# 8.7

# Power Notes

### Gene Mutations:

# Mutations that affect a single gene

- Point Mutation: One nucleotide is substituted for another; may be fixed by DNA polymerase
- Frameshift Mutations: Involves the insertions or deletion of a nucleotide in the DNA sequence; Shifts the entire sequence by one or more nucleotides; throws off the reading frame

### Chromosomal Mutations:

# Mutations that affect an entire chromosome; affects many genes

- Gene Duplication; Caused by the exchange of unequal segments during crossing over; results in one chromosome having two copies of some genes and the other chromosome having no copies of those genes
- Translocation; Movement of a piece of one chromosome to a non-homologous chromosome; they are often reciprocal

### **Mutations**

## Potential:

Chromosomal mutations typically have a large effect on a organism: May result in a disrupted gene or abnormal regulation of genes

Point mutations may result in premature stop codons or amino acids with very different properties from the correct amino acid; May disrupt a splice site; May disrupt a regulatory DNA sequence

Frameshift mutations may result in a completely altered protein or a premature stop codon

## Silent:

Some mutations have no apparent effect.

A point mutation may not change the amino acid that is coded for.

Even if a change occurs, the change may be in an intron that is removed and thus has no effect.

A change may not significantly affect the function of a protein if the new amino acid is similar to the correct one or occurs away from the active site or does not influence protein structure

### Mutagens:

Agents in the environment that can change DNA; Some occur naturally (UV light); some are created by industrial processes such as toxins in factory production (smoke in the air or water etc.)