**Collection of Final term genetics questions**

Note: Many of these questions are collected based on our short-term memory 😂, so plz don't focus that much on the questions wording)

1) The most common pathogenic mutation (associated with human disease): missense mutation.

2) The most common genetic variation: SNPs.

3) Mitochondrial Cristea, which is correct: carries oxidation reduction reactions.

4) Which of the following increases in frequency with advanced paternal age: achondroplasia.

5) All encoded by MtDNA except: None of the above

6) Which is wrong about outer mitochondrial membrane: embeds electron transfer chain.

7) Which of the following urea cycle enzymes is inherited in a X-linked fashion: Ornithine transcarbamylase (OTC).

8) Which of the following forms of inheritance is commonly seen in consanguinity: Autosomal Recessive.

9) Type 1 hypercholesterolemia is caused by mutation in: LDL receptor (the question asks about Type 1 hypercholesterolemia not hperlipoproteinemia , actually there is a difference between the two).

10) True or false CYP1 is the enzyme that metabolizes Codeine: false.

11) True or false: propionic acidemia is associated with low ammonia levels: false.

12) Which one of the following diseases is considered multi factorial disease: Autism.

13) Common genomic variants: All of the above (a and b).

14) Difference between mutation and polymorphism: mutation is permanent change in DNA that is less common while polymorphism is DNA variant that is more common.

15) which of the following Codes represents change in the splicing donor site: c. 1548 +1 G>A.

16) all of the following are considered transversion except: C>T.

17) the third amino acid in this mutation code (p. Ser2_ met3 ins Gln ser met): glutamine.

18) Case of a male child with mental retardation, his mother has ovarian failure, the grandpa has tremor: Grandpa has <80 CGG with FMR1 mutation while the son has > 200 CGG repeats (Fragile X syndrome)?

19) Question talks about a case with mutation in a site about 25 nucleotides upstream to exon 33 in COAL5 gene: this mutation will affect splicing with skipping codons (Sth like that).

20) What do we call change in the DNA sequence: mutation.

21) Noncoding sequence in the DNA that is about 2,3,4 base pairs long: Microsatellite.

22) Retinoblastoma with dysmorphic features: loss of chromosome 13q

23) Which of the following statements about the impact of CYP2C9 polymorphism is NOT correct?

A) Approximately 40% of Europeans exhibit CYP2C9 polymorphism.
B) The metabolism of both isomers of warfarin involves CYP2C9.
C) Variant CYP2C9*3 reduces hydroxylation of warfarin.
D) It is advised that warfarin patients are phenotyped for CYP2C9.
E) None of the above. //// don't be surprised that this question is taken from the internet and its correct answer is B 🥲 (most students chose either A or none of the above 😔)

24) a child with unilateral retinoblastoma, what is the probability that a sibling has the disease: less than 1%.

25) patient with parathyroid hyperplasia, medullary thyroid carcinoma,pheochromocytoma, what is the mutation: gain of function mutation in RET.

26) which is correct: Germline mutations in the reproductive organs and inherited while somatic are in other cells and not inherited.

27) MtDNA is simple ds circular DNA molecule.

28) Loss of heterozygosity, which best applies to this concept: loss of both tumor suppressor genes.

29) Inheritance of Leber hereditary optic neuropathy: mitochondrial DNA.

30) Nonsense mutations most probably will result in non-pathogenic outcome: if it occurs near the 3' end of exons.

31) Human mutations, all are true except: they are protective….

32) Tumor clonality: cells with non-identical genetics that are derived from single progenitor cell.

33) Truncation of proteins is associated with frameshift mutations.

34) Repair mechanisms for alcohol: Base excision repair

35) If there is a substitution in this region (AAAAA) what is the coding mutation: c. 5*…..

36) Genome project achievements: all of the above.

37) Synonymous mutations become pathogenic if: they affect critical splicing and regulatory sequences that lead to decreased protein levels??

38) Which of the following factors once become always active, lead to carcinogenesis: RAS (not sure at all about the wording of the question)

39) True about CYP 450 (Phase 1): All of the above? (Other choices were drug interaction, drug delivery, toxicity).

40) Number of mitochondria in a cell depends on functional state of that cell.

41) Stepwise ……..: none of the above?

42) Nonsense mediated decay (NMD): premature stop codon

43) Transition: very long answers but the correct answer contains (Purines to purines).

44) Which one of the following mutations occur during DNA replication: deletion of 3 nucleotides

#Risk assessment questions:

45) 33 percent.
46) 0 percent.
47) 1/240.
48) 1/32??
49) 1/8?
50) 2/3