



# FINAL GENETICS TEST BANK

DOCTOR 2020

Collected by:  
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## Genetic variation:

### 1. Karyotyping is not used in:

- a. Deletion of a chromosomal segment
- b. Inversion
- c. Insertion of few nucleotides

Answer: C

### 2. Individual A is affected with hearing loss (HL) and his ethnic background is African. Individual E is also affected with HL and his ethnic background is European:

- a. The genomic sequence of individual E is more similar to the genomic sequence of another African who is unaffected with HL than unaffected European
- b. The genomic sequence similarity between individuals A and E is higher than their corresponding ethnic backgrounds
- c. The genomic sequence of individual A is more similar to the genomic sequence of another European who is unaffected with HL than individual E
- d. The genomic sequence of individual A is more similar to the genomic sequence of another African who is unaffected with HL than individual E
- e. The genomic sequence of individual E is more similar to the genomic sequence of another African who is affected with HL than unaffected European

Answer: D

### 3. A mutation leading to:

- a. Edward syndrome due to advanced paternal age is more likely to occur than due to advanced maternal age
- b. Hearing loss due to advanced paternal age is more likely to occur than due to advanced maternal age
- c. Cystic fibrosis due to advanced maternal age is more likely to occur than due to advanced paternal age
- d. Tay-Sachs due to advanced maternal age is more likely to occur than due to advanced paternal age
- e. Down syndrome due to advanced paternal age is more likely to occur than due to advanced maternal age

Answer: B

### 4. Which of the following DNA changes is pathogenic:

- a. a mutation
- b. an epigenetic modification
- c. a variant
- d. any of these choices could be pathogenic

e. a polymorphism

Answer: D

**5. The nomenclature c.2312\_2314delinGCGTGGACAAC G denotes:**

- a. deletion of 12 nucleotides followed by insertion of 3 nucleotides
- b. any of these choices could be correct
- c. deletion of 3 nucleotides followed by insertion of 3 nucleotides
- d. deletion of 3 nucleotides followed by insertion of 12 nucleotides
- e. deletion of 12 nucleotides followed by insertion of 12 nucleotides

Answer: D

**6. Which of the following variants would you expect could to be most common:**

- a. G>C
- b. A>G
- c. C>G
- d. C>T
- e. A>T

Answer: D

**7. The most common occurring mutations in humans are :**

- a. Indels
- b. CNVS
- c. Polyploidy
- d. Aneuploidy
- e. Nucleotide substitution

Answer: d

**8. A chromosomal abnormality that involves 30 megabase region. The most likely abnormality to cause the worse disease is :**

- a. deletion of the region
- b. pericentric inversion
- c. duplication of the region
- d. paracentric inversion
- e. a balanced translocation involving the region

Answer: A

**9. The percentage of DNA sequence variation between humans is the lowest in:**

- a. tandem non coding repeats
- b. introns

- c. transposable elements
- d. exons
- e. interspersed non coding repeats

Answer : D

### Risk assessment:

**10. A unaffected couple who are first cousins request counseling regarding their risk of having a child with alpha-1-antitrypsin deficiency, a rare autosomal recessive trait. Their parents are unaffected. Their shared grandfather is affected with the disorder and their shared grandmother is heterozygous. What is the risk to their child of being HOMOZYGOUS FOR A VARIANT FOR THE CONDITION ? (disregard the population carrier frequency)**

- a. 1/16
- b. 1/8
- c. 1/32
- d. 0
- e. 1/64

Answer: A

**11. A European couple with diagnosis of hearing loss in father's brother in autosomal recessive variant. What is the probability to have an AFFECTED FETUS? Knowing that the carrier frequency of this variant in the European population is around 1/30 .**

- a. 1/4
- b. 1/90
- c. 0
- d. 1/75
- e. 1/180

Answer: E

**12. A couple are both tested, the father is affected with cystic fibrosis and the mother is a carrier of cystic fibrosis gene. What is the chance that they will have a HOMOZYGOUS NORMAL MALE?**

- a. 1/16
- b. 1/8
- c. 0
- d. 1/2
- e. 1/4

Answer: C

13. A couple approached you because of a current pregnancy and a family history with phenylketonuria (PKU). The mother told you that her sister is affected with PKU and the father told you that his uncle is affected with PKU. You performed an ultrasound and the fetus turned out to be a female. What is the risk for the couple to have UNAFFECTED FEMALE with PKU? Assume a population frequency of 1/100? Note: PKU is an autosomal recessive disease .

