Genetics 019 – Final Exam

Collected by: Bara Tanbooz

1) **Rickets disease?**
   X-LINKED DOMINANT

2) **Familial hypercholesteremia?**
   AUTOSOMAL DOMINANT

3) **Myotonic dystrophy?**
   AUTOSOMAL DOMINANT

4) **Duchenne muscular dystrophy?**
   X-LINKED RECESSIVE

5) **A chromosomal analysis is obtained on a young woman with mild signs of Patau syndrome and reveals a 46, XX/47, XX, +13 karyotype. Nondisjunction is most likely to have occurred in?**
   MITOSIS AFTER FERTILIZATION

6) **In 47, XXX individuals, two 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:**
   AFFECTED INDIVISUALS, THE DOSAGE OF THESE NON-SILENCED WILL DIFFER AS THEY ESCAPE X-INACTIVATION

7) **What is the correct representation of chromosomes and chromatids after meiosis 1?**
   23 CHROMOSOMES, 46 CHROMATIDS

8) **Leber's hereditary optic neuropathy?**
   INHERITANCE OF MITOCHONDRIAL GENES

9) **Paternal deletion in chromosome 15q11-13 causes?**
   PRADER-WILLI SYNDROME

10) **Maternal deletion in chromosome 15q11-13 causes?**
   ANGELMAN SYNDROME

11) **Correct about Waardenburg syndrome?**
   SENSORINEURAL HEARING LOSS HAS A PENETRANCE OF 20%

12) **What is the Karyotype for a female that would have 3 Barr bodies due to x-inactivation?**
   47, XXXX

13) **Hemophilia?**
X-LINKED RECESSIVE

14) **A mutation leading to?**
HEARING LOSS IS DUE TO ADVANCED PATERNAL AGE IS MORE LIKELY TO OCCUR THAN DUE TO ADVANCED MATERNAL AGE

15) **Which one of the following syndromes has reduced penetrance?**
RETINOBLASTOMA

16) **The longest phase in the cell cycle is?**
THE INTERPHASE

17) **Which of the following aneuploidies is viable?**
TRISOMY 13

18) **Patau syndrome is diagnosed by?**
CYTOGENETIC ANALYSIS

19) **Which type of mutation needs at least two different chromosomes in order to happen?**
BALANCED TRANSLOCATION

20) **Two different mutations one on FGFR2 and the other on FGFR3, both leading to develop the same disease, example of?**
LOCUS HETEROGENITY

21) **Two different mutations that happen on the same locus, the first mutation leads to sickle cell disease, and the other mutation leads to thalassemia, example of?**
ALLELIC HETEROGENITY

22) **Tyrosinemia is treated by?**
DIETARY RESTRICTION

23) **According to G-banding, which statement is correct?**
G-NEGATIVE BANDS ARE GENE RICH/HAVE MORE GENES

24) **A woman that is a balanced carrier of a reciprocal translocated gene – 45, XX +t(14q,21q), married a normal man, what is the genotype of the child that is least likely to live?**
46, XX -21 +t(14q+21q) – this genotype means trisomy 14- **this question was in both mid and final exams

25) **If one of the parents who carries balanced reciprocal translocation mates with a partner with normal karyotype. What is the chance of having a child with normal karyotype?**
½ -normal+balanced carrier+trisomy21-

26) Two or more distinct cell lines from two zygotes?

CHIMERA

27) Pattern baldness, moustaches and beards in human males are examples of?

SEX-INFLUENCED TRAITS

28) PKU?

AUTOSOMAL RECESSIVE

29) ALBINISM?

AUTOSOMAL RECESSIVE

30) ACHONDROPLASIA?

AUTOSOMAL DOMINANT

31) The genotype of a phenotypically normal women but has infertility?

46, XY – mutation in the SRY gene-

32) CF patient with cancer, immune deficiency and infertility, meanwhile another patient only has infertility, example of?

VARIABLE EXPRESSIVITY

33) We use the genetic banding in order to diagnose?

CRI-DU-CHAT – because we can see the mutated chromosome 5 with it –

34) The genotype of a patient with CML?

46, XY +t (9,22)

35) 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?

ACCORDING TO 018 TEST BANK THE ANSWER IS AUTOSOMAL RECESSIVE, BUT I ANSWERED IT MITOCHONDRIAL, SINCE PYRUVATE DEHYDROGENASE IS A CELL CYCLE ENZYME SO ITS RELATED TO ATP PRODUCTION, WHICH IS MAINLY IN THE MITOCHONDRIA, SO YOU BE THE JUDGE****

36) A male with more than 400 CGG repeats is most likely going to develop?

