Test Bank

Subject:

MG-Mid 018

Collected by:

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Medical Genetics-Mid 018

1-If an embryo with 46 chromosomes develops from an egg that lost its nucleus, it will most probably become :

a. Partial mole
b. Complete mole
c. Normal conception
d. The fertilized egg will die before dividing
e. Fetus that lacks placenta

2-The number of human chromosomes by the end of anaphase of meiosis Il is :

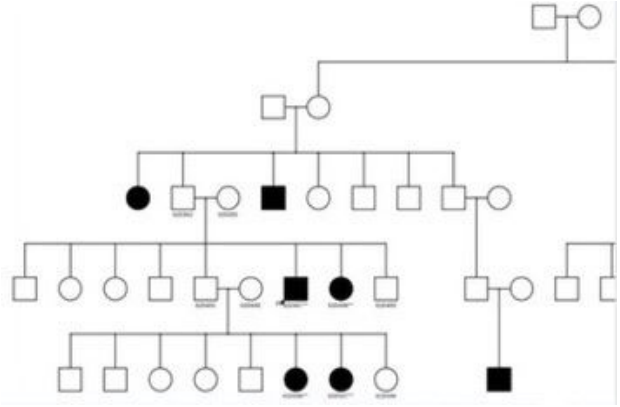
a. 69
b. 23
c. 46
d. 115
e. 92

3-Imagine if the humans' diploid chromosomal complement is 10 instead of 46. What would the number of possible combinations of meiosis be :

a. 64
b. 32
c. 16
d. 4
e. 8

4-What is most probable mode of inheritance for this pedigree :

a. X- lined recessive
b. autosomal recessive
c. autosomal dominant



d. y- linked

e. X- linked dominant

5-Which of the following human triploid is possible to be found in adults :

a. 92 ,XXXY

b. Triploid cannot be found in adult human because it is incompatible with life

c. 23 ,XY

d. 92 ,YY

e. 69 ,XXY

6-Alzheimer's disease is a progressive neurodegenerative disorder that causes memory loss and dementia. In the early 1990s, a number of scientists found that a gene called apolipoprotein E4 was associated with a higher risk of developing Alzheimer's disease (Corder et al, 1993; Saunders et al., 1993; Strittmatter et al, 1993). However, the researchers also noted that while having one or two copies of apolipoprotein E4 increase one's risk of Alzheimer's, not all carriers of apolipoprotein E4 develop the disease. This suggested that other genes and/or gene-gene interactions were involved in the development of Alzheimer's. The concept of this phoenominum is :

a. pelotropy

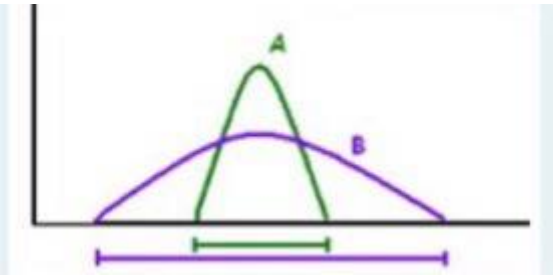
b. anticipation

c. variable age of onset

d. epistasis

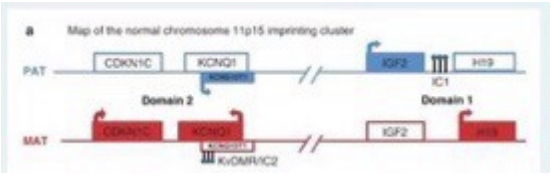
e. parent of origin effect

7-The figure depicts two possible graphs of an assumed population genetic diversity. In comparison to each other:



a. Crossing over occurs during mitosis not meiosis. Therefore, the population genetic diversity is irrelevant and graphs A and B are equally possible to occur whether crossing over happens or not
b. The crossing over does not influence the genetic diversity of the population, therefore the graphs A and B are equally possible to occur regardless whether crossing over occurs or not
c. If crossing over does not occur at all during meiosis, the population will favor graph B
d. If crossing over does occur during meiosis, the population will favor graph B
e. If crossing over does occur during meiosis, the population will favor graph A

