Introduction to Genes

Tuesdays 12.15-12.45 M203
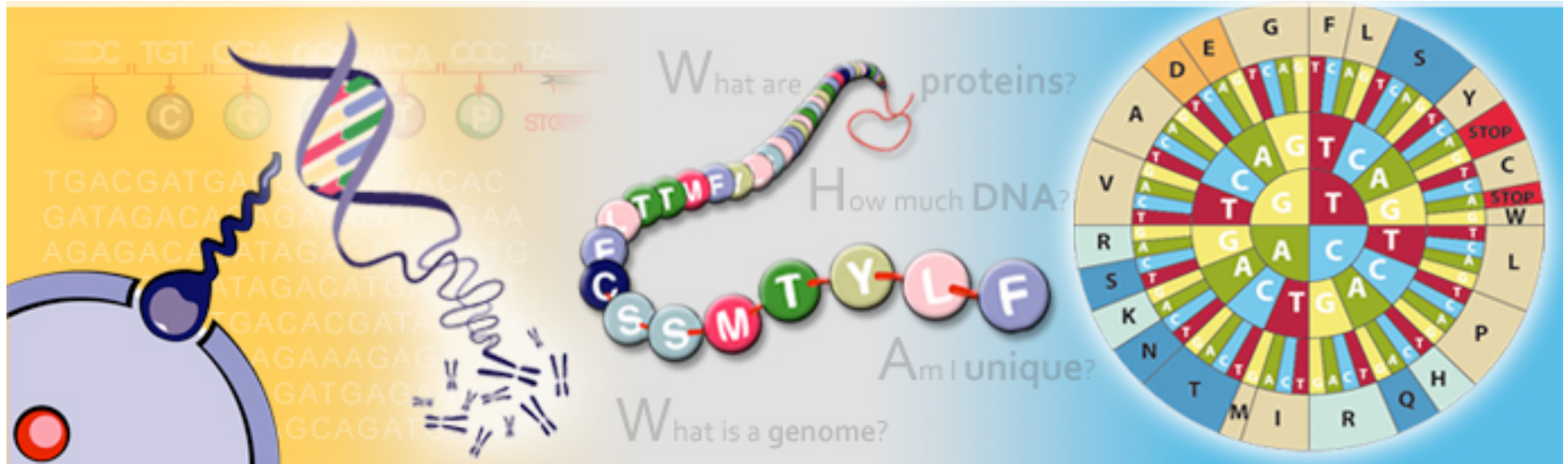
23.02.2010	**What are genes?**
03.03.2010	**How are genes identified?**
29.03.2010	**How do we find out about gene function?**
16.03.2010	**Genes and disease**

http://intweb.sanger.ac.uk/Info/Seminars/genes/
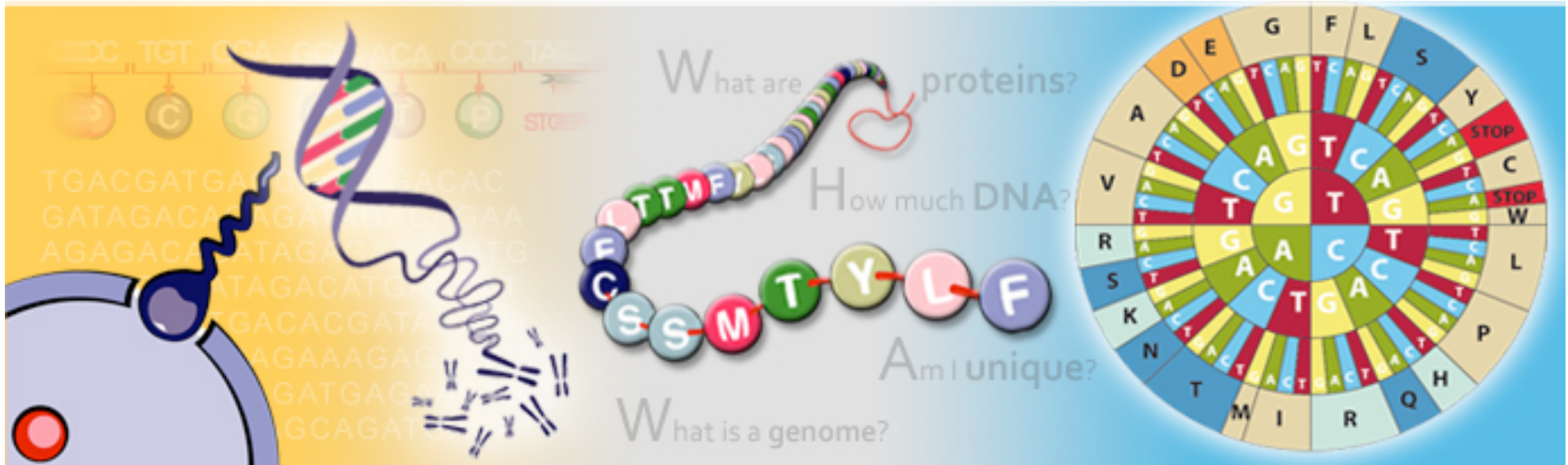
What are genes?

Kerstin Howe

What are Proteins?

essential parts of organisms, participate in virtually every process within cells

- enzymes => 'factories' for chemical reactions
- signals => telling the cells where they are / what's happening
- structural => cell scaffolds, cartilage, hair, nails, feathers, hooves,
- mechanical => muscles

organic compounds made of amino acids in linear chain

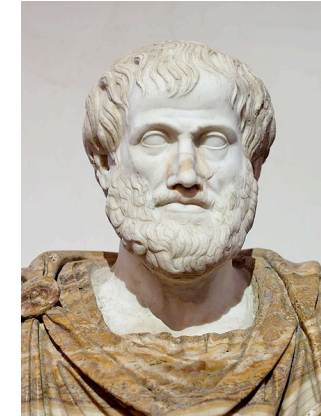folded into globular form
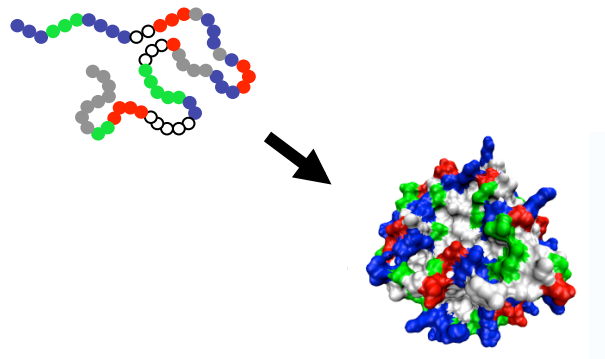
can form complexes
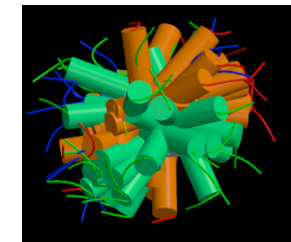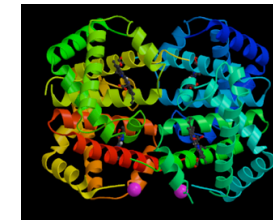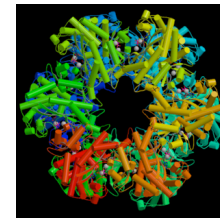
animals can't make all amino acids themselves, so need to take them up with their diet

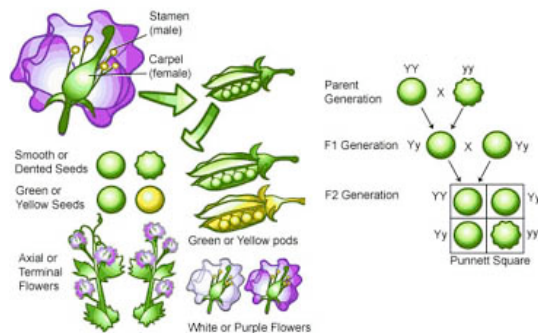# Why do children look like their parents?

Aristotle (384-322 BC)

theory of pangenesis: particles (pangenes) from all parts

of the body come together to form the eggs and sperm,

offspring is a thorough mixture

Gregor Mendel (~1860)

factors convey traits from parents to offspring

these factors are units of inheritance and retain their individuality

=> **GENES** (term established 50 years later!)

