Mutations

Superheroes

How did Cyclops from the X-Men get his superpowers?

- He was born with the mutation

How did the Hulk and Spiderman get their superpowers?

- The Hulk was exposed to gamma radiation and Spiderman was bitten by a radioactive spider

Learning about Mutations

- Types of mutations and how they occur
- How environmental factors influence mutations
- Effects of mutations

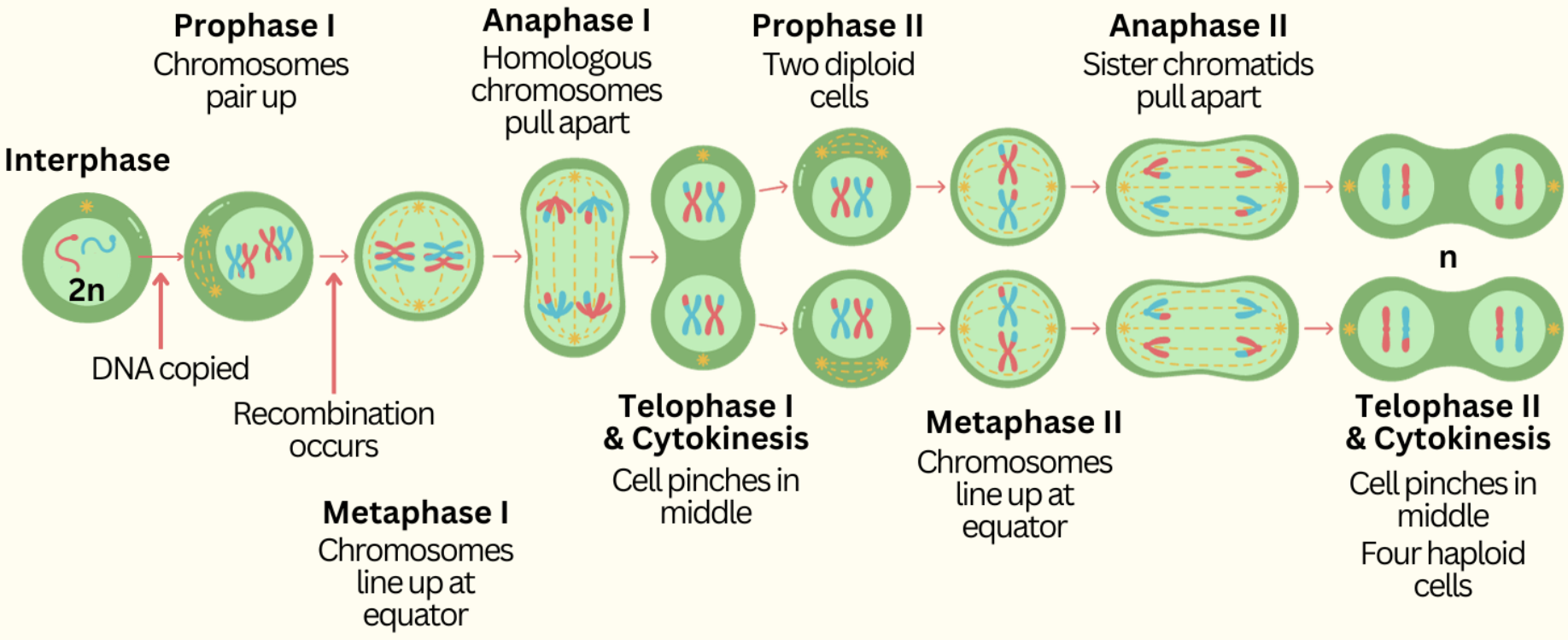
Terminology

- Automsomes: Numbered Chromosomes (22 pairs in humans)
  - Autosomal Disorder: Occurs in genes on one of these chromosomes
  - Recessive: Passed by parents but no shown in parents
- Chromosomal Disorder: Occurs on the sex-chromosomes, usually from an error in cell division

- Diploid: Nucleus contains two complete sets (23 pairs) of chromosomes, one from each parent (2N)- Human somatic cells
- Haploid: Single set of chromosomes (half of diploid)- Only sperm and egg are haploid in humans