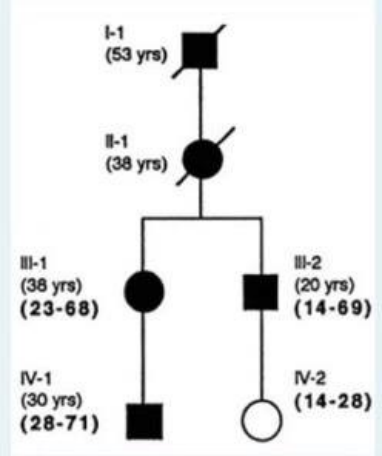
8-Beckwith-Wiedemann syndrome (BWS), which is characterized by an abnormally large birth size and a predisposition to tumors. Genetic testing can be used to confirm the diagnosis of BWS. The Image is a representation of the chromosome 1lp15.5 imprinted region that is associated with BWS. Filled rectangles indicate expressed genes and empty rectangles indicate non-expressed gene. For example: H19 gene is expressed from the maternal allele and IGF2 is expressed from the paternal allele. PAT is paternal and MAT is maternal. Which of the following might be found in BWS patient :



a. Reduced/ lack of expression of maternal CDKNIC
b. Reduced/ lack of expression of maternal CDKNIC
c. Point mutation of paternal H19
d. Nonsense mutation of maternal IGF2
e. Nonsense mutation of paternal KCNQI

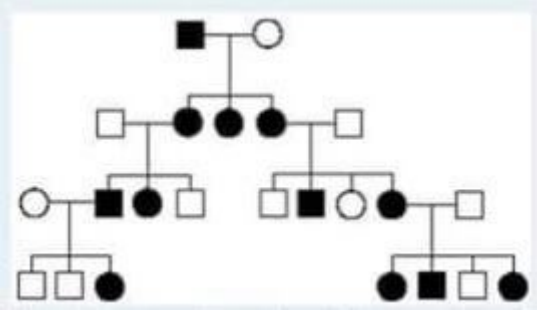
9-Kawagushi et al. (1994) cloned the MJD gene on chromosome 14 and MJD turned out to be a neurodegenerative disease caused by an unstable CAG repeat expansion. The disease is autosomal dominant and is called Machado-Joseph

disease. Age of onset and CAG-repeat numbers for both alleles are shown in brackets. The allele with disease-causing repeat number is:



a. 10
b. 20
c. 100
d. 25
e. 15

10-What is most probable mode of inheritance for this pedigree:



a. y- linked
b. X- linked dominant
c. X- lined recessive
d. autosomal recessive
e. autosomal dominant

11-X-inactivation occurs during:

a. Meiosis I of female embryo
b. Mitosis of male embryo
c. Meiosis Il of female embryo
d. Mitosis of female embryo
e. Meiosis of the female's mother

12-Different mutations in the SMPDI gene cause Niemann-Pick disease. Niemann-Pick disease is an inherited metabolic disorder, in which sphingomyelin accumulates in lysosomes in cells. This is considered:

a. Allelic heterogeneity
b. Variable age of onset
c. X linked recessive
d. Reduced penetrance

e. Variable expressivity

13-In 47,XXX individuals, two X chromosomes are inactive and only one is active, similarly in 46 XX females also one X chromosome is active. Why do 47,XXX individuals express abnormal clinical features? Because in 47,XXX:

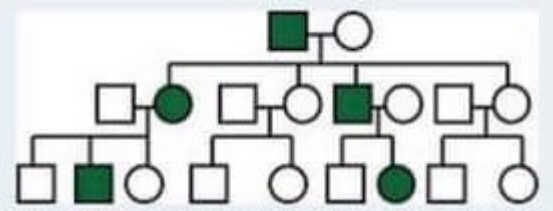
a. three X chromosomes are silent
b. the three X chromosomes are expressed (not silent)
c. one X chromosome is silent and two X chromosomes are expressed
d. the abnormal clinical features are related to autosomal chromosomes
e. affected individuals the dosage of these non- silenced genes will differ as they escape X- inactivation

14-A patient carries a pathogenic mutation in UBE3A that has a paternal parent of origin, will be affected with ?

a. The patient will not be affected with any of the diseases listed in this question
b. Miller-Dieker syndrome
c. Prader-Willi syndrome
d. DiGeorge syndrome
e. Angelman syndrome

15- What is most probable mode of inheritance for this pedigree :

a. autosomal dominant
b. X- lined recessive
c. y- linked
d. X- linked dominant
e. autosomal recessive



16-Trisomy 47,XYY is a syndrome with signs and symptoms that range from being barely noticeable to learning disabilities, speech delay, low muscle tone. How would you expect this syndrome to have occurred ?

a. Dispremy

b. Endomitosis
c. Fertilization by two sperms
d. Chromosomal rescue
e. Nondisjunction of paternal gametes

17-Which of the following karyotype is expected to be associated with abnormal phenotype [Note: "t" is translocation, and "del" is deletion]:

a. Deletion of the "P" arm of chromosome 5. Karyotype is 46,XX,del5p
b. Balanced double Robertsonian translocation between both chromosomes 13 and both chromosomes 14. Karyotype is 44,XX,t(13q;14q) *2 .
c. Deletion of the "P" arm of chromosome 22. Karyotype is 46,XX,de22p
d. Balanced Robertsonian translocation between one chromosomes 13 and one chromosome 14. Karyotype is 45,XX,t(13q;14q)
e. 46,XY

18-Which one of the following karyotypes is most likely to be found in normal human ovarian progenitor cell ?

a. 22 ,Y
b. 46 ,XY
c. 46 ,XX
d. 23 ,X
e. None of the above

19-What is the possibility for a couple to have a child with Edwards syndrome if the fathers' homologous chromosomes 18 fail to disjoin during meiosis 1 ?

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

20-This chromosome is:

a. Metacentric
b. Acrocentric
c. Submetacentric
d. Interphase chromosome
e. Telocentric

21-46 XX male syndrome is a rare condition, described by De la Chapelle et al. in 1964 where the individual has phenotypically male characteristics. It occurs in one out of every 20,000-25,000 newborn males. This condition could be related to:

a. Turner syndrome
b. Translocation of acrocentric chromosomes
c. SRY gene
d. Rb gene
e. Kleinfelter syndrome

22-Which P arm of the following chromosomes carries rDNA genes?

a. 3
b. 15
c. 6
d. 12
e. 9

23-The karyotype where euchromatic regions stain more darkly and the light regions are heretochromatin is:

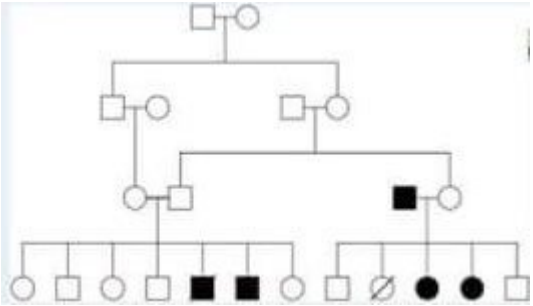
a. Q-banding
b. C-banding
c. G-banding
d. T-banding
e. R-banding

24-If one of the parent who carries balanced reciprocal translocation mates with a partner with normal karyotype. What is the risk of having a fetus with abnormal chromosomal complement ?

a. 1/5
b. 1
c. 1/3
d. 1/2
e. 1/4

25-What is most probable mode of inheritance for this pedigree:

a. autosomal dominant
b. X- linked dominant
c. autosomal recessive
d. X- lined recessive
e. y- linked