What are genes made of?

1910 T.H. Morgan   genes reside on chromosomes

1941 Beadle & Tatum   'one gene – one protein' hypothesis

1944 Avery, MacLeod & McCarty   DNA (!) is the genetic material

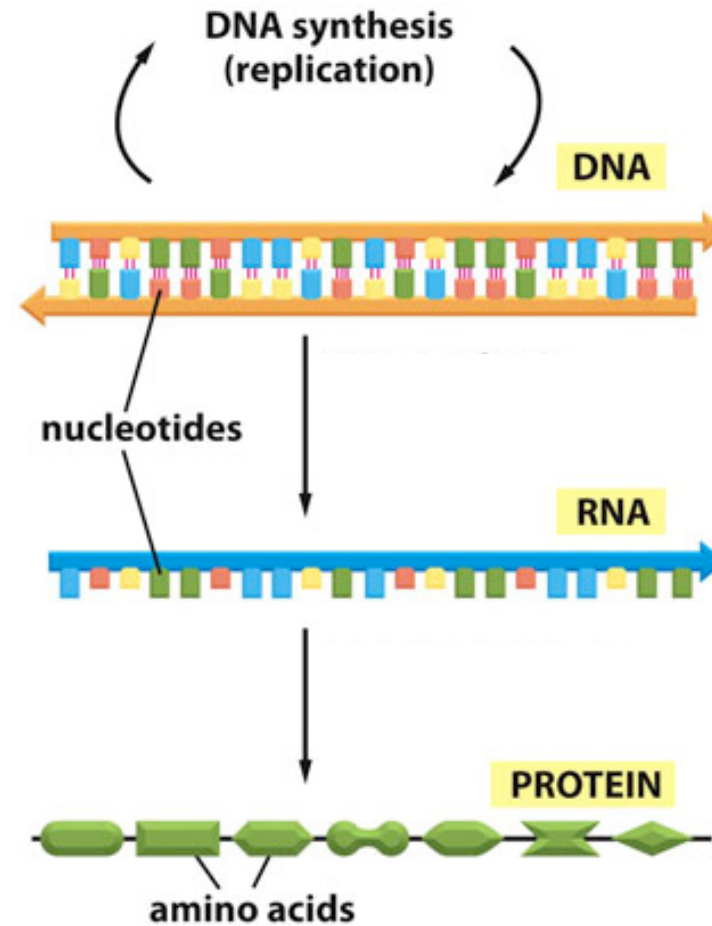**1953 Watson & Crick**   proposed the DNA structure

⇒ **genes are units of DNA on chromosomes, coding for a protein**

⇒ **'one gene – one protein' hypothesis seems right**

The central dogma of molecular biology (Crick 1958)

Genes are **transcribed** into RNA

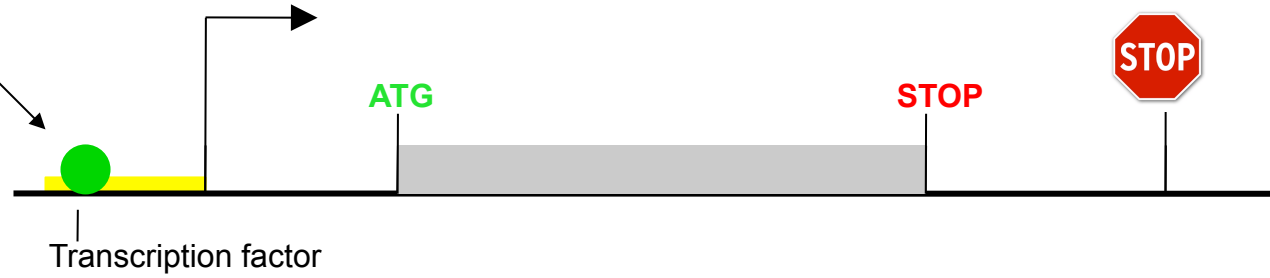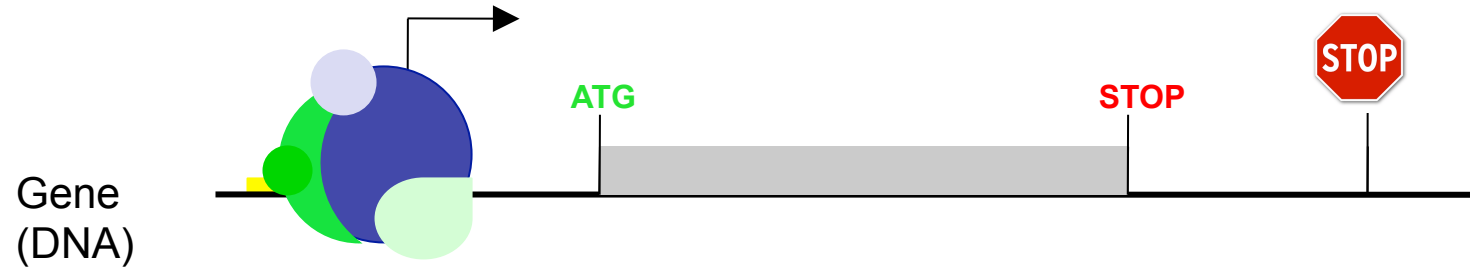The RNA is **translated** into a protein

Simple Genes

Gene
(DNA)

