Glossary:

Allele: One of several alternative forms of a unit (base) of DNA that may be present at a given location on a chromosome

Axon guidance: The neurodevelopmental signaling process by which neuronal projections (axons) extend towards a target

Bioinformatics analysis: The application of computational methods to the study of genetic data

GWAS (Genome-wide association study): An exploratory approach to identify genetic markers of traits and diseases by searching across the genome for locations in which the frequency of a polymorphism is associated with a trait of interest

Haplotype: A combination of genetic variants that are inherited together

Heterozygote/heterozygous: An individual's homologous chromosomes express different genetic variants at a given location

Homozygote/homozygous: Both of an individual's homologous chromosomes express the same genetic variant at a given location

Insertion-deletion polymorphisms (indels): A genetic variation in which one or more units of DNA are added (insertion) or removed (deletion)

Intergenic region: A portion of a chromosome that lies in between genes; these stretches of DNA do not code for proteins and are often of unknown function, though some may have regulatory effects on nearby genes

Intron: A portion of the DNA sequence within a gene that does not code for proteins

Linkage analysis: A method used to identify links between genetic markers (a DNA sequence in a chromosome) and traits or diseases by examining patterns of inheritance among individuals within a family who exhibit the trait of interest. Genetic markers that are specifically carried by family members who exhibit the given trait are inferred to be related to that trait and may be targeted as links to the trait in the general population.

Polymorphism: A varying form of a small structural unit of the DNA molecule (a base or series of bases) which can be expressed in different forms or numbers of repetitions

Sequencing: The process of identifying the order of the small units (bases) that make up DNA and genes

SNP (single nucleotide polymorphism): A genetic variation in which one unit of the DNA molecule (one base) is expressed in an alternative form (e.g., G or Guanine instead of A or Adenine)