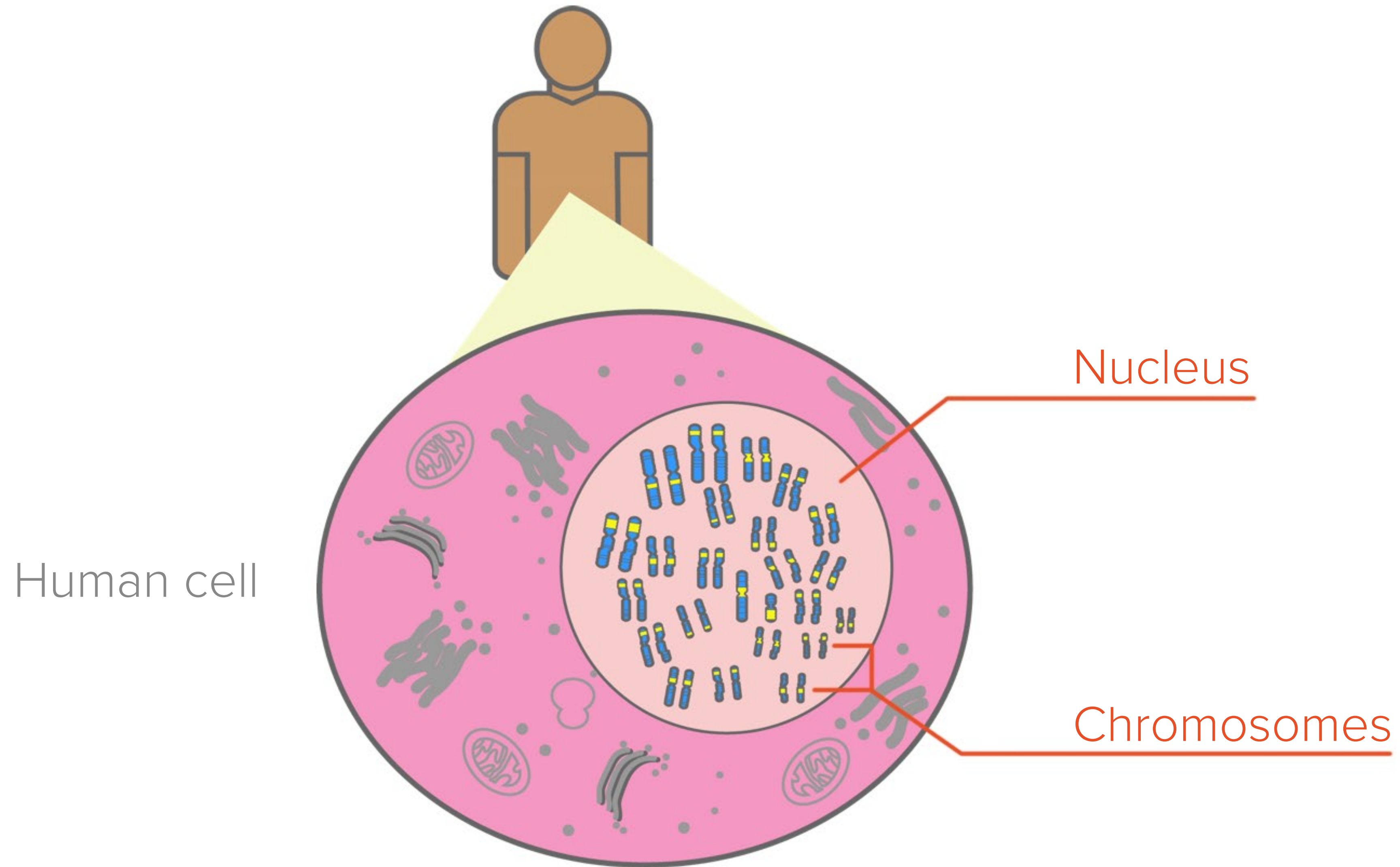


Why? What? and How?

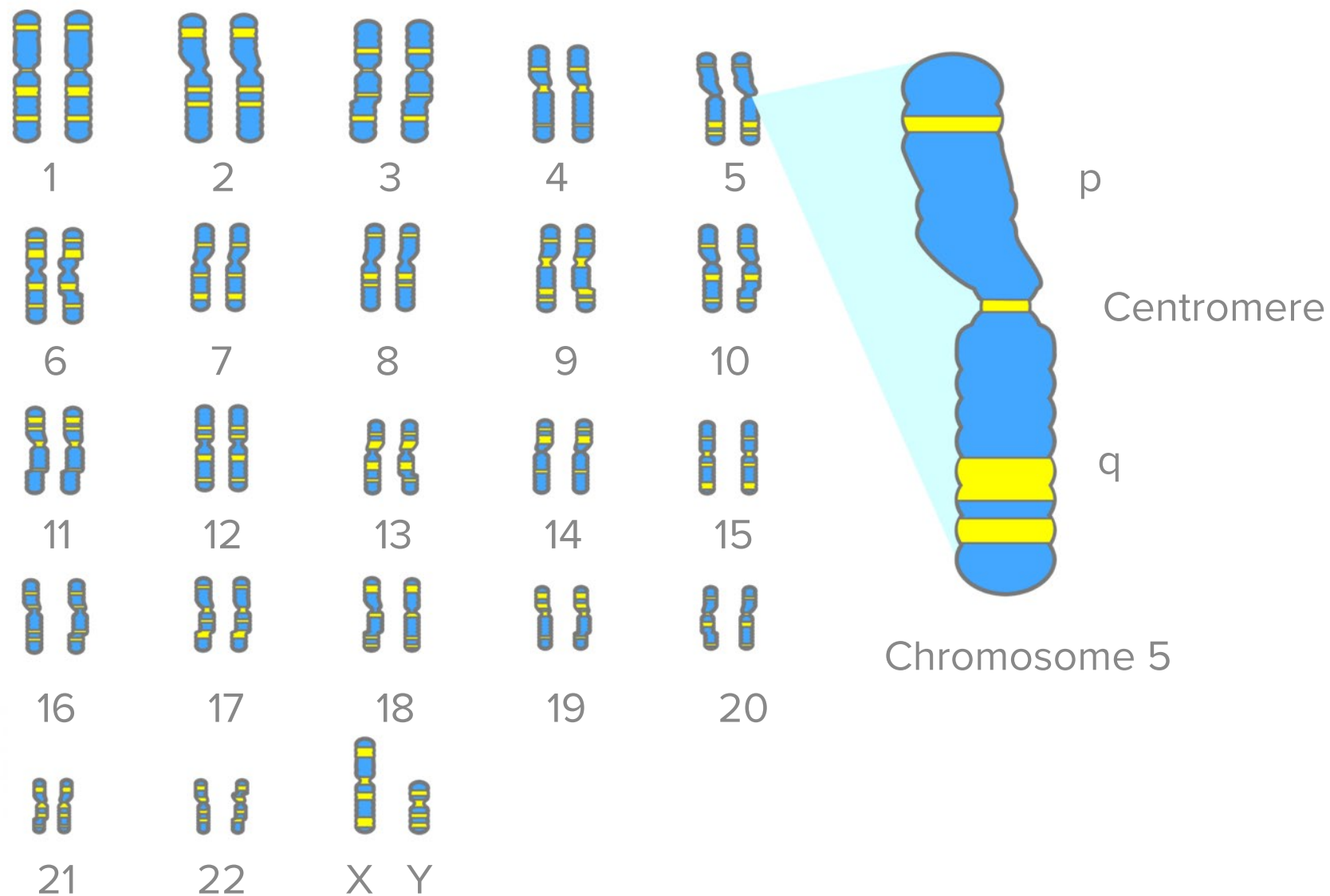
Genome sequencing

- Overview: Basic Concepts of Genome Biology
- Overview of current sequencing technologies:
Second and Third generation platforms
- Major Types of Big Data generated by NGS
- Public on-line resources of Genome Sequencing data

Overview What is the human genome?