26- Which of the following fetal tissues are used for studying the fetal chromosomes :

a. Lymphocytes
b. Check swap
c. Amniotic fluid
d. Skin biopsy
e. Bone marrow

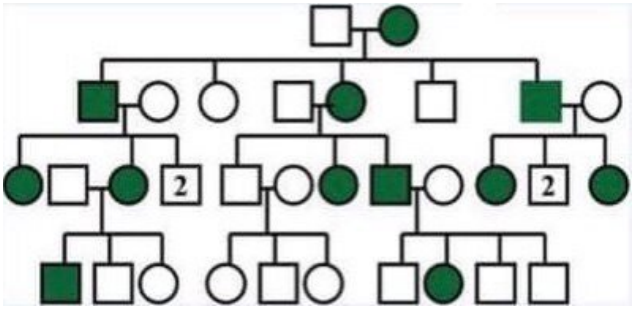
27-A child person with clinical features that include: cardiovascular, brain with neurological, renal, gastrointestinal, respiratory, and skeletal malformations, craniofacial abnormalities such as prominent occiput, hand and feet anomalies including clenched hand. This patient is most probably affected with :

a. Trisomy 18
b. Trisomy 21

c. Turner Syndrome
d. Partial Trisomy 21
e. Klinefelter Syndrome

28-What is most probable mode of inheritance for this pedigree:

a. autosomal recessive
b. X- lined recessive
c. X- linked dominant
d. y- linked
e. autosomal dominant



29-Pathogenic mutations in TBX5 cause Holt-Oram syndrome which includes cardiovascular (atrial septal defect, hypoplastic left heart syndrome), chest (Absent pectoralis major muscle) and skeletal anomalies (vertebral anomalies, triphalangeal thumb and carpal bone anomalies). The TBX5 phenotypic effect is related to which concept :

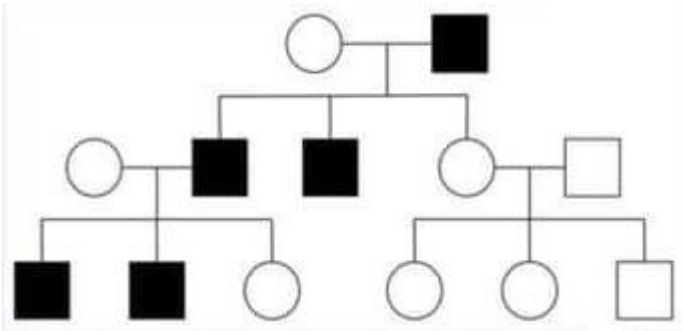
a. Reduced penetrance
b. Sex limited
c. Pleiotropy
d. Multifactorial
e. Genetic heterogeneity

30-How many double stranded DNA molecules are in a somatic human cell that is in present G2 phase:

a. 46
b. 23
c. 92
d. There are no double stranded DNA molecules in G2
e. 69

31-What is most probable mode of inheritance for this pedigree :

a. X- linked dominant
b. y- linked
c. X- lined recessive
d. autosomal dominant
e. autosomal recessive



32-The practical way to visualize a karyotype of a suspected very large chromosomal deletion, is to :

a. Arrest the cells at anaphase
b. Arrest the cells at metaphase
c. Arrest the cells at S phase
d. Arrest the cells at telophase
e. Arrest the cells at prometaphase

33-Inherited retinal dystrophies (IRDS) are a group of diverse hereditary disorders collectively characterized by progressive retinal deterioration and vision loss. More than 250 genes have been identified to cause IRD, where a mutation in any of those genes causes IRD. This is an example of :

a. Multifactorial trait
b. Pleiotropy
c. Sex limited
d. Genetic heterogeneity
e. Reduced penetrance

34-In the routinely performed karyotype (G-banding). Which of the following would you expect to have more clinical impact and lead to a disease ?

