

# PLINX204 PSYCHOLINGUISTICS LANGUAGE ACQUISITION - 2006

## Glossary of terms in genetics (corrected)

Adenine - see Cytosine

Allele - alternative forms of the same gene that occupy the same locus (place) on a chromosome. The forms differ in nucleotide sequence, such that it affects the phenotype: e.g. those responsible for skin colour, as opposed to those invariant ones responsible for e.g. number of legs. Alleles are in fact mutant gene sequences, especially when the change(s) in nucleotide sequences result in a dysfunctional gene product, as is the case in phenylketonuria.

Amino acid - One of the 20 building blocks of proteins, specified by a codon of DNA (3 nucleotides).

Assortative mating - the tendency for like to mate with like

Autosomal - any chromosome which is not a sex (X or Y) chromosome.

Base pair - One step in the spiral staircase of the double helix of DNA, consisting of adenine bonded to thymine or cytosine bonded to guanine.

Chromosome - A structure composed mainly of chromatin, which contains DNA and protein and resides in the nucleus of cells. See also 'Genome'.

Codon - A sequence of 3 bases that code for a particular amino acid.

Concordance - Presence of a particular trait in two family members.

Cytosine - one of the four bases in DNA that make up the letters ATGC, cytosine is the "C". The others are adenine, guanine, and thymine. Cytosine always pairs with guanine.

DNA - 'Deoxyribonucleic acid'. The molecules inside cells that carry genetic information and pass it from one generation to the next: the hereditary material in humans and almost all other organisms. Nearly every cell in a person's body has the same DNA. Most DNA is located in the cell nucleus (where it is called nuclear DNA), but a small amount of DNA can also be found in the mitochondria (where it is called mitochondrial DNA or mtDNA). The information in DNA is stored as a code made up of four chemical bases: adenine (A), guanine (G), cytosine (C), and thymine (T). DNA bases pair up with each other, A with T and C with G, to form units called base pairs. Each base is also attached to a sugar molecule and a phosphate molecule. Together, a base, sugar, and phosphate are called a nucleotide. Nucleotides are arranged in two long strands that form

a spiral called a double helix. The structure of the double helix is somewhat like a ladder, with the base pairs forming the ladder's rungs and the sugar and phosphate molecules forming the vertical sidepieces of the ladder.

Human DNA consists of about 3 billion bases, and more than 99 percent of those bases are the same in all people. The order, or sequence, of these bases determines the information available for building and maintaining an organism, similar to the way in which letters of the alphabet appear in a certain order to form words and sentences. Only about 5% of DNA is transcribed into RNA; the role of the rest is largely a mystery.

An important property of DNA is that it can replicate within an organism, or make copies of itself with the help of complicated protein machinery. Each strand of DNA in the double helix can serve as a pattern for duplicating the sequence of bases. This is critical when cells divide because each new cell needs to have an exact copy of the DNA present in the old cell.

**Dominant** - alleles that produce a particular phenotype when present in the heterozygous state.

**DZ (dizygotic)** - 'fraternal' twins; they share about 50% of their genes

**Effect size** - The proportion of individual differences for a trait in the population accounted for by a particular factor.

**Epigenetic** - factors dealing with the expression of genes rather than their identity. The study of heritable changes in gene function that occur without a change in the sequence of nuclear DNA. This includes the study of how environmental factors affecting a parent can result in changes in the way genes are expressed in the offspring

**Gamete** - The reproductive cells in multicellular organisms, in mammals the egg or sperm. Also known as 'germ cells'. They have a half set of chromosomes.

**Gene** - Segments of DNA usually a few thousand base pairs in length.

**Genome** - All the DNA of an organism. The human genome consists of two sex-chromosomes (males have an X and a Y chromosome, females have two X chromosomes) and 22 pairs of autosomal chromosomes. The autosomal chromosomes are numbered from 1 to 22 according to size and band staining patterns, with chromosome 1 being the largest. Each chromosome has a primary constriction (the centromere), which is used to delineate a short arm (conventionally labelled p) and a long arm (conventionally labelled q). Locations on these arms are specified on the basis of their major staining bands. For example, 15q21 refers to band 21 of the long arm of chromosome 15.