- a. 1/4
- b. 1/2
- c. 1/18
- d. 1/6
- e. 1/12

Answer: D

14. Suppose there's an X-linked recessive disease, if the mother has the disease, while the father doesn't, what's the chance that they'll have an affected female?

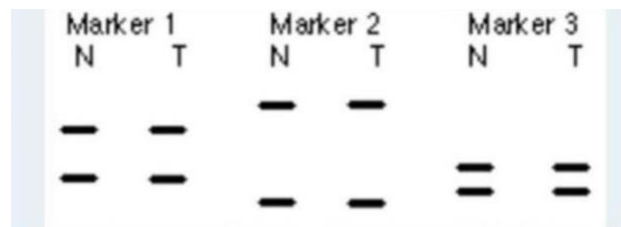
Answer: 0

### Cancer genetics:

15. This is an electrophoresis (southern blots) for an individual with hereditary retinoblastoma; both normal and tumor cells were genotyped for three polymorphic markers flanking the RBI locus on chromosome 13 for this patient (N = normal cells, T = tumor cells).

Based on the figure, which of the following is the %3D most likely to explain the genetic mutation in this patient's cancer ?

- a. Second somatic point mutation hit
- b. Any of those choices could explain the patient's situation
- c. Loss of the normal chromosome 13
- d. Loss of the mutated chromosome 13
- e. Uniparental disomy (UPD) of chromosome 13



Answer: A

16. You observed a patient with a genetic disorder for which there is no previous family history of his disease. which of the following scenarios could explain this situation:

- a. any of those choices could explain the patient's situation
- b. a de novo variant
- c. germline mosaicism in one of the parents anticipation
- d. reduced penetrance

Answer: A

**17. A 6-month-old with unilateral retinoblastoma has genetic testing performed. The results of the tumor and blood analysis at the RBI locus are shown below. What is the likelihood that a future sibling WILL NOT DEVELOP retinoblastoma ? (R is Arginine and X is stop codon)**

- a. < %1
- b. >99%
- c. 50%
- d. 25%
- e. 75%

**Answer: B**

**18. Most cancers cases occur due to:**

- a. Inhibition of cell cycle
- b. Genetic or epigenetic changes in somatic cells
- c. Germline variants
- d. The production of unbalanced gametes because of nondisjunction during meiosis
- e. Delayed cell division during early embryogenesis

**Answer: B**

**19. A patient with retinoblastoma has a single tumor in one eye; the other eye is free of tumors:**

- a. At least one of the parents is most probably affected with retinoblastoma
- b. The patient is more likely affected with heritable retinoblastoma
- c. The tumor formed in the patient during early infancy
- d. The patient is more likely affected with sporadic retinoblastoma
- e. The patient most probably carries a germline hit

**Answer: D**

**20. In an experiment gene X was over expressed in neoplastic cell. Consequently, it acquired malignant traits. Gene X is most likely:**

- a. Cytoskeletal gene
- b. Noncoding repetitive sequence
- c. Tumor suppressor
- d. Gap junction gene
- e. Proto-oncogene

**Answer: E**

**21. Variants in tumor suppressors on the genetic level are——and on the phenotypic level are:———**

- a. recessive, dominate .

- b. dominant, recessive .
- c. dominant, dominant .
- d. recessive, recessive .
- e. codominant, codominant .

Answer: A

**22. Which of the following is NOT a familial cancer syndrome:**

- a. Li-Fraumeni syndrome
- b. Neurofibromatosis type 2
- c. Waardenburg syndrome
- d. Familial adenomatous polyposis
- e. Von Hippel-Lindau syndrome

Answer: C

**23. RET gene:**

- a. drives oncogenesis mainly through loss of function
- b. causes hirschsprung disease mainly through activating variants
- c. follows the two-hit theory
- d. drives the expression of anti-apoptotic genes
- e. is a transcription factor

Answer: D

**24. In a patient with nonhereditary cancer. The driver mutations :**

- a. Exist in the secondary but not in the primary or normal tissue cells
- b. Exist in primary and metastatic tumor tissues but not his normal tissue
- c. Exist in the secondary and normal tissues but not the primary tumor tissue
- d. Exist in primary and normal tissues but not in the metastatic region
- e. Are shared between the primary, secondary and metastatic tissues for the same patient

