Genetics - Past Exam & MNU1 Questions

Organization: Arranged by Lecture Topics, and chronologically by Batch (57 → 58 → 59 → 60 → 61 → MNU1 Questions) within each topic.

1. Down Syndrome

1. All of these disorders are characterized by hypotonia except: (Revision 57)
A. Patau syndrome
B. None of the above
C. Down syndrome
D. Edward syndrome
E. Spinal muscle atrophy
Answer: D

2. Type of leukemia associated with Down syndrome: (End Round 1 - 57, End Round 1 - 58)
A. Acute myeloid leukemia (M7)
Answer: A (Acute myeloid leukemia - M7)

3. Most common congenital disorder associated with Down syndrome: (End Round 1 - 57, End Round 1 - 58)
A. Congenital heart disease (50%)
Answer: A (Congenital heart disease - 50%)

4. Disorder accompanied with polydactyly: (End Round 1 - 57)
A. Trisomy 13 (Patau syndrome)
Answer: A (Trisomy 13 / Patau syndrome)

5. Incidence of Down syndrome with mother age older than 49: (End Round 2 - 57)
A. 1/200
B. 1/300
C. 1/150
D. 1/100
E. 1/50
Answer: E
Note: According to the lecture (Page 7), the highest listed maternal age risk is at 45 years (1/20), while at 40 years it is 1/100; 1/50 was selected here as the highest incidence among the given exam options.

6. In translocated Down syndrome, number of chromosomes is: (End Round 2 - 57)
A. 45 chromosomes
B. 46 chromosomes
C. 47 chromosomes
Answer: B

7. Most common cardiovascular disease associated with Down syndrome: (End Round 2 - 57)
A. AVCD (AVSD)
B. ASD
C. VSD
D. Tetralogy of Fallot
Answer: A

8. Karyotyping of a child with Down syndrome may be one of the following: (End Round 3 - 57)
A. 47, XX, +18
B. 47, XX, +21
C. 46, XX / 47, XX
D. 46, XX
E. 45, XY
Answer: B

9. A female newborn with Down syndrome presents with bilious vomiting shortly after birth. Her abdomen is slightly distended. The most likely diagnosis is: (End Round 3 - 57)
A. Hirschsprung disease.
B. Duodenal atresia.
C. Meconium ileus.
D. Midgut volvulus.
E. Pyloric stenosis.
Answer: B

10. Recurrence risk in t(21q;21q): (End Round 1 - 58)
A. 100%
Answer: A (100%)

11. Commonest type of Down syndrome? (End Round 2 - 58)
A. Nondisjunction
Answer: A (Nondisjunction)

12. Number of chromosomes in translocation type of Down syndrome? (End Round 2 - 58)
A. 46 chromosomes
Answer: A (46 chromosomes)

13. Percent of cardiovascular anomalies in Down syndrome? (End Round 2 - 58)
A. 50%
Answer: A (50%)

14. Down syndrome: (End Round 1 - 59)
A. Trisomy 21
Answer: A (Trisomy 21)

15. Best prenatal diagnostic test in first trimester: **(End Round 1 - 59)**
A. Chorionic villus sampling
Answer: A (Chorionic villus sampling)

16. Most common congenital heart disease in Down syndrome: **(End Round 1 - 59)**
A. AVSD
Answer: A (AVSD)

17. What is the incidence of congenital heart disease in Down syndrome? **(End Round 2 - 59)**
A. 40%
B. 60%
C. 30%
D. 70%
E. 50%
Answer: E

18. You are evaluating the karyotype of an infant with translocation Down syndrome. What is the expected number of chromosomes in this case? **(End Round 2 - 59)**
A. 46 chromosomes
B. 47 chromosomes
C. 45 chromosomes
D. 43 chromosomes
E. 44 chromosomes
Answer: A

19. What is the commonest genotype in Down syndrome? **(End Round 2 - 59)**
A. Reciprocal translocation
B. Mosaic
C. Non-disjunction
D. Robertsonian translocation
E. Isochromosome
Answer: C

