PSC126: Health Psychology Winter 2008
Handout on genetics

thalassemia: a genetic blood disease characterized by absent or decreased production of normal hemoglobin, resulting in anemia.

hemoglobin: a protein produced in bone marrow, and residing in red blood cells; composed of four polypeptides and four heme groups. Each heme group contains a molecule of iron. Hemoglobin transports oxygen to tissues.

genes: the fundamental physical and functional units of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome that encodes a specific functional product.

nucleotides: A subunit of DNA consisting of a nitrogenous base (adenine, guanine, thymine, or cytosine in DNA; adenine, guanine, uracil, or cytosine in RNA), a phosphate molecule, and a sugar molecule (deoxyribose in DNA and ribose in RNA). Thousands of nucleotides are linked to form a DNA or RNA molecule.

DNA: a linear molecule called deoxyribonucleic acid, composed of multiple genes.

chromosome: the self- replicating genetic structures of cells containing the cellular DNA that bears in its nucleotide sequence the linear array of genes.

template strand: the strand of the DNA double helix that is copied by base pair complementarity to make an RNA. The other, non-template strand of the DNA duplex has a sequence that is identical to the synthesized RNA (except in RNA, U replaces T).

triplet: set of three adjacent bases in a DNA molecule, that codes for an amino acid

amino acids: any of a class of 20 molecules that are combined to form proteins in living things.

proteins: large molecules composed of one or more chains of amino acids in a specific order; the order is determined by the base sequence of nucleotides in the gene coding for the protein. Proteins are required for the structure, function, and regulation of the bodys cells, tissues, and organs, and each protein has unique functions. Examples are hormones, enzymes, and antibodies.

meiosis: process of cell division involved in producing reproductive cells

polypeptide: a long chain of amino acids; proteins are built from one or more polypeptides

enzymes: a protein that acts as a catalyst, speeding the rate at which a biochemical reaction proceeds but not altering the direction or nature of the reaction.

catalyst: a substance able to increase the rate of a chemical reaction without itself being consumed or changed

transcription: the process in which the code in DNA is copied to RNA

RNA: a molecule called ribonucleic acid, composed of a phosphate molecule, and the sugar ribose, and 4 bases

RNA polymerase: an enzyme that catalyzes the synthesis of RNA from a DNA template

codons: a set of 3 nucleotide sequences in mRNA

mRNA: messenger RNA carries info for protein construction from DNA to ribosomes

ribosomes: structures where protein synthesis takes place

translation: information from mRNA is translated into sequence of amino acids

tRNA: transfer RNA, involved in translation, contains a 3-base anticodon (which is the complement of the codon) and an amino acid

promoter region: stretch of DNA in front of a gene, where RNA polymerase attaches to begin

transcription

activator protein: a protein that can bind to the promoter, which attracts the RNA polymerase and begins transcription

repressor protein: a protein that can bind to the promoter (or interact with transcription factors), which can prevent attachment of the RNA polymerase, and so inhibit transcription

transcription factors: specific proteins which allow DNA recognition by RNA polymerase

mutation: a change in the base sequence of an organism's DNA

missense mutation: substitution of an incorrect amino acid for a correct one

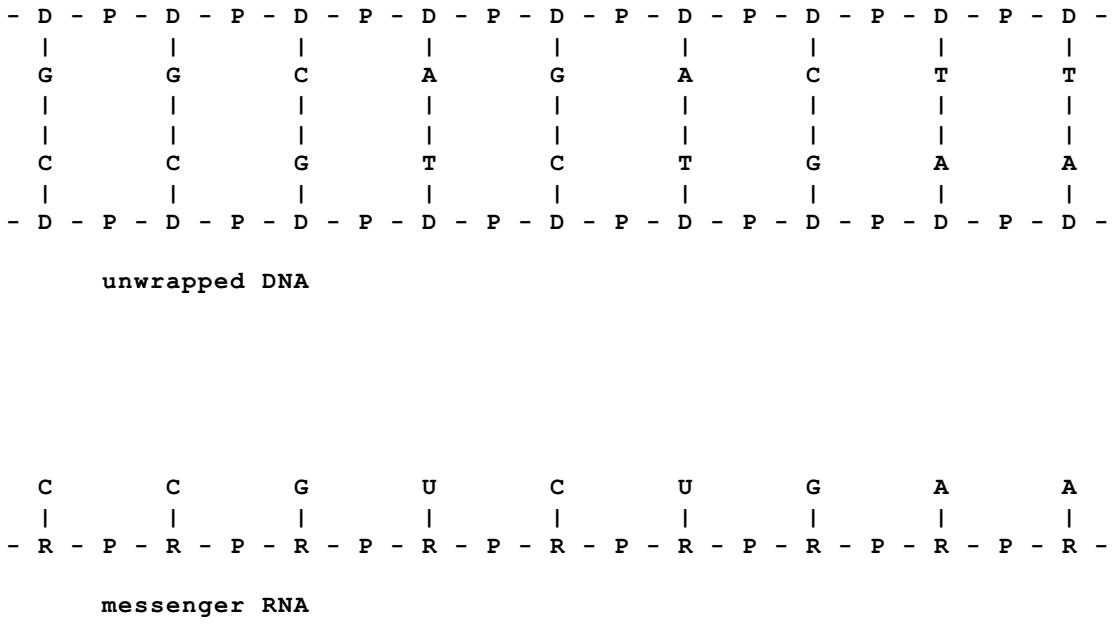
nonsense mutation: substitution of a stop codon for an amino acid, stopping translation, making a shorter protein

