TEST BANK

Doctor 2019

SUBJECT:

Genetics Final 019

COLLECTED BY :

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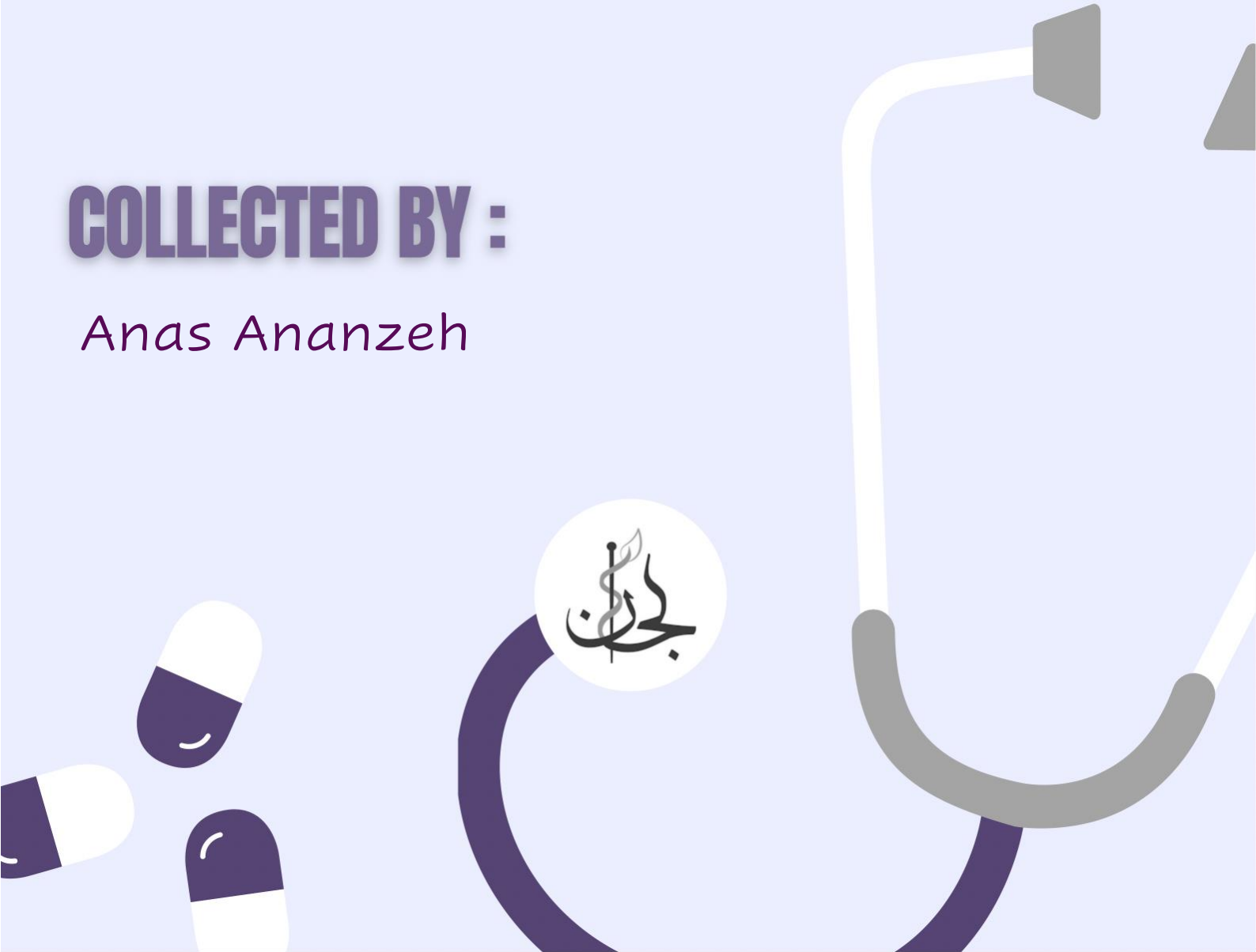


Genetics Final 019

1-Longest phase:

Answer: Interphase

2-True statement:

Answer: G negative (euchromatic region) is richer in genes than G positive (heterochromatic region)

3- A question talking about something having an effect on more than one receptor (Receptor 2 & Receptor 3), what's that called?

