DNA Polymorphism - 2

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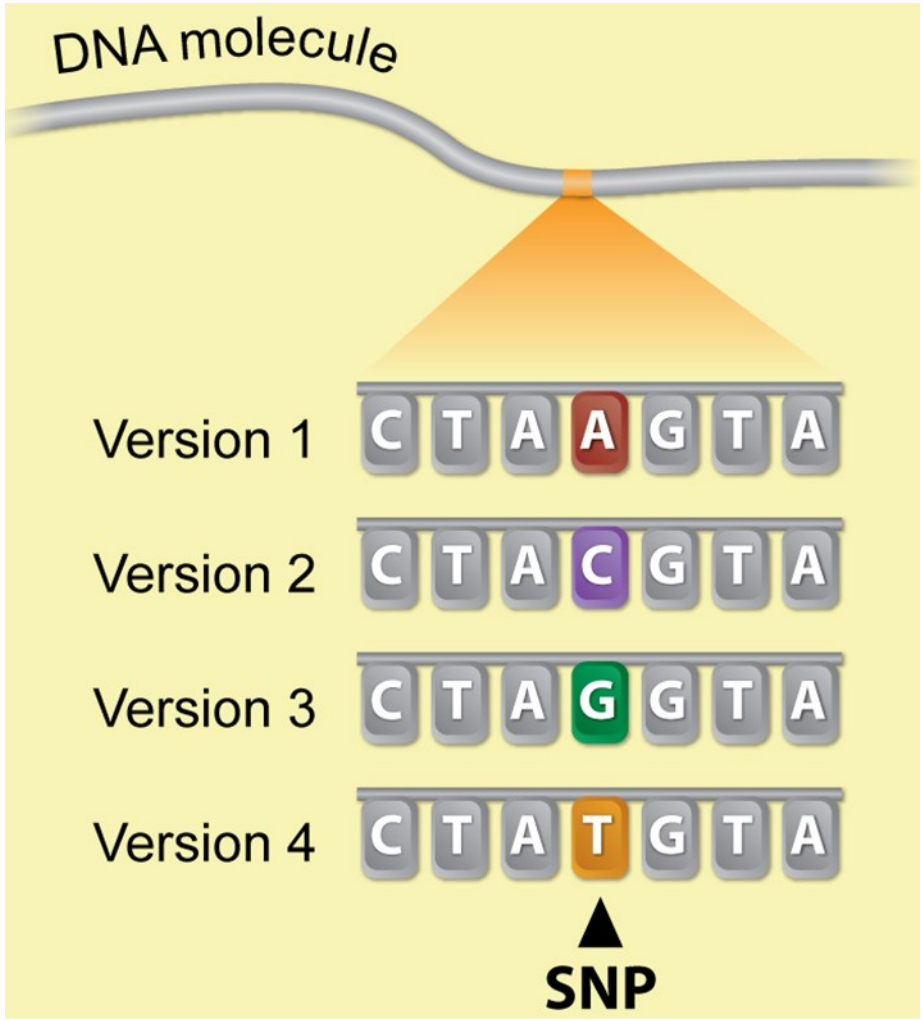
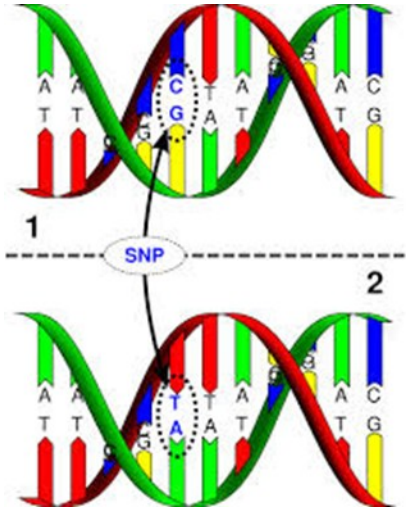
DNA Polymorphism

