COSEE-West lecture & workshop, April 2006
"Genetics"

GLOSSARY

Adenine (A): A nitrogenous base, one member of the *base pair* A-T (*adenine-thymine*).

Alleles: Alternative forms of a genetic *locus*; a single allele for each locus is inherited separately from each parent (e.g., at a locus for eye color the allele might result in blue or brown eyes).

Amino acid: Any of a class of 20 molecules that are combined to form *proteins* in living things. The sequence of amino acids in a protein and hence protein function are determined by the *genetic code*.

Autosome: A *chromosome* not involved in sex determination. The *diploid* human *genome* consists of 46 chromosomes, 22 pairs of autosomes, and 1 pair of *sex chromosomes* (the X and Y chromosomes).

Base: The chemical building blocks of DNA. Named A, T, C, and G (adenine, cytosine, thymine, and guanine), these bases pair up to form the "stairs" of the DNA double helix and always combine in the same patterns: A with T and C with G.

Base sequence: The order of *nucleotide* bases in a DNA molecule.

Cells: The smallest unit of living matter that can operate independently.

Chromosomes: Long strands of DNA on which genes are found. Each human cell has 46 chromosomes in 23 pairs. One member of each pair is inherited from the mother, the other from the father.

Codon: See *genetic code*.

Complementary DNA (cDNA): DNA that is synthesized from a *messenger RNA* template; the single-stranded form is often used as a probe in physical mapping.

Complementary sequences: *Nucleic acid base sequences* that can form a double- stranded structure by matching *base pairs*; the complementary sequence to G- T- A- C is C- A- T- G.

Conserved sequence: A *base sequence* in a DNA molecule (or an *amino acid* sequence in a *protein*) that has remained essentially unchanged throughout evolution.

Cytosine (C): A *nitrogenous base*, one member of the *base pair* G- C (*guanine* and *cytosine*).

Deoxyribonucleotide: See *nucleotide*.

Diploid: A full set of genetic material, consisting of paired *chromosomes* one chromosome from each parental set. Most animal cells except the *gametes* have a diploid set of chromosomes. The diploid human *genome* has 46 chromosomes. Compare *haploid*.

DNA (Deoxyribonucleic Acid): The double helix-shaped molecule that holds an organism's genetic information. DNA is composed of sugars, phosphates, and four nucleotide bases: adenine, guanine, cytosine, and thymine (A, G, C, T). The bases bind together in specific pairs.

DNA replication: The use of existing DNA as a template for the synthesis of new DNA strands. In humans and other *eukaryotes*, replication occurs in the cell *nucleus*.

DNA sequence: The relative order of *base pairs*, whether in a fragment of DNA, a *gene*, a *chromosome*, or an entire *genome*.

Double Helix: The shape of DNA, much like a spiral staircase or twisted ladder. The stairway's railings are composed of sugars and phosphates. Its sides contain the patterned base pairs: A, T, C, and G. When a cell divides for reproduction, the helix unwinds and splits down the middle like a zipper in order to copy itself.

Enzyme: 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.

Eukaryote: Cell or organism with membrane- bound, structurally discrete *nucleus* and other well-developed subcellular compartments. Eukaryotes include all organisms except viruses, bacteria, and blue- green algae. Compare *prokaryote*. See *chromosomes*.

Gene: Segments of DNA that are the basic functional units of heredity. Genes are determined by an ordered sequence of chemical bases found in a unique position on a specific chromosome. Their "blueprint" guides protein production, which determines how different cells in the body function. Inherited genes also control an animal's unique set of physical traits.

Gene expression: The process by which a *genes* coded information is converted into the structures present and operating in the cell. Expressed genes include those that are transcribed into *mRNA* and then translated into *protein* and those that are transcribed into *RNA* but not translated into protein (e.g., *transfer* and *ribosomal RNAs*).

