Roberts syndrome multiple choice questions

1.) The gene ESCO2 is located on which chromosome?

   a. Chromosome 5
   b. Chromosome 8
   c. Chromosome 21
   d. Chromosome 18
   e. Chromosome 4

2.) Siblings of an affected individual with Roberts syndrome have a _____ chance of being a carrier.

   a. 0%
   b. 25%
   c. 50%
   d. 75%
   e. 100%
Review Questions

1. What do the TSC1 and TSC2 genes regulate?
   a. mTOR, a protein that acts as a central regulator of tumor cell division, blood vessel growth, cell metabolism and neuronal migration/wiring.

2. What is a symptom of TSC?
   a. Cortical tubers
1.) Alzheimer’s disease is transmitted in an Autosomal Dominant fashion. Because of this, which of the following is correct?
   a. Two copies of the mutated allele are required for the increased risk of developing Alzheimer’s disease.
   b. Only females are affected by the disease.
   c. Only males are affected by the disease.
   d. One copy of the mutated allele is required to increase the risk of developing Alzheimer’s disease.

2.) Which of the following is not a clinical feature of Alzheimer’s disease?
   a. Plaque formation of beta-amyloid deposits in the brain.
   b. Impaired speech or swallowing in progressed patients.
   c. Cardiovascular problems.
   d. Paranoia

Answers:

1.) D
2.) C
Brochure Review Questions

1. Hereditary nonpolyposis colorectal cancer (HNPCC) is also known as
   a. Lynch Syndrome
   b. colon cancer
   c. proctocolectomy
   d. endometrial cancer
   e. sigmoidoscopy

2. What is the mode of inheritance for hereditary nonpolyposis colorectal cancer?
   a. autosomal recessive
   b. autosomal dominant
   c. x-linked dominant
   d. x-linked recessive
   e. none of the above
Neurofibromatosis Review Questions

1. Which of the following is NOT a clinical feature of neurofibromatosis 1?
   a. Lisch nodules
   b. Neurofibromata
   c. Blindness
   d. Café-au-lait spots

2. At what stage in life do vestibular schwannomas usually begin appearing in an individual that is affected with neurofibromatosis 2?
   a. Early adulthood
   b. At birth
   c. Early childhood
Review Questions: Breast Cancer

1. A mutation in which gene causes an increased propensity for developing breast cancer?

A.) BRCA1 and BRCA2  
B.) FRAXA  
C.) CFTR  
D.) MYOD1

2. What is the mode of inheritance for breast cancer?

A.) X-linked recessive  
B.) autosomal dominant  
C.) X-linked dominant  
D.) autosomal recessive
Multiple Choice Questions

Brochure on Oculodentodigital Dysplasia

1. A deletion on the q arm of what chromosome causes Oculodentodigital Dysplasia?
   a. 1
   b. 9
   c. 6
   d. 20
   e. 17

   Answer = c.

2. What is the mode of inheritance for Oculodentodigital Dysplasia?
   a. Autosomal dominant
   b. Autosomal recessive
   c. Arises from novel mutation
   d. Both a and c are correct
   e. Both b and c are correct

   Answer = d.
Matthew Archevald

Wilson’s Disease Question

1. Wilson’s Disease is characterized by a variety of signs and symptoms, but one aspect in particular is unique to the disease especially for diagnostic purposes. Select the best answer.

   a. Liver failure due to cirrhosis
   b. High levels of copper in the urine
   c. Kayser-Flescher rings due to copper accumulation
   d. Enlarged spleen

2. A mutation in which gene and on which chromosome is responsible for the body’s inability to eliminate excess copper?

   a. ATP7B gene on chromosome 13q14.3
   b. ATP7P gene on chromosome 14q13.4
   c. BRCA2 gene on chromosome 13.12.3
   d. RB1 gene on chromosome 13.14.1
Review Questions for Hemophilia

1. Hemophilia A is caused by a deficiency in or lack of _____ proteins, while Hemophilia B is caused by a deficiency in or lack of _____ proteins.

   A. Factor IX ; Factor III
   B. Factor VIII ; Factor X
   C. **Factor VIII ; Factor IX**
   D. Factor IX ; Factor VIII
   E. None of the Above

2. How is Hemophilia, the blood clotting disorder, inherited?

   A. **X-linked Recessive**
   B. Autosomal Dominant
   C. Random Mutation
   D. X-linked Dominant
   E. Autosomal Recessive
Erika Vance

Non-Hodgkin Lymphoma

1. What is the cause of most lymphomas?
   A. Unknown
   B. Autosomal Dominant Disorder
   C. X Linked Recessive
   D. Autosomal Recessive

   Answer: A

2. What are the 2 main types of treatment for Non-Hodgkin Lymphoma?
   A. Chemotherapy and Multiple different drugs used in combination
   B. Chemotherapy and Radioimmunotherapy
   C. Multiple different drugs used in combination and Radioimmunotherapy
   D. Surgery

   Answer: A
Ovarian Cancer Multiple Choice Questions

1. Which of the following is true of ovarian cancer?

   a) It is the most common gynaecological cancer in the UK
   b) It is the second commonest cancer in females
   c) Approximately 30% are familial
   d) Median age of diagnosis is 53 years

2. Which one of the following symptoms is an uncommon presentation of ovarian cancer?

   a) Abdominal pain
   b) Chest pain
   c) Weight loss
   d) Abdominal distension

Answers

1) d
2) b
Review Questions

1. What is the only known treatment for celiac disease?
   a. Antibiotics
   b. Immunosuppressant drugs
   c. A sugar-free diet
   d. A gluten-free diet
   e. All of the above

2. What genes are associated with celiac disease?
   a. HLA-DQ2
   b. HLA-RO7
   c. HLA-DQ8
   d. All of the above
   e. A and C only