

Source: [KBhBIO101MutationsAllelsInheritance](#)

## 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.

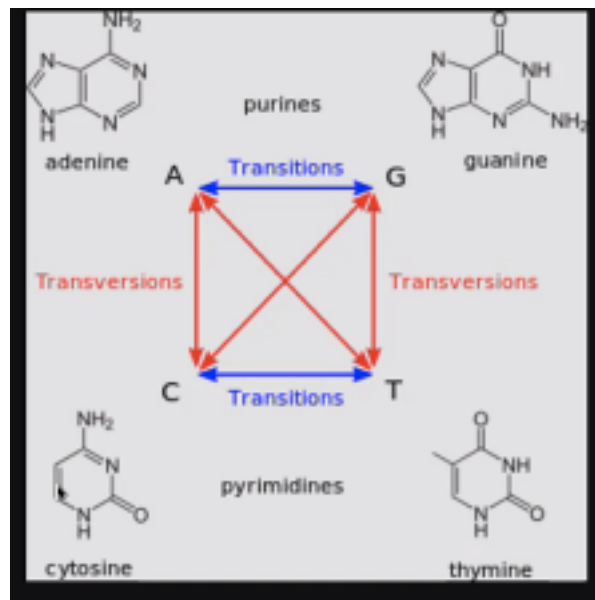


Figure 1: Pasted image 20210331134011.png

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

### **Special Programing!** How does DNA work?

There are two rough types of codons on DNA, namely:

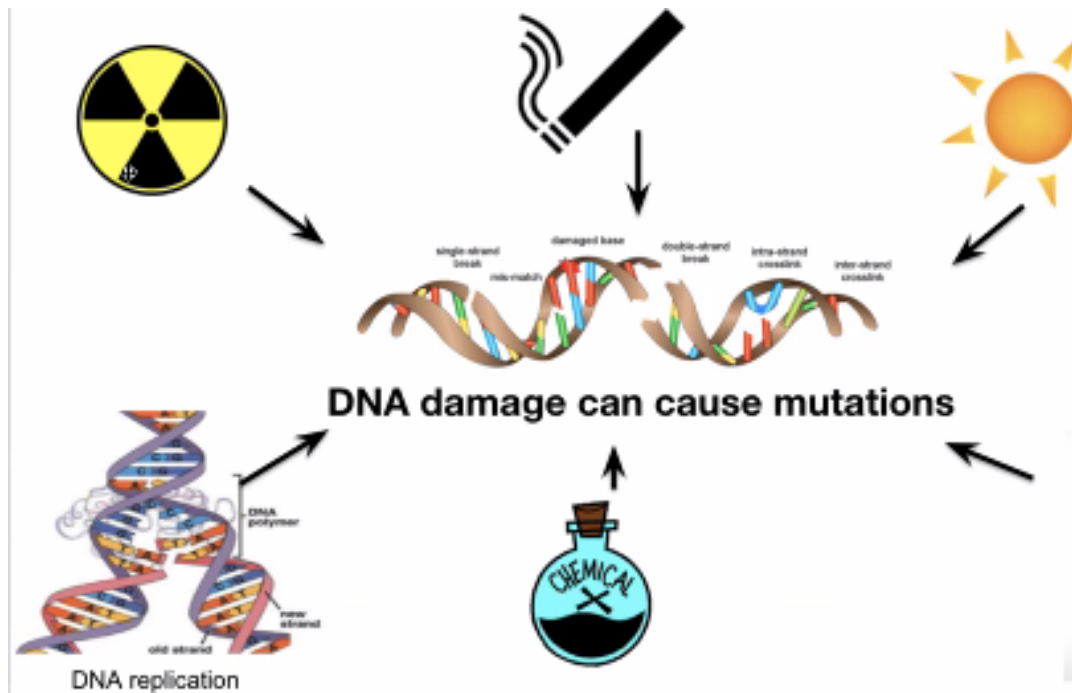
- **Pyrimides** - cytosine + thymine. Single ring.

These are usually paired up with

Purines - adenine + guanine. Double ring.