Initiation of
transcription
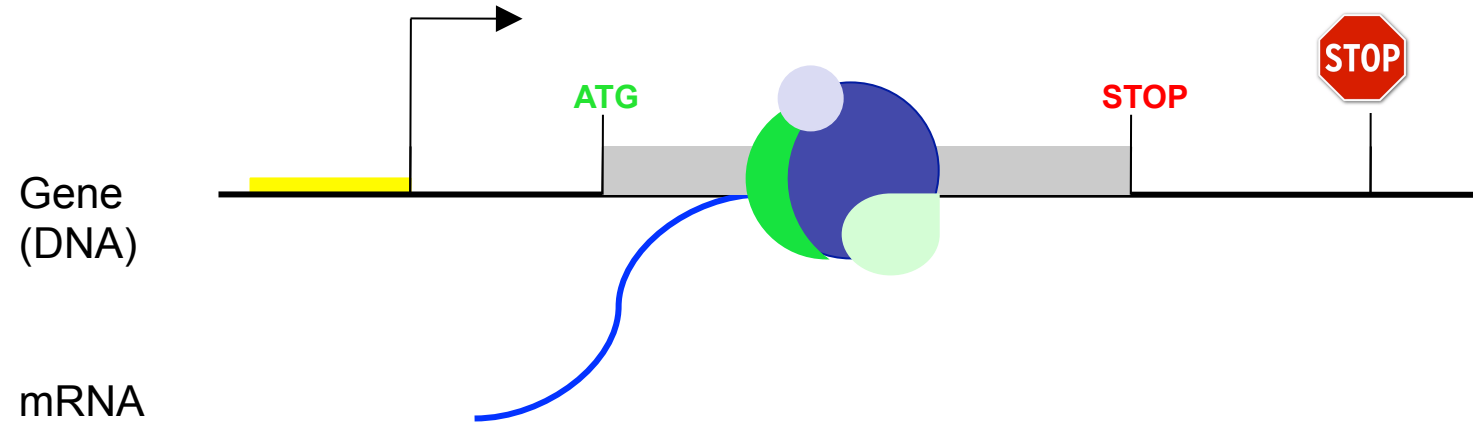
promoter

ATG

open reading frame

STOP

STOP

Termination of
transcription

Simple Genes: Transcription
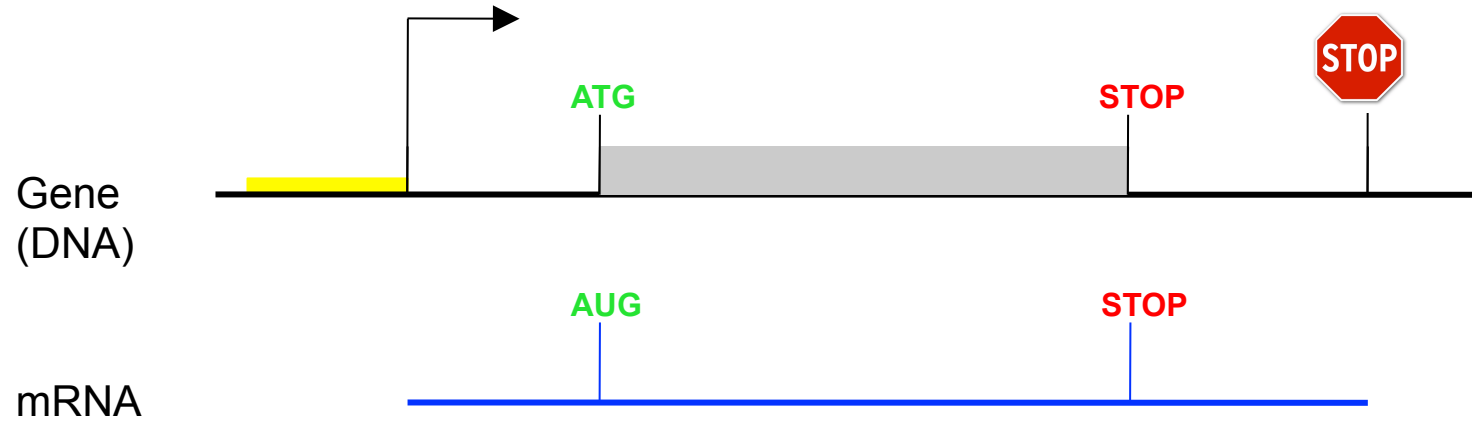
RNA polymerase

Gene (DNA)

Transcription factor

ATG

STOP

STOP

Simple Genes: Transcription

Gene
(DNA)

ATG

STOP

STOP

Simple Genes: Transcription

Gene
(DNA)

ATG

STOP

STOP

mRNA

Simple Genes

Simple Genes: Translation
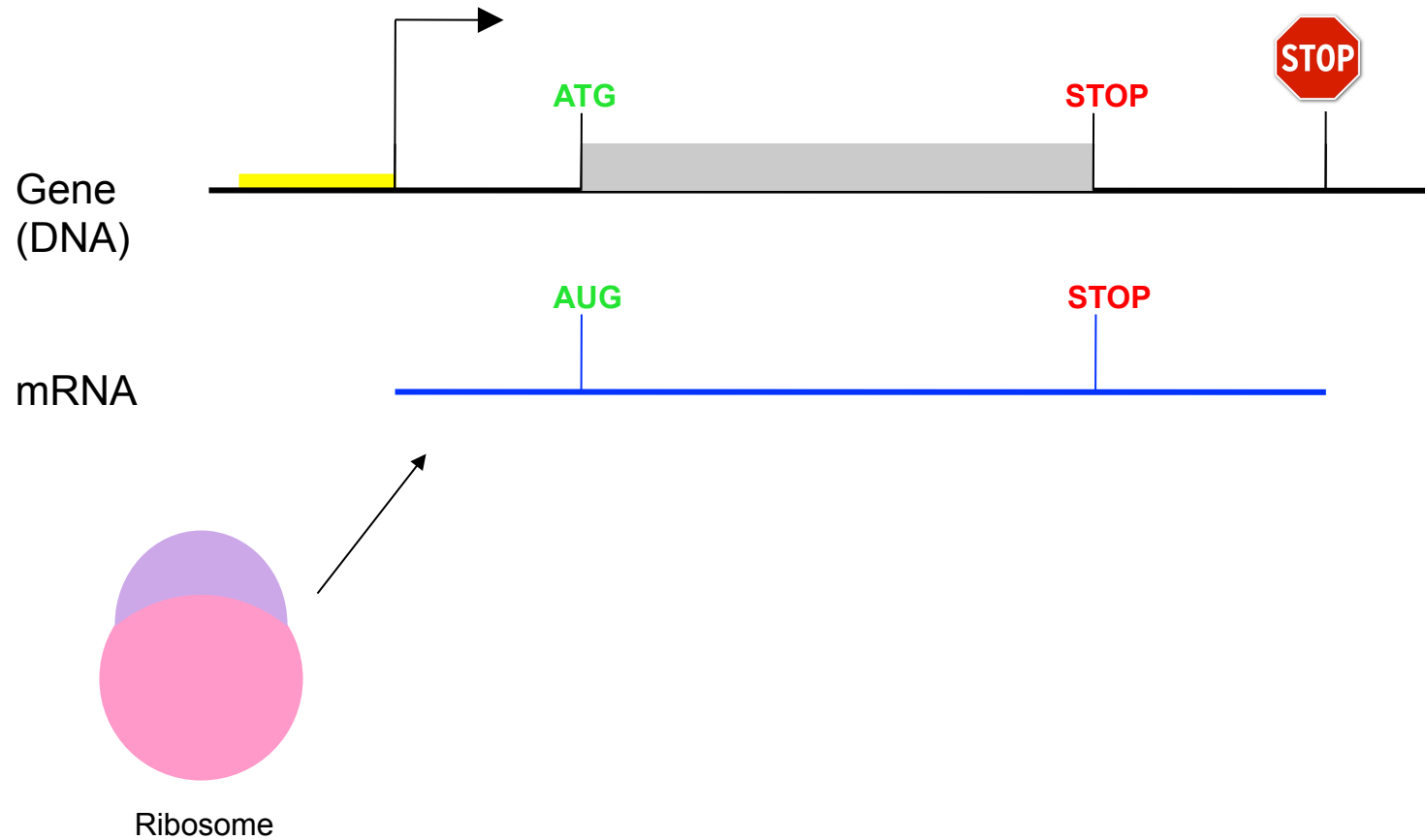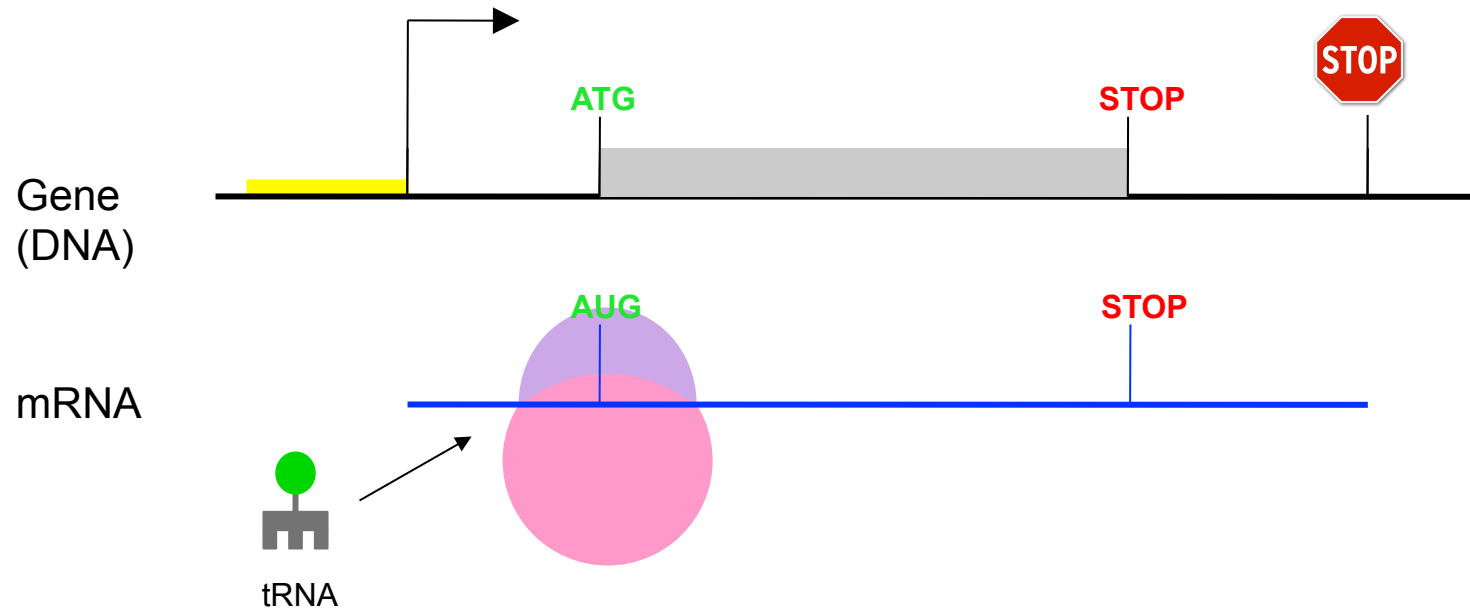
Gene (DNA)

