CONCEPT: TYPES OF MUTATIONS

● There are many different types of _____

□ The first way to classify mutations is to describe how they arise

- **Spontaneous mutations** are changes that randomly occur

- **Induced mutations** are changes caused via natural or artificial agents (ex: radiation, chemicals, etc…)

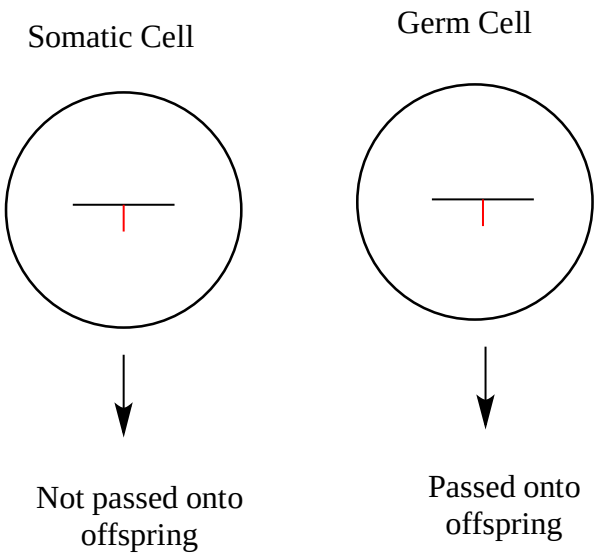
□ A second way to classify mutations is by where the mutation occurs

- **Somatic cell mutations** occur in somatic cells

- **Germ cell mutations** occur in germ cells

- Germ cell mutations are passed onto offspring

EXAMPLE:



□ A **point mutation** is a mutation that affects only a _____ nucleotide

- **Base substitutions** changes on base for another

- **Transitions** replaces the base with one from a similar category (ex: purine to purine)

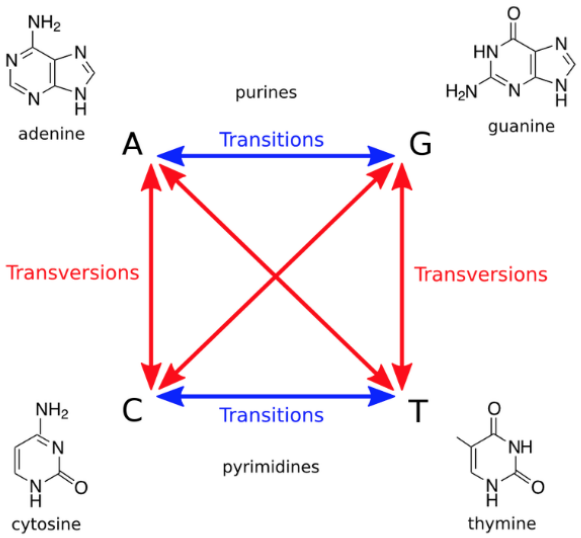
- **Transversions** replaces the base from a different category (ex: purine to pyrimidine)

- **Base insertions** add extra bases, and **base deletions** remove bases

- An **indel mutation** is when both an insertion and a deletion occur

- These can easily effect codons

EXAMPLE:



□ A third way to classify mutations is their effect on _____

- **Synonymous (silent) mutations** change a codon to another codon that codes for the same amino acid
- **Missense mutations** change a codon to another codon that codes for a different amino acid
  - *Conservative* changes one codon to another chemically similar amino acid
  - *Nonconservative* changes one codon to another chemically different amino acid
- **Nonsense mutations** change a codon to a stop codon
- **Frameshift mutations** alter the codon reading frame

EXAMPLE:

	NO mutation	Point mutations			
		Silent	Nonsense	Missense	
				Conservative	non-conservative
DNA level	TTC	TTT	ATC	TCC	TGC
mRNA level	AAG	AAA	UAG	AGG	ACG
Protein level	Lys	Lys	STOP	Arg	Thr

Base Distortions

● Base distortions disturbe the _____ structure of the bases

□ Bases can lose part of their structure

- **Apurinic sites** are regions that have lost their purines

- **Depurination** is the loss of the purine

- **Apyrmidinic sites** are regions that have lost their pyrimidines

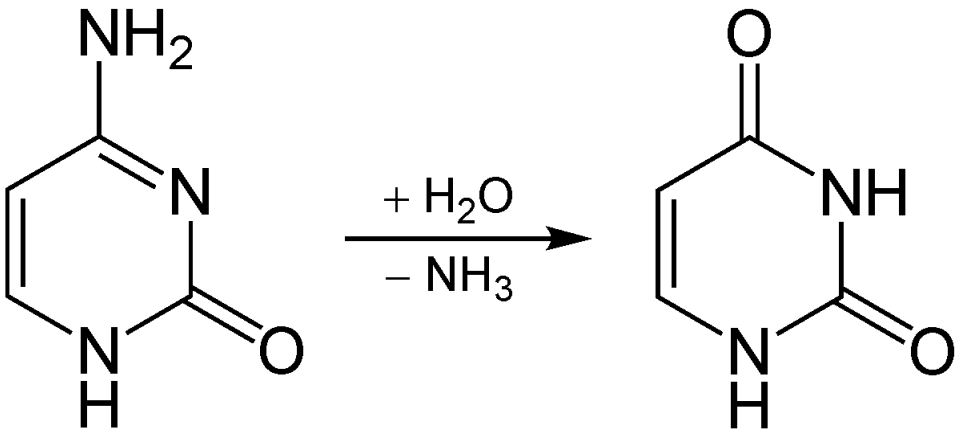
□ **Deamination** is the removal of an amino group from a base or molecule