MEIOSIS

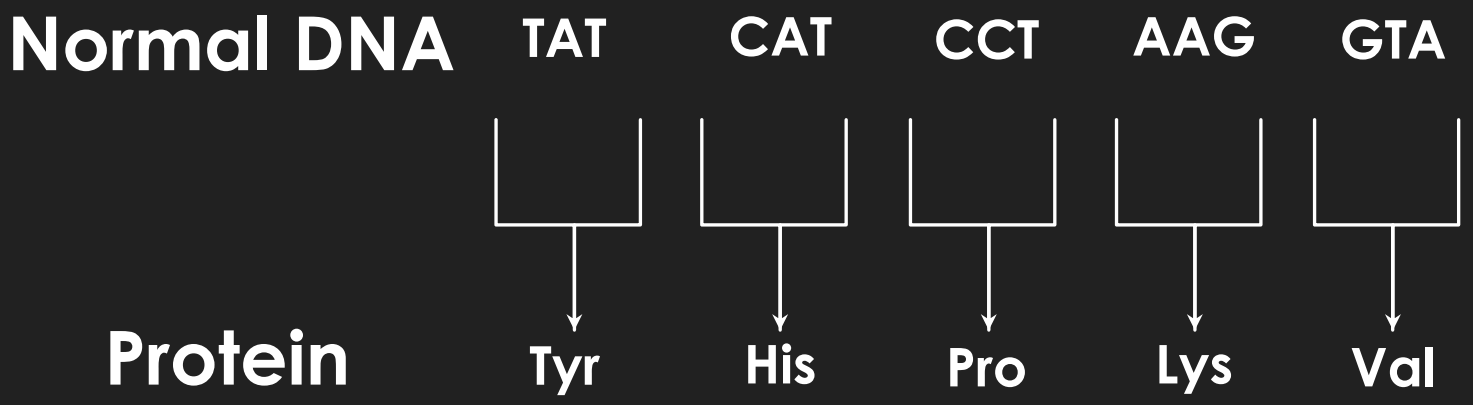
Meiosis is the process where a cell divides twice, forming four cells that each contain half the genetic information (2n→n).



Meiosis forms sperm and egg (sex cells or gametes).

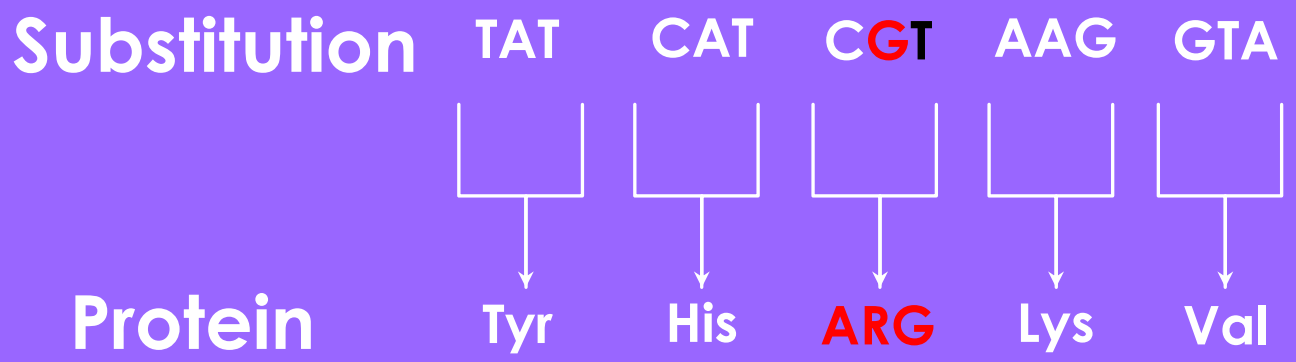
Types of Mutations

- **Small-scale mutations**
  - Affect DNA at the molecular level by changing the normal sequence of nucleotide base pairs
  - Occur during the process of DNA replications (either meiosis or mitosis)



