Faculty of Medicine
Department of Molecular and Medical Genetics
Typical Exam Tests
Genetics 1

1. During telophase all of the above mentioned takes place **EXCEPT**:
   A. New nuclear envelopes form; B. The chromosomes become less dense and more thread-like;
   C. DNA is duplicated; D. The chromosomes are released from the microtubules.

2. After which of the following divisions will there be 23 chromosomes and 46 chromatids in the cell?
   A. Mitosis; B. I meiotic division; C. II meiotic division; D. Amitosis.

3. The human haploid genome is composed of approximately:
   A. 6 billion base pairs of DNA and 20000 protein coding genes;
   B. 6 million base pairs of DNA and 2000 protein coding genes;
   C. 3 million base pairs of DNA and 20000 protein coding genes;
   D. 3 billion base pairs of DNA and 20000 protein coding genes.

4. What is the complementary strand for the DNA of the following RNA strand: 5' AGCCAUUUGCACG 3'?
   A. 3' CGUGCUUUGGCU5'; B. 3' ATGCGTTTACCGA 3';
   C. 5' CGUGCAAUGGCU 3'; D. 3' TCGGTTTCGTGC 5'.

5. Which of the following elements are present in the processed pseudogenes?
   A. Anticodons; B. Introns; C. Exons; D. Enhancers.

6. What effect can methylation of CpG iclands in the promoter region have on gene expression?
   A. Transcription level will be reduced; B. Transcription level is sharply increased;
   C. It will affect translation; D. It will affect splicing process.

7. Which of the following contains protein-encoding genes?
   A. Heterochromatin; B. Centromeric regions; C. Telomeric regions; D. Euchromatin.

8. Which of the following would result in the highest rate of mutation?
   A. Small genes, few hot spots, old age; B. Large genes, many hot spots, old age;
   C. Small genes, many hot spots, young age; D. Large genes, few hot spots, old age.

9. A frameshift mutation is one of the most severe types of mutations because:
   A. More than one gene is affected; B. They occur only in gametes;
   C. Translation is stopped; D. More than one amino acid or entire proteins are affected.

10. Which statement about transcription is true:
    A. Describes the production of polypeptides from the mRNA template;
    B. Produces single-stranded mRNA using the sense DNA strand as a template;
    C. Is regulated by transcription factors that bind to the 3' UTR;
    D. Precedes 5' capping and polyadenylation.

11. Which of the following processes take part in the nucleus of a cell?
    A. RNA processing; B. Posttranslational processing;
    C. Translation; D. Binding of amino acids with transport RNA.

12. During somatic rearrangement which part of the DNA is excised?
A. Exons; B. Introns; C. Gene segments; D. Promotor sequence.

13. Which of the following is **NOT** true about indels?
A. The number of repeats correspond to the number of alleles;
B. Their DNA sequences may vary from 2 pb to 200 bp;
C. There are hundreds to thousands of indels in the genome;
D. Indels are never inherited.

14. In a case when two related individuals in a family have the same disease, they are said to be:
A. Concordant; B. Discordant; C. Quantitative traits; D. Qualitative trait.

15. Which of the following is directly involved in DNA repair:
A. Topoisomerase; B. Ribosomes; C. DNA polymerases; D. Telomerases.

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20. The mutation, causing sickle-cell disease is GAG → GTG in the β-globin gene. This is an example of:
A. Frameshift mutation; B. Missense mutation; C. Nonsense mutation; D. Dynamic mutation.

21. In a case when two related individuals in a family have the same disease, they are said to be:
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22. A polymorphism is:
A. Any change in the DNA sequence;
B. A variation of gene or marker sequence present in >1% of the population;
C The least common variation of a gene or marker sequence;
D. The most common variation of a gene or marker sequence.

23. Which polymprphism is represented with a multiple alleles and used for identity testing:
A. STRs; B. SNPs; C. LINE-1; D. CNVs.