- This process can change cytosine to uracil and adenine to hypoxanthine

□ Oxidative damage occurs when reactive oxygen species affect the _____

- O_2^-, H_2O_2, and OH target the DNA and chemically alter the bases

EXAMPLE: Deamination of cytosine to uracil

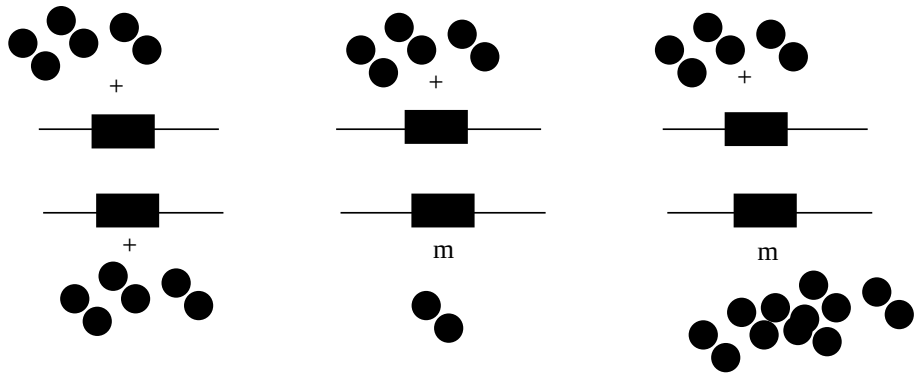


Mutations and Phenotypes

□ A fourth way to classify mutations is by their effect on the protein's _____ or organism phenotype

- **Loss of function** and **gain of function** mutants refer to the activity of the mutated gene
    - **Null mutations** describe mutations that result in a complete loss of function
- **Visible mutations** alter the physical phenotype of the organism
    - **Nutritional mutations** cause loss of an ability to synthesize an amino acid or vitamins
    - **Behavioral mutations** cause changes in behavioral
- **Conditional mutations** are only detectable under certain conditions
    - Ex: temperature sensitive mutants
- **Lethal mutations** cause death of the organism
- **Neutral mutations** have no observable affect on the organism

EXAMPLE:

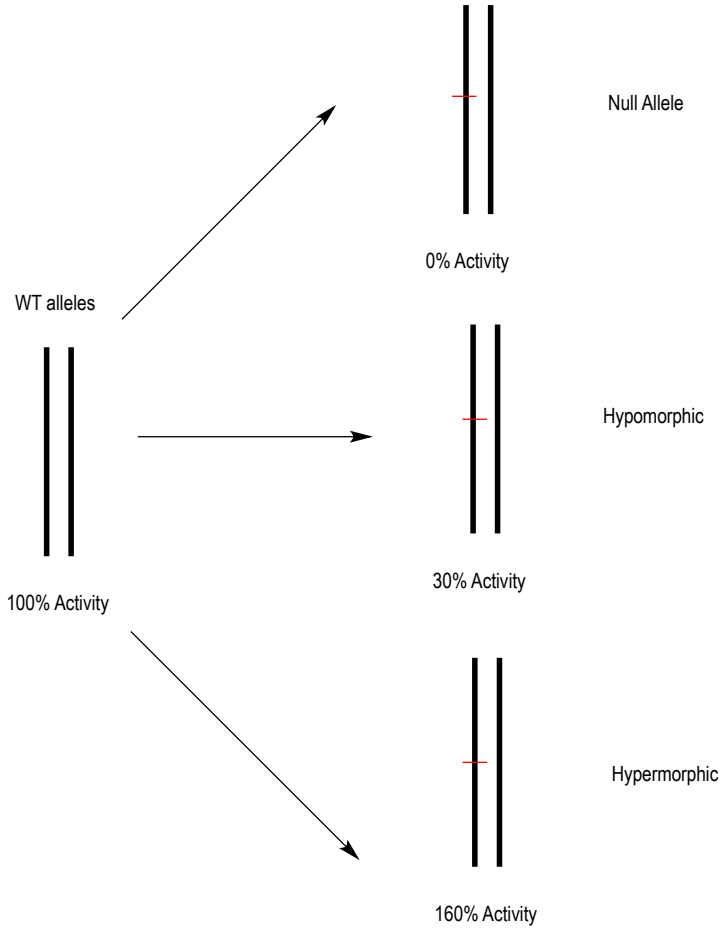


Wild-Type | Loss of function | Gain of function

□ A fifth way to classify mutations is by their effect on individual _____

- **Hypomorphic mutation** is a loss of function mutation that produces a tiny amount of functional protein
- **Haploinsufficiency** describes when one WT allele doesn't provide enough gene product
    - Other allele has been mutated to null or a loss-of-function
- **Dominant-negative** describes when a mutant allele blocks the protein production produced by WT allele
- **Hypermorphic mutations** is a gain of function mutation that produces a more efficient protein than WT
- **Neomorephic mutations** generate a novel phenotype

EXAMPLE:



□ A sixth way to classify mutations is by their _____- activity

- **Suppressor mutations** are genetic changes that suppress the effect of another mutation

  - **Intragenic** are mutations found in the same gene as the mutation being suppressed

  - **Intergenic** are mutations found in a separate gene than the mutation being suppressed