20. What is the most common endocrinal abnormality in Down syndrome? **(Midterm 60, Midterm 61)**
A. Hypopituitarism.
B. Cushing syndrome.
C. Diabetes mellitus.
D. Hypothyroidism.
E. Hyperparathyroidism.
Answer: D (Hypothyroidism)

21. The most common congenital heart disease in Down syndrome is? (Mid 60, Mid 61, Midterm MNU1)
A. TOF
B. PDA
C. AVSD
D. ASD
E. VSD
Answer: C

22. Down syndrome is? (Midterm 60, Midterm 61, Midterm MNU1)
A. Trisomy 22
B. Monosomy X
C. Trisomy 13
D. Trisomy 21
E. Trisomy 18
Answer: D

23. Prenatal diagnosis of Down syndrome in the first trimester is done by? (Midterm 60, Midterm 61, Midterm MNU1)
A. Fetoscope
B. Quadruple test
C. Triple test
D. Chorionic villi sampling
E. Amniocentesis
Answer: D

24. What is the commonest type of leukemia occurring in a child with Down syndrome? (Midterm 60)
A. Acute lymphoblastic leukemia
B. Acute myeloid leukemia, M7
C. Acute myeloid leukemia, M6
D. Acute myeloid leukemia, M5
E. Chronic myeloid leukemia
Answer: B

25. What is the risk of Down syndrome in the next pregnancy, if one of the parents is carrier for translocation t(21q;15q)? (Midterm 61)
A. 23%
B. 33%
C. 60%
D. 43%
E. 50%
Answer: B

26. First-trimester ultrasound screening finding in Down syndrome: (Midterm MNU1)
A. Increased nuchal translucency (transnuchal thickness)
Answer: A (Increased nuchal translucency / transnuchal thickness)

2. Modes of Inheritance

27. Criteria of X-linked recessive inheritance includes all of the following except: (Revision 57)
A. Frequently affect males
B. No male-to-male transmission
C. None of the above
D. From a carrier mother to her son
E. Frequently affect females
Answer: E

28. Autosomal recessive inheritance with heterozygous carrier parents, character of offspring: (End Round 1 - 57, End Round 1 - 58)
A. 25% chance to be affected
Answer: A (25% chance to be affected)

29. Which of the following is considered Autosomal Dominant: (End Round 1 - 57)
A. Achondroplasia
Answer: A (Achondroplasia)

30. Which of the following statements is TRUE regarding autosomal dominant inheritance? (End Round 2 - 57, End Round 2 - 59)
A. The dominant gene shows its effect only in the homozygous state.
B. Each affected individual has an affected parent up to the generation where mutation occurs.
C. Unaffected relatives of affected persons will have affected offspring.
D. Each offspring of one affected parent and one unaffected parent has 25% chance of being affected.
E. Parental consanguinity is usually present.
Answer: B (Each affected individual has an affected parent up to the generation where mutation occurs)

31. X-linked recessive disorder: (End Round 2 - 57)
A. Marfan syndrome
B. Beta thalassemia
C. Duchenne muscular dystrophy
D. Achondroplasia
Answer: C

32. An example of autosomal dominant inheritance is: (End Round 3 - 57)
A. Neurofibromatosis
Answer: A (Neurofibromatosis)

33. Autosomal recessive disease? (End Round 2 - 58)
A. Thalassemia
Answer: A (Thalassemia)

34. In autosomal dominant inheritance? (End Round 2 - 58)
A. Affects all generations
Answer: A (Affects all generations)

35. Autosomal recessive diseases: (End Round 1 - 59)
A. Males & females affected equally
Answer: A (Males & females affected equally)

36. Variable expressivity: (End Round 1 - 59)
A. Striking variation in clinical features
Answer: A (Striking variation in clinical features)

37. Which of the following hereditary disorders is an example of autosomal recessive mode of inheritance? (End Round 2 - 59, Midterm 60)
A. Achondroplasia
B. Duchenne muscular dystrophy
C. Marfan syndrome
D. Beta thalassemia
E. Hereditary spherocytosis
Answer: D (Beta thalassemia)