ATG   STOP   STOP

mRNA

AUG   STOP

Ribosome
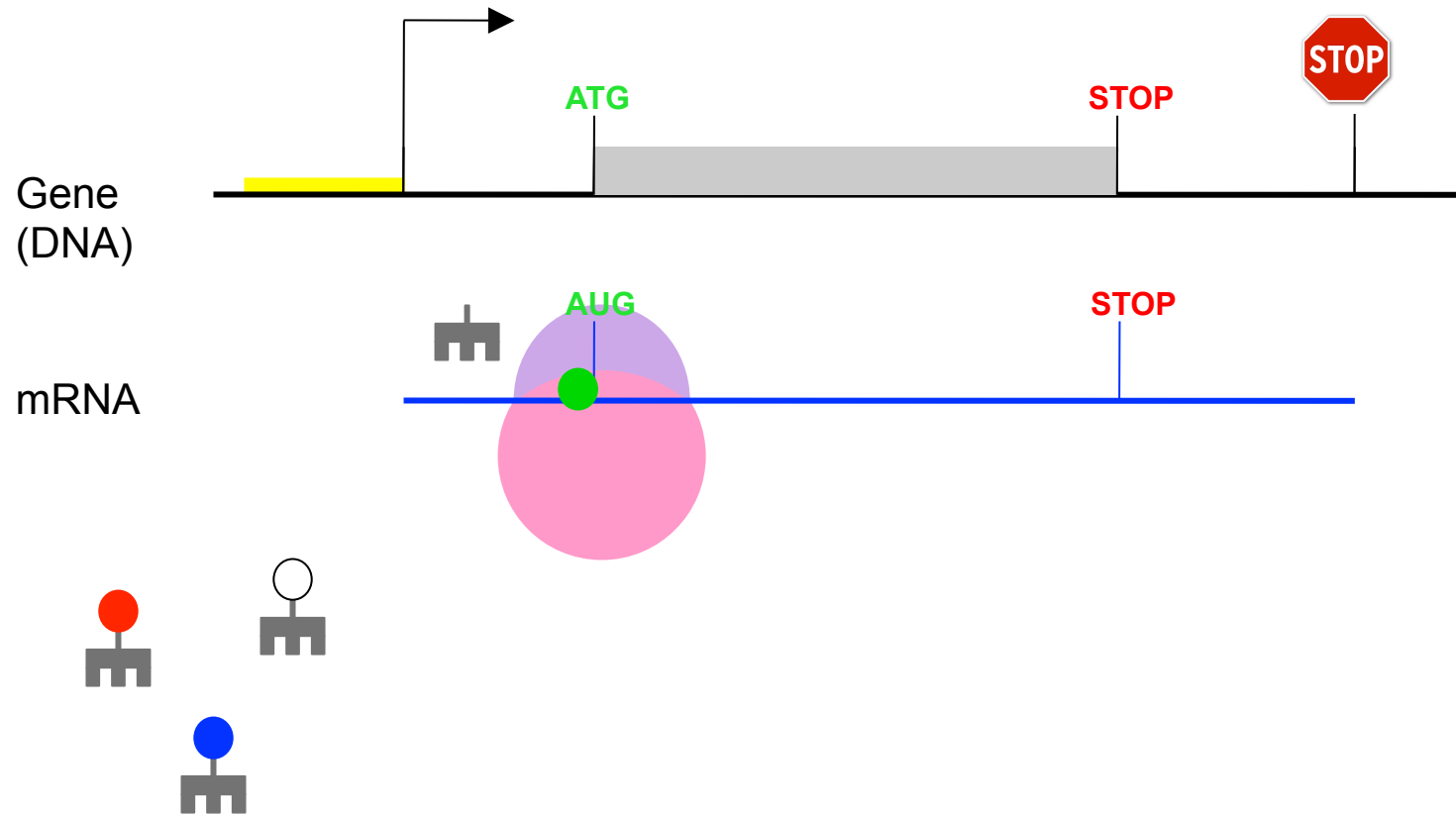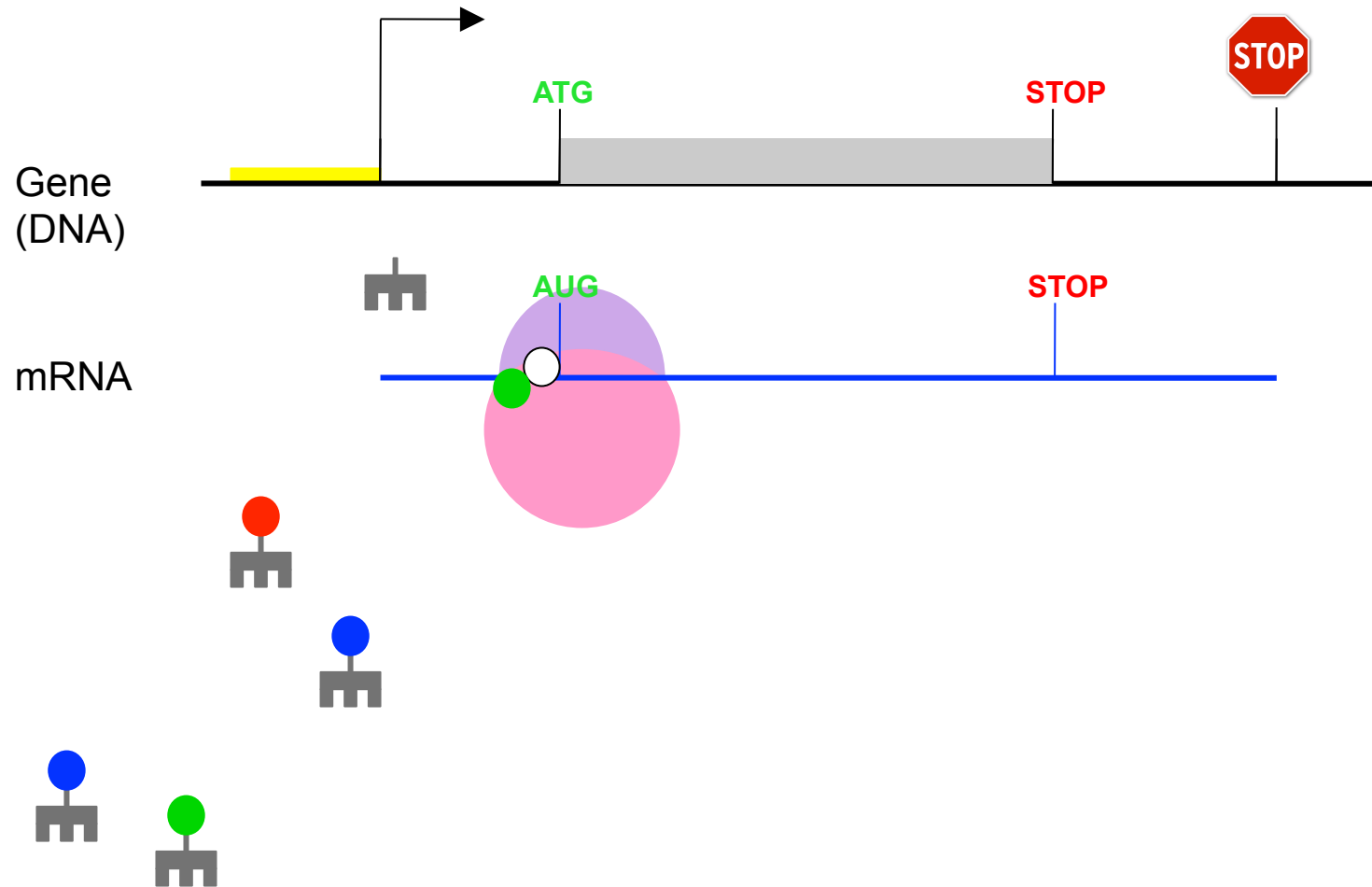