24. Red-green colour-blindness is a relatively benign condition. The gene responsible for this condition is located on the X chromosome and is inherited as a recessive trait. Approximately 7-10% of men and 0.5-1% of women are affected. What is the chance that a colour-blind father and a normal mother will produce a colour-blind son?
A. 0; B. 25%; C. 50%; D. 100%.

25. Robin is affected by an autosomal dominant disorder inherited from her mother. She is married to Chad, who is unaffected and has no history of the disorder in his family. Robin and Chad have two unaffected

children. Studies suggest that for every 100 individuals who inherit mutations in the gene of interest, only 50 actually show symptoms. The new mutation rate for this disorder is essentially zero. Based on this, what is the probability that their next child will present with the clinical signs of the disease?
A. 3/4; B. 1/2; C. 1/4; D. 1/8.

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29. Tay-Sachs disease is inherited as an autosomal recessive, **characterises** deficiency of the enzyme hexosaminidase A. Symptoms include blindness and retardation. Onset of symptoms begins at about six months of age and death results in early childhood. What is the probability that a homozygous normal man and a carrier female will have a child with Tay-Sachs disease?
A. 0%; B. 25%; C. 50%; D. 75%.

30. Hemophilia A is a disease in which patients lack a clotting factor in the blood (factor VIII). It is inherited as an X-linked recessive trait. What is the probability of a haemophilic male and a career female producing a hemophilic son?
A. 0%; B. 25%; C. 50%; D. 75%.

31. Nearly every individual affected with neurofibromatosis type 1 (NF1) exhibits clinical symptoms. Some, however, may present with cafe au lait spots and Lisch nodules, while others have life-threatening tumors surrounding the spinal cord. These represent an example of which of the following?
A. Variable expressivity; B. Reduced penetrance; C. Locus heterogeneity; D. Sex-influenced expression.

32. Which of the following definitions best corresponds to the disease, that is caused by different types of mutations in the same locus?
A. Genetic heterogeneity; B. Allelic heterogeneity; C. Locus heterogeneity; D. Phenotypic heterogeneity.

33. Which of the following is true about incomplete dominance?
A. The hybrid shows a phenotype that is intermediate between the two homozygotes;
B. The dominant allele is expressed only in heterozygotes;
C. The hybrid expresses the traits carried by both alleles;
D. Patients homozygous and heterozygous for a mutant allele will have equally severe disease.

34. Which of the following statements is true about polygenic trait?
A. Several different genes influence one trait; B. One gene influences many traits;
C. Several different genes influence many traits; D. Several alleles exist for one trait.

35. Which of the following is **TRUE** about inheritance of complex diseases?
A. It appears in every second generation of the family;
B. Concordance rate is same in both monozygotic and dizygotic twins;
C. The disease is more common is close relatives of the proband than in far relatives;
D. If a person has a predisposition gene, he will develop the disease.

36. Which of the following is true about conditions that show a multifactorial inheritance pattern?

A. The recurrence risk is lower if more than one family member is affected;
B. If the expression of the disease in the proband is more severe, the recurrence risk is lower;
C. The recurrence risk is higher if the proband is of the less commonly affected sex;
D. The recurrence risk for the disease is quite high even in remotely related relatives.

37. Which of the following is **TRUE** about cleft lip/palate (CL/P)?
A. Syndromic cleft lip/palate is always inherited as a single-gene disorder;
B. It causes mental retardation;
C. Most of the patients with cleft lip/palate are males;
D. In isolated cases of CL/P the recurrence risk in relatives does not always correlate with proband's severity.

38. Which of the following may be a cause of cerebral vein thrombosis?
A. Mutation of prothrombin and FVL genes at the same time;
B. Mutation of prothrombin gene and use of oral contraceptives;
C. Mutation of FVL gene and use of oral contraceptives;
D. Any of the above.

