Review Questions for Final Exam

Medical Genetics

The following topics were discussed after the second exam

I. Chapter 7. Pattern of inheritance: Family studies, Mendelian inheritance, anticipation

II. Chapter 19. Single-gene disorders

III. Southern blotting, DNA methylation

IV. Chapter 21. Prenatal testing and reproductive genetics: techniques used in prenatal diagnosis, Georgia Newborn Screening Program

The materials from the previous 2 exams will be 15-20% of the questions in the final exam. Questions from students’ presentations and brochures will be a part of the questions.

The final exam will be worth 150 points. Good luck with your finals.

Here are some questions from lectures, students’ presentations and brochures.

1. Briefly describe the clinical phenotype of Duchenne muscular dystrophy, and its mode of inheritance and molecular defect.

2. Duchenne muscular dystrophy is caused by
   a. a deletion of the X chromosome.
   b. point mutations of the Dystrophin gene.
   c. a tri-nucleotide expansion within a gene.
   d. a and b are correct
   e. all of the above

3. Which of the following statements about Duchenne muscular dystrophy is correct?
   a. The disease is always transmitted from the carrier mother to the child.
   b. An affected male must have a father who has the disease.
   c. The life expectancy of a diseased individual is much shorter than that of someone with Becker muscular dystrophy.
   d. A sibling of a diseased individual will have a 25% chance to have the disease.
   e. c and d are correct
Simpson-Golabi-Behmel “Bulldog” syndrome:

5. Which of the following is the most prevalent characteristic of Simpson-Golabi-Behmel “Bulldog” syndrome?
   a. abnormal facial features
   b. muscle weakness
   c. The disease is mostly seen in males.
   d. The diagnosis includes muscle biopsy.
   e. a and c are correct

Myotonic dystrophy:
6. Briefly describe the phenotype, the mode of inheritance, and molecular defect.

7. Which of the following statements about myotonic dystrophy is correct?
   a. The patients with the disease will be wheelchair-bound when they become teenagers.
   b. The disease affects mostly males.
   c. Children who inherited the disease gene from the father will have milder phenotypes.
   d. Children who inherited the disease gene from the mother will have milder phenotypes.
   e. Siblings of a diseased individual will have a 25% chance to have the disease.

Hemophilia:
8. Briefly describe the phenotype, the mode of inheritance, and molecular defect.

9. Which of the following statements about hemophilia is correct?
   a. Currently there is no treatment for the disease.
   b. Males and females are equally affected.
   c. Both copies of the factor VIII gene have to be mutated to have the disease phenotype.
   d. a and b are correct
   e. all of the above

Familial Hypercholesterolemia:
10. Briefly describe the phenotype, the mode of inheritance, and molecular defect.
11. Familial hypercholesterolemia is caused by
   a. a defect in the ABCA12 gene.
   b. a defect in the LDL receptor gene.
   c. a deletion of chromosome 13q.
   d. a deletion of chromosome 5p.
   e. an expansion of CAG tri-nucleotide repeats in the LDL receptor gene.

12. Which of the following statements about familial hypercholesterolemia is correct?
   a. It is an autosomal recessive disease.
   b. The individuals who are homozygote for the disease gene will show less severe symptoms.
   c. The individual needs only one single mutation in the gene to show the disease phenotype.
   d. All cases are familial cases (no new mutations).
   e. Mutated gene passing from the father will lead to more severe symptoms.

**Turner syndrome:**

13. Briefly describe the phenotype, the mode of inheritance, and molecular defect.

14. Patients with Turner syndrome were discovered to possess one X chromosome in
   a. 1949
   b. 1959
   c. 1969
   d. 1979
   e. 1989

15. The probability that an individual with Turner syndrome will be fertile is _____.
   a. 0%
   b. 25%
   c. 50%
   d. 75%
   e. 100%

**Wilson disease:**

16. Briefly describe the phenotype, the mode of inheritance, and molecular defect.
17. A slit-lamp is used to diagnose
   a. Turner syndrome.
   b. Wilson’s disease.
   c. familial hypercholesterolemia.
   d. retinoblastoma.
   e. b and d are correct

18. A strict diet, as one of the treatments, is required for individuals with
   a. familial breast cancer.
   b. cystic fibrosis.
   c. Wilson’s disease.
   d. familial hypercholesterolemia.
   e. c and d are correct


20. Which of the following statements is true for follicular non-Hodgkin lymphoma?
   a. It is a childhood cancer.
   b. Patients can have swollen lymph nodes.
   c. It occurs to older people.
   d. a and b
   e. b and c

21. Which of the following disorders illustrates the principle of variable expressivity?
   a. Huntington disease
   b. celiac disease
   c. polycystic kidney disease
   d. cystic fibrosis
   e. Bloom syndrome

22. Tuberous sclerosis can be caused by mutations either in TSC1 or TSC2 gene.
   This is an example of
   a. genomic imprinting
   b. anticipation
   c. late onset
   d. genetic heterogeneity
   e. incomplete penetrance
23. C-banding technique is a useful tool in diagnosing
   a. Prader-willi syndrome
   b. Fragile X syndrome
   c. Bloom syndrome
   d. Robert syndrome
   e. Klinefelter syndrome

24. Mitochondrial DNA is
   a. a linear DNA with a molecular size of ~16,000 base pairs.
   b. a circular DNA with a molecular size of 6,000 base pairs.
   c. a circular DNA encoding for 37 genes.
   d. a linear DNA encoding for 20,000 genes.
   e. none of the above.

25. Jon has a form of mitochondrial myopathy and his wife Cindy is normal. Cindy is pregnant with their first child. The chance of having a child with the myopathy is
   a. 100%
   b. 75%
   c. 50%
   d. 25%
   e. 0%

26. Cystic fibrosis is a common autosomal recessive disease in the ___________ population
   a. African American
   b. Ashkenazi Jewish
   c. Southeast Asian
   d. Caucasian
   e. none of the above

27. SCE (sister chromatid exchange) is a useful cytogenetic technique for diagnosing
   a. hemophilia
   b. fragile X syndrome
   c. Edward syndrome
   d. Bloom syndrome
   e. Angelman syndrome

28. All of the following features can be seen in patients with myotonic dystrophy, except
a. emotionless facial expression.
b. presence of a number of CTG repeats at the 3’ of the DMPK (myotonic dystrophy) gene
c. presence of an expansion of the CTG repeats at the 3’ of the DMPK (myotonic dystrophy) gene
d. higher creatine kinase concentration in the blood
e. Patients with the maternal inherited mutation in the DM1 gene show much more severe symptoms.

**True/False questions.**

29. “All cancers are genetic, but some cancers are more genetic than others”.
   a. true
   b. false

30. Relatives of a patient with breast cancer will have a higher risk of developing breast cancer than other women who are at the same age.
   a. true
   b. false

31. To date, only one gene has been identified to be responsible for the development of colon cancer.
   a. true
   b. false

**Other Review Questions**

List 2 major similarities and 3 major differences between each pair of genetic disorders, including the cardinal clinical phenotype, gene mutation, and diagnostic methods. Here are some examples:

   a. Duchenne muscular dystrophy/Becker muscular dystrophy
   b. Familial breast cancer/ familial colon cancer
   c. Robert syndrome/ Bloom syndrome
   d. Familial hypercholesterolemia/ Wilson disease