frameshift mutation: mutation where reading frame is changed, so true triplet codes are off by one base pair

mutagens: physical and chemical agents that cause mutations

carcinogens: mutagens that result in cancer

chelator: a compound that can remove iron from the body



Hemoglobin Mutants: Missense, Nonsense, and Frameshift

Normal beta chain

```
ATG GTG CAC CTG ACT CCT GAG GAG AAG TCT GCC GTT ACT GCC CTG TGG GGC AAG GTG AAC GTG GAT GAA GTT GGT GGT GAG GCC CTG GGC
   Val His Leu Thr Pro Glu Glu Lys Ser Ala Val Thr Ala Leu Trp Gly Lys Val Asn Val Asp Glu Val Gly Gly Glu Ala Leu Gly
```

HbS Sickle cell (missense)

```
ATG GTG CAC CTG ACT CCT GTG GAG AAG TCT GCC GTT ACT GCC CTG TGG GGC AAG GTG AAC GTG GAT GAA GTT GGT GGT GAG GCC CTG GGC
   Val His Leu Thr Pro Val Glu Lys Ser Ala Val Thr Ala Leu Trp Gly Lys Val Asn Val Asp Glu Val Gly Gly Glu Ala Leu Gly
```

HbC (missense)

```
ATG GTG CAC CTG ACT CCT AAG GAG AAG TCT GCC GTT ACT GCC CTG TGG GGC AAG GTG AAC GTG GAT GAA GTT GGT GGT GAG GCC CTG GGC
   Val His Leu Thr Pro Lys Glu Lys Ser Ala Val Thr Ala Leu Trp Gly Lys Val Asn Val Asp Glu Val Gly Gly Glu Ala Leu Gly
```

HbThalassemia (nonsense)

```
ATG GTG CAC CTG ACT CCT GAG GAG AAG TCT GCC GTT ACT GCC CTG TGG GGC TAG GTG AAC GTG GAT GAA GTT GGT GGT GAG GCC CTG GGC
   Val His Leu Thr Pro Glu Glu Lys Ser Ala Val Thr Ala Leu Trp Gly Stop
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HbThalassemia (frameshift) -AA

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ATG GTG CAC CTG ACT CCT GAG GAG GTC TGC CGT TAC TGC CCT GTG GGG CAA GGT GAA CGT GGA TGA AGT TGG TGG TGA GGC CCT GGG C
   Val His Leu Thr Pro Glu Glu Val Cys Arg Tyr Cys Pro Val Gly Gln Gly Glu Arg Ala Stop
```

The top line above shows the DNA code and amino acid sequence for the first 29 amino acids of the beta globin chain. The next four lines show the same information for four, different mutants. The changed base and amino acid(s) shown in red.

HbS and HbC are both missense mutants at the #6 amino acid position substituting valine or lysine for glutamic acid.

Hb Thalassemia is a general name for a reduction in the amount of hemoglobin produced. The first HbThal mutant shown has a Stop or Termination codon (TAG) at position #17 in place of the codon for lysine. The second HbThal mutant results from the deletion of two adjacent adenines at position #8 causing a shift in the reading frame to produce a series of missense amino acids terminating at a now in-phase, early termination codon.