Source: [KBhBIO101MutationsAllealsInheritance]

1 | **Mutations**

you are probably looking for [KBhBIO101MutationsAllealsInheritance].

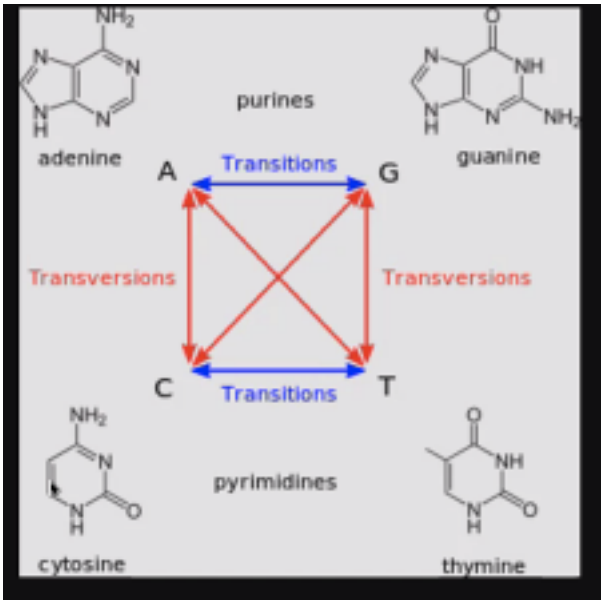


Figure 1: Pasted image 20210331134011.png

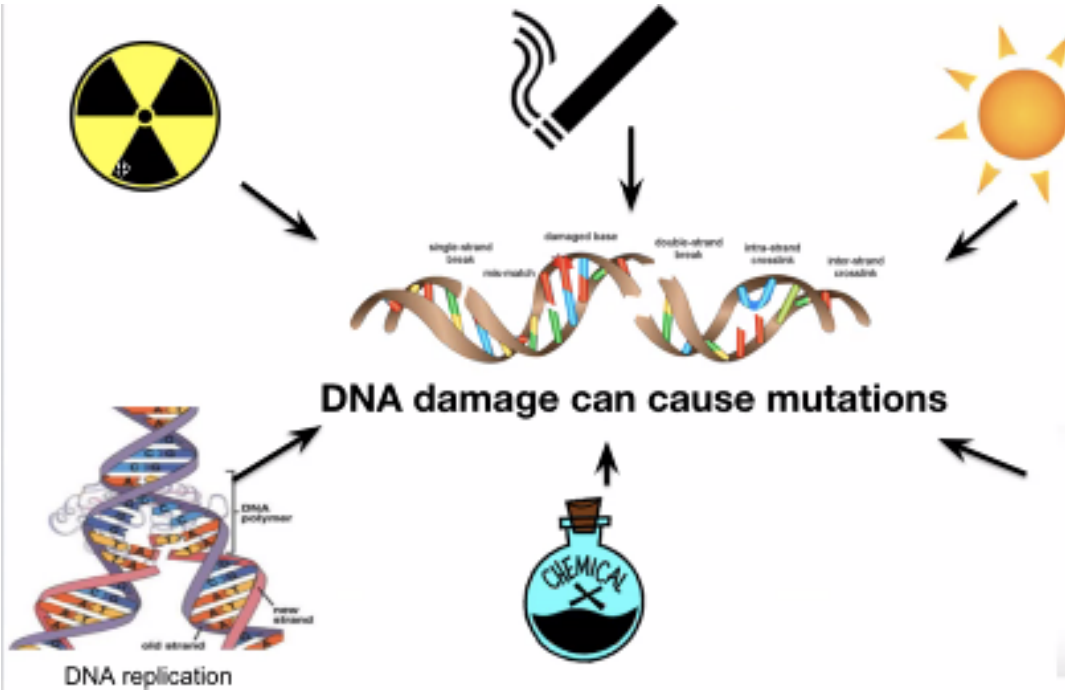
Pyrimides - cytosine + thymine. Single ring.

These are usually paired up 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 isuue because double-double ring is much longer than a traditional single/double match.

Lot's of things cause mutations!