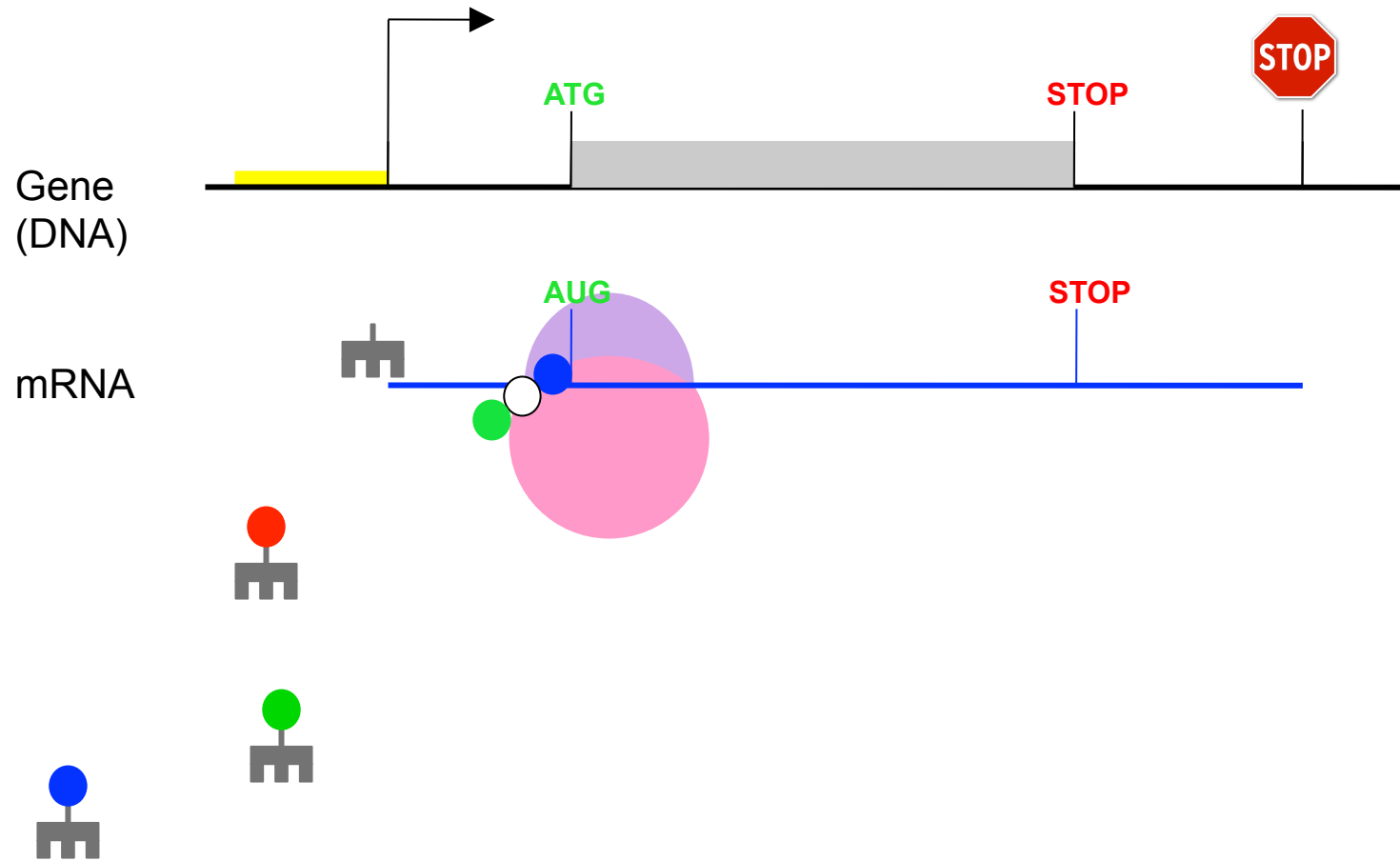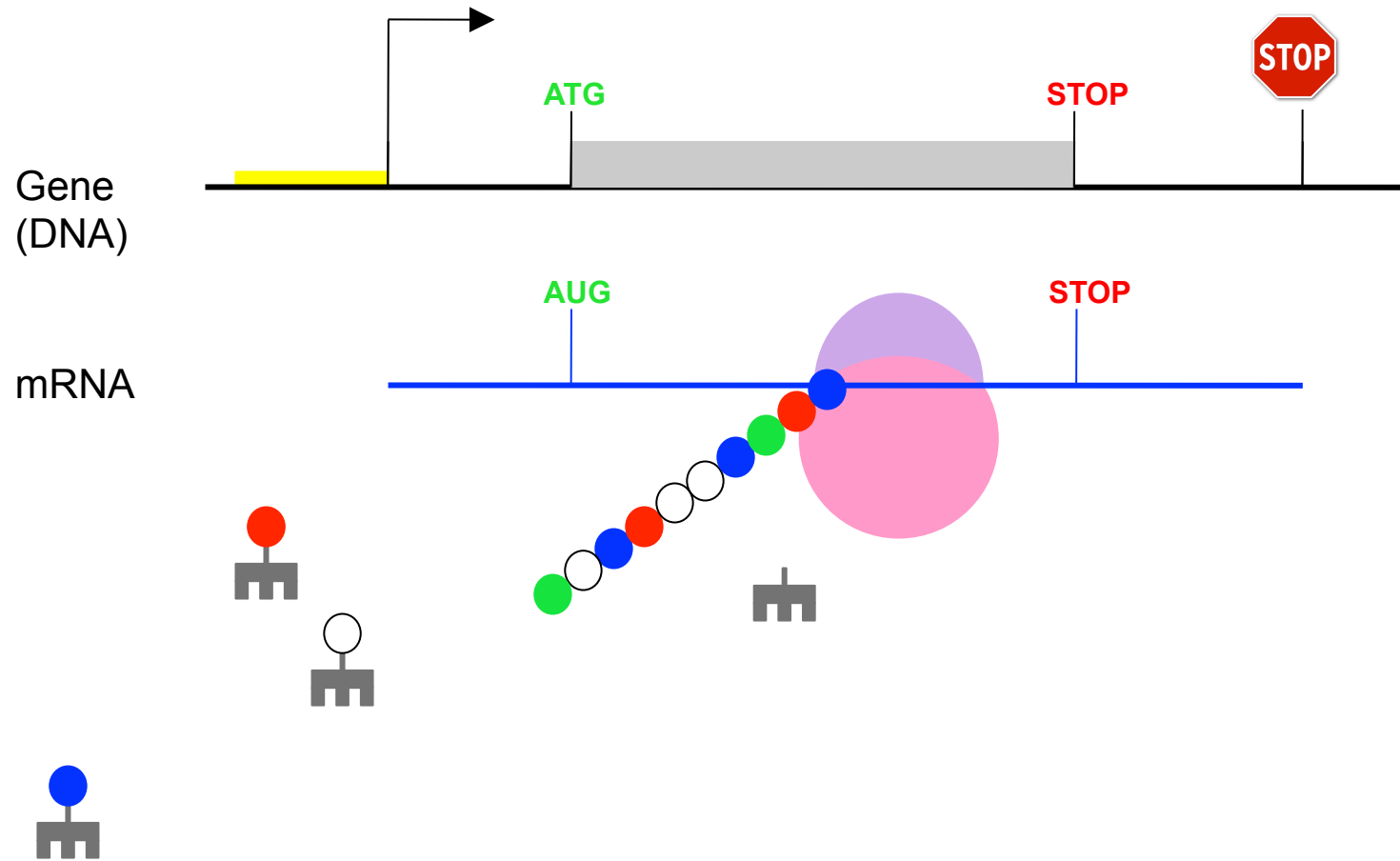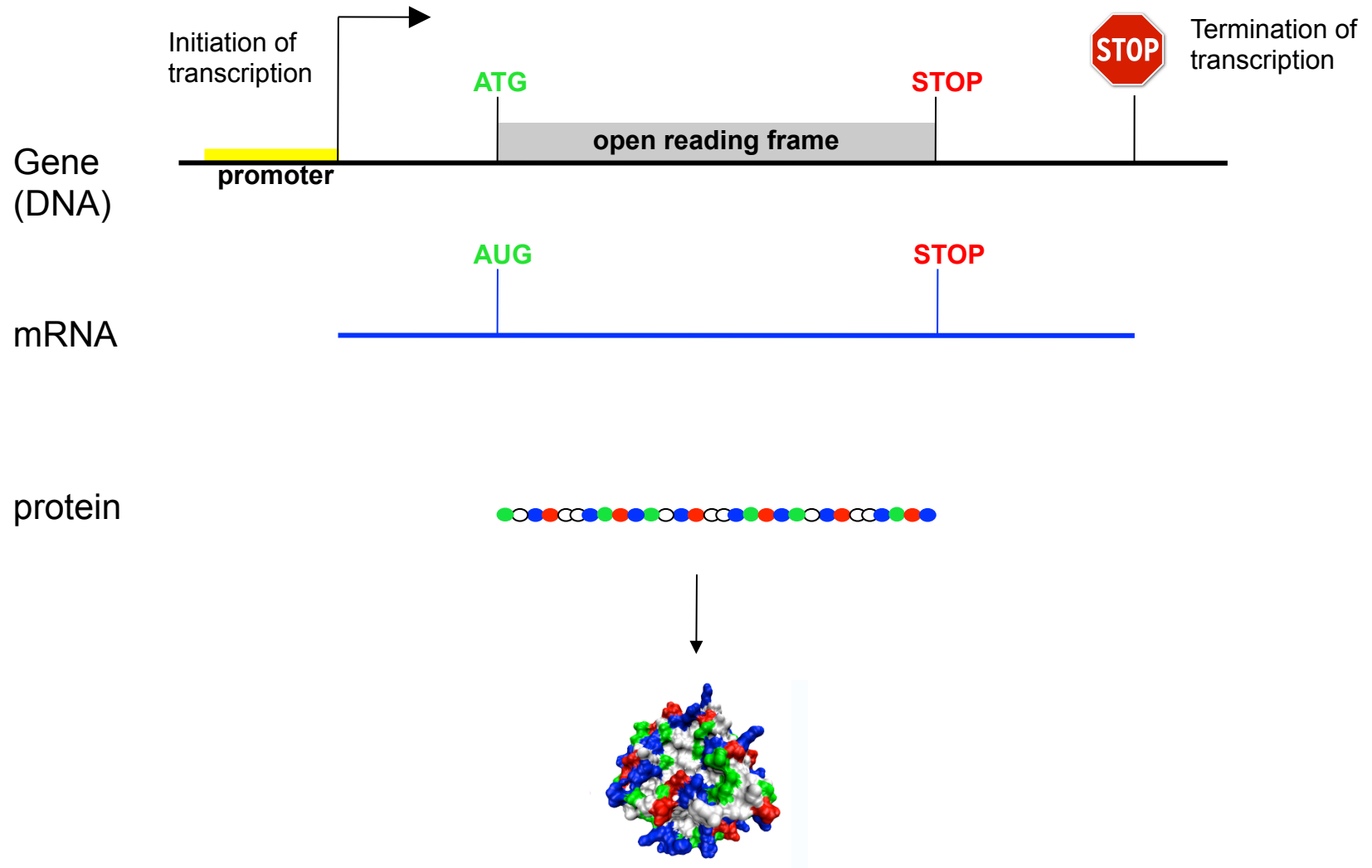
Simple Genes: Translation