39. Which of the following would be most likely to produce an imbalance in the amount of essential genetic material in the carrier?
A. Robertsonian translocation; B. Pericentric inversion; C. Reciprocal translocation; D. Isochromosome.

40. Which of the following types of chromosomes correspond to the dicentric chromosomes?
A. Duplicated; B. Homological; C. Abnormal; D. Diploid.

41. Which of the following does **NOT** cause Down syndrome?
A. Robertsonian translocation between 21q and chromosome 14;
B. Reciprocal translocation between 21q and chromosome 15;
C. Robertsonian translocation between 21q and chromosome 22;
D. Isochromosome 21q21q.

42. Which is **NOT** a feature of Kleinfelter syndrome?
A. Short height; B. Gynecomastia; C. Long, thin fingers, arms and legs; D. Small testes.

43. Marcus and Carmen have a child with ambiguous external genitalia. The internal genitalia are female and chromosome analysis is 46,XX. Biochemical studies reveal a deficiency of 21-hydroxylase. What is the most likely diagnosis?
A. Congenital adrenal hyperplasia; B. Camptomelic dysplasia;
C. Androgen insensitivity; D. Turner syndrome.

44. Which of the following statement is correct:
A. X chromosome inactivation, all the genes of one X are switched off;
B. Male fetal development is solely dependent on normal functioning of the SRY gene;
C. Female fetal development is solely dependent on normal functioning of the SRY gene;
D. X chromosome inactivation may be linked to discordance in monozygotic twin pairs.

45. When is Barr body formed?
A. During oogenesis; B. During spermatogenesis; C. Early embryogenesis; D. After birth.

46. Dosage compensation in mammals typically involves the random inactivation of one of the two X chromosomes relatively early in development. In a such X chromosome inactivation involve:
A. *FMRI* gene; B. *SOX* gene; C. *SRY* gene; D. *XIST* gene.

Genetics 2

1. Which one of the following violates the assumptions upon which the Hardy Weinberg Law is based?
   A. There is no selection against any allele in the population;
   B. There is a constant mutation rate where lethal genes are replaced by new mutations;
   C. There is a large population with assortative mating;
   D. There is no migration into the population.

2. The major abnormal form of hemoglobin that accumulates in a fetus with the severe form of alfa-thalassemia (hydrops fetalis) is composed of:
   A. A tetramer of alpha subunits; B. A tetramer of beta subunits;
   C. A tetramer of delta subunits; D. A tetramer of gamma subunits.

3. Which of the following about Hb Hammersmith is **FALSE**:
   A. It is an unstable Hb that causes denaturation of hemoglobin tetramer;
   B. The polypeptide chain mutation is -beta chain Phe22Ser;
   C. It tends to cristallize in red blood cells;
   D. Mutation allows heme to drop out off its pocket.

4. For a woman living in central Africa, which genotype would be the most advantageous to have?
   A. Homozygous for the sickle cell allele; B. Heterozygous for the sickle cell allele;
   C. Homozygous for the normal hemoglobin allele; D. It doesn't matter; all are equally advantageous.

5. Which of the following about Hb Kempsey is **FALSE**:
   A. It has high oxygen affinity;
   D. It has low oxygen affinity;
   C. Henz bodies are formed that damage the red blood cell membrane;
   D. Mutation prevents oxygen-related movement between the chains and it can not give oxygen to tissues.

6. During which autosomal-recessive disease is mental retardation caused by the accumulation of a certain amino acid in body fluids?
   A. Tay-Sachs; B. Cystic fibrosis; C. Muscle dystrophy; D. Phenilketonuria.

7. Which disease is most commonly associated with a mutation-induced dysfunction of an enzyme involved in removing sugar side-chains from long-chain lipids?
   A. Cystic fibrosis (CF); B. Homocystiuria; C. PKU; D. Tay-Sachs disease.

