Test Bank

Subject:

Medical Genetics-Final 018

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1- NOEY2 gene is located on chromosome 1. Loss of NOEY2 expression is linked to an increased risk of ovarian and breast cancers; in 41% of breast and ovarian cancers the protein encoded by NOEY2 is not expressed, suggesting that it functions as a tumor suppressor gene. Therefore, if a person inherits both chromosomes from the mother, the gene will not be expressed and the individual is put at a greater risk for breast and ovarian cancer. This is because:

a. NOEY2 is a maternally expressed
b. Parent of origin has nothing to do with NOEY2
c. NOEY2 is a paternally expressed
d. NOEY2 is maternally and paternally expressed
e. NOEY2 is an oncogene

2-In a patient with nonhereditary caner. 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

3-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

4-RET gene:

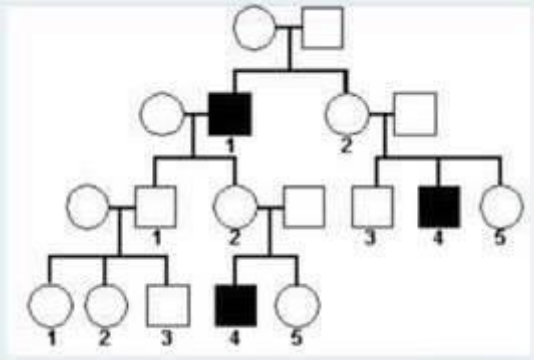
a. drives oncongensis mainly through loss of function
b. causes hirschsprung disease mainly through activating variants
c. follows the two-hit theory
d. drives the expression of anti-apoptotic genes
e. is a transcription factor

5-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

6-What is the most probable mode of inheritance:

a. Autosomal Dominant
b. X-Linked Recessive
c. X-Linked Dominant
d. Autosomal Recessive
e. Y-Linked



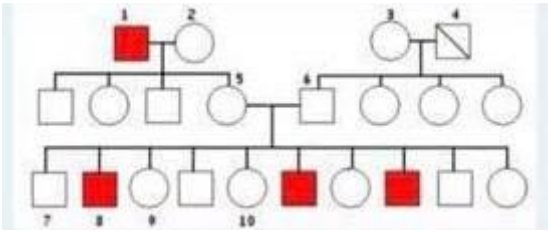
7-You observed a patient with a genetic disorder for which there is no previous family history of his disease. which of the following scenarios could explain this situation :

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

8-What is the most probable mode of inheritance:

a. X-Linked Dominant
b. Autosomal Recessive
c. Y-Linked
d. Autosomal Dominant
e. X-Linked Recessive

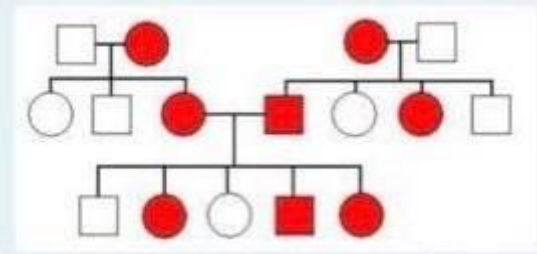


9-A variant that results in genetic mosaicism is :

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

10-What is the most probable mode of inheritance:

a. X-Linked Dominant
b. Autosomal Recessive
c. X-Linked Recessive
d. Y-Linked
e. Autosomal Dominant



11-Which of the following is NOT a familial cancer syndrome:

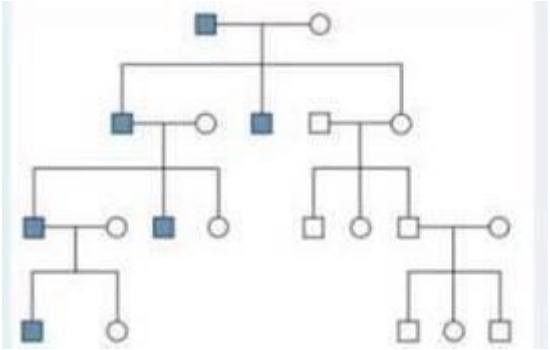
a. Li-Fraumeni syndrome
b. Neurofibromatosis type 2
c. Waardenburg syndrome
d. Familial adenomatous polyposis
e. Von Hippel-Lindau syndrome

12-The most common occurring mutations in humans are :