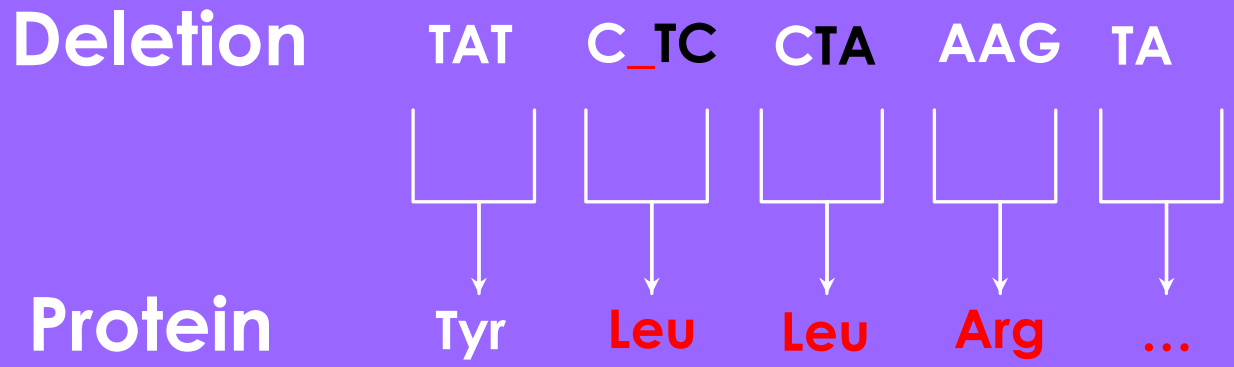
Small-Scale Mutations #1

1. **Substitution** (or a "point" mutation")
   - Substitutions occur when a nucleotide is replaced with a different nucleotide in the DNA sequence
   - This type of mutation only affects the codon for a single amino acid



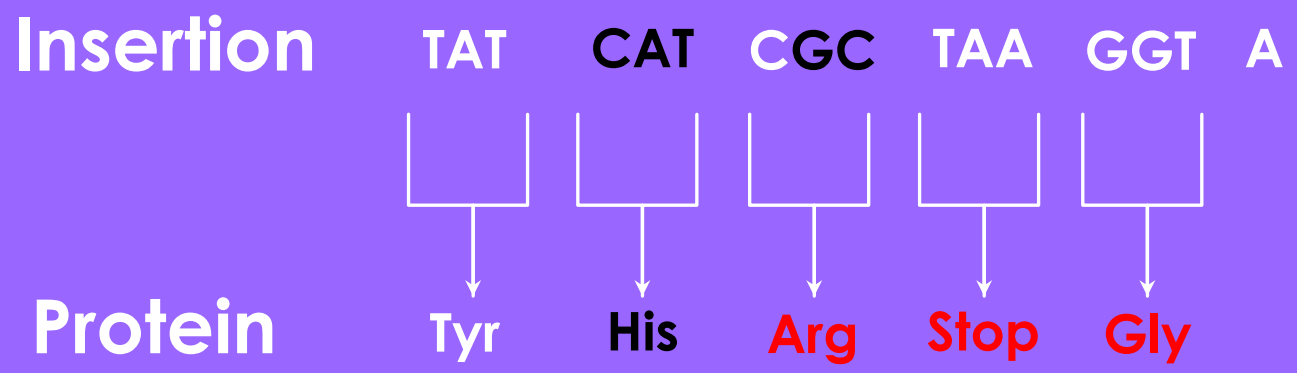
Small-Scale Mutations #2

2. **Deletion** (a "frameshift" mutation)
   - Deletion is the removal of a nucleotide from the DNA sequence
   - Removal of even a single nucleotide from a gene alters every codon after the mutation



Small-Scale Mutations #3

3. **Insertion** (a "frameshift" mutation)
   - Addition of a nucleotide to the DNA sequence
   - Addition of even a single nucleotide to a gene alters every codon after the mutation