- DNA polymorphism is any change in nucleotide sequence at the same point at genome between individuals
- These different nucleotide sequence are common in population (>1% of population has the different sequence)
- If the nucleotide sequence is rare (<1%) in population, it’s considered mutation

DNA Polymorphism

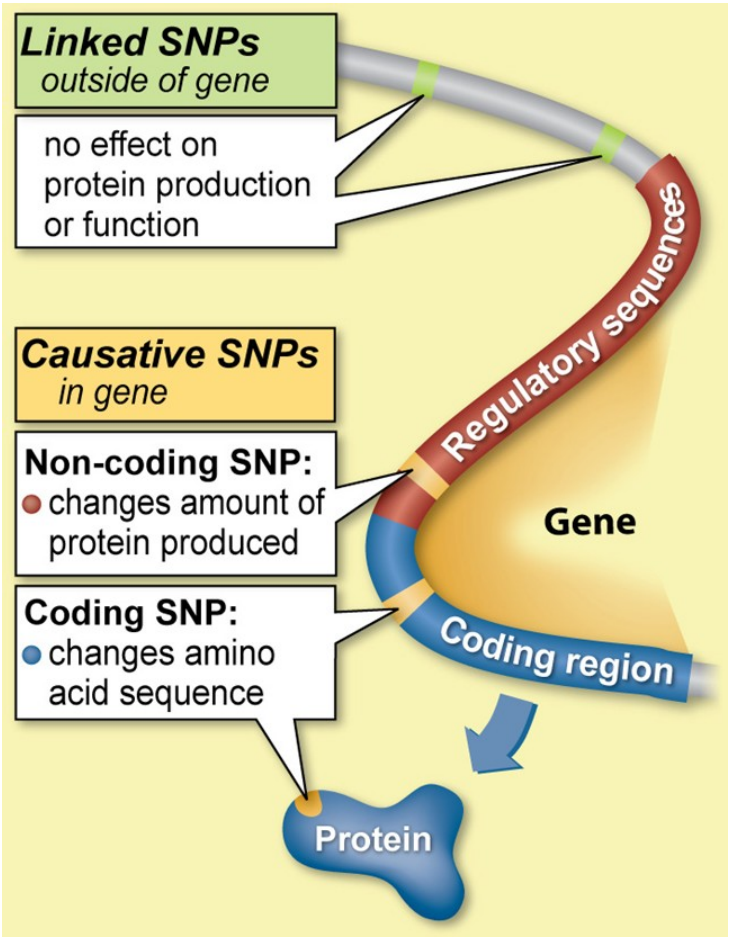
- Most common form of DNA polymorphism is Single Nucleotide Polymorphism (SNP)
- Single base differs between individuals (being A instead of G, for example)
- Not all single-nucleotide changes are SNP
- To be classified as a SNP, two or more versions of a sequence must each be present in at least one





SNP

- SNPs occur throughout the human genome—about one in every 300 nucleotide base pairs
- 10 million SNPs within the 3-billion-nucleotide human genome
- Location of SNP in genome matters, whether on gene or our outside of gene
- Gene make up only ~ 2% of genome, function of rest of DNA unclear (Eukaryotes)



DNA Polymorphism and disease

- *Current estimate: Defects in 5-10 % of gene results in disease*
- *Especially single genes that code of important protein or part of important protein complex*
- *Most of these disease are rare*

Disease	Gene	Defect	Clinical symptoms
Cystic Fibrosis	CFTR	Na/Cl Channel transporter defect	Thick mucus secretion, infection, blocked airways
Huntington's Disease	Huntington's Gene	Increase in neuronal death	Involuntary jerky movement, rigidity, possible dementia, seizures

DNA Polymorphism and disease

Disease	Gene	Defect	Clinical symptoms
Retinoblastoma	RB1 gene	Defect in regulator gene that inhibits cell cycle progression	Eye tumors, cancers in affected individuals
Phenylketonuria	PAH gene	Defect in phenylalanine hydroxylase enzyme	Mental retardation, epilepsy