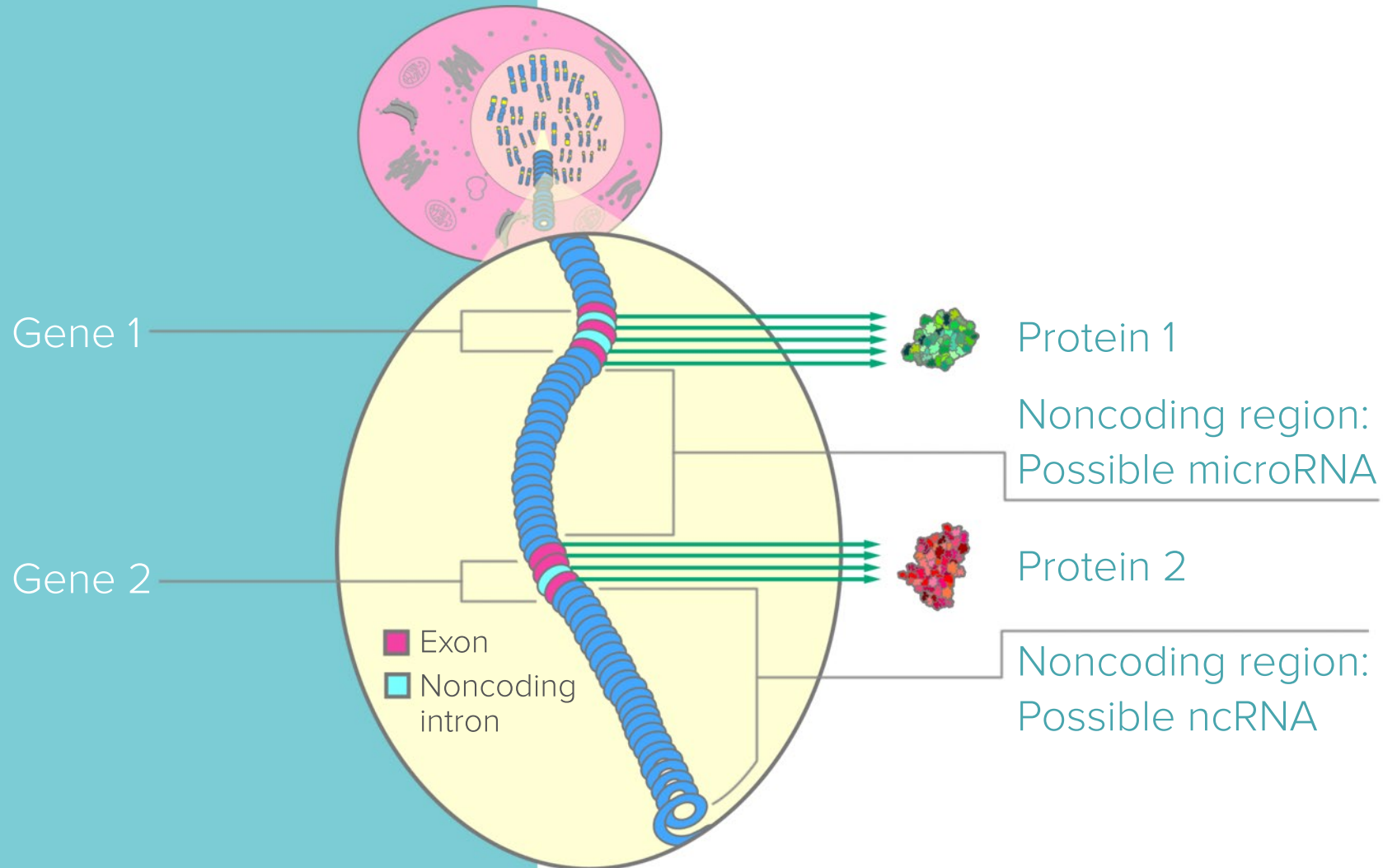


A sample human genome

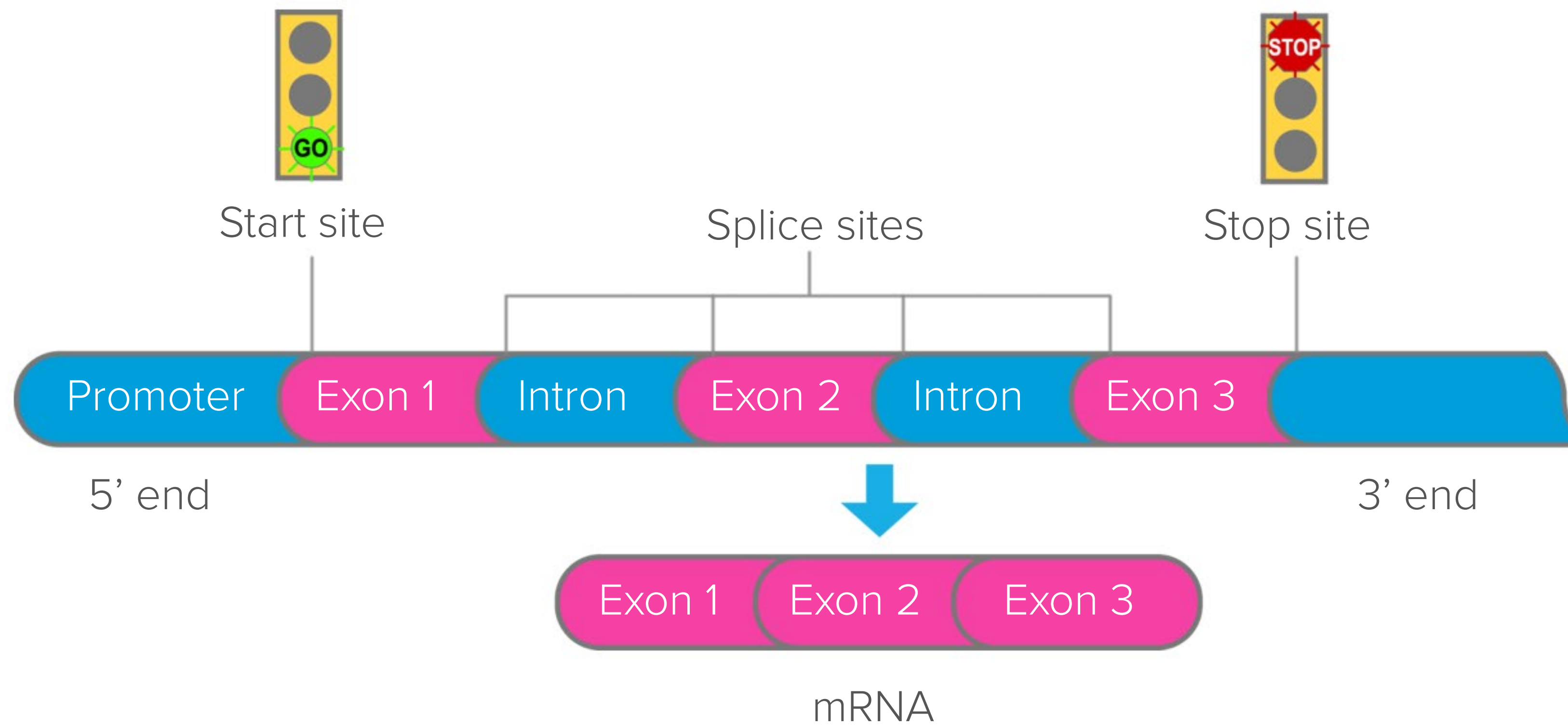


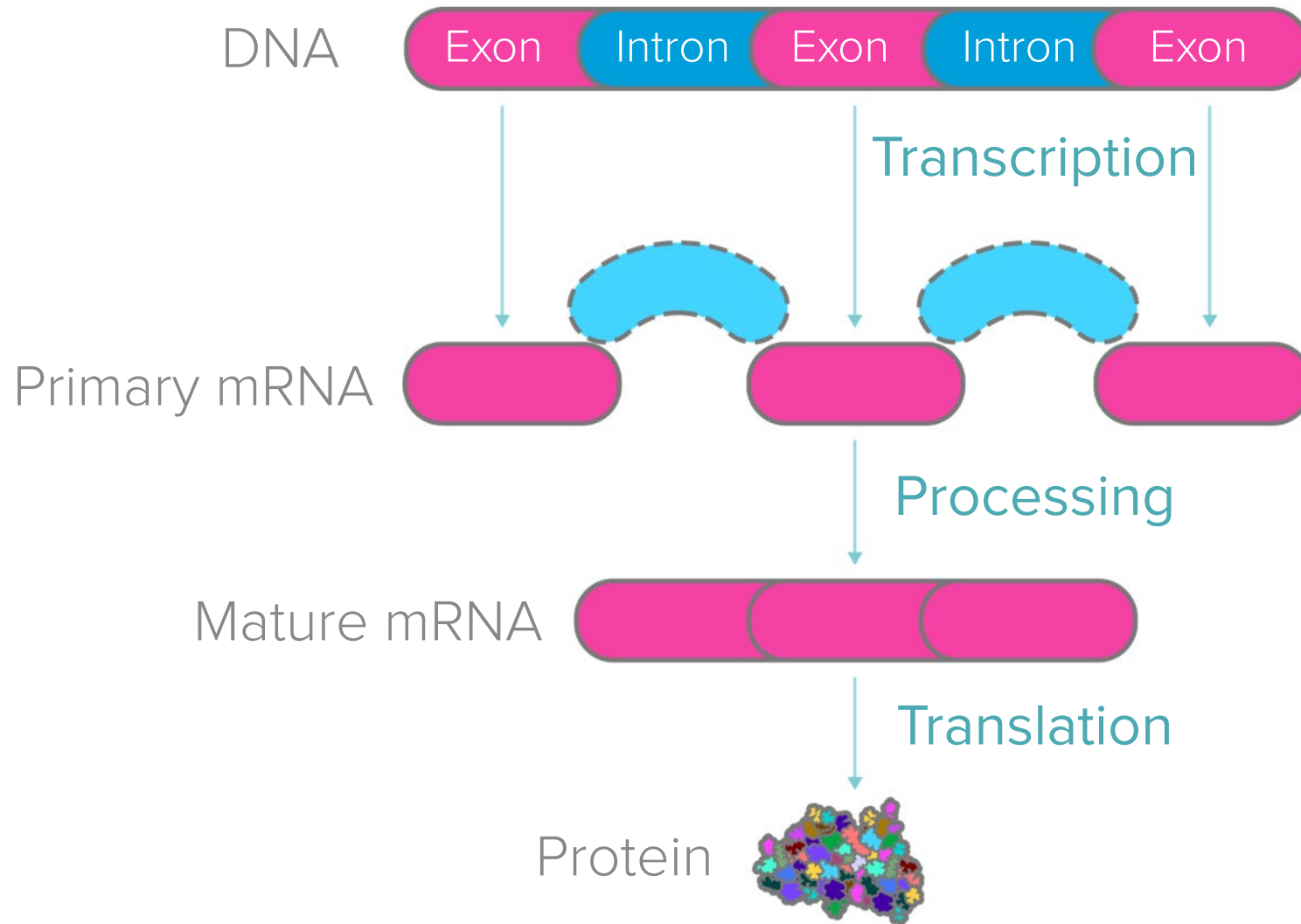
Human genome:
Genes ~2%

The rest: Noncoding regions



