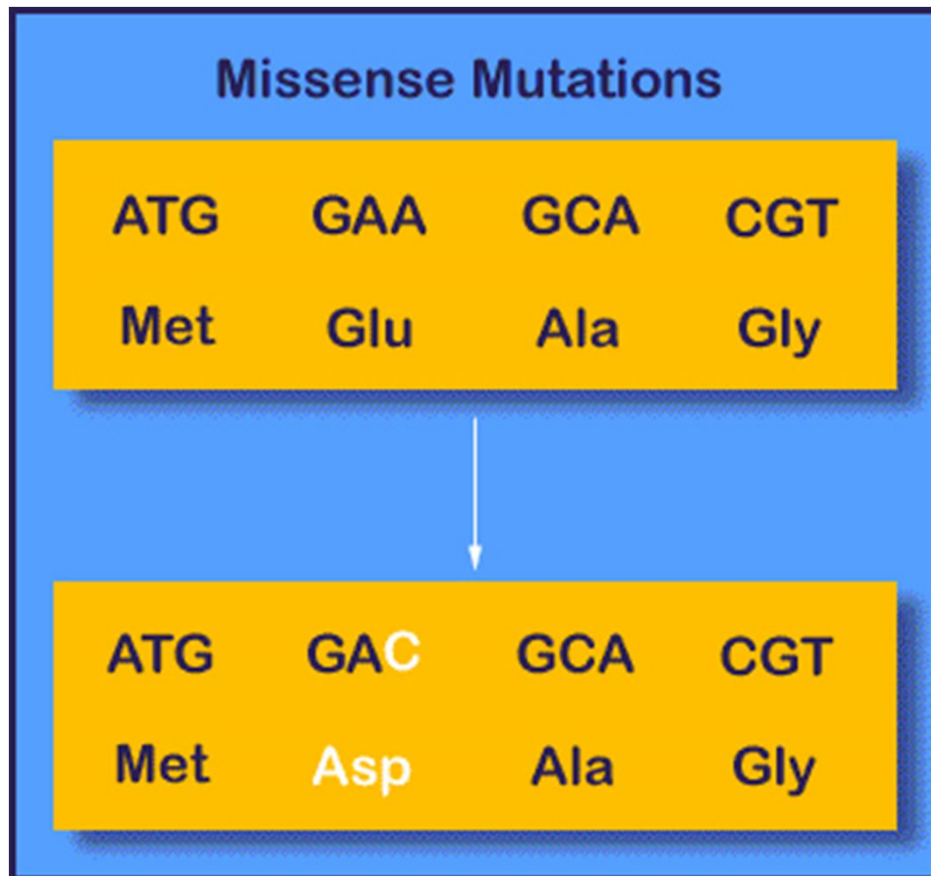


# Mutations

- Cells sometimes make mistakes even with regulations
- Mistakes are usually rare but DO happen
- Cells try to fix when a mistake happens
- When a mistake becomes a permanent change it is a **MUTATION**

# Mutations



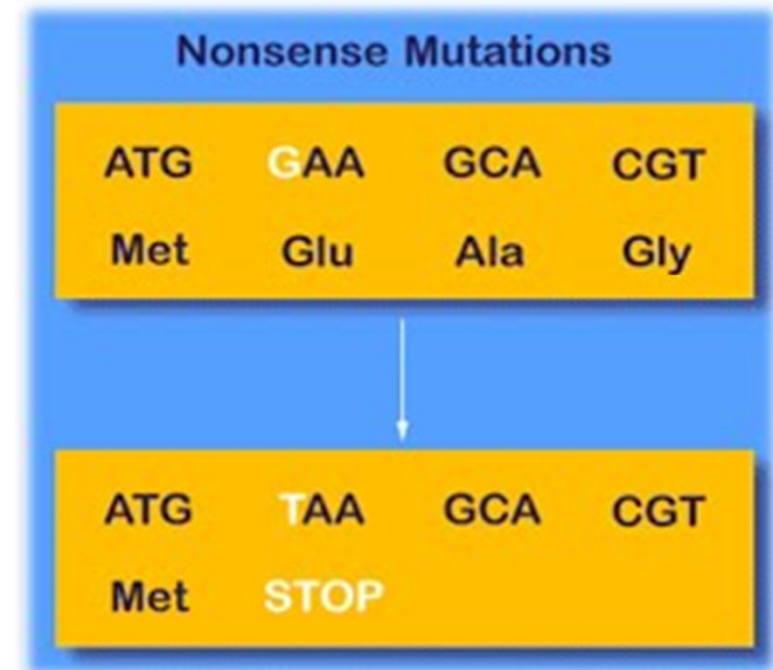
- Types of mutations

- Point mutations

- chemical change in just one base pair
    - Can cause a genetic disorder (ex: sickle cell)
    - When one base is changed with another base = substitution
    - Usually called a missense – DNA codes for wrong amino acid

