Source: ⟦KBhBIO101MutationsAllealsInheritance⟧

1 | **Mutations**

Mutations are one way by which totally random, not controlled for, and fully spontaneous genetic modifications happen to literally anywhere in any cell's DNA during ⟦KBhBIO101CellReproduction⟧. Specifically, it involves an environmental factor or the sheer entropy of things to directly, or indirectly (by causing/creating a oopsie during ⟦KBhBIO101DNAReplication⟧) *mutate* the resulting supposed-to-be-exact copy of DNA.
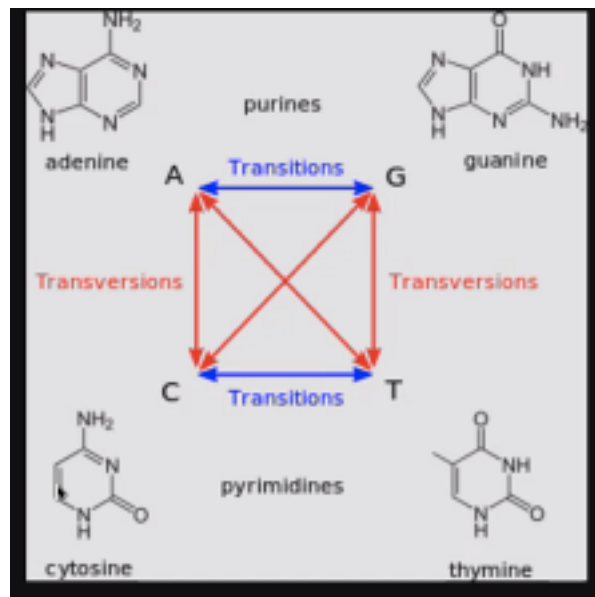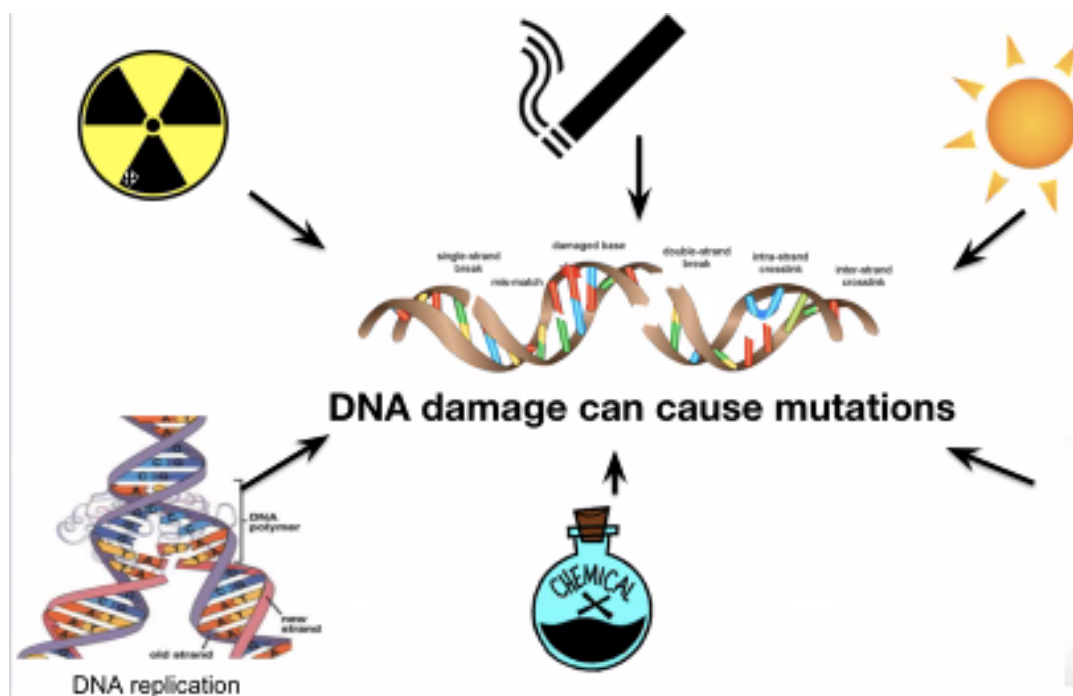

Figure 1: Pasted image 20210331134011.png

Lot's of things cause mutations!

To figure out how mutations work, you first need to know how DNA looks like, so here goes a…

Special Programming! *How does DNA work?*

There are two rought typos of codons on DNA, namely:

- **Pyrimides** - cytosine + thymine. Single ring. Which are usually paired with…
- **Purines** - adenine + guanine. Double ring.

So if a mutation replaces adenine and guanine, it would has less of an effect because a double ring is still matched with a single ring. But if an adenine is replaced by thymine, we could have a bigger issue because double-double ring is much longer than a traditional single/double match.