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

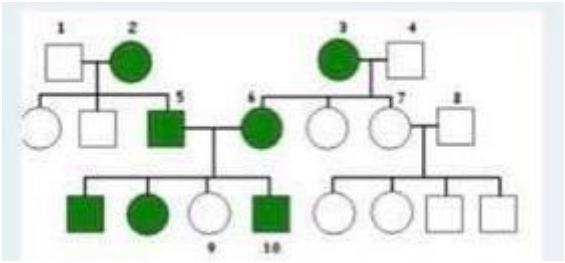
13-What is the most probable mode of inheritance:

a. X-Linked Dominant
b. Autosomal Dominant
c. X-Linked Recessive
d. Autosomal Recessive
e. Y-Linked



14-What is the most probable mode of inheritance:

a. Autosomal Recessive
b. Y-Linked
c. Autosomal Dominant
d. X-Linked Dominant
e. X-Linked Recessive



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

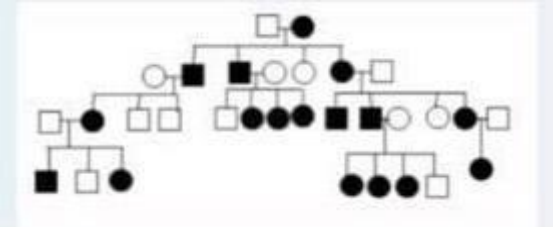
a. deletion of the region
b. pericentric inversion
c. duplication of the region
d. paracentric inversion
e. a balanced translocation involving the region

16-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

17-What is the most probable mode of inheritance:

a. X-Linked Recessive
b. Autosomal Recessive
c. X-Linked Dominant
d. Y-Linked
e. Autosomal Dominant



18-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

19-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

20-Variants in tumor suppressors on the genetic level are———-and on the phenotypic level are:———

a. recessive, dominate .
b. dominate, recessive .
c. dominate, dominate .
d. recessive, recessive .
e. codominant, codominant .

21-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

22-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

23-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

24-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

25-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

26-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

27-Adult polycystic kidney disease (APKD), an autosomal dominant disorder in which a progressive accumulation of renal cysts is seen. APKD can be caused by

mutations in genes on either chromosome 16 (PKDI) or chromosome 4 (PKD2). In one family, the disease may be caused by a PKDI mutation, whereas in another family it may be caused by a PKD2 mutation. The disease states produced by mutations in these two genes may be clinically indistinguishable. This is an example of :

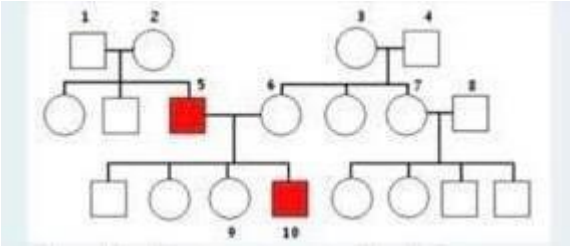
a. Penetrance
b. Pleiotropy
c. Allelic heterogeneity
d. Anticipation
e. Locus heterogeneity

28-45,X/46,XX karyotype was revealed in the peripheral lymphocytes of a Turner syndrome patient. Which of the following is the underlying cause ?

a. Reciprocal translocation
b. Nondisjunction in meiosis II
c. Nondisjunction in mitosis
d. Nondisjunction in meiosis |
e. Robesonian translocation

29-What is the most probable mode of inheritance:

a. Autosomal Dominant
b. X-Linked Recessive
c. Y-Linked
d. Autosomal Recessive
e. X-Linked Dominant



30-47 XYY occurs due to :

a. chimeric event
b. nondisjunction event from both parents
c. paternal nondisjunction
d. uniparental disomy (UPD)

e. maternal nondisjunction

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

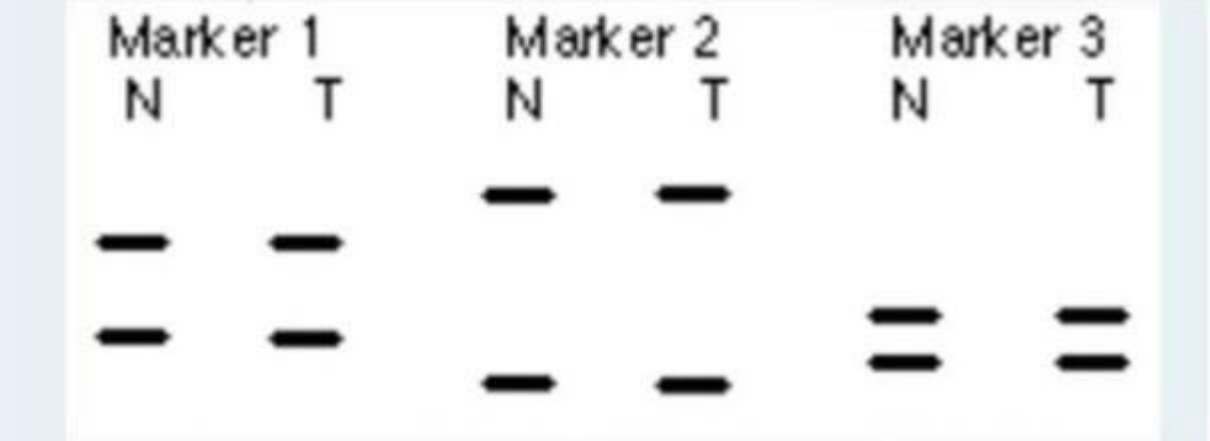
32-A de novo variant occurs due to an error in :

