

POPULATION GENETICS DEFINITIONS

1. Polymorphism is the occurrence of two or more clearly different morphs or forms, also referred to as alternative phenotypes, in the population of a species.
2. Polymorphic means that the data at that locus can have more than one possible variant.
3. An autosome is any chromosome that is not a sex chromosome.
4. The genotype of an organism is its complete set of genetic material.
5. The phenotype is the set of observable characteristics or traits of an organism.
6. Structural variants include many types of chromosomal changes, including rearrangements, duplications, translocations inversions, deletion or insertions of genetic material.
7. A haplotype (haploid genotype) is a group of alleles in an organism that are inherited together from a single parent and they appear on the same chromosome.
8. Phasing is the task or process of assigning alleles (the As, Cs, Ts and Gs) to the paternal and maternal chromosomes.
9. Mendelian randomization (commonly abbreviated to MR) is a method using measured variation in genes to interrogate the causal effect of an exposure on an outcome.
10. Mendel's second law - "It states that the alleles of one gene sort into the gametes independently of the alleles of another gene."
11. The objective of MR is testing causal relationship between intermediate phenotype and disease outcome, by testing association between genotypic instrument and disease outcome.
12. Pleiotropy describes the genetic effect of a single gene on multiple phenotypic traits.
13. The curse of dimensionality refers to various phenomena that arise when analyzing and organizing data in high-dimensional spaces that do not occur in low-dimensional settings such as the three-dimensional physical space of everyday experience.
14. The expectation-maximization algorithm is an approach for performing maximum likelihood estimation in the presence of latent variables.
15. Haplotype diversity is the probability that two randomly chosen haplotypes are different.