

Examples of Genetics Test Questions

*** Marks correct answer**

Topic: Clinical Cytogenetics: You need to know

1. The major clinical features of the three classical trisomies i.e., trisomy 21, 18 and 13.
2. How to interpret a written description of a karyotype and take appropriate action such as ordering additional tests, doing parental chromosomes.
3. How to determine the source of the extra chromosome in a karyotype from data showing RFLP
4. Approximate incidence of chromosomal abnormalities in various clinical samples such as abortuses, malformed newborns, and individuals with mental retardation. You should be able to look at a karyotype, identify a trisomy and select from a series of descriptions the phenotype that matches the karyotype.

Topic: Cancer Genetics and DNA repair defects / chromosomal breakage syndromes:
You should know

1. The genetic models (Vogelstein model for colorectal cancers and the Knudson two-hit hypothesis) of the origin and dissemination of cancers
2. The tumor suppressor genes Rb 1
3. Loss of heterozygosity
4. Steps involved in the progression of colon polyps to cancer and metastasis
5. Breast and ovarian cancers and the role of BRVA genes
6. Role of the p 53 gene in cancers
7. Clinical features and key pathological features of Xeroderma pigmentosa, AT and Bloom syndromes

Topics: Multifactorial inheritance and Genetic screening: You should know

1. The liability and threshold model and examples of conditions that fit that model e.g. pyloric stenosis, cleft lip and palate, congenital hip dysplasia
2. Gene environmental interactions e.g. neural tube defects
3. Use of twins in the study of heritability of complex, multifactorial phenotypes
4. Interpretation of correlation data from twin studies to calculate heritability
5. Newborn screening laws, regulations, process and the conditions screened for in South Carolina

Practice questions.

1. Which of the following would suggest the presence of an extra chromosome 13 in a newborn?
 - A. Upward slanted palpebral fissure, epicanthal folds, hypotonia and simian crease
 - B. Polydactyly, cleft lip and palate and microphthalmia *
 - C. Rocker bottom feet, overlapping fingers and intrauterine growth retardation
 - D. Kitten-like cry, microcephaly and round face

2. Which of the following conditions is not screened for in South Carolina?
 - A. Sickle cell anemia
 - B. Galactosemia
 - C. Hypothyroidism
 - D. PKU
 - E. Cystic fibrosis *

3. Which of the following is due to a defective excision repair?
 - A. Ataxia telangiectasia
 - B. Bloom syndrome
 - C. Fanconi anemia
 - D. Xeroderma pigmentosa *

4. Which of the following is the most likely cause of trisomy 21?
 - A. An error in maternal first meiotic division *
 - B. Faulty second meiotic division during spermatogenesis
 - C. Defective second meiotic division during oogenesis
 - D. None of the above

6. Prenatal diagnosis of neural tube defects may involve all of the above except
 - A. Maternal serum AFP levels
 - B. Amniotic fluid AFP levels
 - C. Fetal karyotyping *
 - D. Fetal sonography