Simple Genes: Translation

Simple Genes: Translation

Simple Genes: Translation

Gene (DNA)

mRNA

Simple Genes: Translation

Simple Genes: Translation

Simple Genes

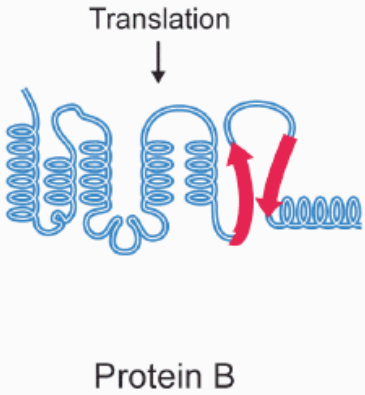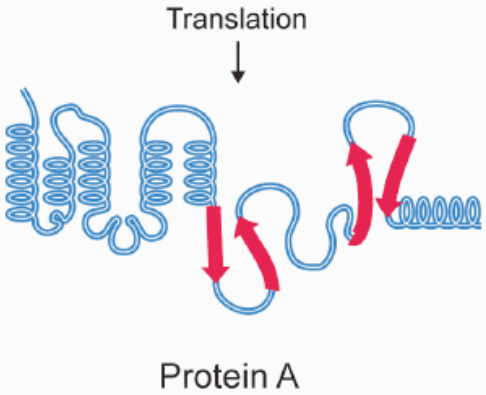Initiation of transcription

Termination of transcription

ATG

STOP

STOP

open reading frame

promoter

Gene (DNA)

AUG

STOP

mRNA

protein

Translation: The Codon Table

Second base of codon

		U		C		A		G		
U	UUU UUC	Phenylalanine phe	UCU UCC	Serine ser	UAU UAC	Tyrosine tyr	UGU UGC	Cysteine cys	U C
	UUA UUG	Leucine leu	UCA UCG		UAA UAG	STOP codon	UGA UGG	STOP codon Tryptonphan trp	A G
C	CUU CUC	Leucine leu	CCU CCC	Proline pro	CAU CAC	Histidine his	CGU CGC	Arginine arg	U C
	CUA CUG		CCA CCG		CAA CAG	Glutamine gin	CGA CGG		A G
A	AUU AUC	Isoleucine ile	ACU ACC	Threonine thr	AAU AAC	Asparagine asn	AGU AGC	Serine ser	U C
	AUA AUG	Methionine met (start codon)	ACA ACG		AAA AAG	Lysine lys	AGA AGG	Arginine arg	A G
G	GUU GUC	Valine val	GCU GCC	Alanine ala	GAU GAC	Aspartic acid asp	GGU GGC	Glycine gly	U C
	GUA GUG		GCA GCG		GAA GAG	Glutamic acid glu	GGA GGG		A G