8. All of the following are true about homocystinuria **EXCEPT**:
   A. It is a vitamin-responsive disease and the administration of large amounts of pyridoxine usually ameliorates the disease;
   B. Deficiency of homocysteine causes death before the age of 5 years;
   C. It is characterized with dislocation of the lens, mental retardation, osteoporosis, long bones, and thromboembolism of both veins and arteries;
   D. It is often confused with Marfan Syndrome.

9. Which of the following is true about alpha $_1$-Antitrypsin (alpha 1AT) deficiency: A. It is X-linked recessive disorder; B. It mainly affects respiratory and digestive systems; C. It is autosomal recessive condition associated with a substantial risk of emphysema and cirrhosis of the liver; D. It is autosomal recessive disorder associated with mental retardation.

10. In alpha $_1$-Antitrypsin (alpha 1AT) deficiency which organ is a site of alpha *1AT* gene expression:
    A. Liver; B. Lungs; C. Brain; D. Muscles.

11. In which of the following populations is alpha 1AT deficiency most prevalent?

A. Asians; B. White populations; C. African Americans; D. Ashkenazy Jews.

12. Alexander has congenital myotonic dystrophy, which is an autosomal dominant myopathy. Alex's mother is mildly affected with moderate facial weakness and myotonia. What is the most likely molecular mechanism for congenital myotonic dystrophy?
    A. A splice-site mutation near the 5~ end of the DMPK (myotonic dystrophy protein kinase) gene;
    B. A point mutation in the promoter region of the DMPK gene;
    C. An expansion of a CTG triplet repeat in the 3' region of the DMPK gene;
    D. A deletion that encompasses the entire DMPK coding region.

13. Which of the following diseases is characterized with X-linked inheritance, is severe, progressive, quite common and incurable?
    A. Miotonic dystrophy; B. Cystic fibrosis;
    C. Duchenne muscular dystrophy; D. Xeroderma Pigmentosum.

14. Mark is affected with Duchenne muscular dystrophy, an X-linked disorder. Mutation analysis in his family identifies a single base pair substitution in exon 1 of the DMD gene, introducing a premature stop codon. The most likely consequence of this mutation is a reduction in which of the following?
    A. Amount of DMD DNA; B. Length of DMD DNA;
    C. Length of DMD mRNA; D. Length of DMD-encoded protein.

15. Which statement about sickle-cell disease is true:
    A. The sickling effect of red blood corpuscles is the result of abnormal Hb binding with the red blood cell membrane;
    B. Hb S differs from normal Hb A by a single amino-acid substitution;
    C. Splenic infarction may occur but this has little clinical consequence;
    D. Point (missense) mutations are the usual cause of abnormal Hb in the sickling disorders.

16. Which of the following statements is true:
    A. Cystic fibrosis and hemophilia are unlikely candidates for gene therapy;
    B. Neurofibromatosis is characterized with de novo mutations;
    C. Scoliosis can be a feature of both NF1 and Marfan syndrome;
    D. Cataracts can be a feature of NF1 but not NF2.

17. Which change in the polipeptide chain is characteristic to Hb S:
    A. Beta chain: Glu6Lys; B. Beta chain: Glu6Val;
    C. Beta chain: Glu6Tyr; D. Beta chain: Glu6 His.

18. The worldwide distribution of beta thalassemia, sickle cell anemia, and glucose-6-phosphate dehydrogenase (G6PD) deficiency coincides with that of:
    A. Influenza; B. Malaria; C. Cholera; D. Multiple sclerosis.

19. Which disease is most commonly associated with a mutation-induced cell-membrane transport protein alteration:
    A. Galactosemia; B. Huntington disease; C. Phenilketonuria (PKU); D. Cystic fibrosis (CF).

20. Which disease is most commonly associated with a mutation-induced alteration of a protein by expansion of an unstable repeat:
    A. Cystic fibrosis (CF); B. Huntington disease; C. Phenilketonuria (PKU); D. Tay-Sachs disease.

