

## DEFINITIONS OF COMMONLY USED TERMS AND STATISTICAL TERMS IN POPULATION GENETICS

1. Population genetics is a subfield of genetics that deals with genetic differences within and among populations, and is a part of evolutionary biology.
2. A genome is all the genetic information of an organism.
3. A chromosome is a long DNA molecule with part or all of the genetic material of an organism.
4. A single-nucleotide polymorphism (SNP /snip/; plural SNPs /snips/) is a germline substitution of a single nucleotide at a specific position in the genome that is present in a sufficiently large fraction of the population (generally regarded as 1% or more).
5. A genome-wide association study (GWA study, or GWAS), is an observational study of a genome-wide set of genetic variants in different individuals to see if any variant is associated with a trait.
6. Polymorphic means that the data at that locus can have more than one possible variant.
7. Allele frequency, or gene frequency, is the relative frequency of an allele (variant of a gene) at a particular locus in a population, expressed as a fraction or percentage.
8. Heterozygous. A diploid organism is heterozygous at a gene locus when its cells contain two different alleles (one wild-type allele and one mutant allele) of a gene.
9. Homozygous. A cell is said to be homozygous for a particular gene when identical alleles of the gene are present on both homologous chromosomes.
10. In population genetics, the Hardy–Weinberg principle, also known as the Hardy–Weinberg equilibrium, model, theorem, or law, states that allele and genotype frequencies in a population will remain constant from generation to generation in the absence of other evolutionary influences.
11. Identical by descent (IBD) is a term used in genetic genealogy to describe a matching segment of DNA shared by two or more people that has been inherited from a common ancestor without any intervening recombination.
12. The coefficient of inbreeding ( $F$ ) of an individual is the probability that two alleles at any locus in an individual are identical by descent from the common ancestor(s) of the two parents.

13. Population structure (also called genetic structure and population stratification) is the presence of a systematic difference in allele frequencies between subpopulations. In a randomly mating (or panmictic) population, allele frequencies are expected to be roughly similar between groups.
14. Population substructure refers to features of a population which result in variation of expected allele frequencies across individuals in a population.
15. Population stratification is the presence of a systematic difference in allele frequencies between subpopulations in a population, possibly due to different ancestry, especially in the context of association studies.
16. The “admixture model” of STRUCTURE assumes that each individual has ancestry from one or more of  $K$  genetically distinct sources.
17. Population Inbreeding is defined as mating between individuals that are related by ancestry and is more likely in populations that are, or have been, small.
18. Population admixture refers to a situation where individuals in population have a mixture of different genetic ancestries due to the mixing of two or more populations at a previous point in time.
19. In genetics, coalescent theory is a retrospective model of population genetics that traces all alleles of a gene in a sample from a population to a single ancestral copy shared by all members of the population, known as the most recent common ancestor (MRCA)
20. Wright defined the basic inbreeding coefficient,  $F_{IT}$  or  $F$ , as the correlation between genes on uniting gametes relative to the total array of those in random derivatives of the foundation stock.
21. The Wright–Fisher model assumes a randomly mating population of finite size reproducing in discrete non-overlapping generations, by allowing the individuals in generation to choose parents at random from the previous generation.
22. Natural selection is the differential survival and reproduction of individuals due to differences in phenotype.
23. Genetic drift, also known as allelic drift or the Wright effect, is the change in the frequency of an existing gene variant (allele) in a population due to random chance.
24. A set of closely linked genetic markers or DNA variations on a chromosome that tend to be inherited together.
25. Haplotype inference is the process of recovering the haplotypes that explain the genotypes.