Glossary for Anth 3234: Genes, Health, and History

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allele When several variants exist at a genetic locus, they are said to be *alleles*. For example, Mendel's gene for seed coat texture had two alleles, one for round and the other for wrinkled seeds.

centromere A portion of each chromosome, which is essential during the production of haploid gametes—sperm and ova.

chromosome A molecule of DNA. A single chromosome may contain thousands of genes, and humans have 23 pairs of chromosomes.

diploids and haploids Humans (and most multicellular animals and plants) are *diploid*: they have two copies of each chromosome and (consequently) of each gene. Human gametes (sperm and ova) are *haploid*: they contain just one copy of each chrosome (23 chromosomes in all). Thus, the human life cycle contains both diploid and haploid stages.

coalesce When we trace two lineages backwards in time, they will eventually join, or *coalesce*, at their most recent common ancestor.

coalescent event the point at which two lineages coalesce, as we tract their ancestry backwards in time.

gene A stretch of DNA that begins with a start sequence and ends with a stop sequence. Such stretches and only such stretches are used in protein synthesis. We used to teach that a gene is a piece of DNA that codes for a single protein. However, we now know that some proteins are assembled from smaller proteins whose genes are widely separated on the chromosome, or even lie on different chromosomes altogether. Thus, it is not necessarily the case that, for every protein their is a single Mendelian gene. Furthermore, some genes code for RNA molecules that are never translated into protein. Thus, it seems best to say only that a gene is a stretch of DNA that codes for a single function.

dominance If the phenotype of individuals with genotype Aa is identical to that of genotype AA, but not to aa, then A is said to be *fully dominant* to a. If the heterozygote, Aa, is intermediate between the two homozygotes, then there is *no dominance.* If the value of the heterozygote exceeds that of either homozygote, there is *overdominance*. If the heterozygote's value is less than that of either homozygote, there is *underdominance*.

genotype The genotype of individual is a description of the genes that he carries.

heterozygote An individual with copies of two different alleles at a given locus.

homozygote An individual with two copies of the same allele at a given locus.

independent assortment of chromosomes. Consider an individual whose 2-locus genotype is Aa/Bb (This means that alleles A and a are present at the first locus, B and b at the second). He is capable of producing four kinds of gametes: AB, Ab, aB, and ab. Mendel's principle of independent assortment says that these gamete types are produced with equal frequency. This principle holds when the two loci are on different chromosomes, but not when they are close together on the same chromosome.

introgression the process by which alleles are transmitted from one population to another.

lineage Trace the history of a nucleotide backwards in time, from the observed nucleotide to its parent to its parent and so on. This path is called a *lineage* or a *line of descent*.

line of descent see "lineage."

linkage of loci. Loci that are close together on the same chromosome are *linked* and do not assort

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independently. Suppose that the Aa/Bb individual above inherited an AB gamete from his father and an ab gamete from his mother. Then the AB and ab gamete types are referred to as *parental* gamete types, while Aa and aB are *recombinant* gamete types. In general, recombinant gametes are produced less frequently than parental gametes unless the loci are on different chromosomes, or very far apart on the same chromosome. If recombinant gametes are produced very rarely, linkage is said to be tight. If they are as common as parental gametes, the loci are said to be *unlinked*.

linkage disequilibrium (LD) When an allele at one locus tends to co-occur with one at another locus, the two loci are in *linkage disequilibrium*. The opposite case—*linkage equilibrium*—occurs when alleles at two loci assort randomly on chromosomes. The term is confusing, because linkage disequilibrium can occur without genetic linkage, and linkage equilibrium does not entail the usual meaning of "equilibrium"—a state with no tendency to change.

locus A location on a chromosome.

Mesolithic The Middle Stone Age, lasting in Europe from the end of the last Ice Age, 10 ky ago, until the rise of agriculture around 7 ky ago. The end date varies from place to place.

Neolithic The New Stone Age, lasting in Europe from the appearance of agriculture until the appearance of metal tools.

Paleolithic The Old Stone Age, lasting from about 2.5 my ago until the end of the last Ice Age, 10 ky ago.

phenotype The detectable properties of an individual.

recombination Chromosomes are usually not passed on just as they were inherited. As the strands of homologous chromosomes intertwine, they occasionally break and reattach. Sometimes the left end of the maternal chromosome gets attached to the right end of the paternal chromosome. This is called *crossing over*, or *recombination*. Recombinant gametes may be produced if crossing over has occurred between the two loci. Since this is less likely to occur if the loci are close together on the chromosome, such loci seldom produce recombinant gametes. This principal underlies the classical methods for mapping chromosomes.

segregation of alleles. Consider a heterozygote whose genotype is Aa. When this individual produces gametes, the two genes *segregate*, so that some gametes carry A and some a. Mendel's law of segregation says that these types occur with equal frequency. This law usually holds, but there are rare exceptions that need not concern us here.

/selective sweep] A *selective sweep* occurs when a favorable allele arises (either by mutation or by introgression from another population) and increases in frequency under the influence of natural selection.

telomere The ends of a chromosome. Telomeres contain repetitive DNA, some of which is lost each time the cell divides.