Gene mapping: Determination of the relative positions of *genes* on a DNA molecule (*chromosome* or *plasmid*) and of the distance, in *linkage* units or physical units, between them.

Genetic code: The sequence of *nucleotides*, coded in triplets (*codons*) along the *mRNA*, that determines the sequence of *amino acids* in *protein* synthesis. The DNA sequence of a *gene* can be used to predict the mRNA sequence, and the genetic code can in turn be used to predict the *amino acid* sequence.

Genetic marker: Random mutations in the DNA sequence which act as genetic milestones. Once markers have been identified they can be traced back in time to their origin—the most recent common ancestor of everyone who carries the marker.

Genetics: The study of the patterns of inheritance of specific traits.

Genome: The total DNA sequence that serves as an instruction manual for all proteins created in our body. Two copies of the genome are found inside each of our cells.

Guanine (G): A nitrogenous base, one member of the *base pair* G- C (guanine and *cytosine*).

Haploid: A single set of *chromosomes* (half the full set of genetic material), present in the egg and sperm cells of animals and in the egg and pollen cells of plants. Human beings have 23 chromosomes in their reproductive cells. Compare *diploid*.

Heredity: The total sum of genetic information that humans pass on from generation to generation.

Homologies: Similarities in DNA or *protein* sequences between individuals of the same species or among different species.

Homologous chromosomes: A pair of *chromosomes* containing the same linear *gene* sequences, each derived from one parent.

Linkage: The proximity of two or more *markers* (e.g., *genes, RFLP* markers) on a *chromosome*; the closer together the markers are, the lower the probability that they will be separated during DNA repair or replication processes (binary fission in *prokaryotes, mitosis* or *meiosis* in *eukaryotes*), and hence the greater the probability that they will be inherited together.

Linkage map: A map of the relative positions of genetic *loci* on a *chromosome*, determined on the basis of how often the loci are inherited together. Distance is measured in *centimorgans (cM)*

Karyotype: A photomicrograph of an individuals *chromosomes* arranged in a standard format showing the number, size, and shape of each chromosome type; used in low- resolution *physical mapping* to correlate gross chromosomal abnormalities with the characteristics of specific diseases.

Marker: An identifiable physical location on a *chromosome* (e.g., *restriction enzyme cutting site, gene*) whose inheritance can be monitored. Markers can be expressed regions of DNA (genes) or some segment of DNA with no known coding function but whose pattern of inheritance can be determined. See *RFLP, restriction fragment length polymorphism*.

Meiosis: The process of two consecutive cell divisions in the *diploid* progenitors of sex cells. Meiosis results in four rather than two daughter cells, each with a *haploid* set of *chromosomes*.

Messenger RNA (mRNA): RNA that serves as a template for protein synthesis. See genetic code.

Mitochondria: A remnant of an ancient parasitic bacteria that now helps to produce energy inside the cell. A mitochondrion has its own genome, present in only one copy, which does not recombine in reproduction. This genetic consistency makes mitochondrial DNA a very important tool in tracking genetic histories.

Mitochondrial DNA or mtDNA: Genetic material found in the mitochondria. It is passed from females to their offspring without recombining, and thus is an important tool for geneticists.

Mutation: Any heritable change in DNA *sequence*. Compare *polymorphism*.

Nucleic acid: A large molecule composed of *nucleotide* subunits.

Nucleotide: A subunit of DNA or RNA 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. See *DNA, base pair, RNA.*

Nucleus: The cellular organelle in *eukaryotes* that contains the genetic material.**Nucleotide** A DNA building block which contains a base, or half of a "staircase step," and sugars and phosphates which form the "railing." Nucleotides join together to form DNA's distinctive double helix shape.

Phylogeny: The evolutionary development of a species. Phylogeny is sometimes represented as a tree that shows the natural relations and development of all species.