First base of codon

Third base of codon

Mosaic Genes: Exons and Introns
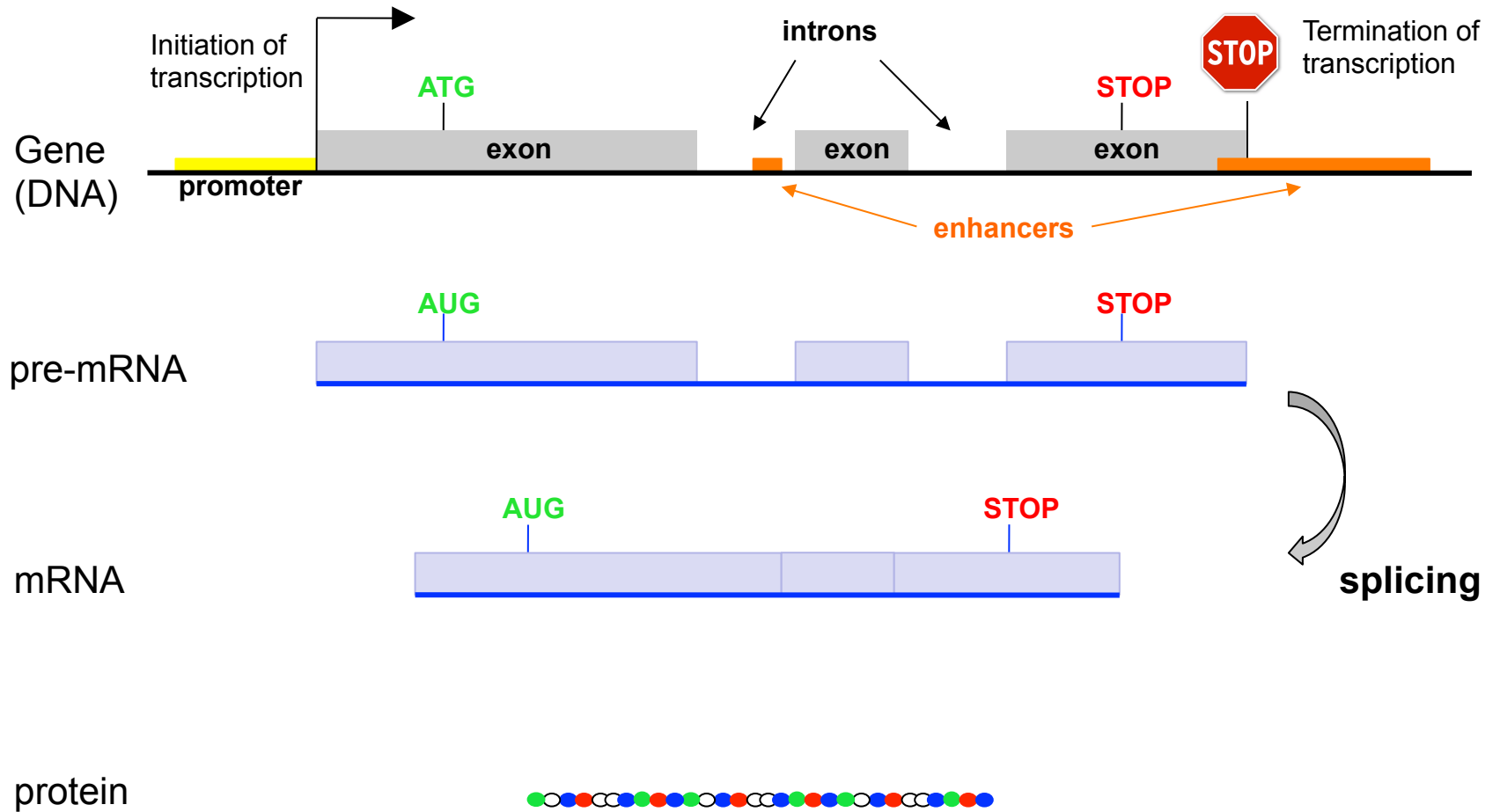
Alternative Splicing
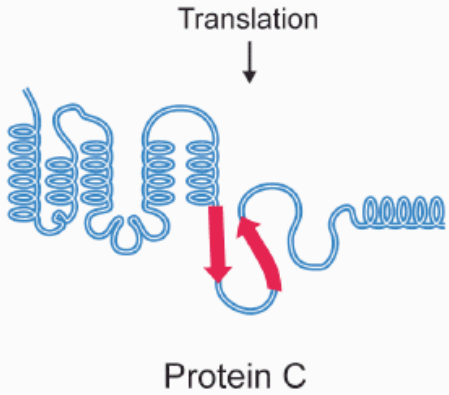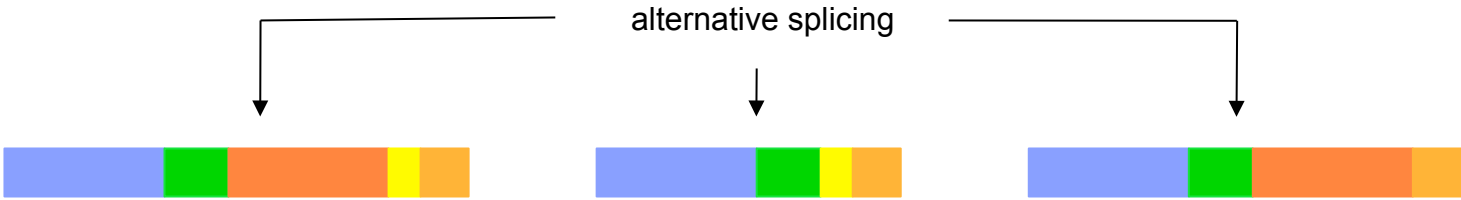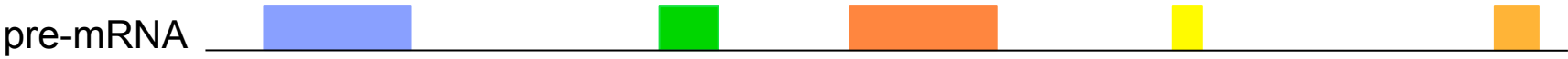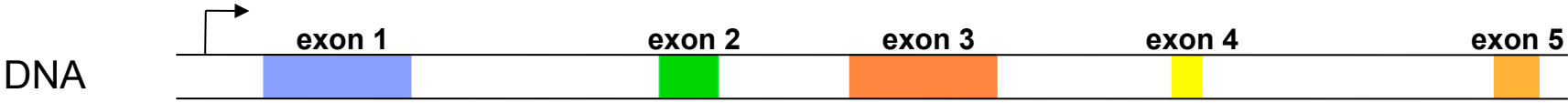
How many human genes?

Ensembl! 56

23,621 protein coding genes

73,516 protein coding transcripts

12,346 pseudogenes

6,407 RNA genes

Only about 2% of the human genome is covered by protein-coding exons!

The rest:

- introns

- regulatory sequence

- repeats   'structural' DNA (telomeres, centromeres)

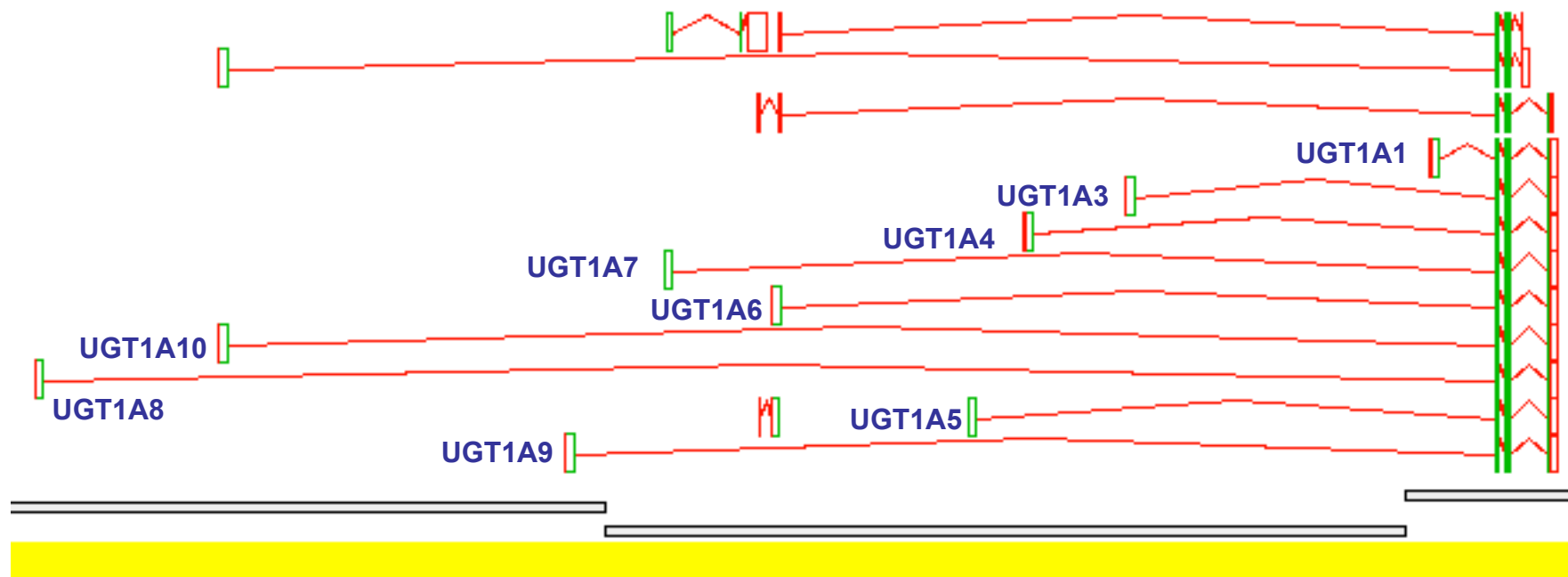   transposons

                        other

What is a gene?