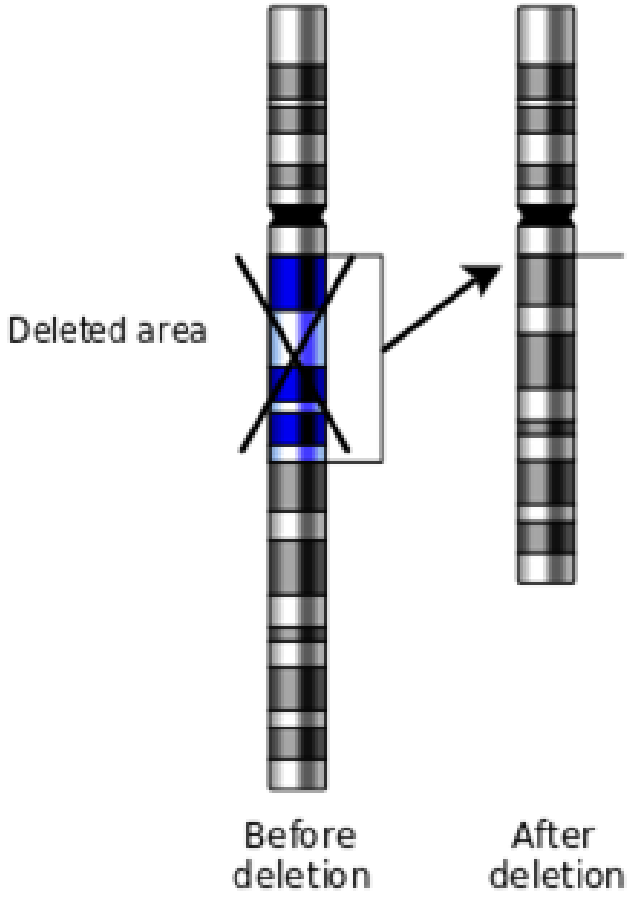


Large-Scale Mutations

- **Affect entire portions of the chromosome**
  - Some large-scale mutations affect only single chromosomes, others occur across nonhomologous pairs
  - Entire genes or sets of genes are altered rather than only single nucleotides of the DNA
  - Mutations involving multiple chromosomes are likely to occur in meiosis, during the prophase I

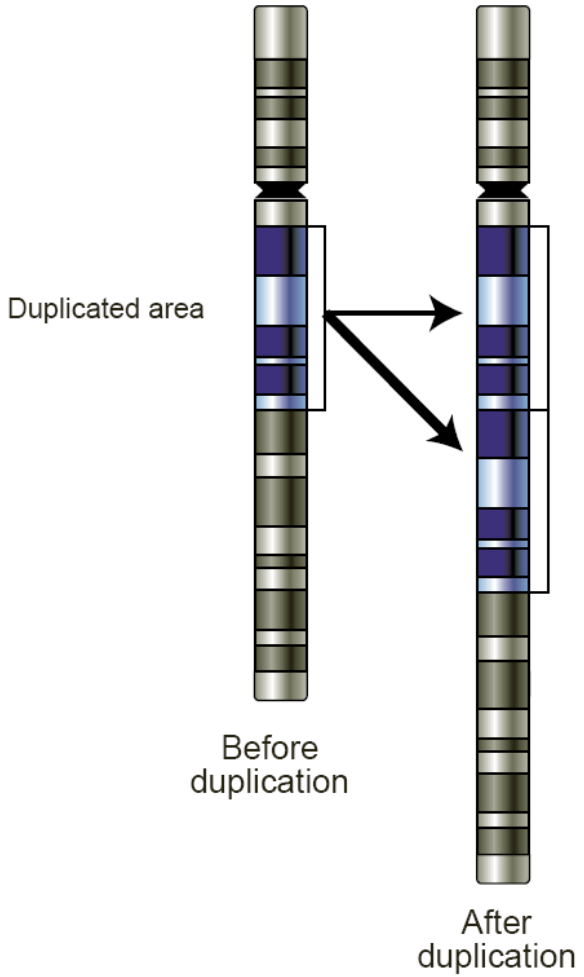
Large-Scale Mutations

- Deletion
  - Single chromosome mutation
  - The loss of one or more gene(s) from the parent chromosome
  - Ex: DiGeorge Syndrome (deletion of portion of c-22)
  - Ex: Cri du Chat Syndrome (deletion of portion of c-5)