**Genomics** - The study of genes and their function.

**Genotype** - The entire genetic identity of an individual, including alleles, or gene forms, that do not show as outward characteristics.

Guanine - see Cytosine

Hemizygous - having only one allele of a gene or chromosome instead of the usual two.

Heterozygous - pertaining to the presence of two different alleles at a given locus.

Homozygous - pertaining to the presence of two of the same allele at a given locus.

Imprinting - the process by which maternally and paternally derived chromosomes are uniquely chemically modified leading to different expression of a certain gene or genes on those chromosomes depending on their parental origin.

Independent Assortment - see Mendel's second law

Linkage - An association between characters in inheritance, such that if one parent has a pair of characters, there is a probability greater than 50% that any offspring inheriting one of the characters will also inherit the other. The effect is due to the two characters being controlled by alleles of genes located in close proximity on the same chromosome.

Locus - The site of a specific gene on a chromosome.

LoD score - Log of the odds – a statistical indication of whether two loci are linked or unlinked.

Meiosis - a special method of cell division, occurring in maturation of the germ cells, by means of which each daughter nucleus receives half the number of chromosomes characteristic of the somatic cells of the species.

Messenger RNA (mRNA) - Processed RNA that leaves the nucleus of the cell and serves as a template for protein synthesis.

Mitosis - the process of nuclear division in cells that produces daughter cells that are genetically identical to each other and to the parent cell.

Mendel's first law: Segregation – A gene may exist in two or more different forms (alleles). The two alleles (one from each parent) separate (*segregate*) during gamete formation.

Mendel's second law: Independent Assortment: the inheritance of one gene is not affected by the inheritance of another gene. Violation of Mendel's second law indicates that genes are inherited together on the same chromosome - the basis for linkage analysis.

Methylation - the attachment of methyl groups to DNA at cytosine bases; correlated with reduced transcription of the gene and thought to be the principal mechanism in X-chromosome inactivation and imprinting.

Mutation - a heritable change in DNA base pair sequences.

MZ (monozygotic) - 'identical' twins; they are (almost) genetically identical.

Nucleotide - see DNA

Penetrance - the extent to which the properties controlled by a gene are expressed in the phenotype.

Phenotype - the appearance of an individual that results from the genotype and the environment.

PKU (phenylketonuria) - a single gene recessive disorder, caused by any of 100 mutations in a gene named PAH, and which results in an enzyme which is unable to break down phenylalanine (an amino acid found in all food containing protein).

Pleiotropy - the production by a single gene of two or more apparently unrelated phenotypic effects. (e.g. PKU)

Polygenic - referring to the production by a number of genes of a single phenotypic effect. (e.g. autism).

Polymorphism - a locus with two or more alleles; i.e. multiple possible states for a single property, often described as mutations.

Quantitative Trait Locus (QTL) - a region of DNA that is associated with a particular trait (e.g., plant height). Though not necessarily genes themselves, QTLs are stretches of DNA that are closely linked to the genes that underlie the trait in question.

Recessive - alleles that produce a particular phenotype only when present in the homozygous state.

Ribosome - An organelle that translates mRNA into a protein (a polypeptide chain)

RNA - Ribonucleic acid. The information carried by DNA is decoded in two steps: transcription of DNA into RNA, and translation of RNA into proteins. In the transcription process the sequence of bases in one strand of the DNA double helix is copied to RNA, specifically messenger RNA (mRNA). mRNA is single stranded and is formed by a process of base pairing similar to the replication of DNA except that uracil substitutes for thymine, so that A pairs with U instead of T. In the second step mRNA is translated into amino acids that form proteins. Another form of RNA ('transfer RNA' or tRNA) transfers amino acids to the ribosomes.

Segregation - see Mendel's first law

Sex-linked - associated with the sex chromosomes. Red-green colour-blindness is sex-linked because the genes for red and green colour perception are located on the X chromosome, of which men have only one, whereas women have two.

Thymine - see Cytosine

Uracil - see RNA

Zygote - the cell resulting from the union of a sperm and an egg.

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