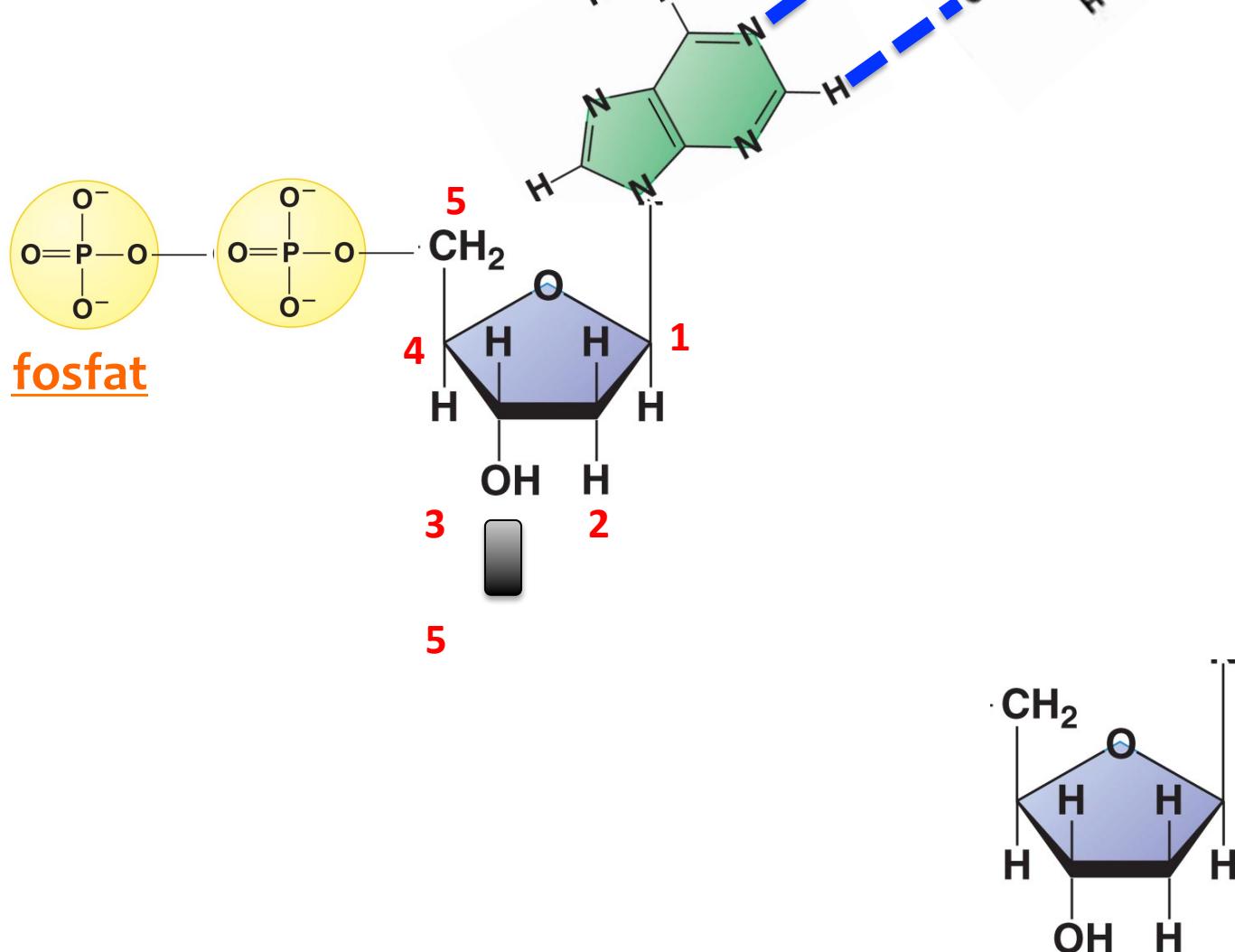
Seker

Baz



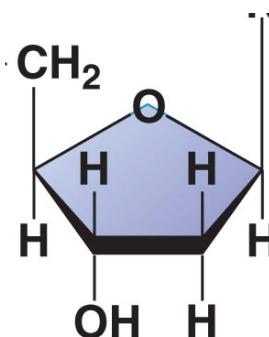
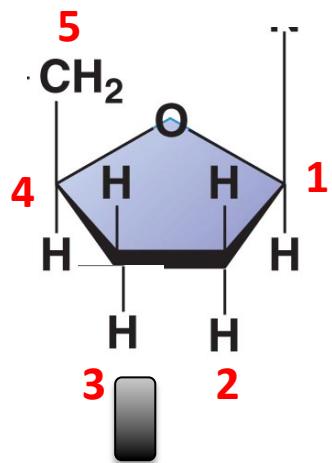


Deoksinükleotid\_DNA





## DiDeoksinükleotid



# Sanger Sekans

TATGGAGACGCTGGACAATGAGTCCTTTACAGCAAAGC GTCTGCAGGCAAA

# Sanger Sekans

TATGGAGACGCTGGACAATGAGTCCTTTACAGCAAAGC GTCTGCAGGCAAA



A T T A T A T  
  C G C G G C  
   G C G C G

# Sanger Sekans

TATGGAGACGCTGGACAATGAGTCCTTTACAGCAAAGC GTCTGCAGGCAA

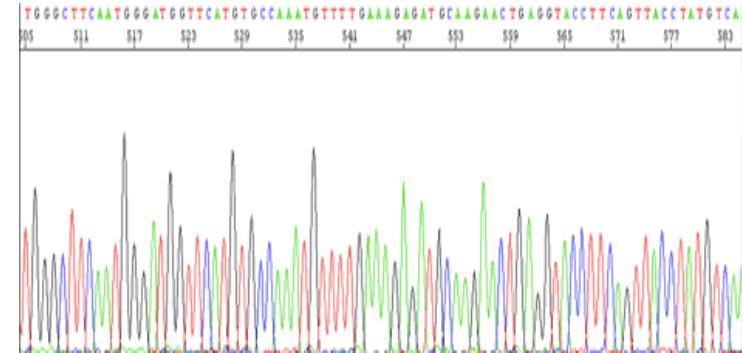
TGC

TGCGA

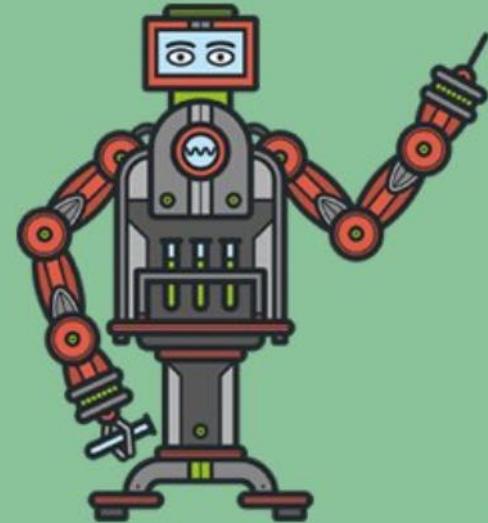
TGCGACCTGTTACTCAGGAAAATGTCGTTCG

T

TG



- <http://www.ensembl.org/index.html>
- <https://genome-euro.ucsc.edu/index.html>