21. Which substance is highly elevated in blood serum in the preclinical stage of Duchenne muscular dystrophy? A. Dystrophin; B. Creatin-kinase; C. Cholesterol; D. Phenilalanine.

22. Gene Mutation of which substance can cause Duchenne and Becker muscular dystrophies?
    A. Kreatin-kinase; B. Dystrophin; C. Cholesterol; D. Phenilalanine.

23. John is a 42-year-old man with hemophilia A. John's daughter, Susan, is married to Fred, who also has hemophilia A. Susan and Fred are expecting their first child, and ultrasound demonstrates the fetus is a boy. The probability that Fred and Susan's new son will have hemophilia A is closest to which of the following? A. 3/4; B. 2/3; C. 1/2; D. 1/4.

24. In normal conditions tau-protein is involved in the formation of which of the following structures?
A. Membrane; B. Microtubules; C. Organelles; D. Myelin.

25. Which of the following types of mutation causes Alzheimer disease in Down syndrome?
A. Inversion; B. Translocation; C. Duplication; D. Deletion.

26. Which of the following protein is responsible for the age at onset of Alzheimer disease?
A. Preselin; B. Amyloid; C. Apolipoprotein; D. Alfa -secretase.

27. Which of the following genes are NOT associated with Familial Hypercholesterolemia:
A. *LDL* receptor; B. *HEXA* and *HEXB*; C. Apoprotein *B-100*; D. *ARH* adaptor protein.

28. Enhanced synthesis of Hyperphosphorylated forms of which of the following substances can increase the risk of Alzheimer's disease?
A. Alfa –secretase; B. Phenylalanine; C. Tau-protein; D. Kreatin-kinase.

29. In which of the following diseases the first-degree relative has a high risk of developing a disease?
A. Tay-Sachs; B. Fragile X-syndrome; C. DMD; D. Alzheimer disease.

30. How are mtDNA distributed to the daughter cells?

A. Regularly; B. Randomly; C. Equally; D. In a specific order.

31. Which of the following paternally inherited genetic information is eliminated from the embryo?
A. X chromosome; B. mtDNA; C. Excess DNA; D. Unbalanced translocation.

32. Which of the following mutation has NOT been identified in mtDNA?
A. Missense; B. Point; C. Duplication; D. Robertsonian translocation.

33. Which of the following is NOT a triplet repeat disorder?
A. Fragile X syndrome; B. Huntington disease; C. Myotonic dystrophy; D. Duchenne muscular dystrophy.

34. Expansion of which of the following trinucleotide sequences causes Fragile X syndrome?
A. CTC; B. TGG; C. CGG; D. CAG.

35. Which of the following processes causes inhibition of transcription in Fragile X syndrome?
A. Deletion; B. Acetilisation; C. Methylation of cytosines; D. Inversion.

36. Fragile X syndrome and Huntington disease are caused by:
A. Tandem duplication; B. Fusion gene; C. Expanding triplet repeats; D. Deletion.

37. How does CGG expansion influences the function of FMRP protein in Fragile X syndrome?
A. FMRP gains the function; B. FMRP aquires a novel property;
C. FMRP is expresses ectopically; D. FMRP loses the function.

38. Expansion of which of the following trinucleotide sequences causes Friedreich ataxia?
A. GAA; B. CGG; C. CGT; D. TAT.

39. Which of the following proteins loses its function in Friedreich ataxia?
A. Hemoglobin; B. Hexosaminidase; C. Frataxin; D. Hydroxilase.

40. Which protein is encoded by the gene that causes myotonic dystrophy?
A. Dystrophin; B. Protein kinase; C. Creatine kinase; D. Cholesterol.

41. All of the following is notorious for muscular dystrophy EXCEPT:
A. Lack of penetrance; B. Pleiotrophy; C. Variable expression in clinical severity and age of onset;
D. Constant expression in clinical severity and age of onset.

