

The Basic Genome

After Module 1, you should be able to explain the following terms and ideas.

KEY TERMS

Base pair, Nucleotide, Strand, Single nucleotide polymorphism (SNP), Haplotype, Genotype, Indel, Variant, Mutation, Allele, Genotyping, Sequencing, Phased/Unphased Genotypes, Chromosome

CONCEPT QUESTIONS

What is the basic unit of DNA? What are the four types and how are they abbreviated?

What is a base pair? Why is it a *pair*? What does this have to do with strands? When we write out the genome, why do we only write out one of the strands?

What is an allele? How many does a normal human have for each position in the genome?

What is a genotype?

What is a variant? Is this the same as a mutation or a SNP?

What does SNP stand for? What about SNV? How are they different?

What is an indel? How is it different from a SNP?

What is genotyping? How is it different from sequencing?

What does it mean to have phased genotypes? How do you phase genotypes?

What is a haplotype? How is it different from chromosomes?