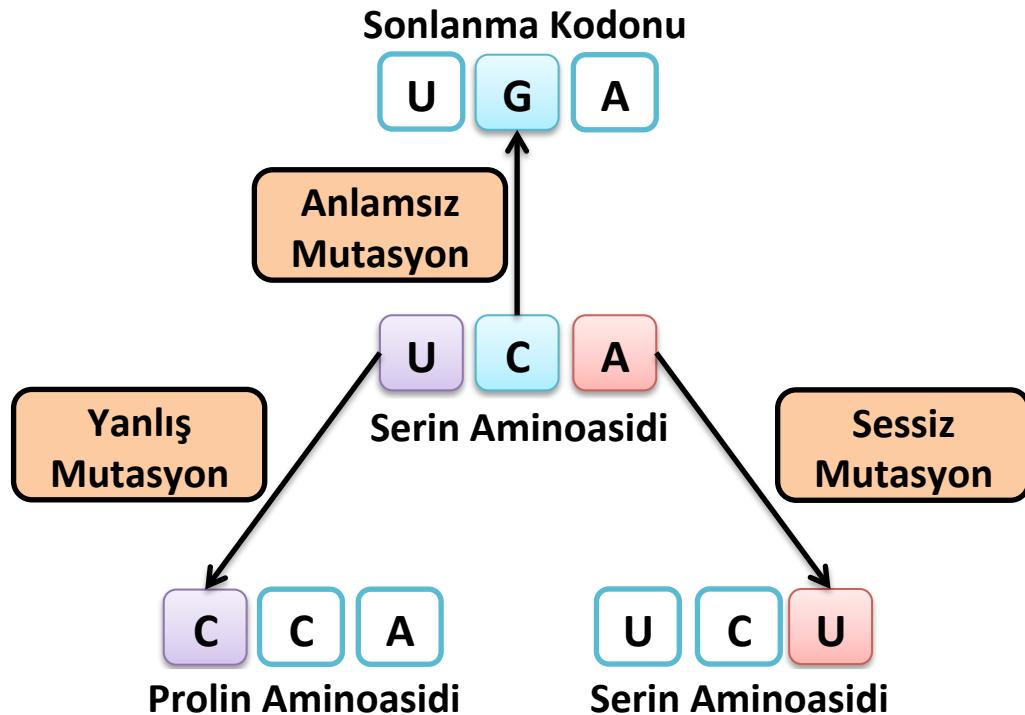


**nature**

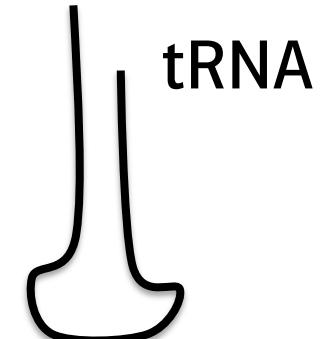
[bit.ly/OnwardsUpwards](http://bit.ly/OnwardsUpwards)

**“By 2040, 1 billion people will have their whole genome sequenced and get constant updates of their immunomes and microbiomes.”**