Polymerase chain reaction (PCR): A method for amplifying a DNA *base sequence* using a heat- stable *polymerase* and two 20- base *primers*, one *complementary* to the (+)- strand at one end of the sequence to be amplified and the other complementary to the (-)- strand at the other end. Because the newly synthesized DNA strands can subsequently serve as additional templates for the same primer sequences, successive rounds of primer annealing, strand elongation, and dissociation produce rapid and highly specific amplification of the desired sequence. PCR also can be used to detect the existence of the defined sequence in a DNA sample.

Polymerase, DNA or RNA: *Enzymes* that catalyze the synthesis of *nucleic acids* on preexisting nucleic acid templates, assembling RNA from ribonucleotides or DNA from deoxyribonucleotides.

Polymorphism: Difference in DNA sequence among individuals. Genetic variations occurring in more than 1% of a population would be considered useful polymorphisms for genetic *linkage* analysis. Compare *mutation*.

Population genetics: The study of genetic variation in a species.

Prokaryote: Cell or organism lacking a membrane- bound, structurally discrete *nucleus* and other subcellular compartments. Bacteria are prokaryotes. Compare *eukaryote*. See *chromosomes*.

Proteins: Linear sequences of amino acids that are the building blocks of cells. Each protein has a specific function that is determined by the "blueprint" stored in DNA.

Recombination: The process by which each parent contributes half of an offspring's DNA, creating an entirely new genetic identity. This process mixes genetic signals, so that non-recombining DNA, passed intact through the generations, is most important to population genetics.

Replication: The process by which two DNA strands separate, with each helping to duplicate a new strand. During reproduction, the DNA double helix unwinds and duplicates itself to pass on genetic information to the next generation. Because bases always form established pairs (AT and CG), the sequence of bases on each strand will attract a corresponding match of new bases. Only occasional errors occur—about one for every billion base-pair replications.

Restriction fragment length polymorphism (RFLP): Variation between individuals in DNA fragment sizes cut by specific *restriction enzymes; polymorphic sequences* that result in RFLPs are used as *markers* on both *physical maps* and genetic *linkage maps*. RFLPs are usually caused by *mutation* at a cutting site. See *marker*.

Ribonucleic acid (RNA): A chemical found in the *nucleus* and cytoplasm of cells; it plays an important role in protein synthesis and other chemical activities of the cell. The structure of RNA is similar to that of DNA. There are several classes of RNA molecules, including *messenger RNA, transfer RNA, ribosomal RNA*, and other small RNAs, each serving a different purpose.

RNA (Ribonucleic Acid): Transfers the genetic "blueprint" that is stored in DNA during protein production. RNA has a single-stranded linear structure and a slightly different chemical composition from DNA.

Sequencing: Determines the order of nucleotides for any particular DNA segment or gene. The order of a DNA string's base pairs determines which proteins are produced, and thus the function of a particular cell.

Single Nucleotide Polymorphism: Small, infrequent changes which help to create an individual's own unique DNA pattern. When a single nucleotide (A, T, G, or C) is altered during DNA replication, due to a tiny "spelling mistake," the genome sequence is altered.

Thymine (T): A nitrogenous base, one member of the *base pair* A- T (*adenine*- thymine).

Trait: The physical characteristics, like eye color or nose shape, which are determined by inherited genes.

Transcription: The synthesis of an *RNA* copy from a *sequence* of DNA (a *gene*); the first step in *gene expression*. Compare *translation*.

Transfer RNA (tRNA): A class of *RNA* having structures with triplet *nucleotide* sequences that are *complementary* to the triplet nucleotide coding sequences of *mRNA*. The role of tRNAs in protein synthesis is to bond with *amino acids* and transfer them to the ribosomes, where proteins are assembled according to the genetic code carried by mRNA.

Transformation: A process by which the genetic material carried by an individual cell is altered by incorporation of exogenous DNA into its *genome*.

Translation: The process in which the genetic code carried by mRNA directs the synthesis of *proteins* from amino acids. Compare *transcription*.