Answer: B

**25. A de novo variant occurs due to an error in:**

- a. DNA replication
- b. Post translational modification
- c. DNA transcription
- d. Translation
- e. Reverse transcription

Answer: A

**26. A gain of abnormal function rather than excessive activity in an oncogene could most probably occur through:**

- a. A point mutation in the exon
- b. A point mutation in the promoter
- c. Any of those choices could equally cause a gain of abnormal function
- d. Gene amplification
- e. A translocation of a proto-oncogene under a weak promoter

**Answer: A**

**27. A variant that results in genetic mosaicism is:**

- a. a germline variant
- b. a somatic variant
- c. a dynamic variant
- d. a permutation
- e. a missense variant

**Answer: B**

**28. Translocation between ch 9 and 22---> Chronic myelogenous leukemia**

**29. A 6-month-old with unilateral retinoblastoma has genetic testing performed. The results of the tumor and blood analysis at the RBI locus are shown below (the photo is missing, but you don't need it anyway!). What is the likelihood that a future sibling WILL DEVELOP retinoblastoma?**

- a. < %1
- b. >99%
- c. 50%
- d. 25%
- e. 75%

**Answer: a**

**30. Which of the following diseases has reduced penetrance?**

**Answer: Retinoblastoma**

**31. Next Generation Sequencing (NGS) can perform Whole Exome Sequencing (WES) for the entire coding regions in one reaction. WES will be more successful**

**in detecting a candidate disease-causing change in :**

- a. Promoter region
- b. Imprinting of a coding region



- c. Distal enhancer element
- d. Synonymous Splicing variant
- e. Telomere region

Answer: D

**32. Wilms' tumor (nephroblastoma), an embryonal malignancy of the kidney, is the most common renal tumor of childhood. The tumor suppressor WTI is associated with familial Wilms Tumor and its locus is 11p15.5. DNA was extracted from the normal tissue and tumor of a patient with Wilms' tumor, and from the blood of the patient's father and mother. Electrophoresis (southern blot) was performed for a polymorphic marker (A) which is close to WTI. Based on the figure, which of the following is the most likely to explain the mutation in this patient's cancer?**

- a. Loss of the mutated chromosome 11
- b. Loss of the normal chromosome 11
- c. Imprinting mutation driving the second mutation
- d. Any of those choices could equally explain the patient's situation
- e. Second somatic point mutation hit

Answer: B

# Genetics Questions

First 14 Questions are from Medical genetics 5e textbook, and they're mostly about risk assessment. Notice that the underlined questions are not explained in our lectures and you won't likely see them in the exam, yet you can answer them if you scratch your head a little.

Question 15 is by Nabil Sweis

Questions 16 – 45 are from Step 1 USMLE Q bank, they include our entire material, even the midterm diseases and their clinical aspect. Don't bother yourself with clinical questions if you are here for exam purposes only.

Good luck,

**Mothana Olimat**

PS: I looked in both books, Medical genetics & New clinical genetics, but the later didn't include questions that I could turn into MCQs & had no model answer either.

1) A man who has achondroplasia (Autosomal dominant) marries a phenotypically normal woman. If they have four children, what is the probability that none of their children will be affected with this disorder? What is the probability that all of them will be affected? Respectively.

A.  $\frac{1}{4}, \frac{1}{4}$

B.  $\frac{1}{16}, \frac{1}{8}$

C.  $\frac{1}{16}, \frac{1}{16}$

D.  $\frac{1}{8}, \frac{1}{16}$

2) The estimated penetrance for familial retinoblastoma is approximately 90%. If a man has had familial retinoblastoma and mates with a woman who does not have a retinoblastoma mutation, what is the risk that their offspring will develop retinoblastoma?

A. 0.5

B. 0.45

C. 0.25

D. 0.333

3) A 30-year-old woman had a sister who died from infantile Tay–Sachs disease, an autosomal recessive disorder that is fatal by age 6 years. What is the probability that this woman is a heterozygous carrier of the Tay–Sachs mutation?

A.  $\frac{2}{3}$

B.  $\frac{1}{2}$

C.  $\frac{1}{4}$

D.  $\frac{3}{4}$

4) A man has neurofibromatosis type 1 (AD). His mother also has this condition. What is the probability that his sister also has this disease? In the absence of knowledge of his sister's phenotype, what is the probability that his sister's daughter has neurofibromatosis type 1? Respectively.