# **TRANSLASYON (PROTEİN SENTEZİ)**



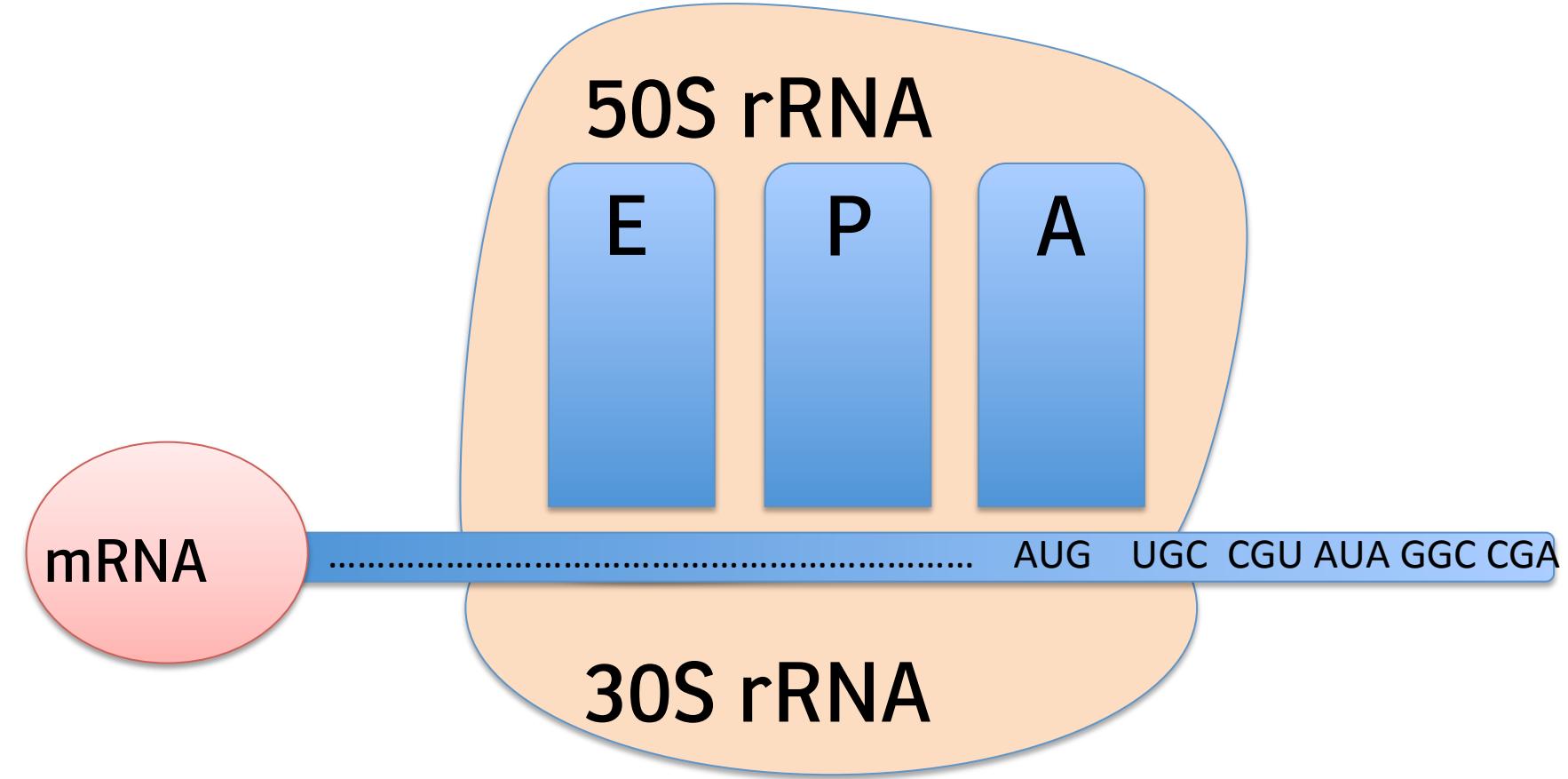
50S rRNA

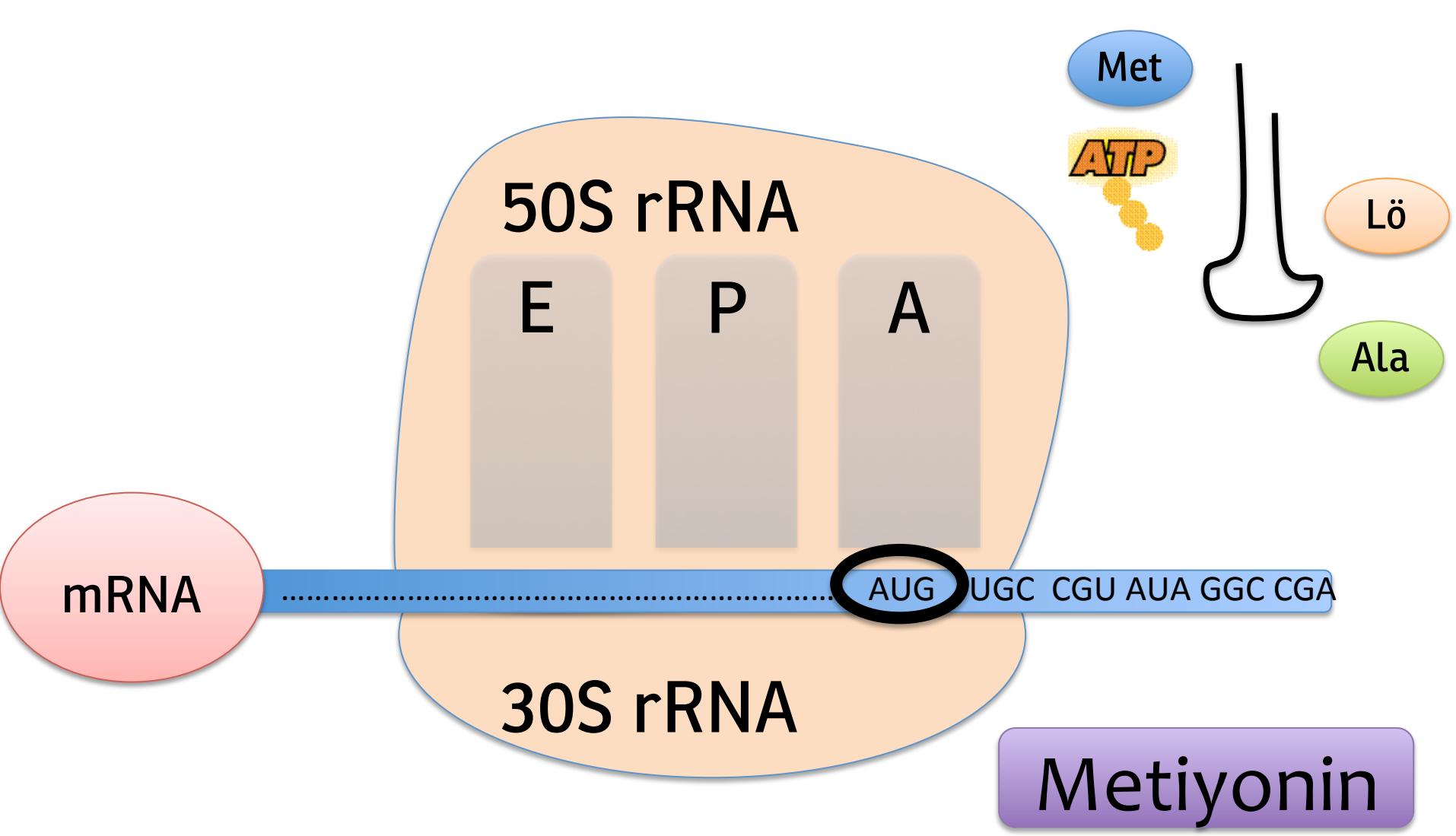


mRNA

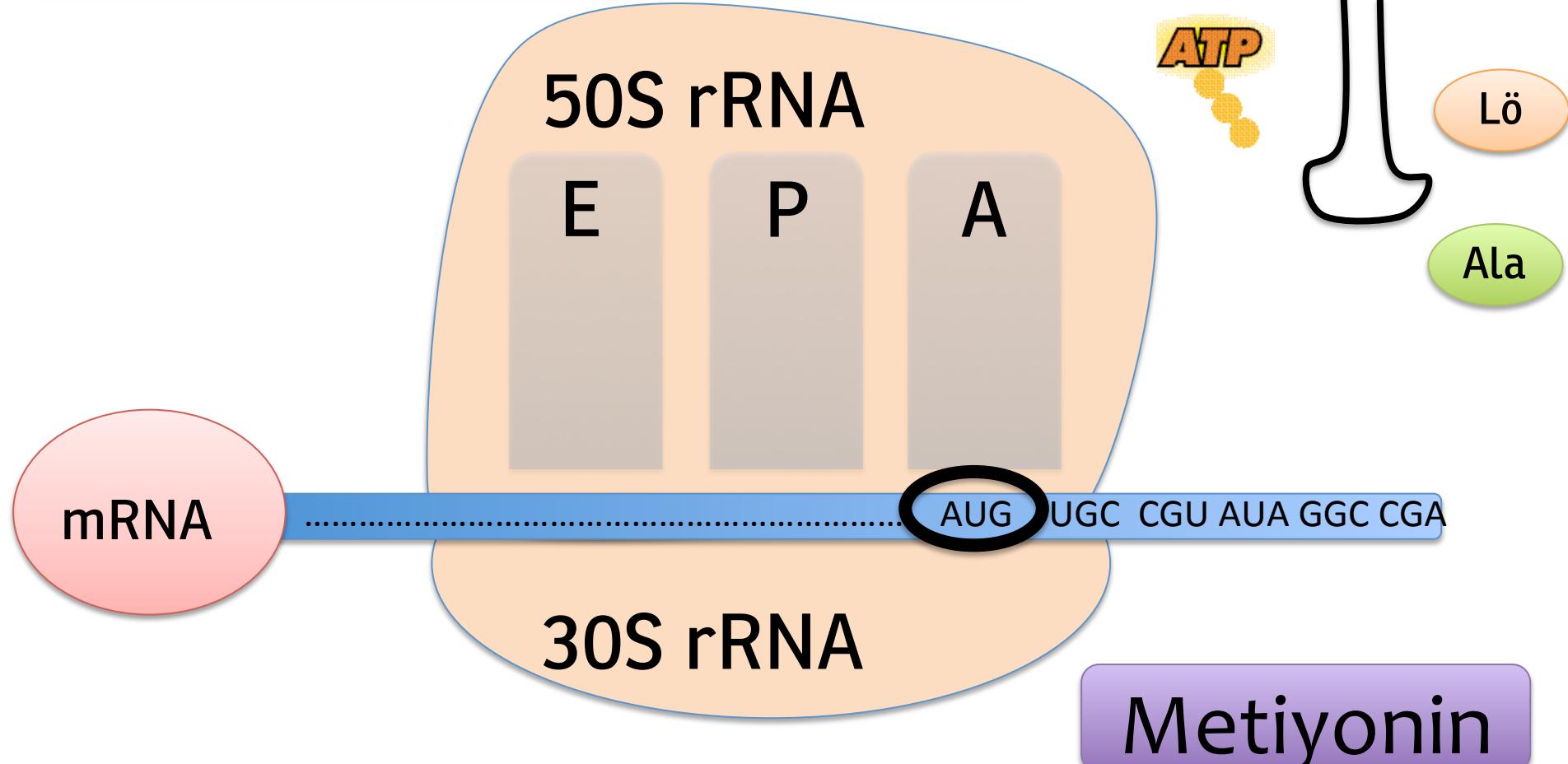
AUG UGC CGU AUA GGC CGA

30S rRNA

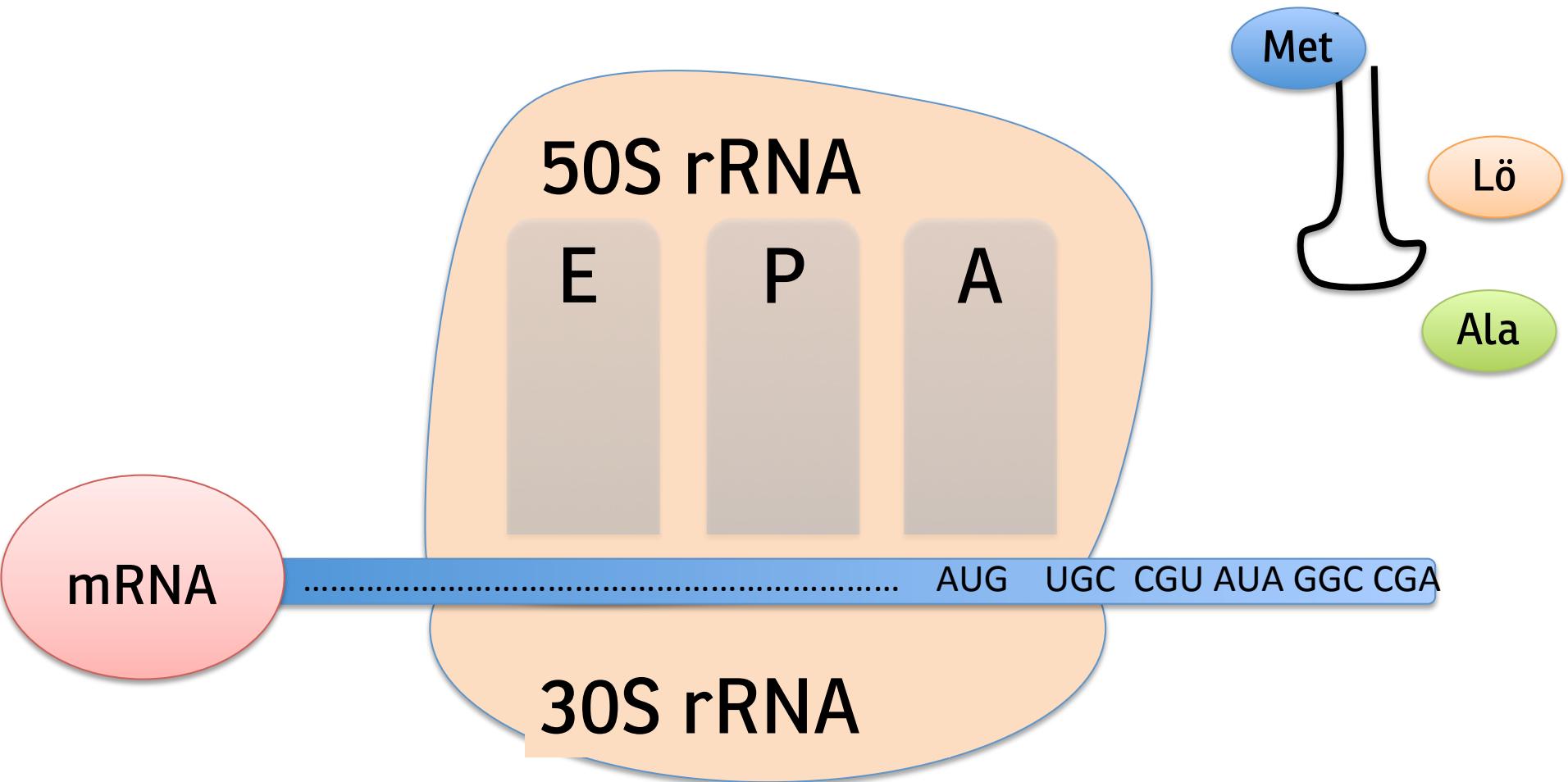