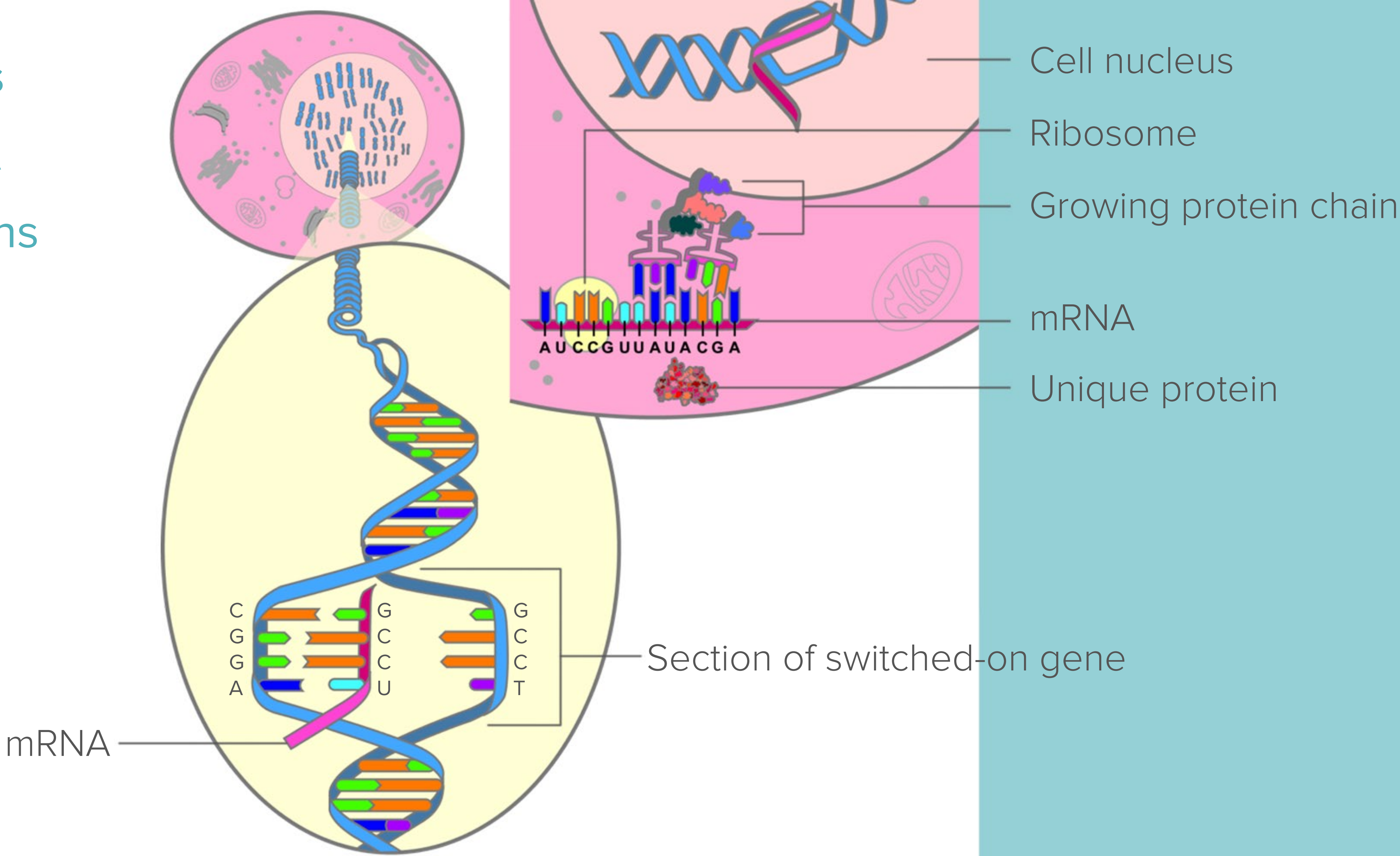
Genes: Keepers of the code





RNA processing
before
translation

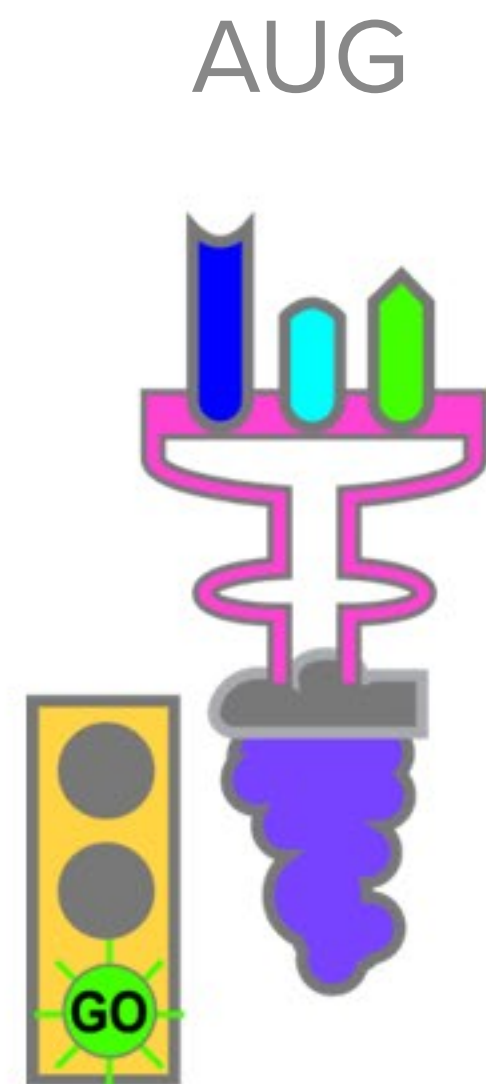
Genes
to
mRNA
to
proteins



Triplet code

A codon is made of 3 mRNA nucleotides
64 codons total