a. A duplication of a region with a light band
b. A duplication of a region with a light band
c. No clinical consequences will be observed due to a single band chromosomal aberration

d. A deletion of a region with a light band
e. A deletion of a region with a dark band

35-A chromosomal analysis is obtained on a young woman with mild signs of Patau syndrome and reveals a 46,XX/47,XX +13 mosaic karyotype. Nondisjunction is most likely to have occurred in :

a. Mosaic Patau are not possible to happen because they are incompatible with life
b. mitosis after fertilization
c. Parental meiosis II
d. Parental meiosis I
e. Parental progenitor cells S Phase

36-A person whose karyotype is 45,XX,t(15q;21q). If she mates with a 46, XY normal individual. What is the possibility that they will have a zygote which will develop into Down syndrome? ("t" is translocation):

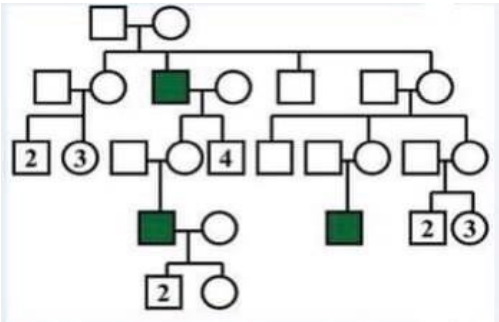
a. 1/6
b. 1/4
c. 1
d. 1/2
e. 1/3

37-You are asked to consult about a 2-month-old girl with hypotonia, seizures, and an elevated plasma lactate (8 mM/L, normal <2). Brain MRI shows a thin corpus callosum but no other abnormalities. You suspect pyruvate dehydrogenase deficiency. Which of the following is the most likely mode of inheritance in this infant ?

a. X-linked
b. Autosomal dominant
c. Autosomal recessive
d. Mitochondrial
e. Sporadic

38-What is most probable mode of inheritance for this pedigree:

a. autosomal recessive
b. X- lined recessive
c. X- linked dominant
d. y- linked
e. autosomal dominant

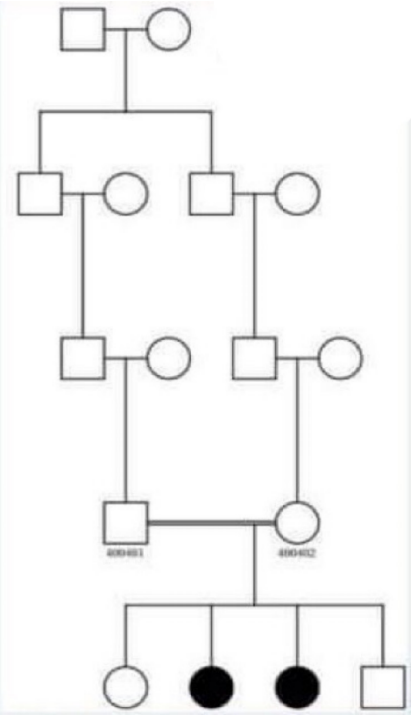


39- When a pair of twins is conceived and one embryo dies in the womb. The surviving fetus may absorb some of the cells of its deceased twin. The surviving twin will be :

a. Tetraploid
b. Triploid
c. Mosaic
d. Chimeric
e. Partial trisomy

40-What is most probable mode of inheritance for this pedigree:

a. autosomal recessive
b. X- lined recessive
c. X- linked dominant
d. y- linked
e. autosomal dominant



Answers

1	B	11	D	21	C	31	B
2	C	12	A	22	B	32	B
3	B	13	E	23	E	33	D
4	B	14	A	24	D	34	D
5	B	15	A	25	C	35	B
6	B	16	E	26	C	36	A
7	D	17	A	27	A	37	C
8	A	18	C	28	C	38	B
9	C	19	C	29	C	39	D
10	B	20	C	30	C	40	A

اذكرونا بدعوة. ✨