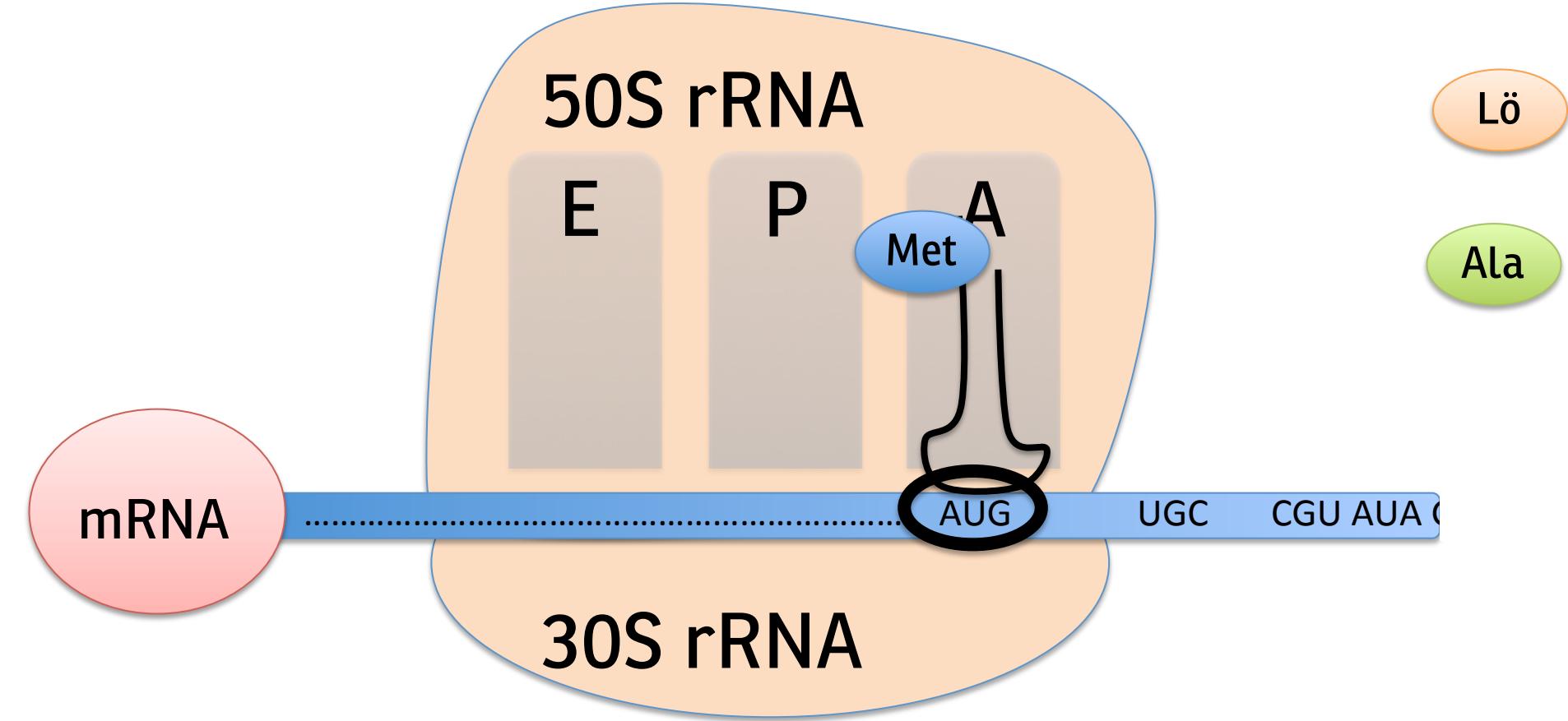


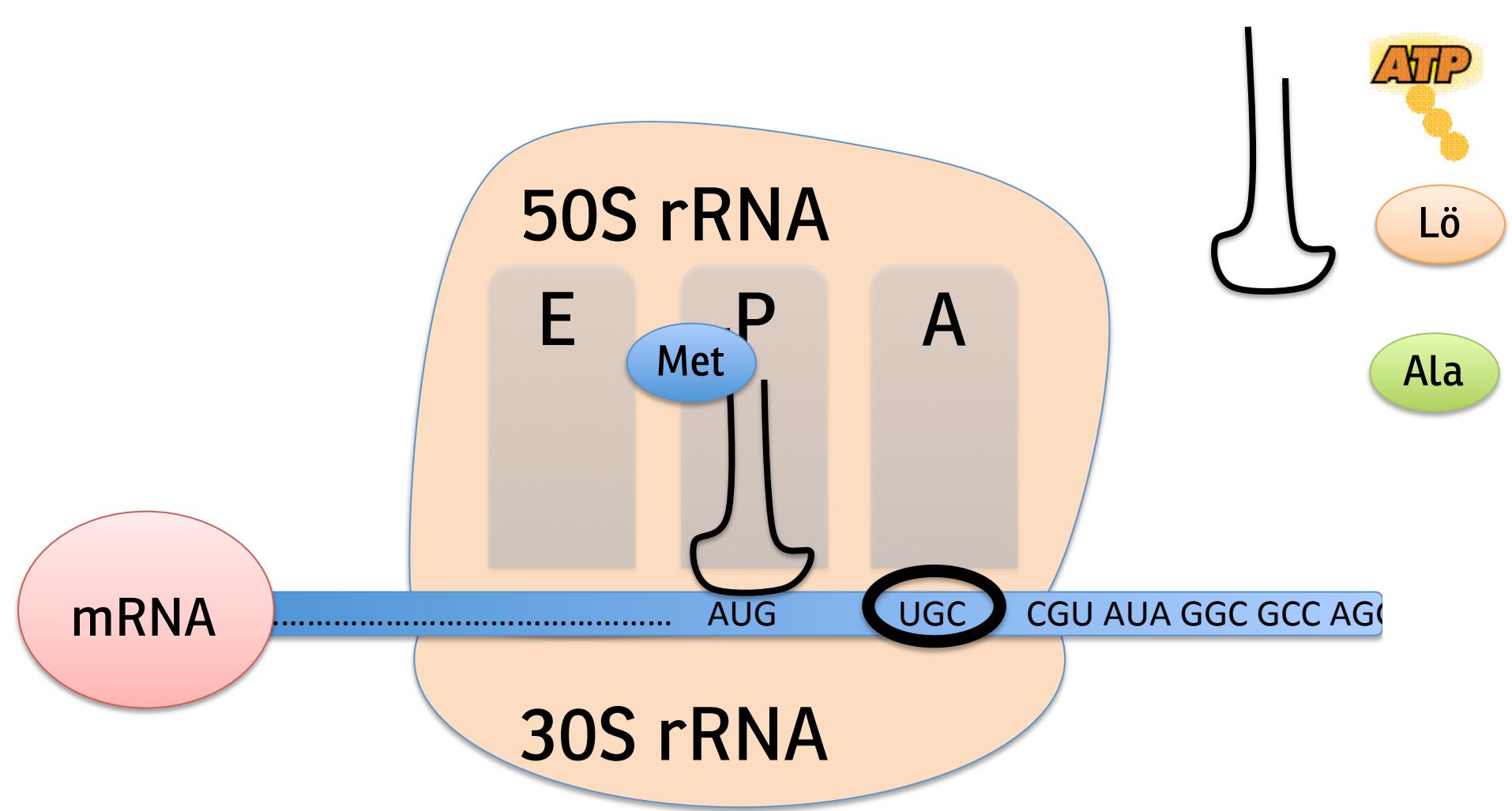
Başlangıçta Amino Asit Aktive Edilir

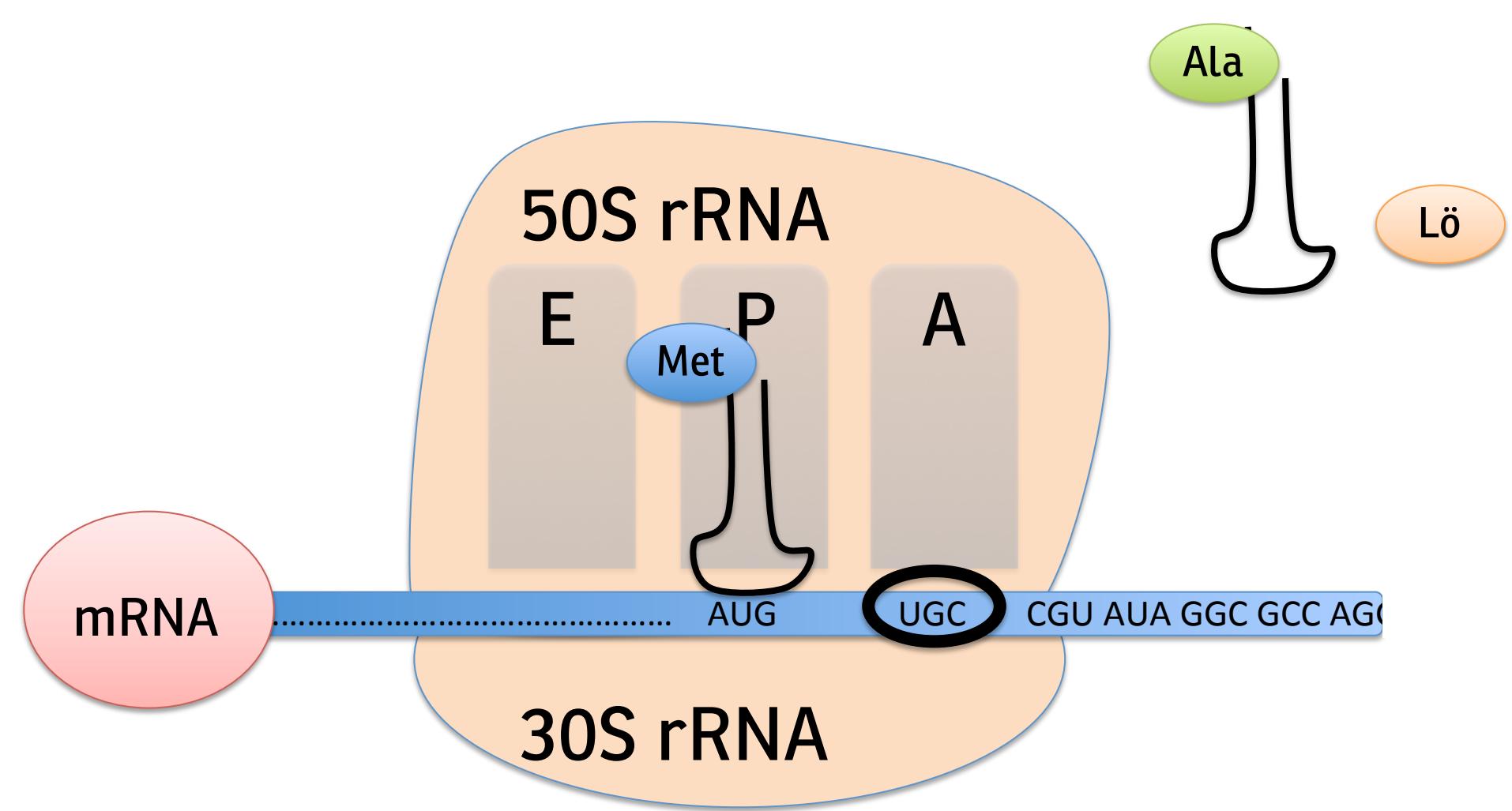


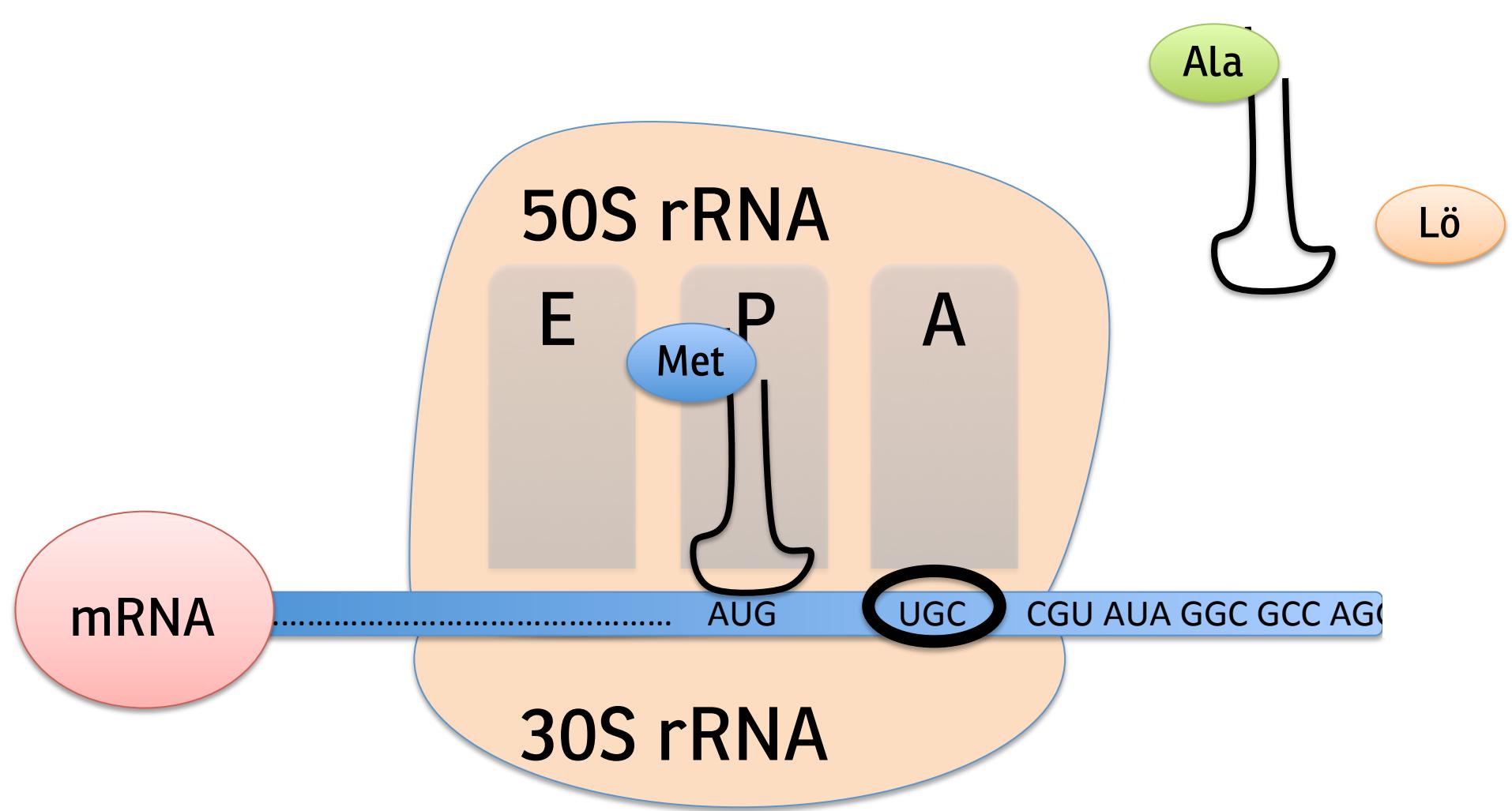
Metiyonin







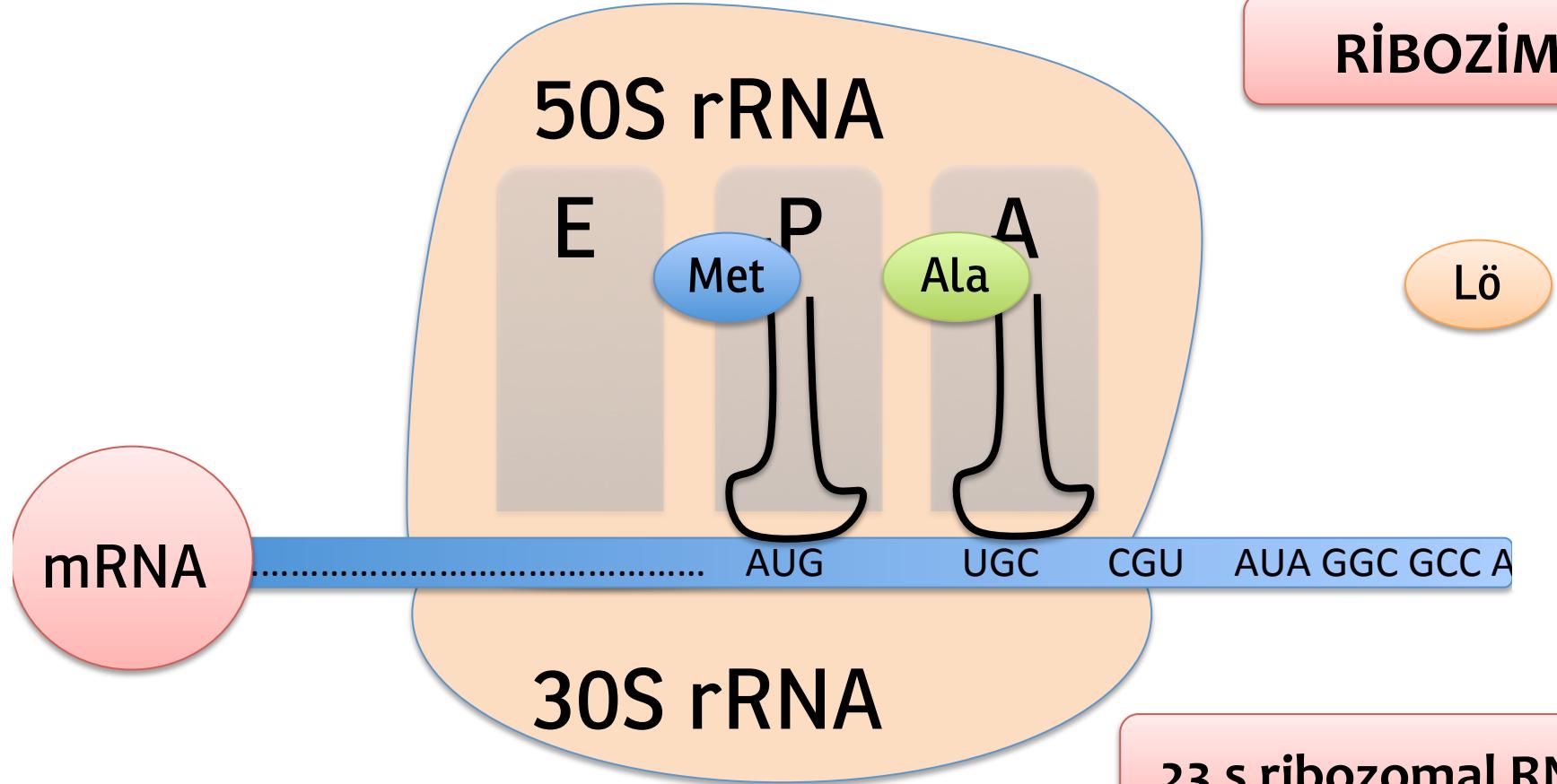




