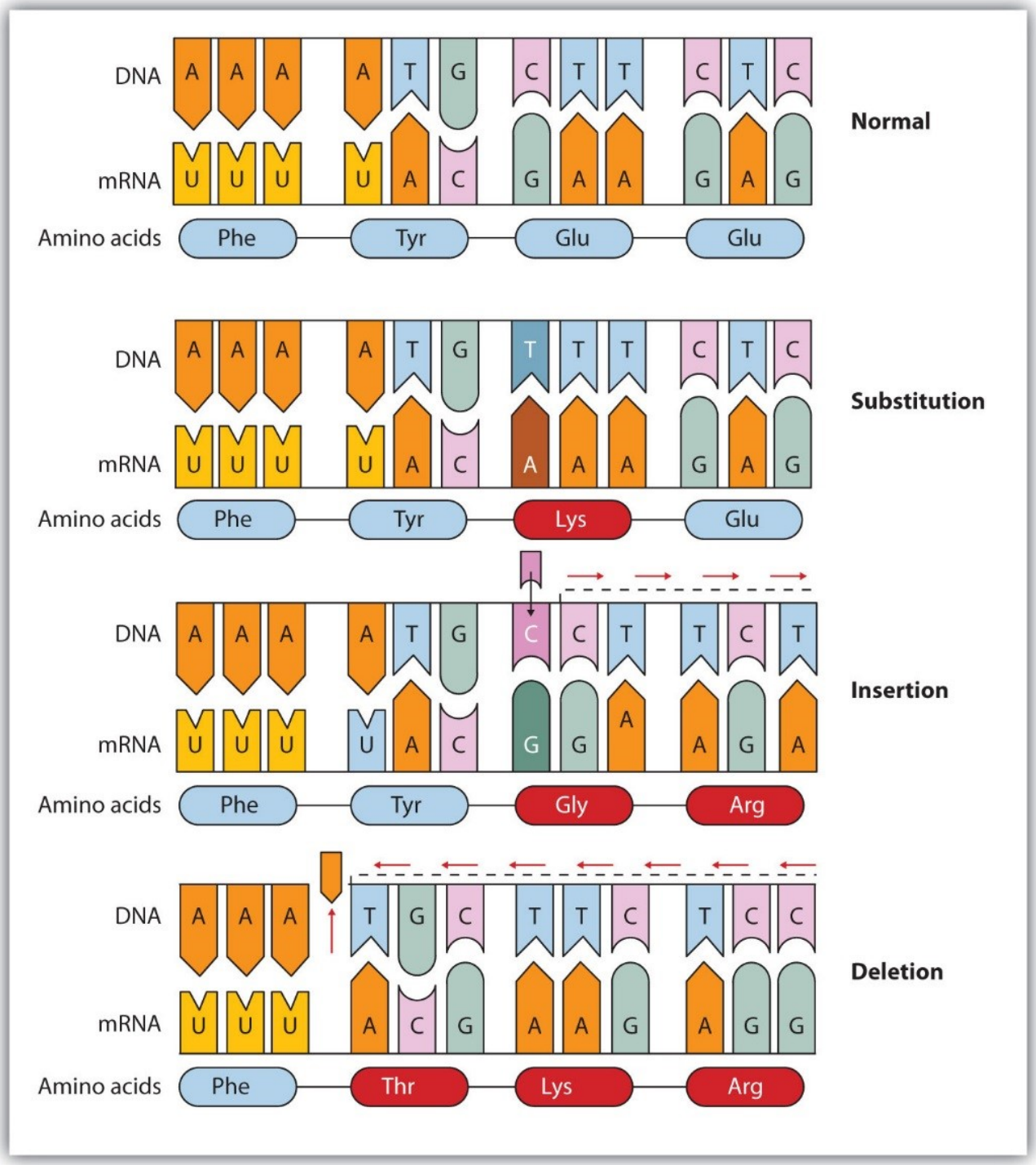
Gene Regulation and Mutation

Mutations

Defintion

Mutation: A **permanent change** or mistake **in DNA** that happens **during replication**