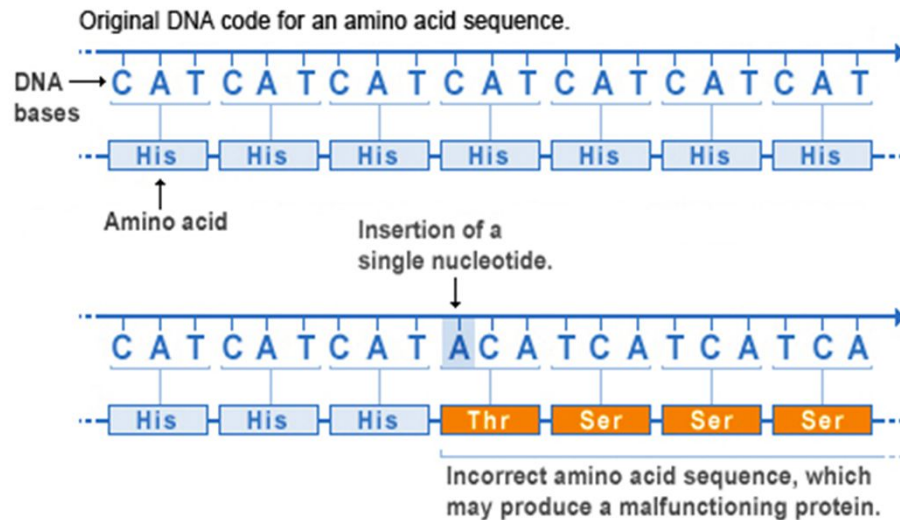
# Mutations

- Point mutations
  - Nonsense mutation changes a codon from an amino acid to a stop codon
  - Causes translation to stop early
  - Leads to proteins that cannot function normally
  - Leads to possible genetic disorders (ex: blood disorders)



# Mutations

## Insertion mutation



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## ■ Insertions

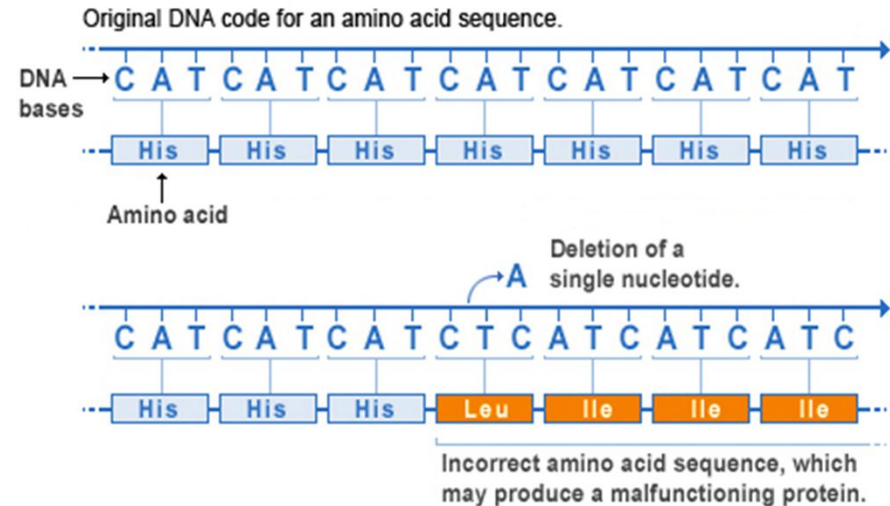
- Additions of a nucleotide to the DNA sequence
- Change the multiple of three
- These can also be called a frameshift
- Can cause disorders like Crohn's disease

# Mutations

## ■ Deletions

- Loss of a nucleotide in the DNA sequence
- Changes the multiples of three in the code
- Also called a frameshift
- Can cause disorders like cystic fibrosis

### Deletion mutation



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# Mutations

Mutations		
Mutation Type	Analogy Sentence	Example of Associated Disease
Normal	THE BIG FAT CAT ATE THE WET RAT	
Missense (substitution)	THE BIZ FAT CAT ATE THE WET RAT	Achondroplasia: improper development of cartilage on the ends of the long bones of arms and legs resulting in a form of dwarfism
Nonsense (substitution)	THE BIG RAT	Muscular dystrophy: progressive muscle disorder characterized by the progressive weakening of many muscles in the body
Deletion (causing frameshift)	THB IGF ATC ATA TET HEW ETR AT	Cystic fibrosis: characterized by abnormally thick mucous in the lungs, intestines, and pancreas
Insertion (causing frameshift)	THE BIG ZFA TCA TAT ETH EWE TRA	Crohn's disease: chronic inflammation of the intestinal tract, producing frequent diarrhea, abdominal pain, nausea, fever, and weight loss
Duplication	THE BIG FAT FAT CAT ATE THE WET RAT	Charcot-Marie-Tooth disease (type 1A): damage to peripheral nerves leading to weakness and atrophy of muscles in hands and lower legs
Expanding mutation (tandem repeats) Generation 1 Generation 2 Generation 3	THE BIG FAT CAT ATE THE WET RAT THE BIG FAT CAT CAT CAT ATE THE WET RAT THE BIG FAT CAT CAT CAT CAT CAT CAT ATE THE WET RAT	Huntington's disease: a progressive disease in which brain cells waste away, producing uncontrolled movements, emotional disturbances, and mental deterioration