A.  $\frac{2}{3}, \frac{1}{4}$

B.  $\frac{1}{2}, \frac{1}{2}$

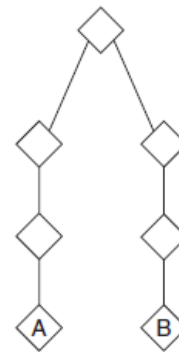
C.  $\frac{1}{4}, \frac{1}{4}$

D.  $\frac{1}{2}, \frac{1}{4}$

- 5) Consider a woman who is a known heterozygous carrier of a mutation that causes PKU (autosomal recessive). What is the probability that her two grandchildren, who are first cousins, are both heterozygous carriers of this PKU-causing allele? Suppose instead that the woman is affected with PKU. Now what is the probability that both of her grandchildren carry the disease-causing allele?

- A.  $\frac{1}{4}, \frac{1}{2}$   
 B.  $\frac{1}{8}, \frac{1}{4}$   
 C.  $\frac{1}{16}, \frac{1}{4}$   
 D.  $\frac{1}{16}, \frac{1}{8}$

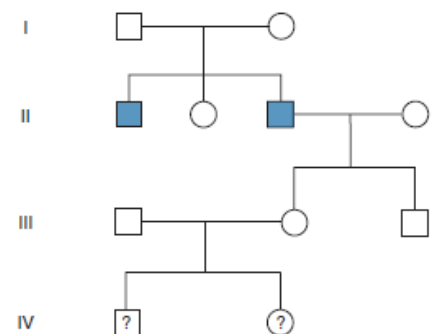
- 6) Two mating individuals, labelled A and B in this figure, share a single great-grandparent. Suppose that one member of this couple is a heterozygous carrier for PKU. What is the probability that this couple will produce a child affected with PKU?



- A.  $\frac{1}{64}$   
 B.  $\frac{1}{4}$   
 C.  $\frac{1}{256}$   
 D.  $\frac{1}{32}$

- 7) The figure aside shows the inheritance of hemophilia A (X-linked recessive) in a family. What is the risk that the male in generation IV is affected with hemophilia A? What is the risk that the female in generation IV is a heterozygous carrier?

- A. 50%, 50%  
 B. 100%, 50%  
 C. 100%, 100%  
 D. 50%, 0%



8) A boy and his brother both have hemophilia A (X-linked recessive). If there is no family history of hemophilia A in previous generations, what is the probability that the boys' aunt (the mother's sister) is a heterozygous carrier of the disease gene?

- A.  $\frac{1}{4}$
- B.  $\frac{2}{3}$
- C. 1
- D.  $\frac{1}{2}$

9) Rank the following individuals, from lowest to highest, in terms of the risk of producing a child with Down syndrome:

Person 1] 45-year-old woman with no previous family history of Down syndrome

Person 2] 25-year-old woman who has had one previous child with Down syndrome.

Person 3] 25-year-old male carrier of a 21/14 Robertsonian translocation

Person 4] 25-year-old female carrier of a 21/14 Robertsonian translocation

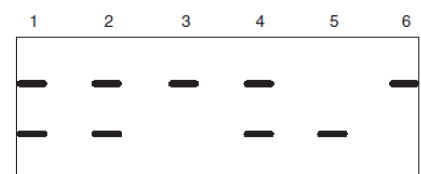
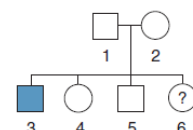
- A. 2<3<4<1
- B. 2<3<1<4
- C. 3<4<1<2
- D. 1<3<4<2

10) A man with hemophilia A (X-linked recessive) and a normal woman produce a child with Turner syndrome (45,X). The child has normal factor VIII activity. Which of the following is true?

- A. The meiotic error must have occurred in the mother
- B. The father must have passed his X chromosome to the child
- C. The meiotic error must have occurred in the father
- D. None of the above

**\*\*SNP (Single nucleotide polymorphism) & STR (short tandem repeat polymorphism) are biomarkers used to track the inheritance of disease genes within families.**

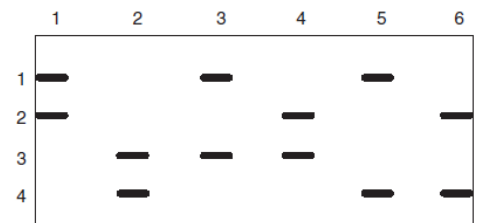
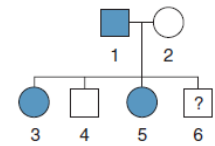
11) Study the family shown in the pedigree. Individual 3 has PKU, an autosomal recessive disease. A two-allele SNP closely linked to the PKU locus has been assayed for each family member, and the figure shows the **genotypes** of each individual. Based on the genotypes of the linked marker, individual 6