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

Changes in gene structure cause a lack of sythesis for purple pigment

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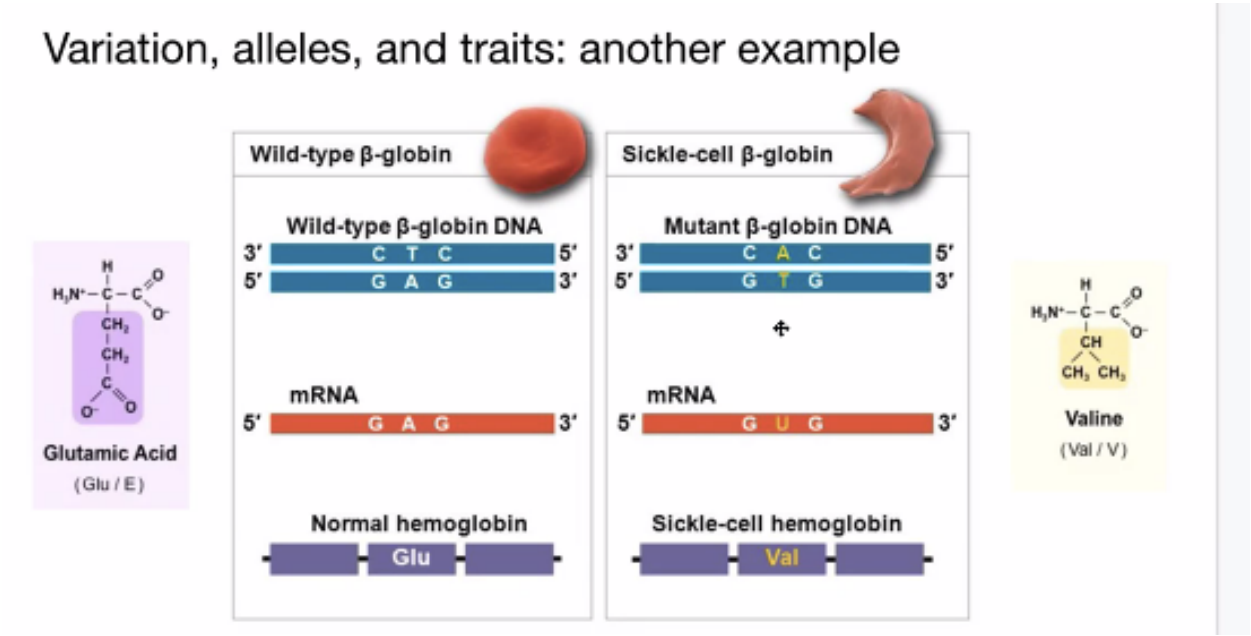


Mutant hemoglobin could… 1) with one mutation, cause a slight change in the RBC but cause resistance to malaria 2) with two mutation, cause sickle-cell.

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.

1.1 | Types of Mutations

1.1.1 | 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)

1.1.2 | 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.

1.2 | Mutations in other places

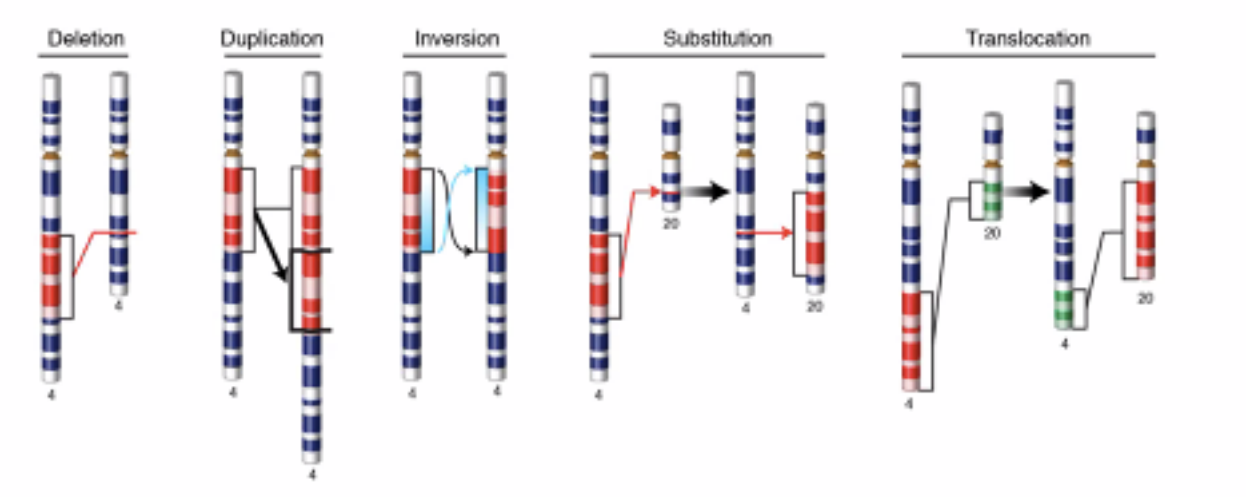
Promoter/Enhancer mutation: control the level of expression for genes, which could relate to cancer (over-activation) or a protein deficiency (lack of activation)

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

Ribosome binding sites: prevents the ribosome from binding

1.3 | 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.



1.4 | **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