Codon (AUG)
encodes
methionine and
starts translation
of all proteins



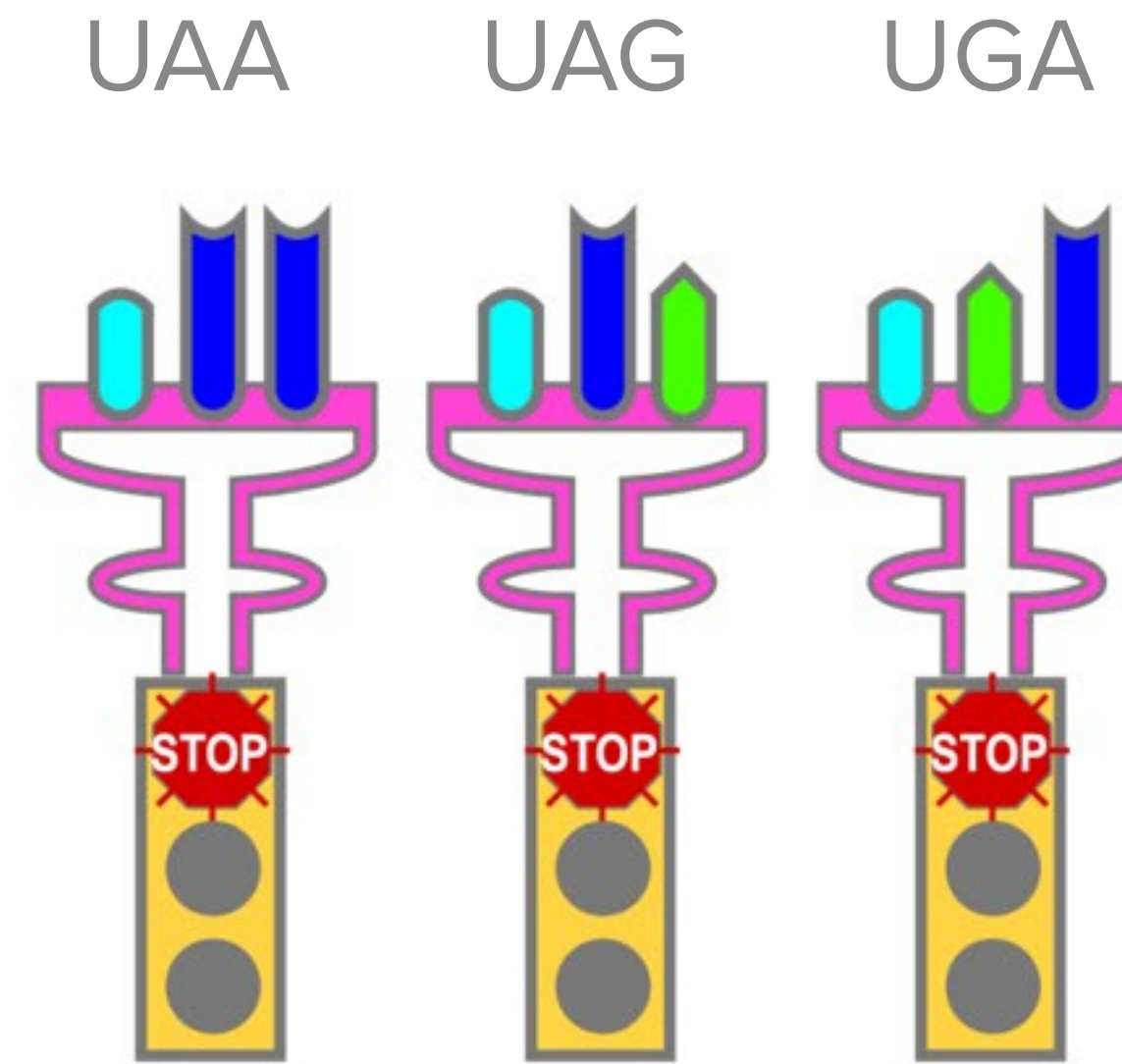
Methionine

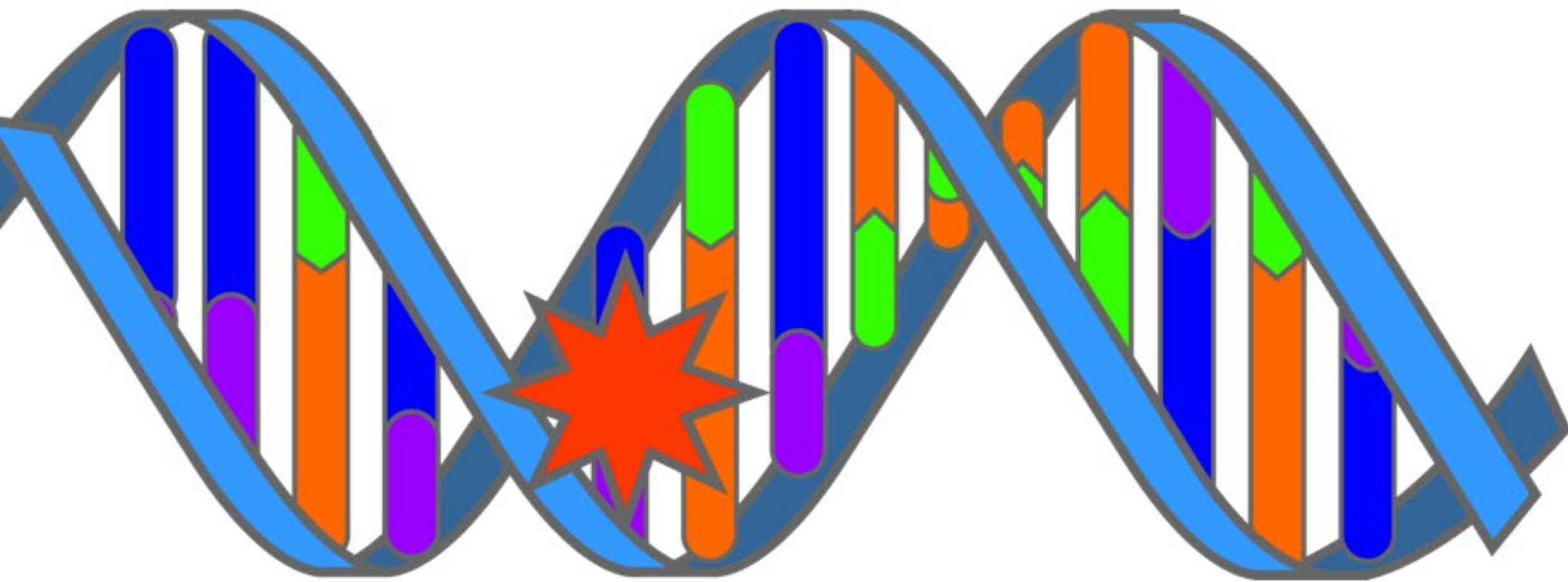
61 codons
encode
20 amino acids
(redundant
code)