So if a mutation replaces adenine and guanine, it would have 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 assembly. You could leave now. \*\*\*

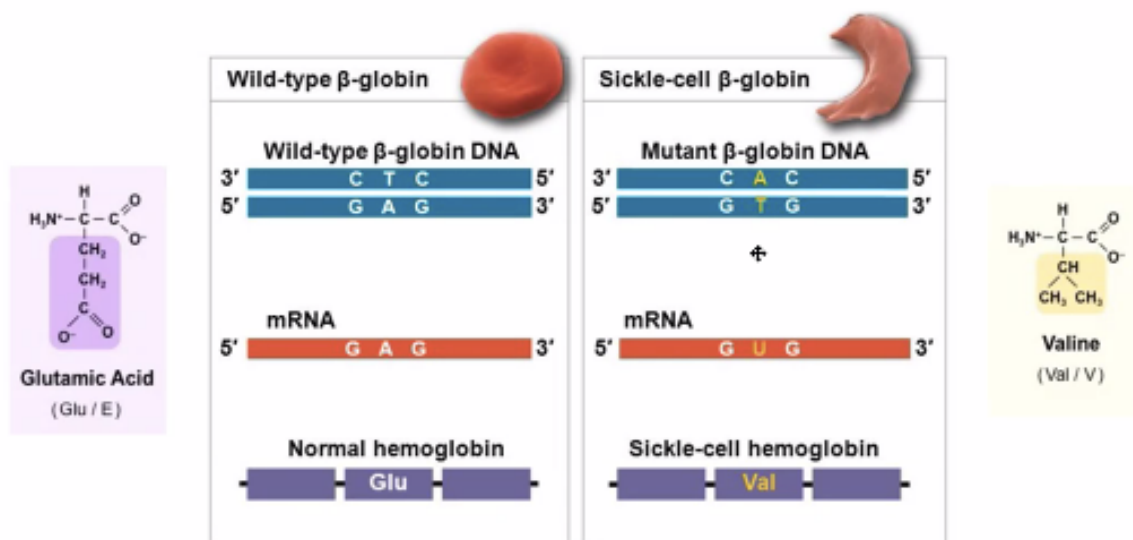


**Trait:** characteristic of organism influenced by its genes & modified by its environment

**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 synthesis for purple pigment

## Variation, alleles, and traits: another 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-chromosome sickle-cell mutation, a protein is changed but the result is not necessarily a different RBC.

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## 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.

- Silent 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/subtracting codons and shift the gene. Everything downstream to the point of mutation will be completely incorrect.

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## 1.2 | Mutations in other places

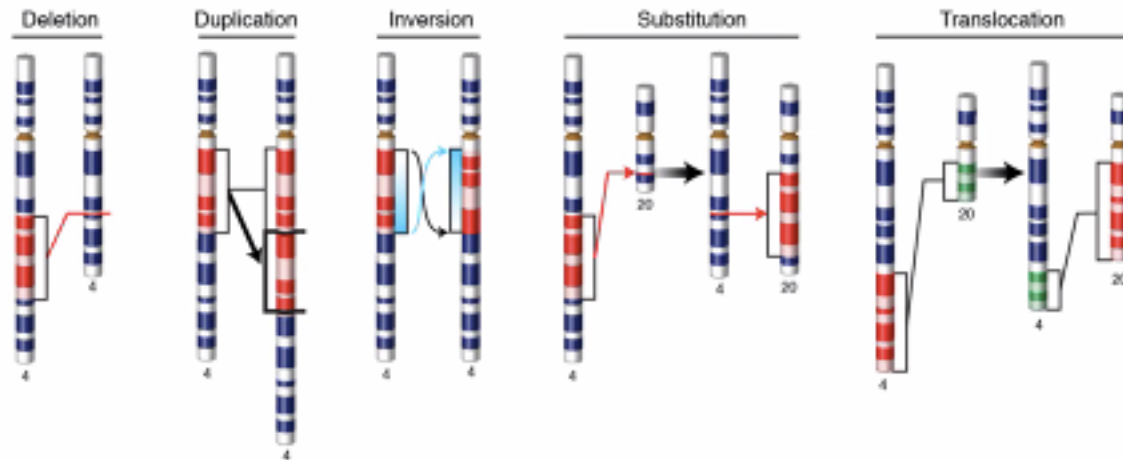
**Promoter/Enhancer mutation:** control the level of expression for genes, which could relate to cancer (over-activation) or a protein deficiency (lack of activation)

**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 machinery 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