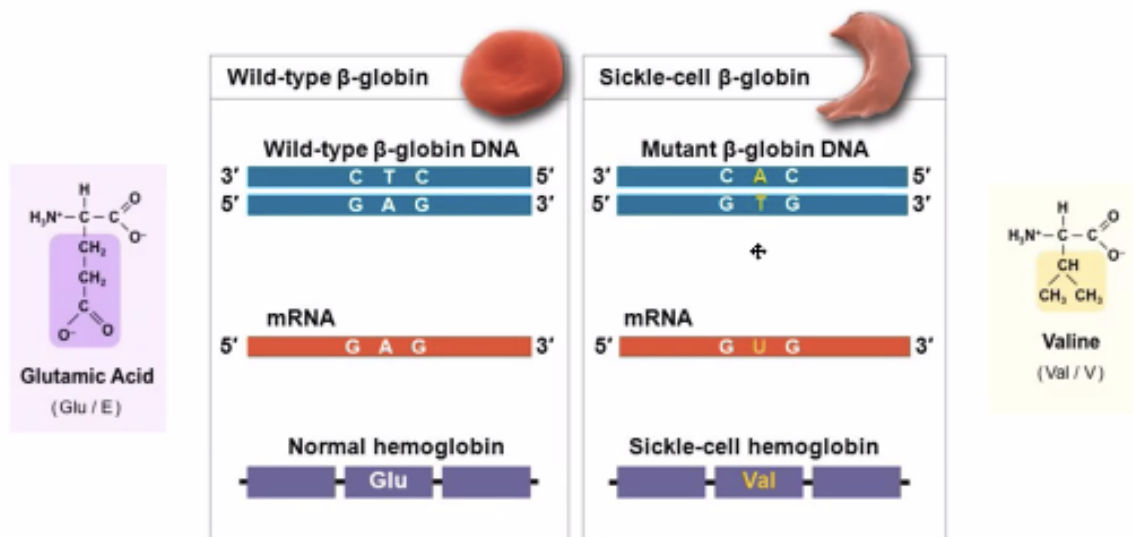Thank you for coming to this assembly. You could leave now. ***

1.1 | **Mutation Vocab**

Trait: characteristic of organism influnenced by its genes & modified by its enviroment

Phenotype: a collective subset of all the traits ("that looks different from wild type") in an organism

1.2 | **And now, an example**

Mutant hemoglobin could… 1) with one mutation, cause a slight change in the RBC but cause resistance to malaria 2) with two mutation, cause sickle-cell.

Remember that DNA codes for proteins, so mutations in DNA will cause different proteins BUT not necessarily different traits. In the case of 1-chromasome sickle-cell mutation, a protein is changed but the result is not nocessarily a different RBC.

1.3 | **Types of Mutations**

There are many types/methods by which DNA mutate. See…

1.3.1 |**By Place**

Germline mutations mutate the egg/cell causes no/local problems but pass the mutated gene down to the children fully

Somatic mutations mutated somatic cell causes local mutations that does not influence much (cancer, but)

1.3.2 |**By Method**

Point mutations

Change one codon on the gene and potentially cause something.

- Slient mutation: has no effect on protein
- Missense: result in amino acid substitution
- Nonsense: substitutes a stop codon for an amino acid

Indel/Frameshift mutation

Shift by adding/substracting codons and shift the gene. Everything downstream to the point of mutation will be completely incorrect.

## 1.4 |**Mutations in other places**

Promoter/Enhancer mutation: control the level of expression for genes, which could relate to cancer (over-activation) or a protein deficiency (lack of activation)

Splice donor and acceptor site mutation: including extra intron or cutting out required exon

Ribosome binding sites: prevents the ribosome from binding

1.5 |**Large scale DNA changes**

Taking whole chunks of DNA or swapping them; usually caused by your DNA wholly breaking (Radioactivity? Incorrectly functioning enzymes?) and then your repair machinary stitching it up wrongly.
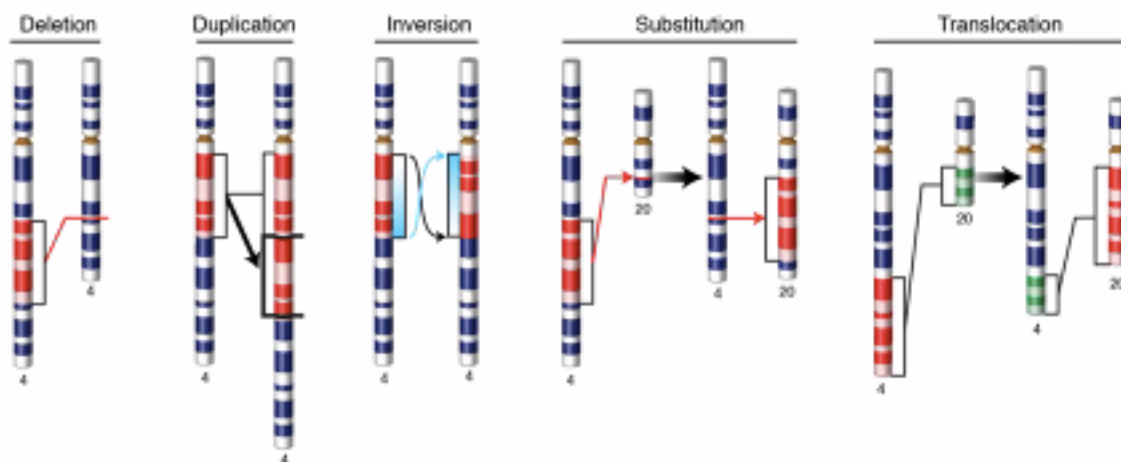
1.6 | **Impacts of mutations**

Loss of function mutations

- Complete loss of a proteins
- Reduction of a protein's ability to function

Gain of function mutations

- Increase the function of a protein
- Aquire new protein function
- Expression of protein in new location/time

Neutral function

Does nothing