42. Which disease is most commonly associated with a mutation-induced alteration of a protein by expansion of a segment encoded by an amplified region of trinucleotide repeats?
A. Cystic fibrosis; B. Huntington disease; C. Phenilketonuria; D. Tay-Sachs disease.

43. What is the probability that a mother, how contains 41 CGG repeats in the exon of one of the genes on her X chromosome will have a child with Huntington disease? A. 0%; B. 25%; C. 50%; D. 75%.

44. The first apoptotic gene associated with cancer was identified in which of the following types of cancer.
A. Lymphoma; B. Lypoma; C. Sarcoma; D. Myoma.

45. What is the name of a normal gene, which, when activated by a mutation, is transformed to an oncogene.
A. Supressor; B. Regulator; C. Modificator; D. Protooncogene.

46. Which of the following properties of a gene is characteristic to many types of cancer?
A. Duplications; B. Amplification; C. Insertions; D. Translpocations.

47. Which of the following statements is true concerning the function of proto-oncogenes:
A. Proto-oncogenes serve as a signal for cellular apoptosis;
B. Proto-oncogenes are components of cell growth pathways;
C. Proto-oncogenes are cell checkpoint regulators;
D. Proto-oncogenes repair DNA damage across the genome.

48. What change(s) in the retinoblastoma (Rb) gene are required for the formation of bilateral retinoblastomas in a young child?
A. Somatic occurrence of a single mutation in one Rb allele in an otherwise genetically normal cell;
B. Somatic occurrence of mutations in both Rb alleles in an otherwise genetically normal cell;
C. Loss of function of one copy of the Rb gene due to an inherited mutation;
D. Loss of function of both copies of the Rb gene due to inherited and somatic mutations.

49. Which of the following is true:
A. The two-hit hypothesis predicted that a tumor would develop when both copies of a critical gene were mutated;
B. TP53 mutations are only found in Li-Fraumeni syndrome;
C. The RET proto-oncogene is implicated in all forms of multiple endocrine neoplasia (MEN);
D. One mutation in the somatic cells is sufficient to cause cancer.

50. Telomerase loses its functions during the process of cell differentiation and it becomes:
A. Long;
51. B. Short; C. Thick; D. Thin.

52. Telomerase expression gives cancer cells the property of:
A. Limited proliferation; B. Unlimited proliferation;
C. Local proliferation; B. Partial proliferation.

53. Which of the following statements is true:
A. Cystic fibrosis and hemophilia are unlikely candidates for gene therapy;
B. Cataracts can be a feature of NF1 but not NF2;
C. Scoliosis can be a feature of both NF1 and Marfan syndrome;
D. Neurofibromatosis is characterized with variable expressivity.

54. In cancer, loss of heterozygosity among tumor cells suggests that the region 'lost' may contain which gene type?
A. Acute renal failure; B. A receptor for a growth factor; C. A tumor suppressor; D. An oncogene.

55. Sarah has sickle cell anemia, caused by a single nucleotide change in the beta-globin chain of hemoglobin. She has splenic infarctions, limb pain, early renal failure, splenomegaly, weakness and lassitude. The effect of this single nucleotide change on multiple organ systems is an example of:
A. Pleiotropy; B. Penetrance; C. Locus heterogeneity; D. Anticipation.

56. In retinoblastoma, the "second-hit" mutation is:
A. Always inherited; B. Always somatic; C. Sometimes somatic; D. An activating mutation.

57. Methods currently used to treat genetic disease include:
A. Germ-cell gene therapy; B. Stem-cell transplantation;
C. Nock-out of mutant gene; D. In situ repair of mutations by cellular DNA repair mechanism.

58. Gene therapy may **NOT** be delivered by:
A. Liposomes; B. Adeno-associated viruses;
C. Antisensc oligonucleotides; D. Injection of plasmid DNA.