P bölgesinde Peptid Bağı Yapılır

Peptidil Transferaz

RİBOZİM

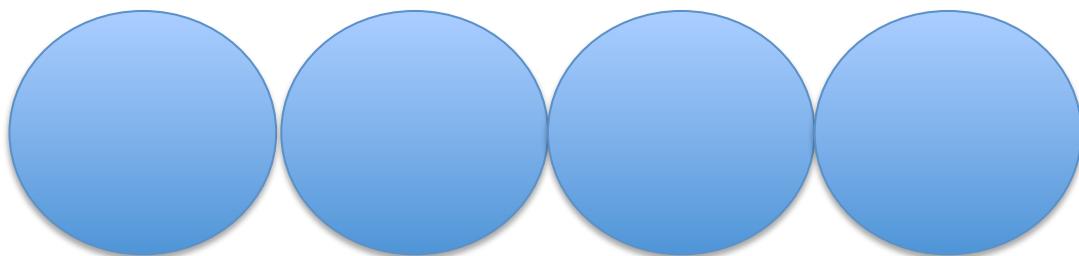


23 s ribozomal RNA

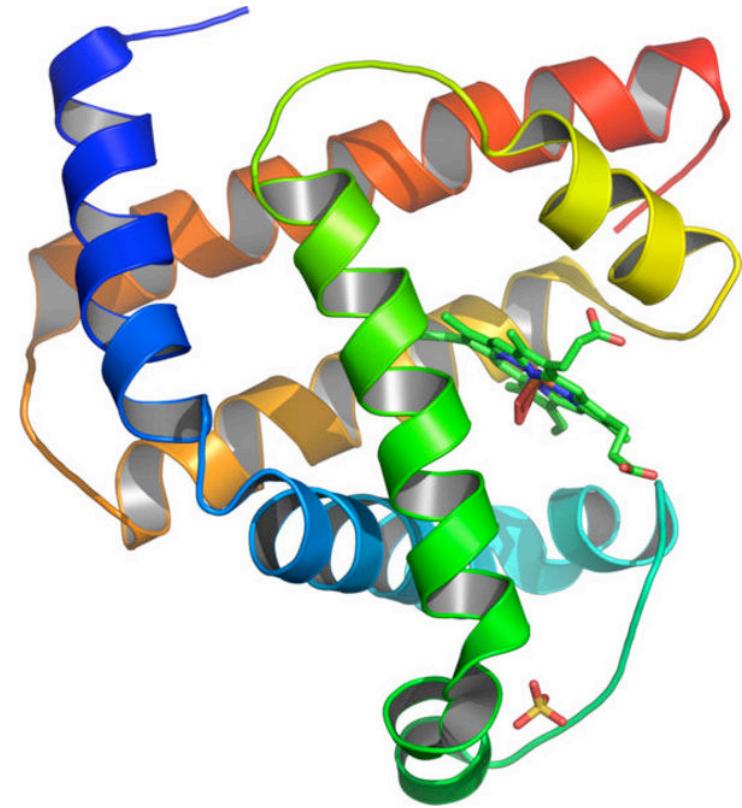
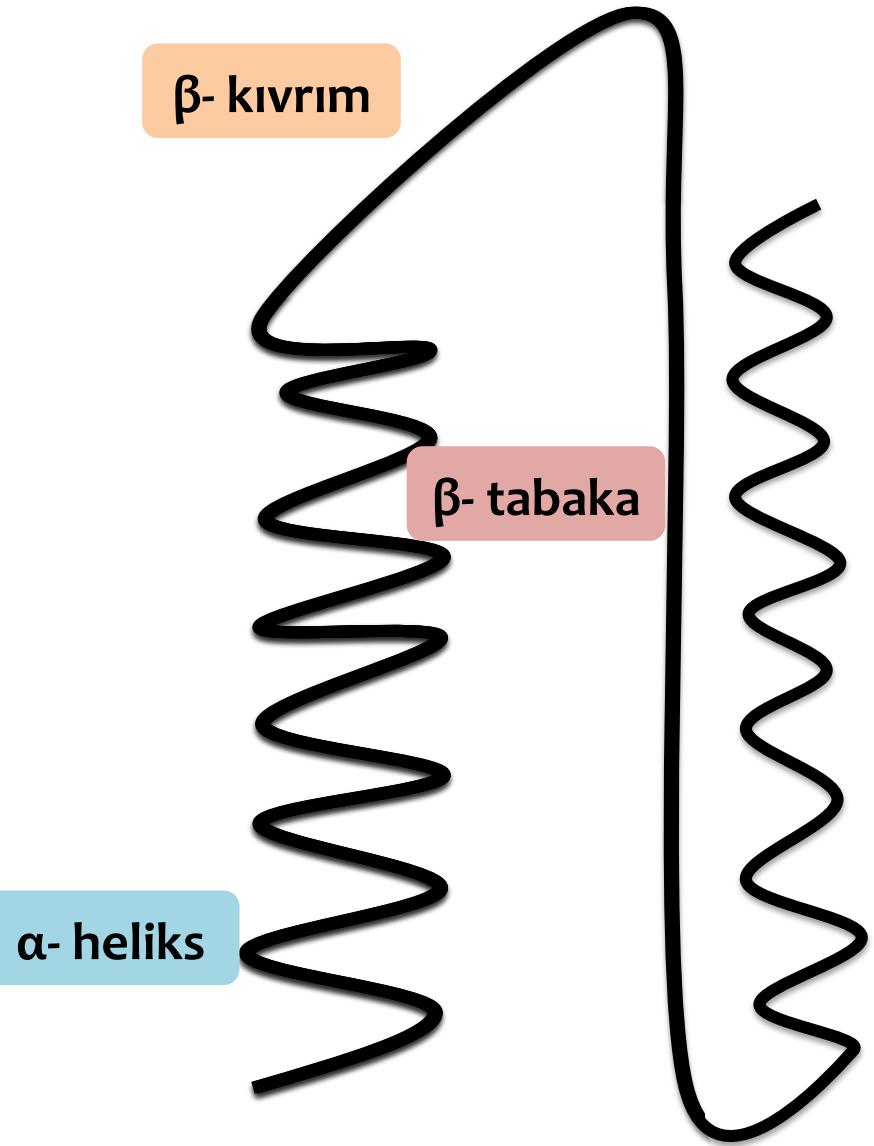
28 s ribozomal RNA

