Robbins Review Questions - Chapter 5

1. Approximately what percentage of spontaneous abortions during the early months of gestation are due to genetic abnormalities?
    a. 20%
    b. 30%
    c. 40%
    d. 50%
2. Permanent changes to the DNA are known as _____.
3. Mutations arising within _____ (somatic/germ) cells are more likely to give rise to hereditary diseases while mutations arising within _____ (somatic/germ) cells give rise to cancers.
4. Transcription of DNA is initiated and regulated by promoter and enhancer sequences. Point mutations or deletions involving these regulatory sequences may interfere with binding of transcription factors and thus lead to a marked reduction in or total lack of transcription. This is referred to a
    a. Trinucleotide repeat mutation
    b. Point mutation within a coding sequence
    c. Mutation within a noncoding sequence
    d. Insertion or deletion
5. Missense mutations are single nucleotide substitutions that
    a. Alter the meaning of the sequence of the encoded protein
    b. Do not alter the meaning of the sequence of the encoded protein
    c. Amplify the sequence of three nucleotides
    d. All of the above
6. A distinguishing feature of _____ is they are characterized by amplification of a sequence of nucleotides and this change is dynamic (i.e., the degree of amplification increases during gametogenesis).
    a. Nonconservative missense mutations
    b. Conservative missense mutations
    c. Trinucleotide-repeat mutations
    d. Deletions and insertions
7. Mutations involving _____ (single/multiple) genes typically follow one of three patterns of inheritance: autosomal dominant, autosomal recessive, and X-linked.
8. Differentiate between penetrance and expressivity.
9. Autosomal _____ (dominant/recessive/codominant) disorders are manifested in the heterozygous state.
10. When a single mutant gene may lead to many end effects, this is termed
    a. Variable penetrance
    b. Pleiotropism
    c. Variable expressivity
    d. Genetic heterogeneity
11. Variation in the degree of phenotypic expression influenced by a single gene is
    a. Penetrance
    b. Pleiotropism
    c. Variable expressivity

    d. Genetic heterogeneity
12. Mutations at several genetic loci producing the same trait is known as
    a. Penetrance
    b. Pleiotropism
    c. Variable expressivity
    d. Genetic heterogeneity
13. Mendelian genetic disorders that usually result in a loss of function and show clinical manifestations later in life are typically
    a. Autosomal dominant
    b. Autosomal recessive
    c. X-linked disorders
    d. Epigenetic mutations
14. Mendelian disorders result from alterations involving _____ (single/multiple) genes.
15. This disorder involves connective tissues, manifested principally by changes in the skeleton, eyes and cardiovascular system. It is related to a defect in fibrillin-1.
16. This disorder results from a mutation in the synthesis and structure of fibrillar collagen.
17. Common symptoms of Ehlers-Danlos syndrome include all of the following except
    a. Fragile, hyperextensible skin vulnerable to trauma
    b. Hypermobile joints,
    c. Mitral valve prolapse
    d. Ruptures of the colon or large arteries
    e. Stiff joints
18. Mutations in the gene encoding LDL receptors lead to
    a. Familial hyperbilirubinemia
    b. Familial hypercholesterolemia
    c. Tay-Sachs disease
    d. Non alcoholic fatty liver disease
19. An accumulation of GM2 gangliosides because of a deficiency in hexosaminidase involves neurons of the central and autonomic nervous systems and retina. This commonly gives rise to a cherry red spot on the macula. What lysosomal storage disease does this describe?
20. The lysosomal disease that is recognized by an inability to catabolize gangliosides is
    a. Tay-Sachs
    b. Niemann Pick A-C
    c. Gaucher
    d. All of the above
    e. Not enough information to tell
21. Niemann-Pick disease types A and B are due to lysosomal accumulation of _____ due to a deficiency in _____ (an enzyme).
22. What is the most common lysosomal storage disease?
23. Gaucher disease is a reduction in the activity of what enzyme?
    a. Sphingomyelinase
    b. Glucocerebrosidase
    c. Hexosaminidase
    d. Glycosaminoglycansidase