Large-Scale Mutations

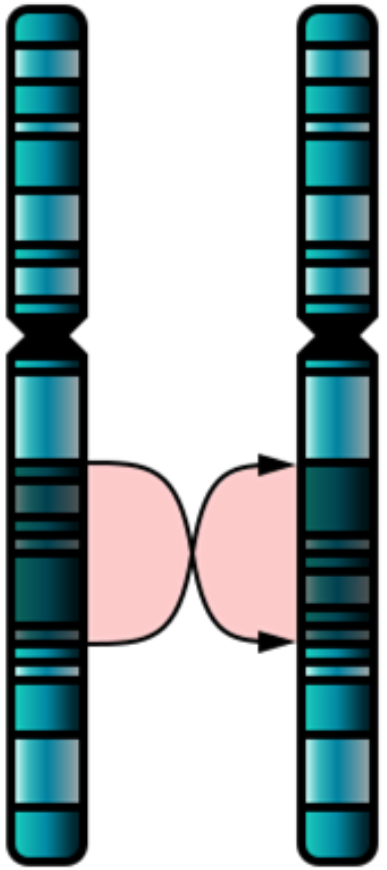
- Duplication (amplifications)
  - Single chromosome mutation
  - The addition of one or more gene(s) that are already present in the chromosome



Large-Scale Mutations

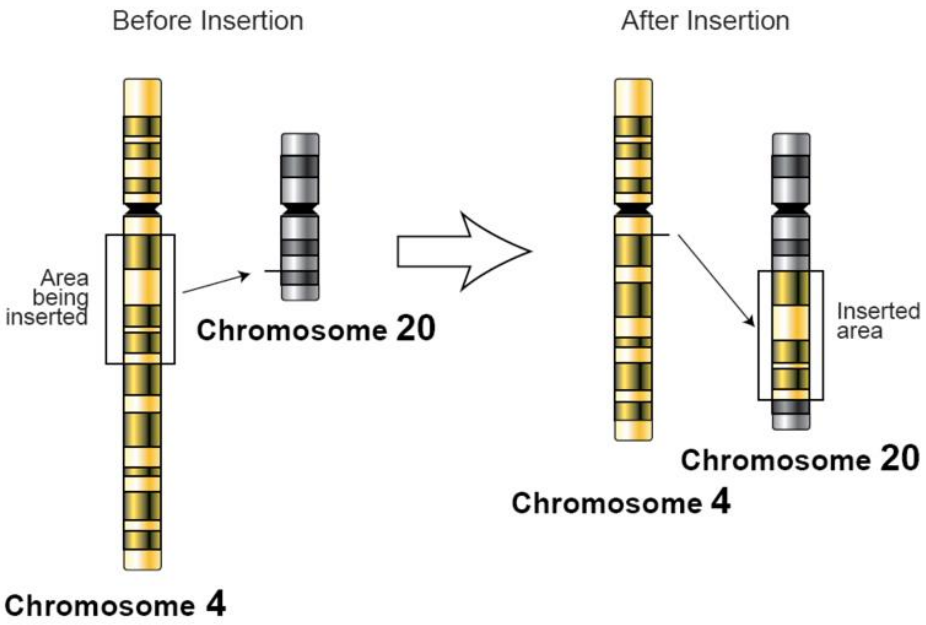
- Inversion (chromosomal inversions)
  - Single chromosome mutation
  - The complete reversal of one or more gene(s) within a chromosome; the genes are present, but the order is backwards from the parent chromosome