FRAGILE X SYNDROME

37) 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 de novo variant +germline mosaicism in one of the parents anticipation+ reduced penetrance

38) **The presence of a genotype in one locus is required for the presence of another genotype in other locus is?**

EPISTASIS

39) **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?**

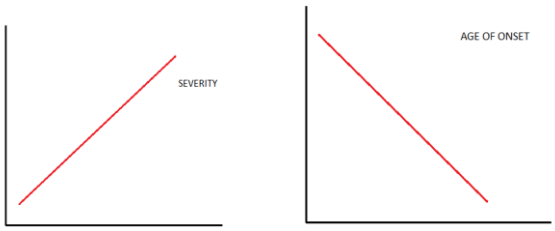
NOEY2 IS PATERNALLY EXPRESSED

40) **Multiple phenotypical changes due to a single mutation?**

PLEIOTROPY مش ذاكر السؤال صراحه بس هيك كان بما معناه

41) **Alzheimer's disease is a progressive neurodegenerative disorder that causes memory loss and dementia. In the early 1990s, several 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 phenomenon is?**

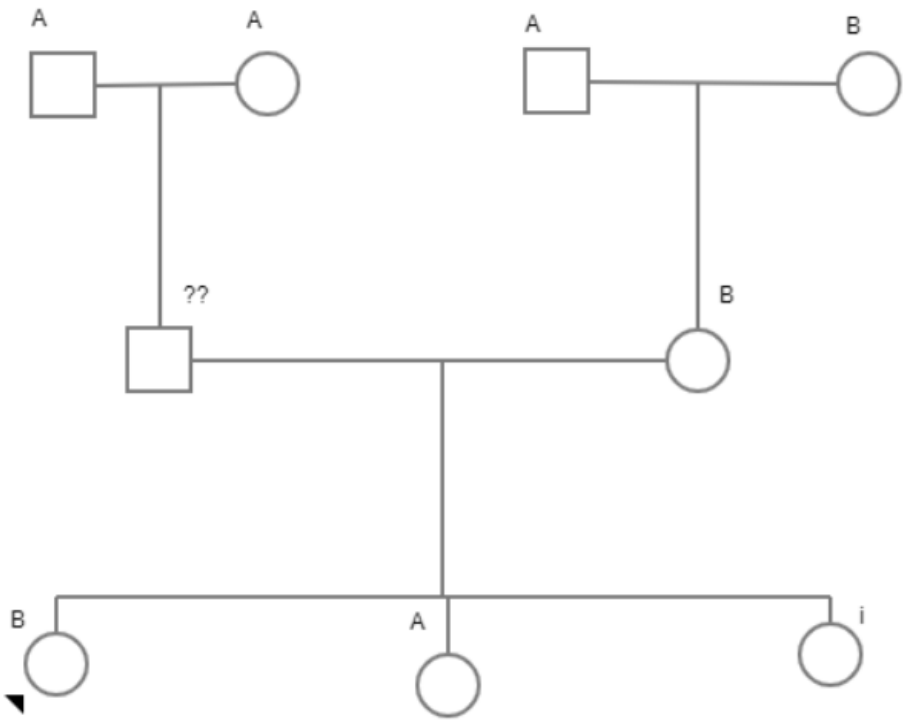
EPISTAISIS

42) 

these graphs resemble?

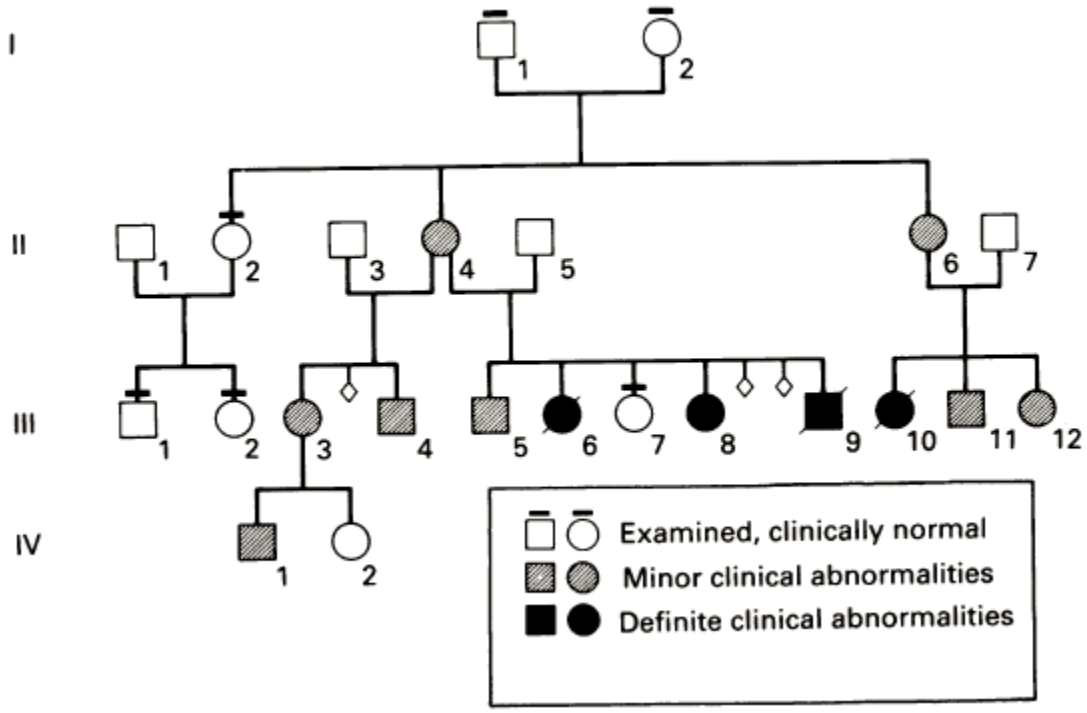
ANTICIPATON

43) This is a pedigree about the ABO blood system, we know that blood type has 3 alleles which are IA, IB, I , what is the genotype of the unknown individual?



AO- (IA,i)

44) This graph resembles which phenomena?



VARIABLE EXPRESSIVITY

THE REST OF THE QUESTIONS WERE PEDIGREES

واللي بدرس بعرف يحلهم