24. The disorder mucopolysaccharidoses is a deficiency in the enzyme that degrades _____.
25. Inherited deficiency of enzymes involved in glycogen metabolism can result in storage of normal or abnormal forms of glycogen, predominantly in _____ or _____ (i.e. tissues that most commonly store glycogen) but also in other tissues as well.
26. Briefly outline the normal human karyotype.
27. Karyotyping is done by arresting the cell cycle during which phase of the cell cycle?
28. Any exact multiple of the haploid number of chromosomes (23) is called _____(euploid/diploid/aneuploid). If an error occurs in meiosis or mitosis and a cell acquires a chromosome complement that is not an exact multiple of 23, it is referred to as _____ (haploidy/diploidy/aneuploidy).
29. What is the consequence of a Robertsonian translocation?
30. Mitotic errors in early development give rise to two or more populations of cells with different chromosomal complement, in the same individual, a condition referred to as _____?
31. What is the most common chromosomal disorder?
32. Trisomy 18 and 13 are more commonly known as _____ and _____.
33. Chromosome 22q11.2 deletion results in what syndrome?
34. DiGeorge syndrome results in
    a. Marrow hypoplasia and resultant anemia
    b. Hepatic hypoplasia, metabolic deficiency and hepatic encephalopathy
    c. Neural crest hypoplasia and cognitive deficiency
    d. Thymic hypoplasia and T-cell deficiency
35. Imbalances (excess or loss) of sex chromosomes are much better tolerated than are similar imbalances of autosomes. True or false?
36. This is most often defined as male hypogonadism and occurs when there are two or more X chromosomes and one or more Y chromosomes.
37. Complete or partial monosomy of the X chromosome is known as _____, resulting in hypogonadism in phenotypic females.
38. What is the most common cause of male sterility?
39. Bilateral neck webbing, cardiovascular abnormalities, failure to develop normal secondary sex characteristics, amenorrhea and streak ovaries are associated with what genetic disease?
40. What characteristic determines genetic sex?
41. Compare/contrast a hermaphrodite with pseudohermaphrodite.
42. Trinucleotide repeat mutations are an important cause of
    a. Cardiovascular disease
    b. Neurodegenerative disease
    c. Sterility
    d. Muscular dystrophies
43. Down syndrome is the most common genetic cause of mental retardation. What is the second most common cause?
44. Mitochondrial DNA is inherited entirely from which parent?
45. What is genomic imprinting?

1. D (pg 137)
2. Mutations (pg 138)
3. Germ. Somatic (pg 138)
4. C (pg 138-139)
5. A (pg 138)
6. C (pg 138-139)
7. Single (pg 140)
8. **Penetrance** - The percentage of individuals with the mutant gene and who show phenotypic signs of the mutation. Some individuals inherit the mutant gene but are phenotypically normal. This is referred to as incomplete penetrance. Penetrance is expressed in mathematical terms. Thus, 50% penetrance indicates that 50% of those who carry the gene express the trait. **Expressivity** - if a trait is seen in all individuals carrying the mutant gene but is expressed differently among individuals, the phenomenon is called variable expressivity (pg 140)
9. Dominant (pg 140)
10. B (pg 140)
11. C (pg 140)
12. D (pg 140)
13. A (pg 140-141)
14. Single (pg 142)
15. Marfan syndrome (pg 144)
16. Ehlers Danlos (pg 144)
17. E (pg 145-146)
18. B (pg 147)
19. Tay-Sachs (pg 151)
20. A (pg 151)
21. Sphingomyelin. Sphingomyelinase (pg 152)
22. Gaucher disease (pg 153)
23. B (pg 153)
24. Glycosaminoglycans (pg 154)
25. Liver or muscles (pg 157)
26. Human males and females have 46 chromosomes; these comprise 22 homologous pairs of autosomes (ie somatic chromosomes) and two sex chromosomes, XX in the female and XY in the male (pg 158)
27. Metaphase (pg 158)
28. Euploid. Aneuploid (pg 159)
29. Typically the translocations/breaks occur close to the centromeres of each chromosome. Transfer of the segments between the chromosomes then leads to one very large chromosome and one extremely small one. Usually the small product is lost and the large chromosome is retained (pg 161)
30. Mosaicism (pg 159)
31. Trisomy 21 or Down syndrome (pg 162)
32. Edward and Patau syndrome (pg 163)
33. Chromosome 22 deletion (pg 163)
34. D (pg 163)
35. True (pg 164)

36. Klinefelter syndrome (pg 165)
37. Turner syndrome (pg 166)
38. Klinefelter syndrome (pg 165)
39. Turner syndrome (pg 166)
40. Presence of a Y chromosome dictates testicular development and the genetic male gender (pg 167)
41. Both ovarian and testicular tissue are present in a true hermaphrodite whereas a pseudohermaphrodite represents a disagreement between the phenotypic and gonadal sex (eg female hermaphrodite has ovaries but male external genatalia) (pg 167)
42. B (pg 168)
43. Female (pg 169)
44. The mother (pg 171)
45. Differences between the paternal and maternal alleles can result in inactivation of either the maternal or paternal allele. Maternal imprinting refers to transcriptional silencing of the maternal allele, whereas paternal imprinting implies that the paternal allele is inactivated (pg 172)