Inversion



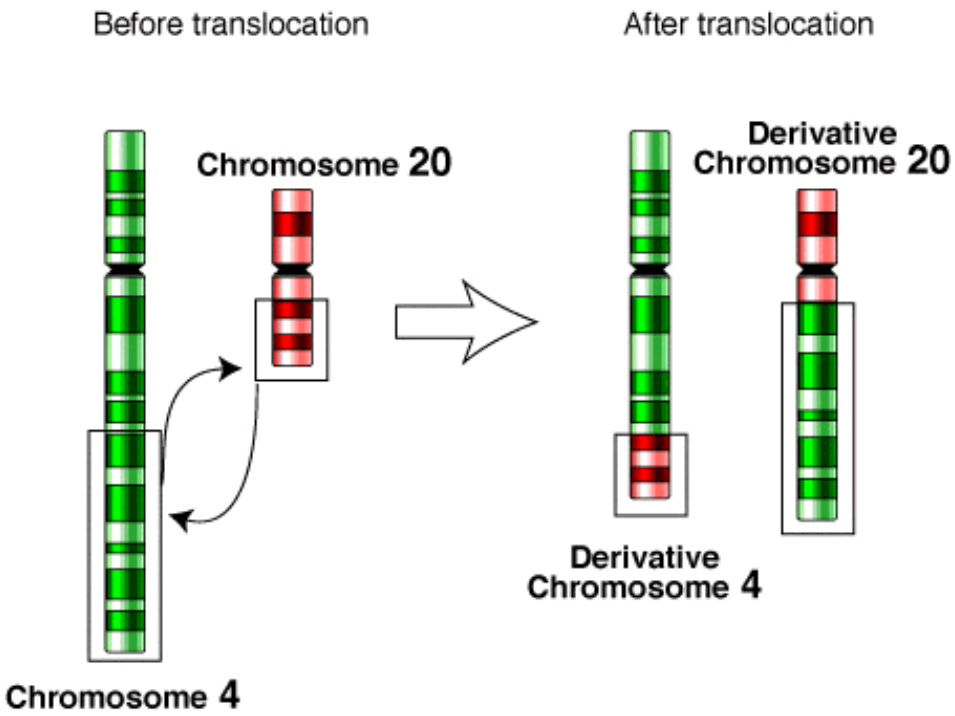
Large-Scale Mutations

- Insertion
  - Multiple chromosome mutation
  - One or more gene(s) are removed from one chromosome and inserted into another nonhomologous chromosome
  - Can occur by an error during the prophase I of meiosis when the chromosomes are swapping genes to increase diversity



Large-Scale Mutations

- Translocation
  - Multiple nonhomologous chromosome mutation
  - Chromosomes swap one or more gene(s) with another chromosome



Large-Scale Mutations

- Nondisjunction
  - Does not involve any errors in DNA replication or crossing-over
  - Mutations occur during the anaphase and telophase when the chromosomes are not separated correctly into the new cells
  - Common nondisjunctions are missing or extra chromosomes

Effects of Mutations

- The effects of mutations may range from nothing to the unviability of a cell
- All mutations affect the proteins that are created during protein synthesis, but not all mutations have a significant impact

Small-Scale Mutation Effects

1. Silent
   - The nucleotide is replaced, but the codon still produces the same amino acid
2. Missense
   - The codon now results in a different amino acid, which may or may not significantly alter the protein's function
3. Nonsense
   - The codon now results in a "stop" command, truncating the protein at the location where the mutated codon is read; this almost always leads to a loss of protein functionality

Large-Scale Mutation Effects

- Effects of large-scale mutations are more obvious than those of small-scale mutations
- Duplication of multiple genes causes those genes to be overexpressed while deletions result in missing or incomplete genes
- Mutations that change the order of the genes on the chromosome—such as deletions, inversions, insertions and translocations—result in genes that are close together

Large-Scale Mutation Effects

- When certain genes are positioned closely together, they may encode for a "fusion protein"
  - A fusion protein is a protein that would not normally exist but is created by a mutation in which two genes were combined
  - The new proteins give cells a growth advantage, leading to tumors and cancer

Large-Scale Mutation Effects

- Often, large-scale mutations lead to cells that are not viable
  - The cell dies due to the mutation