- A. is affected

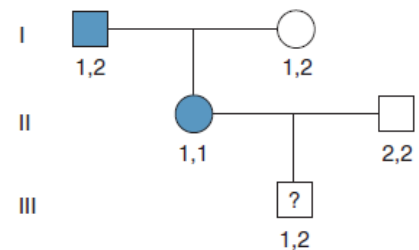
- B. is a heterozygous carrier
- C. is a normal homozygote
- D. Data given isn't enough

12) Study the family shown in the pedigree. The affected individuals have neurofibromatosis type 1 (NF1), an autosomal dominant condition. A four-allele STR locus closely linked to the NF1 locus has been typed for each family member. Based on the genotypes shown in the accompanying figure, is individual 6



- A. is affected
- B. is normal

13) In this pedigree for an autosomal dominant disorder, a tightly linked two-allele SNP has been typed in each family member. Based on this information, what is the risk that the offspring in generation III will develop the disorder?



- A. 100%
- B. 25%
- C. 0%
- D. 50%

14) Mary's two brothers and her mother's brother all had Duchenne muscular dystrophy (DMD) (X-linked recessive) and are now dead. Based on only this information, what is the probability that Mary is a heterozygous carrier for this disorder? What is the probability that she will produce affected offspring? Respectively

- A.  $\frac{1}{2}, \frac{1}{4}$
- B.  $\frac{2}{3}, \frac{3}{4}$
- C.  $\frac{1}{2}, \frac{1}{2}$
- D.  $\frac{1}{2}, \frac{3}{4}$

15) **(By Nabil)** Assuming a gene gives rise to an mRNA consisting of 4 exons. Exon 1 codes for 80 amino acids, Exon 2 codes for 100 amino acids, Exon 3 codes for 40 amino acids, Exon 4 codes for 70 amino acids. If a p. Trp90Ter mutation occurs, what is the most likely fate of the mutated mRNA?

- A. Translated into a truncated protein product
- B. Non-sense mediated decay (NMD)

18) A couple goes to a genetic counselor for family planning advice. Both male and female partners are of northern European ancestry and are concerned about offspring with cystic fibrosis, even though neither has a family history of the disease. The counselor learns that they recently heard of a family in their community whose son has cystic fibrosis. The counselor explains that the incidence of cystic fibrosis among populations of northern European ancestry is  $1/2,500$ . Which of the following is the likelihood that this woman is a carrier for the cystic fibrosis gene mutation?

A.  $\frac{1}{25}$

B.  $\frac{1}{50}$

C.  $\frac{2}{2500}$

D.  $\frac{1}{2500}$

E.  $\left(\frac{1}{2500}\right)^2$



42) In a given population, a disease is almost exclusively observed in males and has an incidence of 1 out of every 2,500 live male births. Which of the following is the frequency of carriers within that population?

- A.  $\frac{1}{25}$
- B.  $\frac{1}{50}$
- C.  $\frac{1}{1250}$
- D.  $\frac{1}{2500}$
- E.  $\frac{49}{1250}$

43) A 10-month-old boy is brought to the physician because of listlessness, apparent mental deterioration, atrophy, and difficulty in swallowing. He appeared normal at birth, but at age 6 months, he began to startle at loud noises. He was previously able to sit, but can now no longer sit, roll over, or recognize his parents. Ophthalmologic examination shows a central red area of the retina surrounded by white tissue. Genetic testing reveals a mutation in the HEXA gene. If this patient's parents have another child, what is the most likely risk of the child having the same disease?

- A.  $\frac{1}{4}$
- B.  $\frac{1}{2}$
- C.  $\frac{2}{3}$
- D.  $\frac{3}{4}$
- E. Not elevated above that of the general population

45) An autosomal recessive disease has a carrier frequency of  $\frac{1}{25}$  in a specific population. Which of the following is the most likely frequency of individuals expressing the disease in this population?

- A.  $\frac{1}{25}$
- B.  $\frac{1}{50}$
- C.  $\frac{1}{625}$
- D.  $\frac{1}{2500}$
- E.  $\frac{1}{5000}$