Types of mutations



Point Mutations

Defintion

Point mutations: A small mutation involving a singular nucleotide pair
substitution (a pair being exchanged for something else)

1. **Missense** substitution - DNA is altered to code for the **wrong amino acid**
2. **Nonsense** substitution - DNA is **altered** to code **for a stop codon**

3. **Silent** substitution - DNA is altered, although **produces the same amino acid**

Frameshift Mutations

Defintion

Framshift Mutation: One base pair is inserted or deleted, making **all pairs afterwards read incorrectly**

1. **Insertion** - A base pair is added, making all base pairs afterwards shifted towards the end, this leads to the last base pair not making it into a codon
2. **Deletion** - A base pair is deleted, making all base pairs afterwards shift forwards, this leads to the last codon missing a pair

Duplication Mutations

Defintion

Duplication Mutation: A codon is repeated, leading to the protein to be longer

Expanding Mutations

Defintion

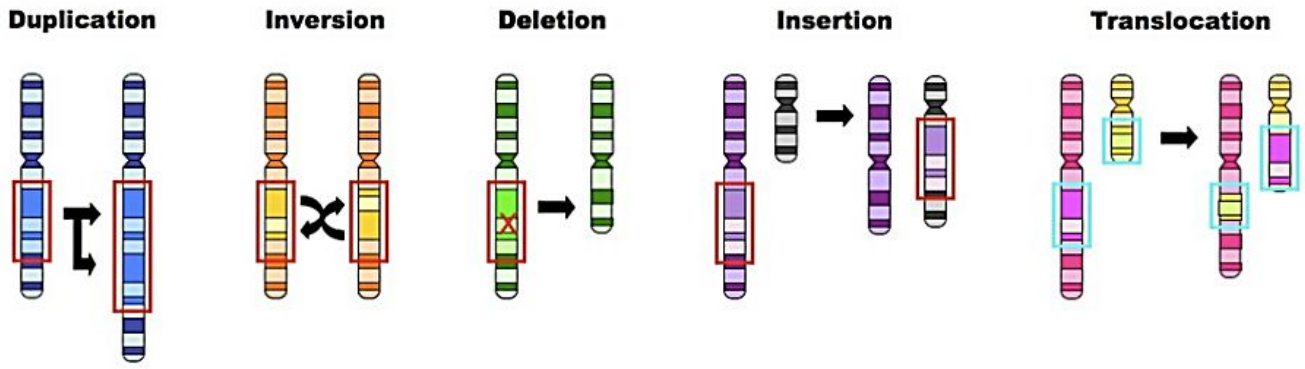
Expending mutation: A particular codon is repeated **several times**, repeating more and more over generations

Chromosomal mutations

Defintion

Chromosomal mutation: An entire portion of a chromosome being copied incorrectly

- Leads to large drastic effects on the expression of such genes



1. **Deletion**
    - A section of the chromosome is completely deleted
2. **Duplication**
    - A section of the chromosome is duplicated next to itself
3. **Inversion**
    - A section of the chromosome is flipped upside down, reversing the DNA
4. **Insertion**
    - A section of the chromosome is moved to a completely different chromosome
5. **Translocation**
    - A section of two completely different chromosomes are swapped

Protein folding and stability

Both small and large mutations can cause **proteins to misfold**, making them function incorrectly

Example

Sickle blood cells

- A single codon changes from **gutamic acid (GAA)** → **valine (GUA)**
- This leads to the protein being folded incorrectly

Causes of mutation

Small mutations

Small mutations may just happen spontaneously, sometimes DNA polymerase adds the wrong nucleotide.

Larger (Chromosomal) mutations

> **Defintion**
>
> **Mutagen**: A substance which may cause mutations, might be a chemical or radiation

Chemicals

They may:

- **Bond in place of a nucleotide**, making further replication unsuccessful
- Alter the chemical structure of DNA leading it to **mispair**

Radiation

1. High energy rays may cause **electrons to be misplaced**
2. The misplaced electron (free radical) is free from any atom
3. This **free radical can react violently** with other molecules including DNA