# PROTEİN

# **1-PRIMER YAPI**

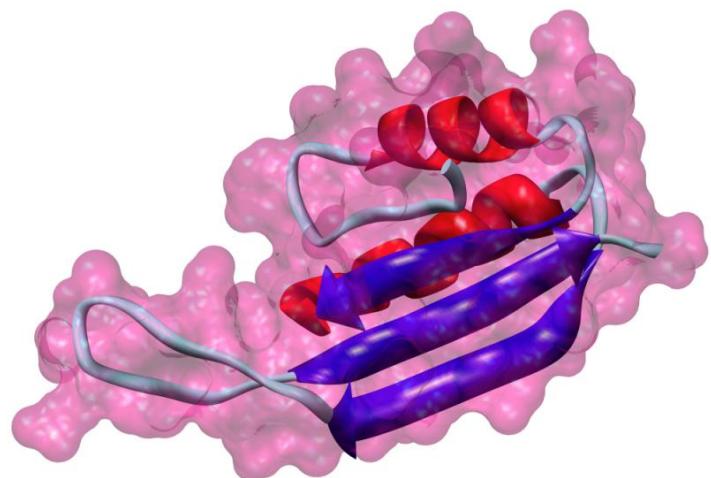


**2– SEKONDER YAPI**



<http://www.uniprot.org/>  
<http://swissmodel.expasy.org/>  
<http://www.rcsb.org/pdb/home/home.do>

## 3– TERSİYER YAPI



# **POST TRANSLASYONEL KONTROL**

# Post Translasyonel Kontrol

- Fosforilasyon
  - Serin treonin tirozin
- Glikolizasyon
- Karboksillenme
  - K vit, gama karboksi glutamat, varfarin
- Hidroksilasyon
  - Kollojen, lizin prolin
- Metillenme
  - Metil histidin
- Sulfatasyon
  - PAPS
- Prenilasyon

# **EPİGENETİK KONTROL**

# Epigenetik Mekanizmlar

- DNA baz dizisinde değişiklik (**mutasyon**) olmaksızın gen ifadesindeki değişimlere epigenetik mekanizmlar denir.

- 1-DNA metilasyonu
- 2-Histon modifikasyonu
- 3-miRNA ve siRNA
- 4-Genomik imprinting

