

Genes and Genetic Diseases

1. Define and give examples of the following genetic terms: progeny, chromosomes, gene, allele, gamete, homozygous, heterozygous, karyotype, genotype, phenotype, dominant traits, recessive traits, pedigree chart, penetrance, and expressivity,
2. List the cause and possible outcome for the following mutations: base pair substitution, frameshift substitution, spontaneous mutation, and mutational hotspots.
3. Define and give examples of the following chromosome terms: euploid, haploid, diploid, polyploidy, aneuploid, trisomy, monosomy, disjunction, and nondisjunction.
4. Describe the following deviations in normal chromosome structure: deletion, duplication, inversion, translocation and fragile sites.
5. Define and describe the following elements of inheritance: autosomal, sex-linked, carrier, dominant, and recessive.
6. Evaluate pedigree charts for the inheritance of genetic diseases.
7. Describe the genetic abnormalities and resulting clinical abnormalities associated with the following diseases: Down syndrome, Turner syndrome, Klinefelter syndrome, Cri du Chat syndrome, Huntington disease, cystic fibrosis, neurofibromatosis, hemophilia, and Duchenne muscular dystrophy.