Alanine

3 codons stop
protein translation





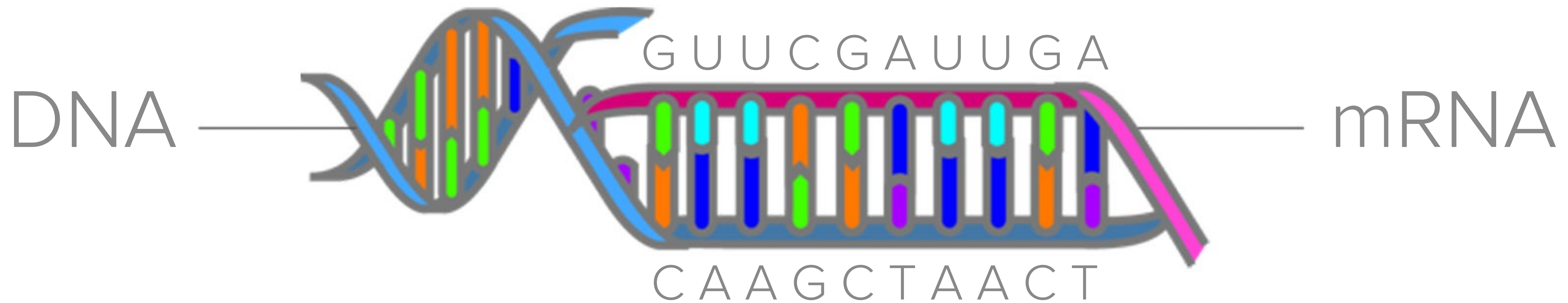
Mutations

A mutation is

- A change in the normal base pair sequence
- Commonly used to define DNA sequence changes that alter protein function

