Genetics final Exam

1)All of the following about Telomeres is correct except?
It codes for important genes

2)One of the following is true about Telomeres ?
Whole Telomeres has the same sequence

3)A cell in G0 phase,how many chromosomes?
46

4)Which of the following is mismatched?
Down syndrome,xx,+13

5)pattern baldness,moustaches,and beard in human males are example of:
Sex-influenced traits

6)A female with a flattened face, small head, short neck, protruding tongue, small ears, and a poor muscle tone (hypotonia). She probably has a genetic disorder that's caused by:
Trisomy 21

7)Which of the following genetic changes is associated with a female whose karyotype is 46,XY ?
SRY gene mutations

8)The main treatment for PKU includes:
A lifetime diet with very limited intake of protein

9)Chronic myeloid leukemia is caused by: t(9:22); translocation that fuses part of the ALB1 gene from Chromosome 9 with part of the BCR gene from chromosome 22,creating a gene called BCRALB1

10)The most common aneuploidy that infants can survive with is _ (most compatible with life);
Trisomy 21 (down syndrome)

11)Which of the following statements is NOT associated with multifactorial disease inheritance:
Increased incidence of the sibling having disease if a person has the disease late in life

12)Angelman syndrome (AS) and Prader-Willi syndrome (PWS) are examples of disorders that can be caused by:
Uniparental disomy

13)The most commonly used stain for metaphase chromosomes is:
Giemsa stain

14)The study of chromosomes and cell division is called:
Cytogenetics

15)The most stable chromosome of the following is:
45 XY t(14,21)

16)A patient with klinefelter syndrome can be seen as:
Male with 47 XXY

17)Expressivity can be described as:
The degree of which a gene is expressed depending on other genetic or environmental factors

18)Which of the following is incorrect regarding Angelman syndrome?
It's caused by a micro deletion

19)Which of the following is incorrect regarding mitochondrial inheritance?
The genes involved are inherited paternally

20)A study about a particular disease showed that monozygotic twins had 40% correlation in disease status, while normal siblings had only 10%. The best statement describing this gene abnormality is:
Genetic abnormality contributes but doesn't determine the disease occurrence

21)Karyotyping is not used for:
Insertion of few nucleotides

22)If an embryo with 46 chromosomes develops from an egg that lost its nucleus, it will most probably become:
Complete mole

23)The number of human chromosomes by the end of anaphase of meiosis II is:
46

24)Imagine if the humans' diploid chromosomal complement is 10 instead of 46. What would the number of possible combinations of meiosis be :
32

24)Which of the following human triploidy is possible to be found in adults :
Triploidy cannot be found in adults because it's incompatible with life

25)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 :
Anticipation

26)X-inactivation occurs during:
Mitosis of female embryo

27)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:
Allelic heterogensity

28)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: affected individuals the dosage of these non silenced genes will differ as they escape X inactivation

29)A patient carries a pathogenic mutation in UBE3A that has a paternal parent of origin, will be affected with ?
The patient will not be affected with any of the disease listed

30)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 ?
Non disjunction of paternal gametes

31)Which of the following karyotype is expected to be associated with abnormal phenotype [Note: "t" is translocation, and "del" is deletion]:
Deletion of the p arm of chromosome 5,karyotype is 46 XX del5p

32)Which one of the following karyotypes is most likely to be found in normal human ovarian progenitor cell ?
46/XX

33)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 ?
50%

34)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:
SRY gene

35)Which P arm of the following chromosomes carries rDNA genes?
15
[euchromatic region that is lightly stained and codes for rRNA]

36)The karyotype where euchromatic regions stain more darkly and the light regions are heretochromatin is:
R banding

37)How many double stranded DNA molecules are in a somatic human cell that is in present G2 phase:
92

38)The practical way to visualize a karyotype of a suspected very large chromosomal deletion, is to :
Arrest the cell in metaphse

39)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 deletion of a region with a light band

40)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 :
Mitosis after fertilization

42)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):
1/6

43)You are asked to consult about a 2-month-old girl with hypotonia, seizures, and an elevated plasma lactate

44)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 ?
1/2

45)Which of the following fetal tissues are used for studying the fetal chromosomes :
Aminotic fluid

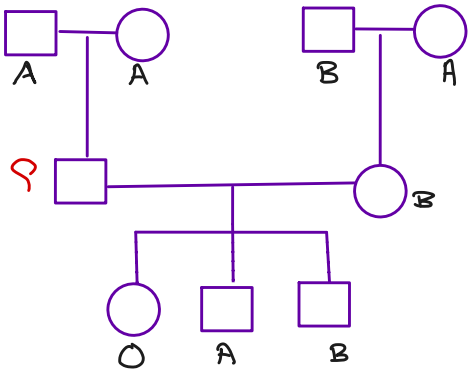
46)A child person with clinical features that include: cardiovascular, brain with neurological, renal, gastrointestinal, respiratory, and skeletal malformations, craniofacial abnormalities such as prominent occiput, hand and feet anomalies including clenched hand. This patient is most probably affected with :
Trisomy 18 (patau)

47) 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

48) The longest phase of the cell cycle ?
Interphase

49)Chimera definition

50) blood type for the known person
A



51)in G -banding :
g negative > more genes

[in GC-rich areas, giemsa will bind weakly therefore this area will have light color]

The exam also contains 9 pedigree questions

Good luck all