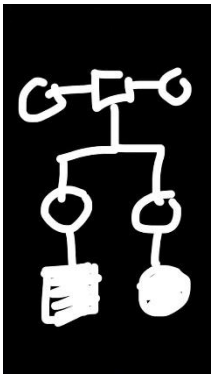
Answer: Locus heterogeneity

4- a question talking about a person having sickle cell anemia, and another one having hemophilia, due to effects on the same gene… :

Answer: Allele heterogeneity

5- True regarding this disease shown in the pedigree:

Answer: autosomal dominant with paternal imprinting



6- Repeat expansions in Huntington disease occurs in:

Answer: in the exons

7- 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

8- repeat expansion in 3' UTR:

Answer: They say it's myotonic dystrophy, but most people answered Fragile x syndrome as it's not mentioned in the course (probably deleted from the exam)

9- Asking about Hirschsprung disease:

Answer: a mutation in the RET gene

10- Large deletion in a chromosomal segment results:

Answer: Prader-Willi syndrome

11- Leber's hereditary optic neuropathy:

Answer: mitochondrial inheritance

12- We need at least 2 chromosomes in:

Answer: Reciprocal translocation

13- Female (phenotype normal but infertile):

Answer: 46,XX/ 45,XO

14- A female with genotype (XXXX) has:

Answer: 3 Barr bodies

15- X linked less severe than autosomal:

Answer: X inactivation

16- exchange of cells between these 2 different populations of embryonic cells:

Answer: Chimera

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 (the photo is missing, but you don't need it anyway!). What is the likelihood that a future sibling WILL DEVELOP retinoblastoma?

Answer: <1%

18- Lisch nodules can be found in:

Answer: neurofibromatosis type 1

19- True about X inactivation:

Answer: happens during embryonic development

20- Symptoms of Turner Syndrome:

Answer: XO

21- Symptoms of Patau Syndrome:

Answer: Trisomy 13

22- Which of the following diseases has reduced penetrance:

Answer: Retinoblastoma

23- Vitamin D deficiency:

Answer: X - linked dominant

24- Duchenne muscular dystrophy-DMD:

Answer: X - linked recessive

25- Hemophilia:

Answer: X - linked recessive

26- Color blindness:

Answer: X - linked recessive

27- Huntington disease:

Answer: Autosomal dominant

28- Marfan syndrome:

Answer: Autosomal dominant

29- Myotonic dystrophy:

Answer: Autosomal dominant

30- Albinism:

Answer: Autosomal recessive

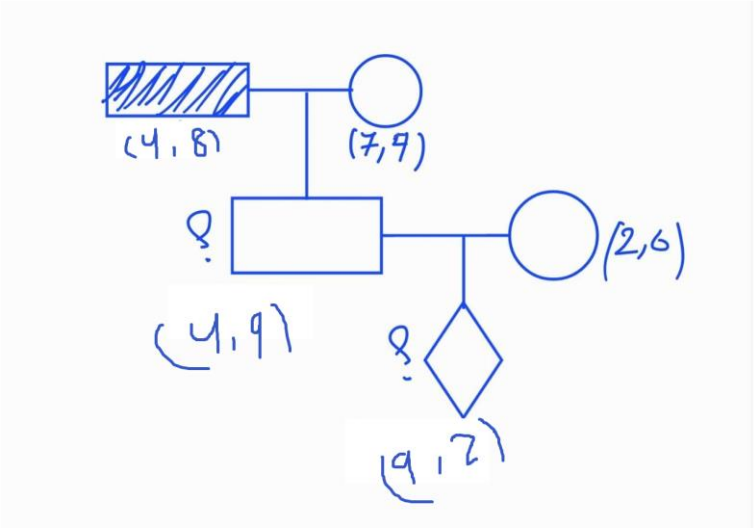
31- Cystic fibrosis:

Answer: Autosomal recessive

32- In metaphase II, what's the number of chromosomes & chromatids:

Answer: 23 chromosomes, 46 chromatids

33- Rob's father has a Huntington disease and rob himself doesn't want to know if he is affected or not. His wife has no history with the disease, and she is pregnant. What is the probability that the child is affected?



Answer: 1/2

34- Which of the following diseases can chromosomal studies be done on?

Answer: Cri-du-chat

35- 2 Brothers (one of them is affected), married 2 sisters, the unaffected brother married one of the girls (her sister's child is affected) …

What's the probability that they'll have an affected child. Giving that the disease is AR

Answer: 1/12

36- A question talking about mild symptoms Vs. severe symptoms for the same disease:

Answer: Variable expressivity

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)

Answer: 1/16

38- Genes affecting another gene phenotyping:

Answer: Epistasis

39- Asking about the most probable cause of Achondroplasia:

Answer: None of the parents is affected (Not sure)

40- Question about cystic fibrosis having more than one phenotypic effect, what is this called?

Answer: Pleiotropy

41- 60y/o man presents to neuro clinic with late onset ataxia and intention tremor his daughter has a son affected with fragile x MR, the cause of his disease:

Answer: 80 CGG

Note: Pedigrees & most of risk assessment questions aren't collected in this file, as they're hard to remember exactly, but all of them were so easy & straight forward.