'One gene - one protein' hypothesis?

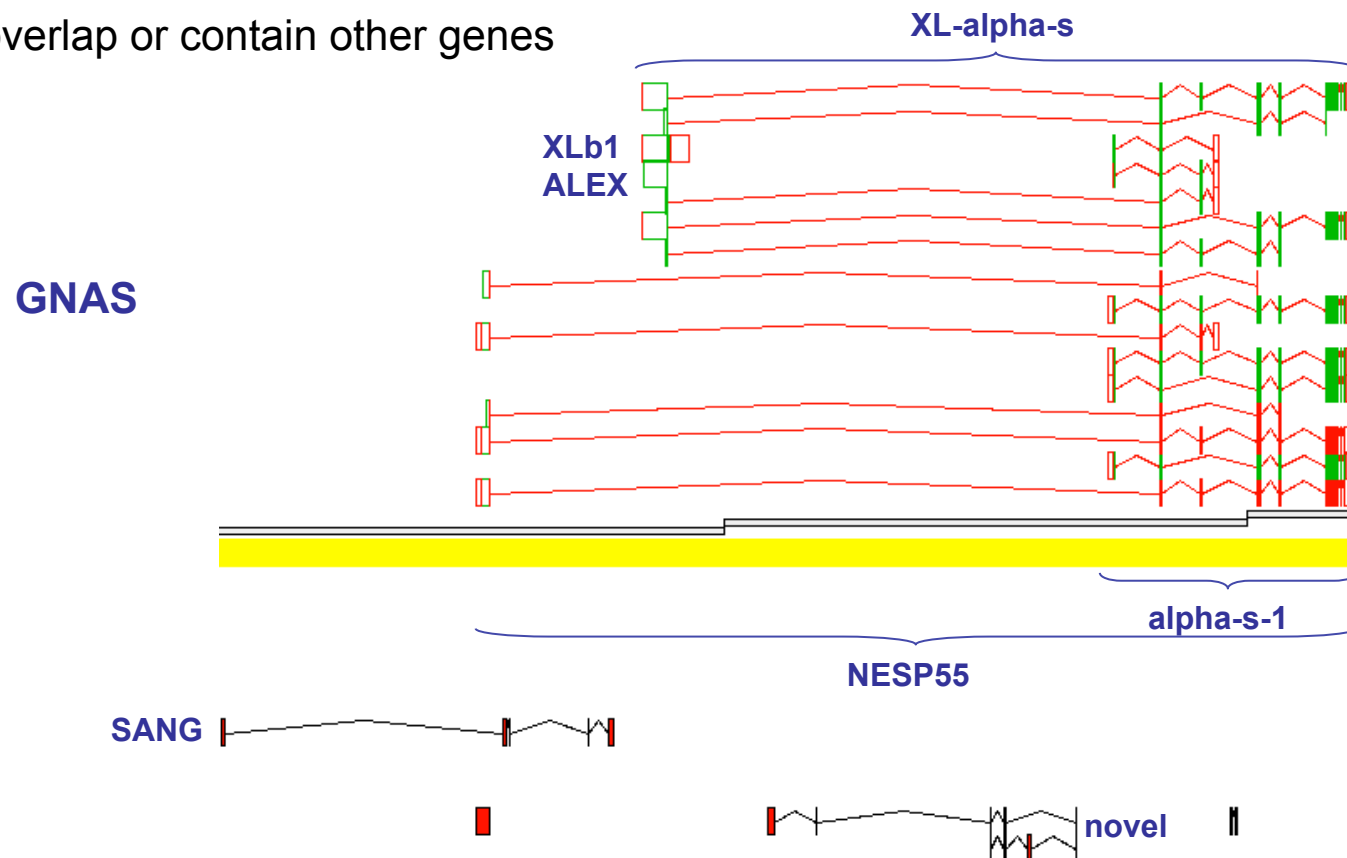• a gene can code for many different proteins with different function

UDP glucuronosyltransferase 1 family

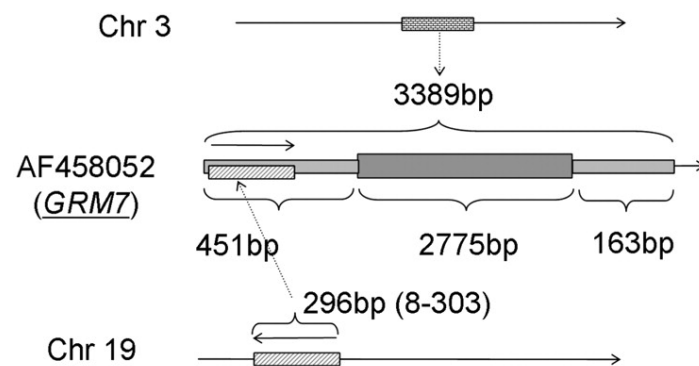# What is a gene?

'One gene - one protein' hypothesis?

- a gene can code for many different proteins with different function

- genes can overlap or contain other genes

What is a gene?

'One gene - one protein' hypothesis?

• a gene can code for many different proteins with different function

• genes can overlap or contain other genes

• splicing can happen between exons of different genes

What is a gene?

'One gene - one protein' hypothesis?

• a gene can code for many different proteins with different function

• genes can overlap or contain other genes

• splicing can happen between exons of different genes

• some (most?) genes don't code for proteins at all
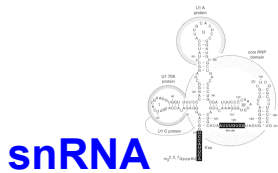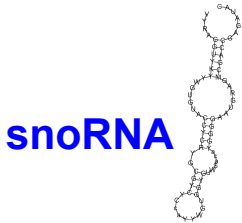
snRNA

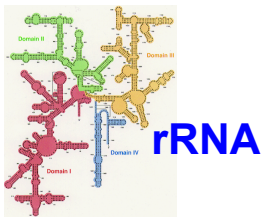rRNA

snoRNA

aRNA

tRNA

SmY

7SL RNA

miRNA

siRNA

RNAse P
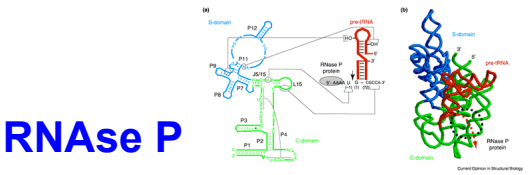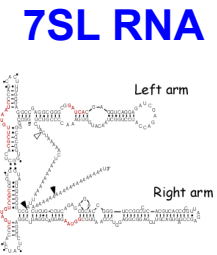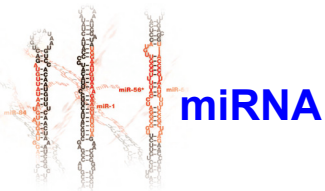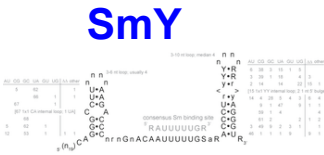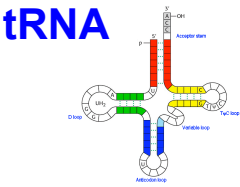
What is a gene?

'One gene - one protein' hypothesis? Not really…

• a gene can code for many different proteins with different function

• genes can overlap or contain other genes

• splicing can happen between exons of different genes

• some (most?) genes don't code for proteins at all

A locatable region of genomic sequence, corresponding to a unit of inheritance, which is associated with regulatory regions, transcribed regions, and/or other functional sequence regions

Helen Pearson (2006) Genetics: What is a gene? Nature 441, 398-441

Mosaic Genes: Exons and Introns

yourgenome.org

Animations
Video interviews
Downloadable activities
Contemporary topics & issues

Introductions to:
DNA, genes and genomes
Human Genome Project