Mutation Influences

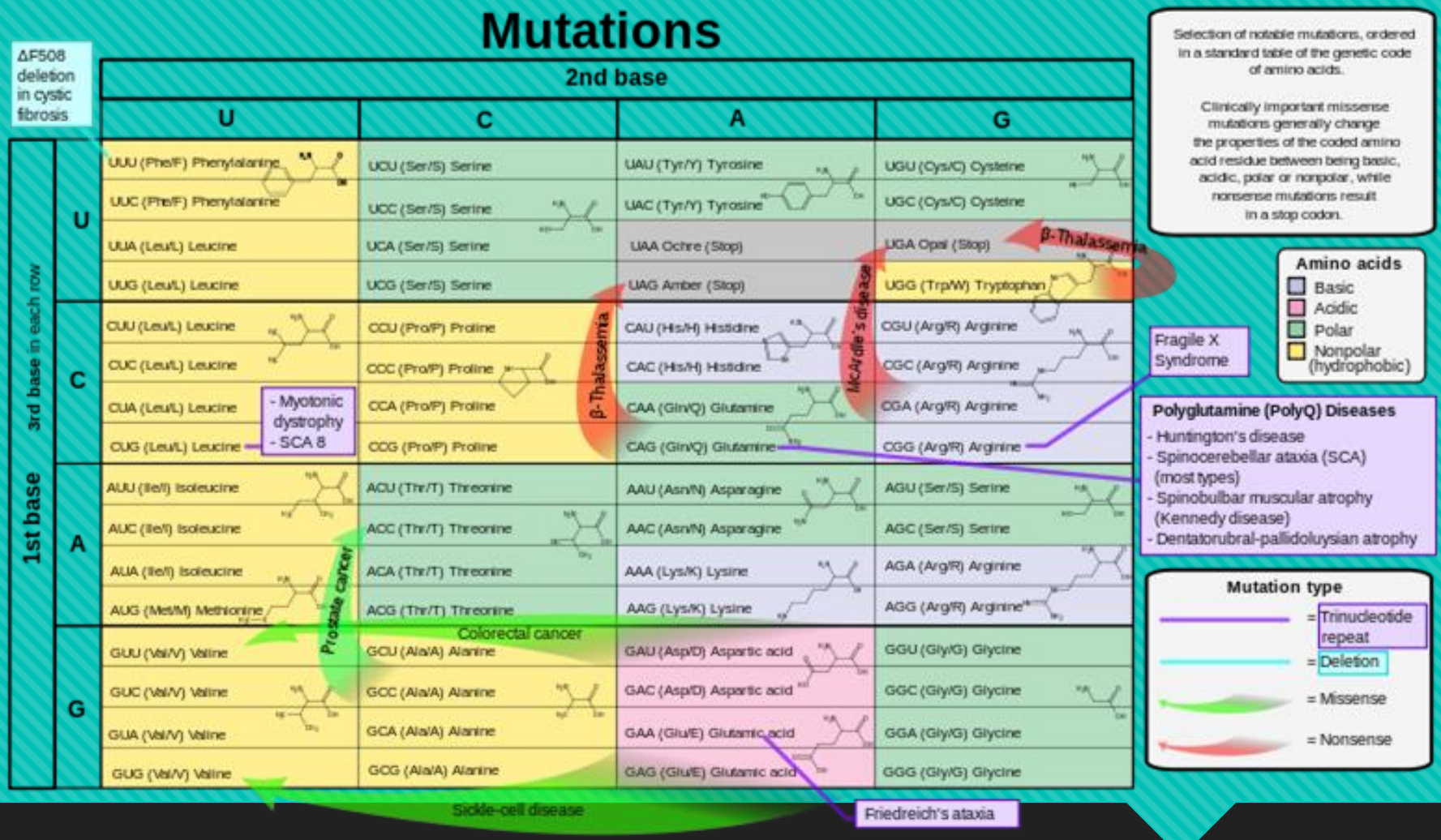
- Exposure to certain chemicals
  - Carcinogenic chemicals may cause cancer
- Exposure to radiation
- Retroviruses
  - Retroviruses such as HIV naturally experience mutations at a much higher rate than other organisms

Engineering Connection

- Humans have been genetically modifying plants and animals for thousands of years
  - Example: Breeding watermelons to be larger and have fewer seeds
  - Example: Breeding chickens to have more white meat and more breast meat

Engineering Connection

- Engineers can directly manipulate the genetic code of plants and animals (controversial)
  - Examples: Disease-resistant papaya, vitamin A-rich rice, and drought-tolerant corn
- Engineers and scientists are currently studying gene editing in the womb
  - May prevent the child from having diseases and disabilities



Examples of Notable Mutations