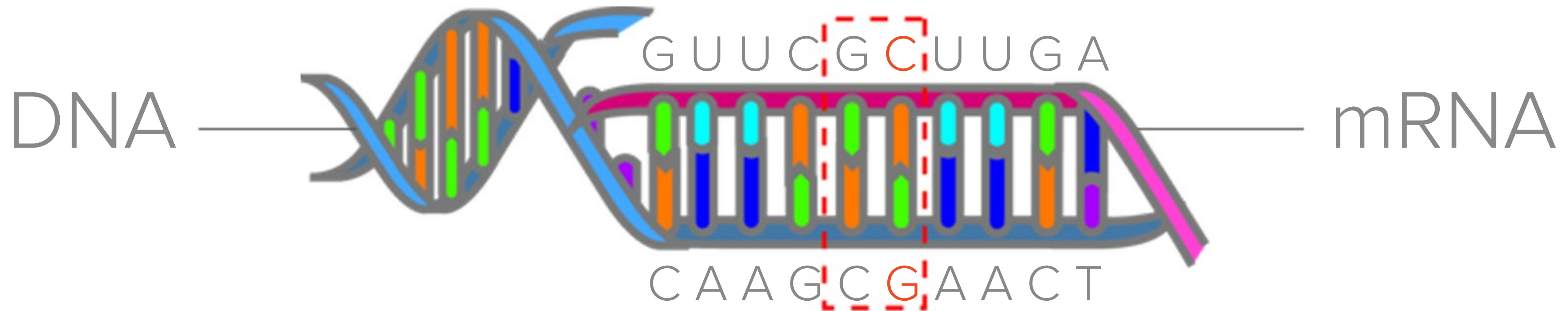
Point Mutations

Normal



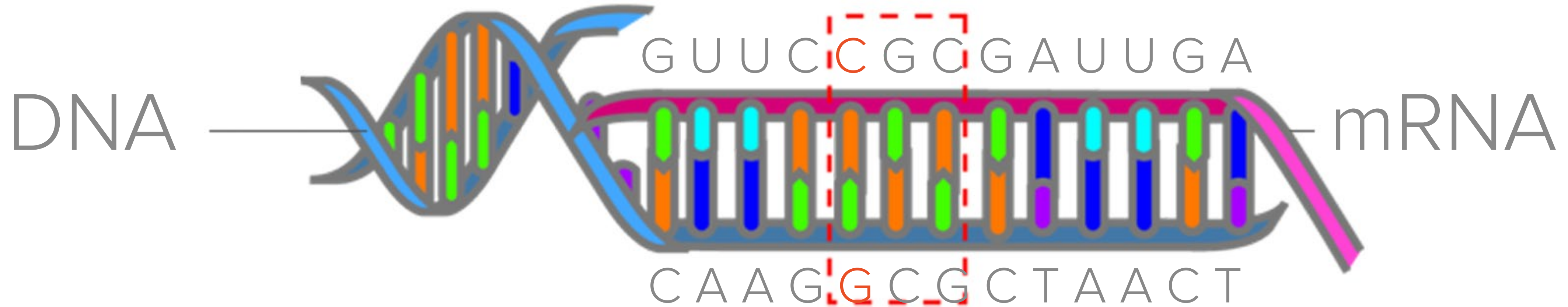
Point Mutations

Missense



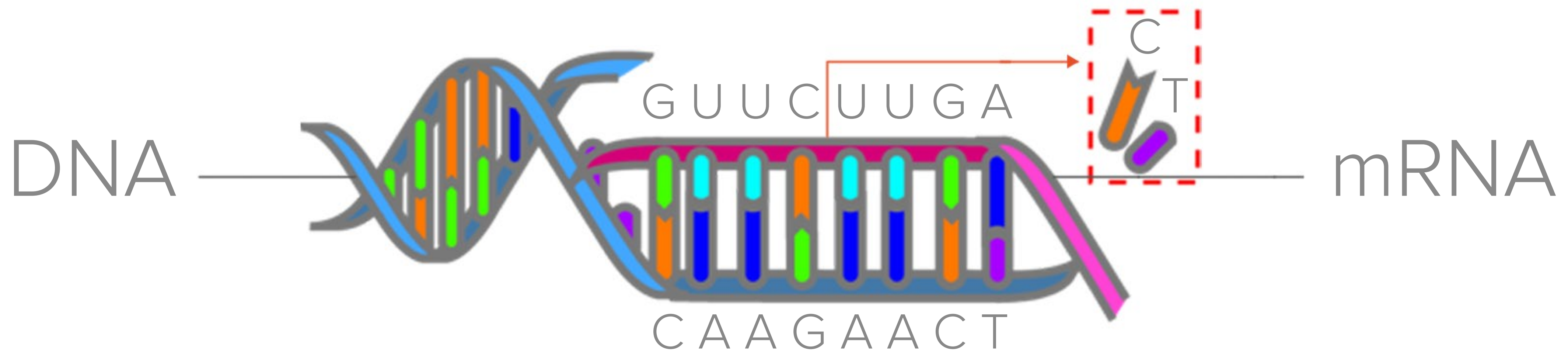
Point Mutations

Frameshift insertion



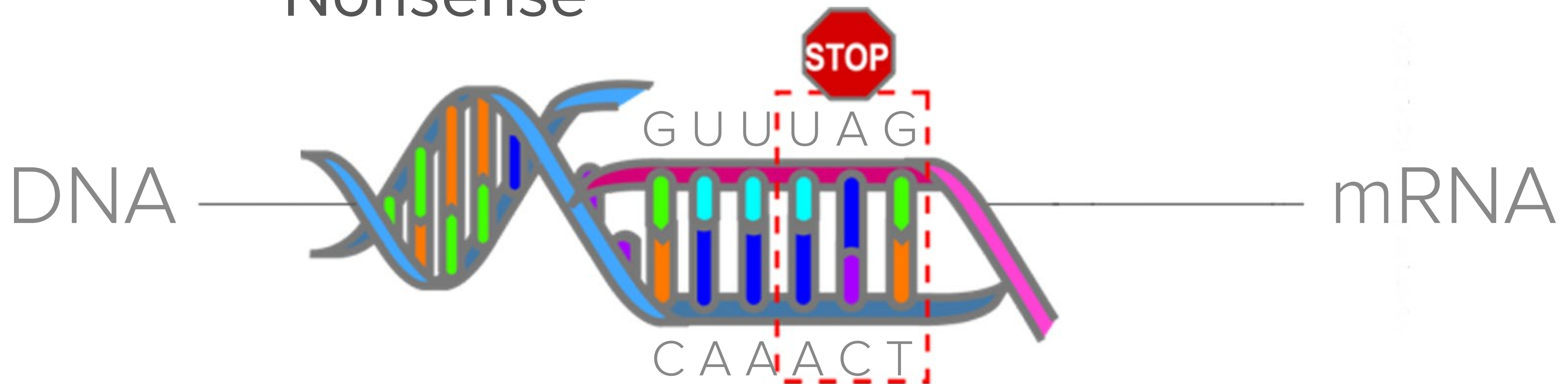
Point Mutations

Frameshift insertion

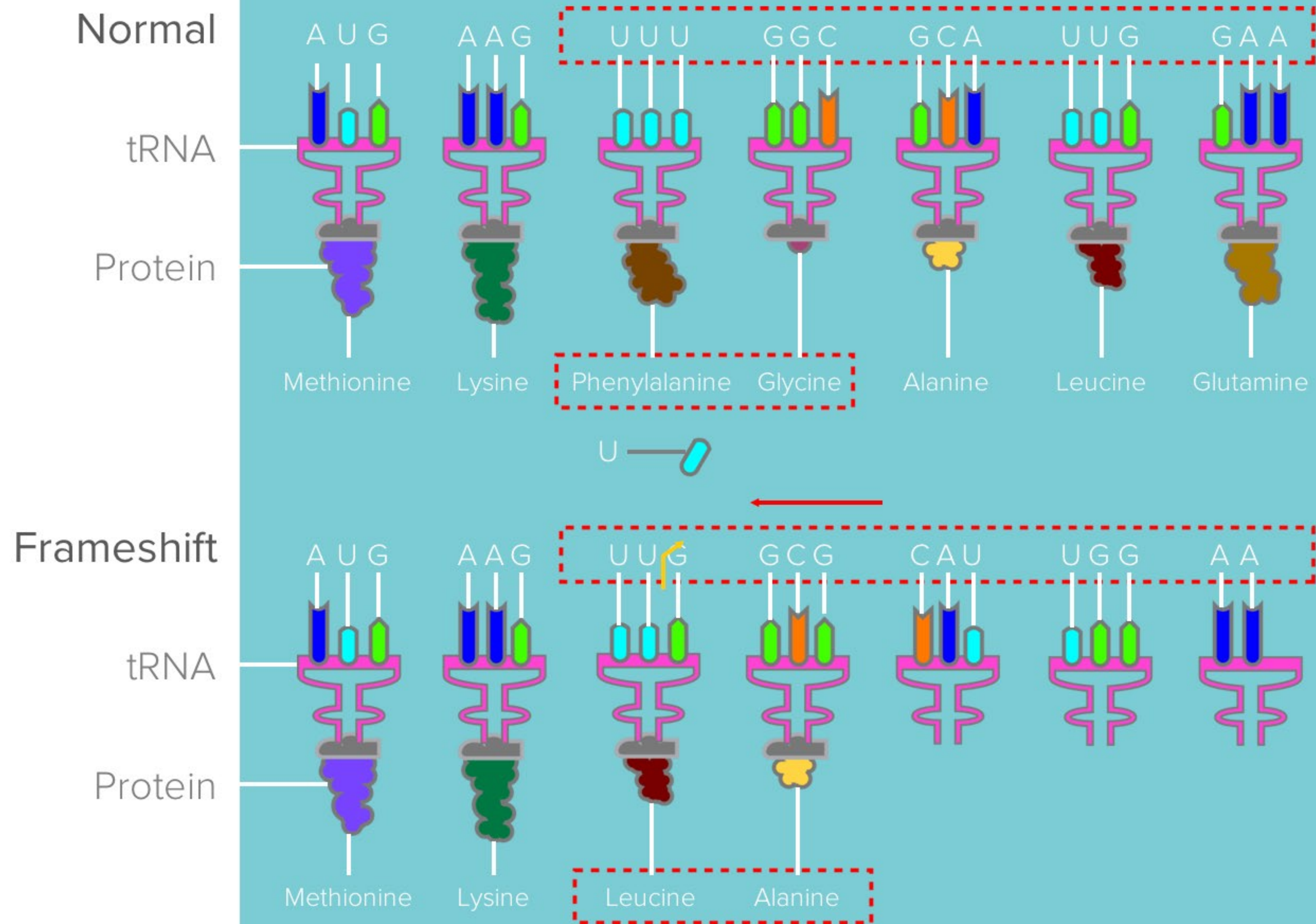


Point Mutations