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

33-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

34-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 ?



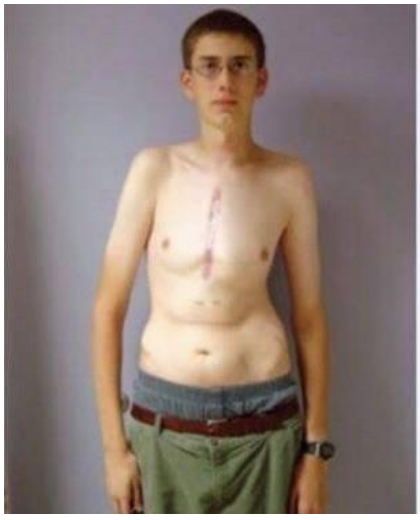
a. Second somatic point mutation hit
b. Any of those choices could explain the patient's situation
c. Loss of the normal chromosome 13
d. Loss of the mutated chromosome 13
e. Uniparental disomy (UPD) of chromosome 13

35-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%

36-The clinical features of patients with Marfan syndrome are caused by unusually stretchable connective tissue. The most observed features in Marfan syndrome affect the eye, the skeleton, and the cardiovascular system. This is an example of :



a. Allelic heterogeneity

b. Pleiotropy
c. Anticipation
d. Penetrance
e. Locus heterogeneity

37-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

38-The main features of Silver Russell syndrome (SRS) are pre- and postnatal growth restriction and a characteristic small, triangular face. SRS is also accompanied by other dysmorphic features including fifth finger clinodactyly and skeletal asymmetry. SRS could be caused by uniparental disomy (UPD) for chromosome 7. The underlying genetic etiology for the UPD pathogenicity is most probably related to :

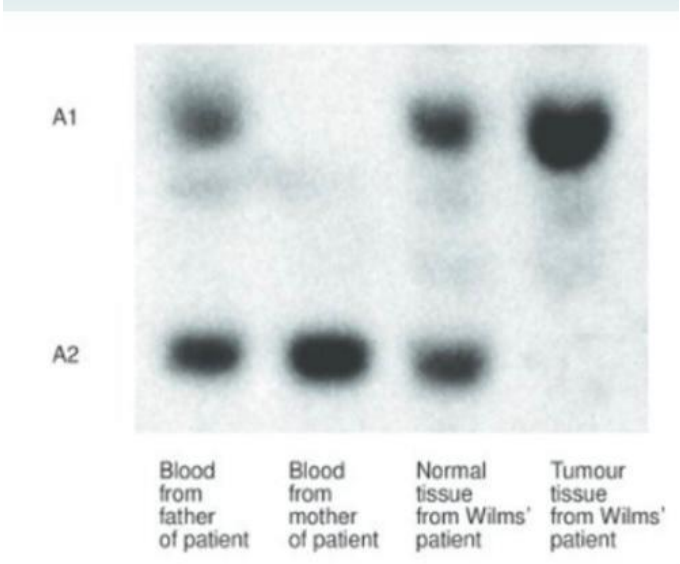
a. Missense variants
b. Synonymous variants
c. Imprinting
d. Splice site variants
e. Non-synonymous variants

39-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

40-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 llp15.5. 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



Answers

1	c	11	c	21	d	31	d
2	b	12	d	22	d	32	a
3	d	13	e	23	e	33	b
4	d	14	c	24	d	34	a
5	d	15	a	25	d	35	b
6	b	16	c	26	b	36	b
7	a	17	c	27	e	37	a
8	e	18	e	28	c	38	c
9	b	19	a	29	omitted	39	d
10	e	20	a	30	c	40	b

اذكرونا بدعوة. ✨