Genetic Inheritance and Human Health

Genetics And Genealogy · Answer Key · 15 Questions

1. Which specific inheritance pattern describes a condition caused by a mutation in a gene located on the X chromosome, where males are more likely to be affected than females?

A) Autosomal dominant
B) X-linked recessive
C) Mitochondrial inheritance
D) Y-linked dominant

2. What is the term for the phenomenon where an individual has two different alleles at a particular gene locus, often acting as a carrier for recessive health traits?

A) Homozygosity
B) Hemizygosity
C) Heterozygosity
D) Polyploidy

3. In mitochondrial inheritance, which parent is responsible for passing the mitochondrial DNA to all of their offspring?

A) The father only
B) The mother only
C) Both parents equally
D) Neither parent

4. Which genetic condition is characterized by a trinucleotide repeat expansion in the HTT gene, leading to progressive neurodegeneration?

A) Huntington's disease
B) Cystic fibrosis
C) Sickle cell anemia
D) Hemophilia

5. What is the primary physiological mechanism that causes Sickle Cell Disease, often traced through genealogical lineage in populations with high malaria prevalence?

A) Deficiency in Vitamin B12
B) Abnormal hemoglobin S polymerization
C) Excessive production of white blood cells
D) Genetic deletion of chromosome 21

6. Which type of genetic testing assesses the likelihood of developing a disease based on the cumulative effect of multiple genetic variants across the genome?

A) Monogenic risk scoring
B) Polygenic risk scoring
C) Karyotype analysis
D) Exome sequencing

7. In autosomal recessive inheritance, what is the probability that the offspring of two carrier parents will be affected by the disorder?

A) 25%
B) 50%
C) 75%
D) 100%

8. What is the scientific term for the proportion of phenotypic variation in a population that is attributable to genetic variation among individuals?

A) Penetrance
B) Expressivity
C) Heritability
D) Epistasis

9. Which gene mutation is most famously associated with an increased genealogical risk for hereditary breast and ovarian cancer syndrome?

A) BRCA1/BRCA2
B) CFTR
C) LDLR
D) F8

10. What occurs when a gene's expression is modified by environmental factors or chemical tags without changing the underlying DNA sequence?

A) Epigenetic modification
B) Genetic recombination
C) Chromosomal translocation
D) Point mutation

11. Which condition is an example of an autosomal dominant disorder that typically manifests in adulthood, characterized by the growth of cysts in the kidneys?

A) Polycystic kidney disease
B) Phenylketonuria
C) Tay-Sachs disease
D) Thalassemia

12. What is the clinical significance of 'consanguinity' in a genealogical health context?

A) Increased risk of autosomal recessive disorders
B) Reduced risk of polygenic traits
C) Higher probability of spontaneous mutation
D) Neutral impact on health

13. In the context of pharmacogenomics, what does the CYP450 gene family primarily regulate in the human body?

A) Bone density regulation
B) Drug metabolism in the liver
C) Insulin production
D) Visual pigment synthesis

14. What structural change in a chromosome occurs when a segment of the chromosome breaks off and reattaches in reverse orientation?

A) Inversion
B) Duplication
C) Deletion
D) Translocation

15. Which syndrome is caused by a trisomy of chromosome 21, often linked to maternal age but not strictly hereditary in most instances?

A) Down syndrome
B) Turner syndrome
C) Klinefelter syndrome
D) Williams syndrome