Nonsense

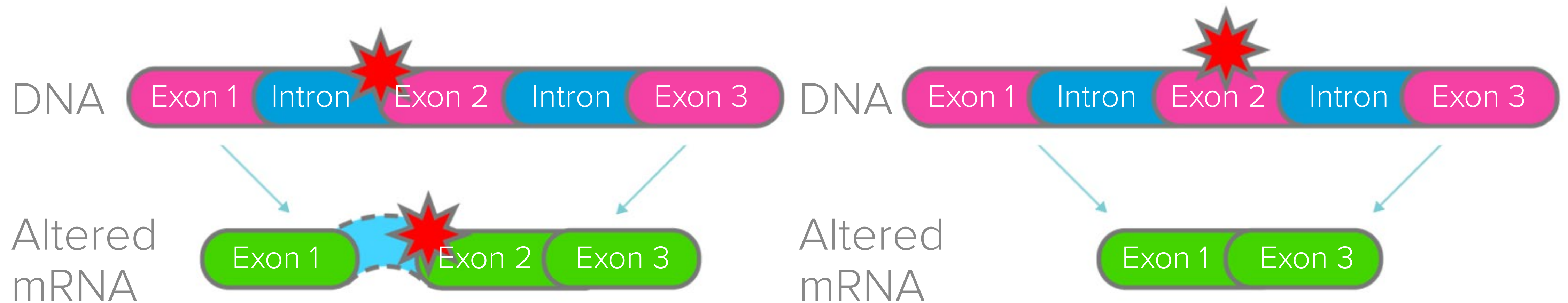


Frameshift mutations

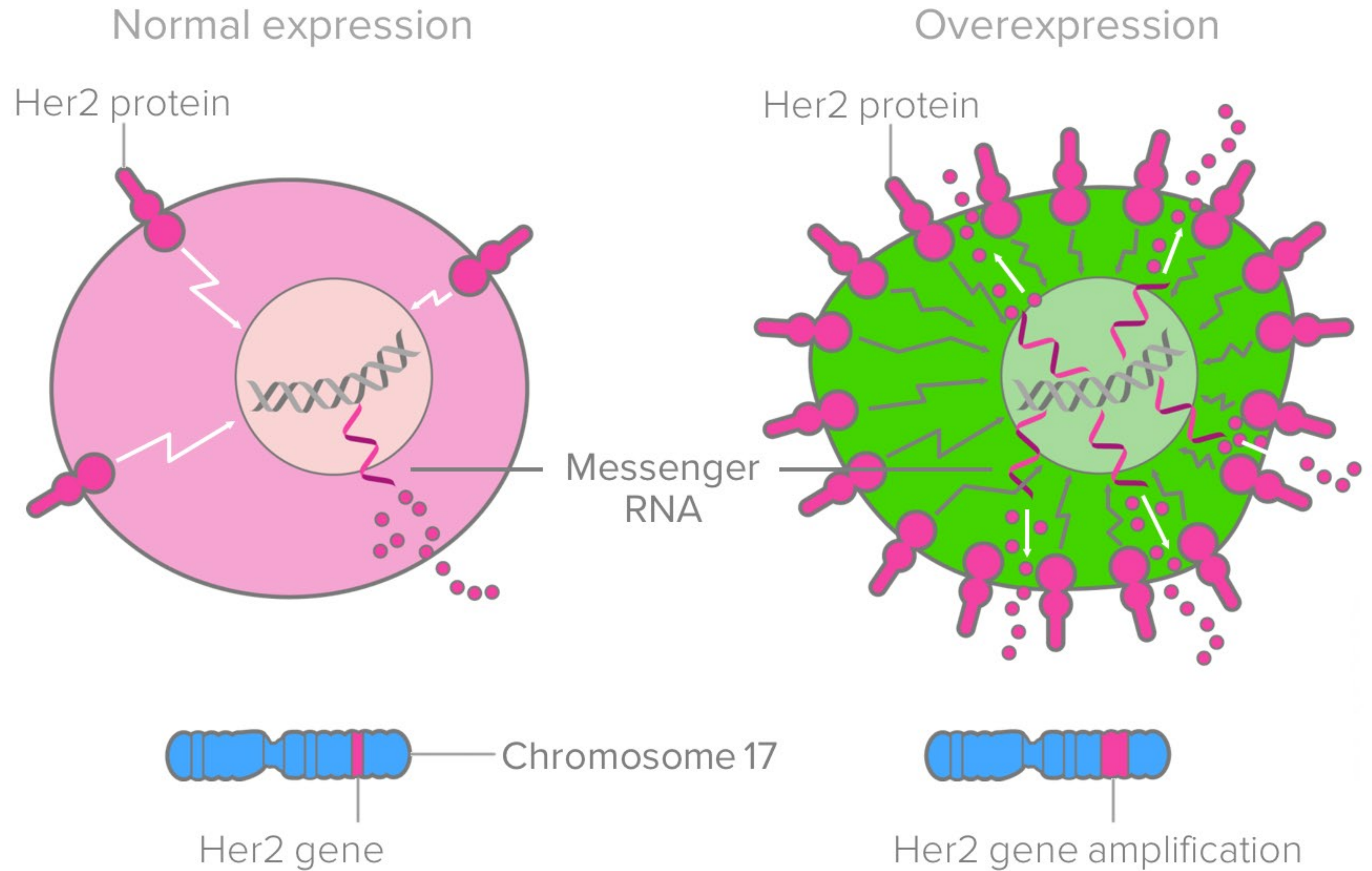


Splice-site mutations

Lombardi | Georgetown



Regulatory mutations

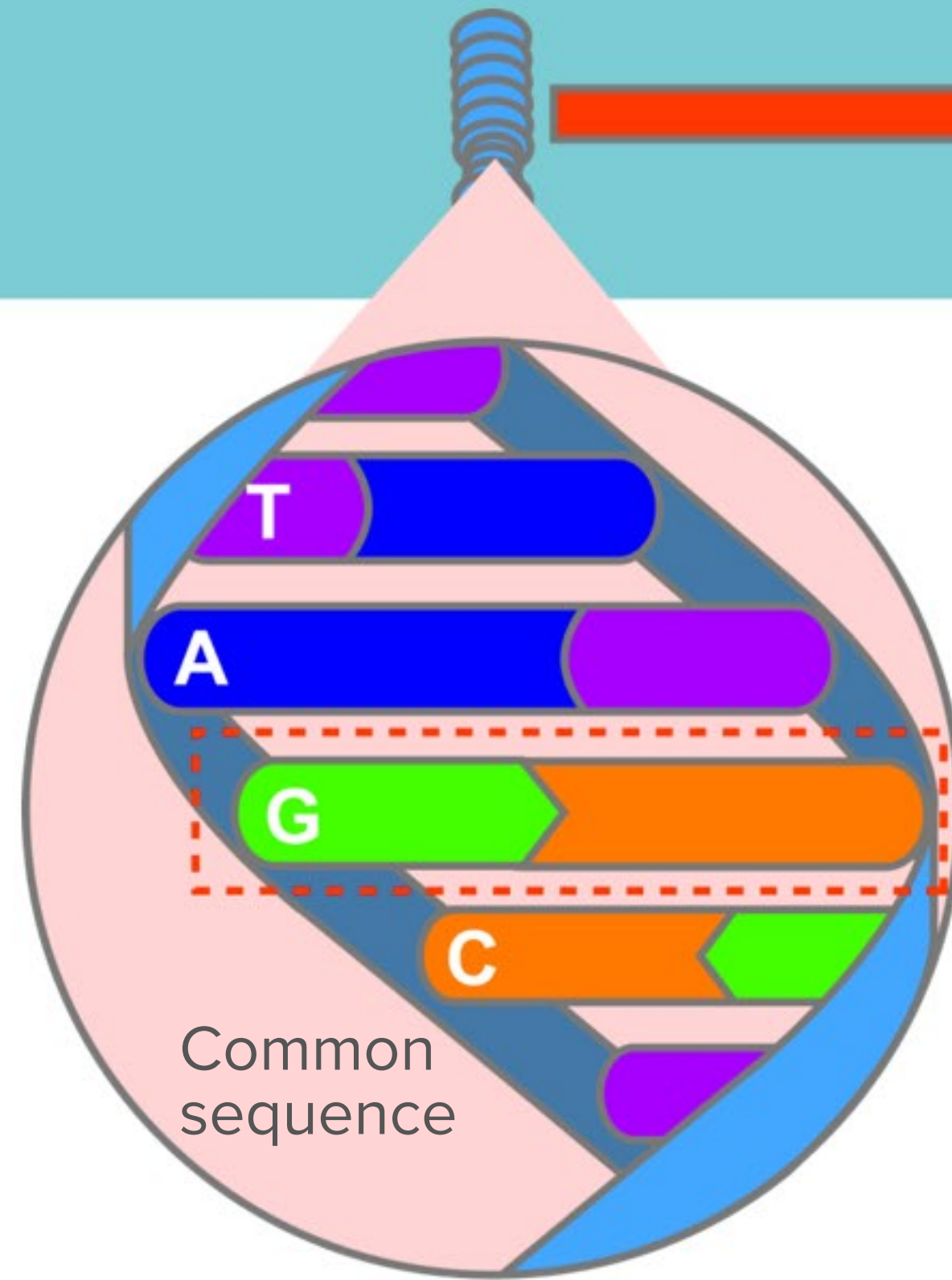


SNPs: Frequently occurring genetic variants

Most of the population

At least 1 percent of the population

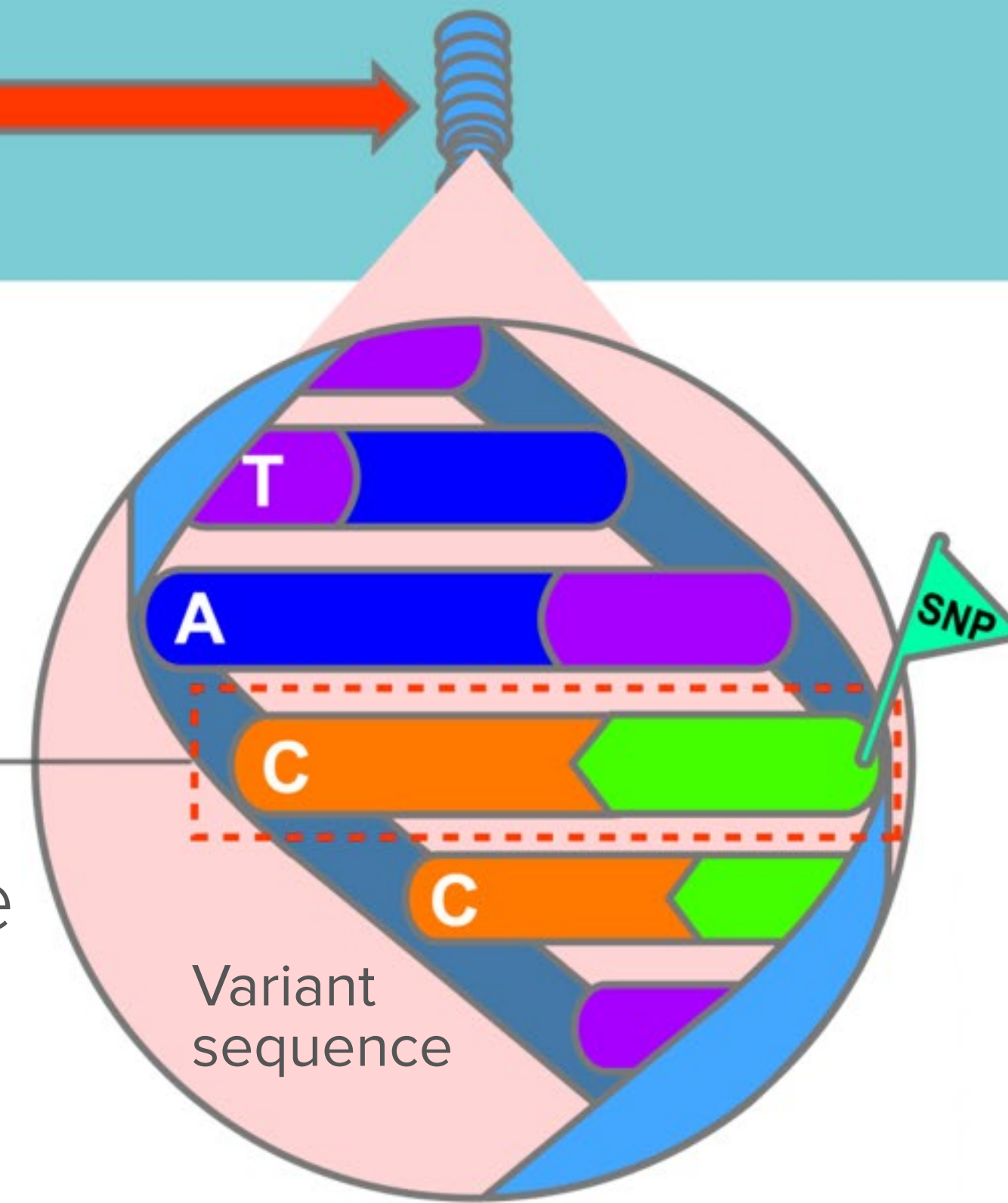
