QBIO490 Genomics/Sequencing Worksheet

1. Mutations in DNA
2. What is a point mutation?

A point mutation is when a single base in a DNA strand is mutated or changed.

1. What are three different types of point mutations?

Deletion (removing a single base, causing a frameshift), Insertion (adds a single base, causing a frameshift), and Substitution (swapping one base for another, may have a serious effect or may have no effect).

1. How does a frame shift mutation affect a resulting protein?

A frame shift causes the reading frame for the DNA to move. This typically causes the amino acid chain that’s produced from this DNA to be significantly modified (and likely leads to a nonfunctioning or damaging protein) from the intended sequence, as a frameshift influences all 3-base codon frames downstream from it.

2/3. A short history of genomics & Next Generation Sequencing

1. Explain the steps of NGS in your own words.

NGS starts with the breakage of DNA fragments into pieces. Two different oligonucleotides are attached to the ends of these fragments, allowing DNA fragments to stick to a chip with other oligonucleotides. Using PCR techniques (specifically bridge amplification), the chip will become saturated with the DNA fragments. Next, the DNA fragments are sequenced by using tagged nucleotide bases that emit signals, allow us to figure out which bases are being used to match our chip strands. Lastly, the small fragments are overlayed onto each other using their overlap (from each part of the DNA), which allows the fragments to be reconciled as a whole DNA strand again.