G to C



Functional protein



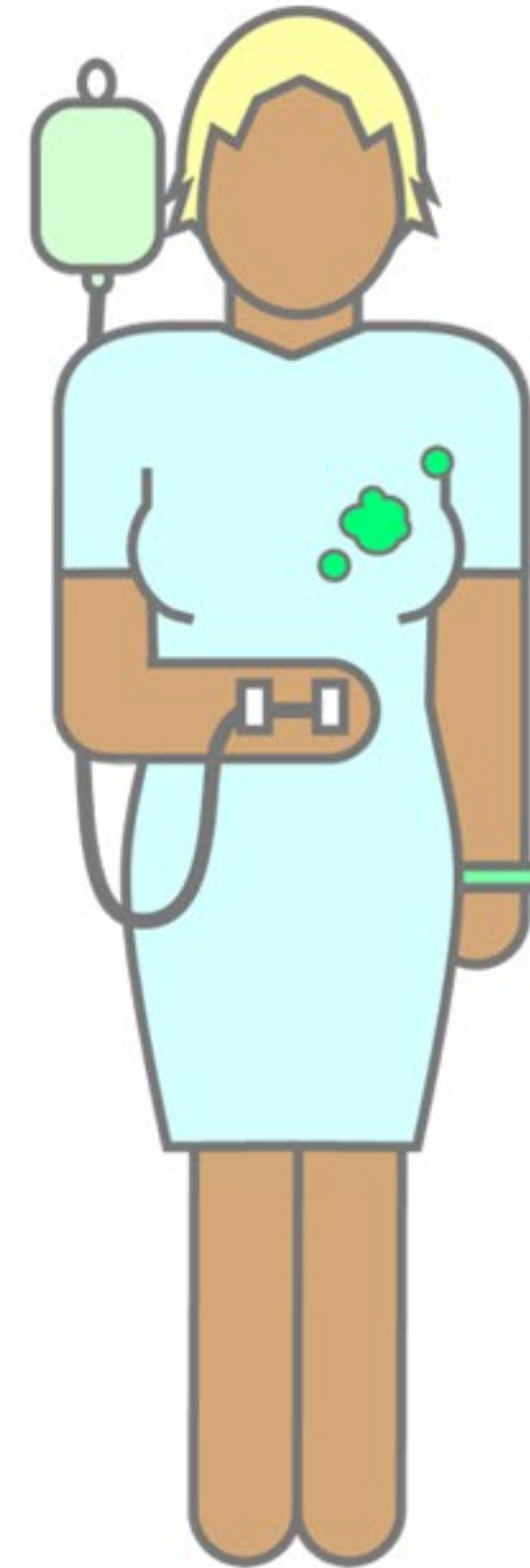
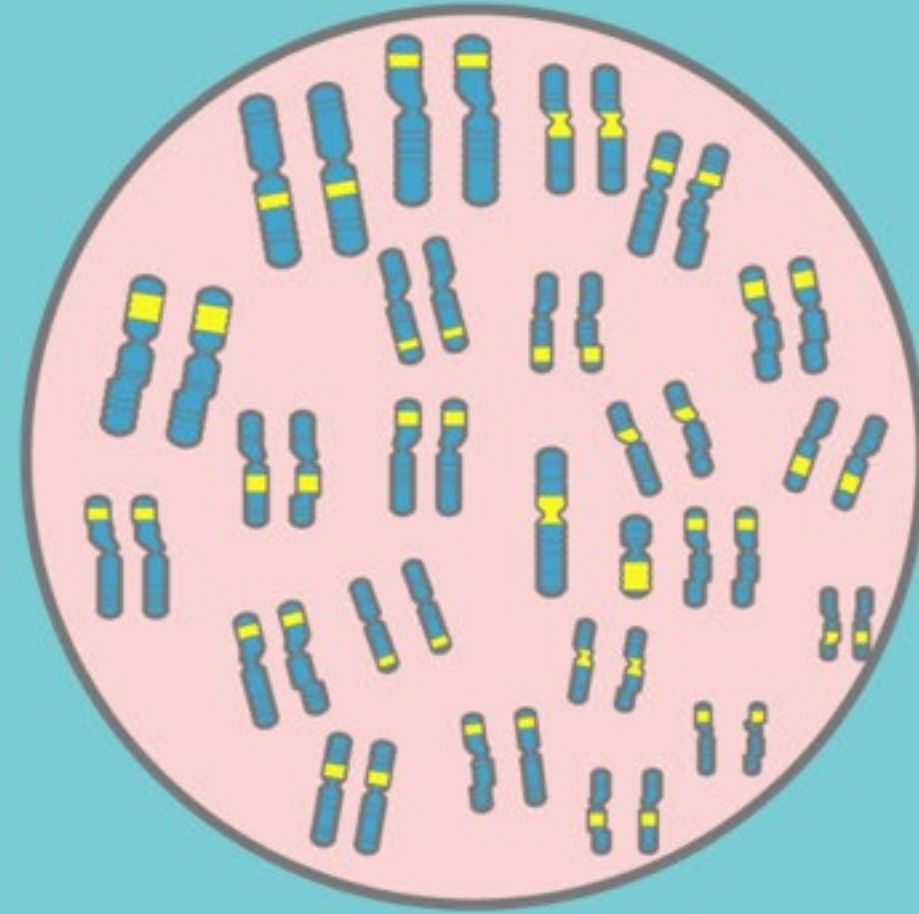
SNP site



Functional protein



A **genotype** is the genetic makeup of a person



A **phenotype** is the physical manifestation of an inherited trait or disease

In cancer, both genotype and phenotype keep changing over time.