38. Variable expressivity is best described by? (Midterm 60, Midterm 61, Midterm MNU1)
A. A single gene (autosomal recessive) may give rise to two or more apparently unrelated effects.
B. A single gene (autosomal dominant) may give rise to two or more apparently unrelated effects.
C. Striking variation in the clinical features of the disorder from person to person, even in the same family.
D. The frequency with which phenotypic manifestations of a gene are expressed.
E. De novo occurrence of the disease within the family.
Answer: C (Striking variation in the clinical features of the disorder from person to person, even in the same family)

39. Which of the following hereditary disorders is an example of autosomal dominant mode of inheritance? (Midterm 60, Midterm 61, Final MNU1)
A. Achondroplasia.
B. Hypophosphatemic rickets.
C. Beta thalassemia.
D. Lignac syndrome.
E. Duchenne muscular dystrophy.
Answer: A (Achondroplasia)

40. Autosomal recessive inheritance is characterized by? (Midterm 61, Midterm MNU1)
A. The disease is transmitted from one generation to the next generation
B. The offspring of two heterozygotes have a chance of 50% to be affected
C. It manifests in the heterozygous state
D. Females and Males are equally affected
E. Hemophilia A is a classic example of this mode of inheritance
Answer: D

3. Prevention of Genetic Diseases (Genetic Counseling & Prenatal Diagnosis)

41. Genetic counseling is the initial step in prevention of genetic disease aiming at: (Revision 57)

A. All of the previous
B. Prevention of recurrence.
C. Construction of pedigree.
D. None of the above
E. Diagnosis of genetic basis

Answer: A

42. When genetic counseling is NOT indicated: (End Round 1 - 57)

A. In non-consanguinity

Answer: A (In non-consanguinity)

43. The most important step in any genetic counseling is: (End Round 2 - 57)

A. Risk assessment.
B. Establishing a diagnosis.
C. Understanding the options.
D. Choosing a course of action.
E. Long-term contact and support.

Answer: B

44. Indication of Genetic counseling: (End Round 1 - 58)

A. Exposure to teratogens

Answer: A (Exposure to teratogens)

45. Which of the following indicates prenatal diagnosis and genetic counselling? (End Round 2 - 58)

A. Advanced maternal age

Answer: A (Advanced maternal age)

46. Which of the following is an indication for genetic counselling? (End Round 2 - 59)

A. A woman with previous baby with congenital toxoplasmosis
B. Non-consanguineous marriage
C. A woman with previous baby with birth injury
D. A woman who had spontaneous abortion in her first pregnancy
E. Advanced maternal age

Answer: E

47. Which of the following is an indication for genetic counselling? (Midterm 60, Midterm 61)
A. Non-consanguineous marriage
B. A woman who had spontaneous abortion in her first pregnancy
C. A child with mental retardation
D. A woman with previous baby with birth injury
E. A woman with previous baby with congenital toxoplasmosis
Answer: C (A child with mental retardation)

48. Definition of genetic counseling (Midterm MNU1)
Recall topic only - no answer provided in source

49. Indication of genetic counseling includes all of the following EXCEPT: (Midterm MNU1)
A. Child with infection
Answer: A (Child with infection)

4. Newborn Screening (General Principles, PKU & Congenital Hypothyroidism)

50. Newborn screening as a national program is applied in Egypt for: (Revision 57)
A. All inborn errors of metabolism.
B. Four diseases.
C. Two diseases.
D. Seven diseases.
E. Ten diseases.
Answer: C

51. Time of Newborn screening in Egypt: (End Round 1 - 57, End Round 1 - 58)
A. 3-7 days after delivery
Answer: A (3-7 days after delivery)

52. Phenylketonuria is considered as: (End Round 1 - 57)
A. Aminoacidopathy
Answer: A (Aminoacidopathy)

53. Heel prick test TSH = 5 uIU/ml, diagnosis and decision: (End Round 1 - 57, End Round 1 - 58)
A. Normal baby & reassure parents
Answer: A (Normal baby & reassure parents)

54. The Guthrie test is a diagnostic tool to test infants for: (End Round 2 - 57)
A. Maple syrup urine disease.
B. Homocystinuria.
C. Galactosemia.
D. Phenylketonuria.
E. Biotinidase deficiency.
Answer: D

55. Phenylketonuria is due to: (End Round 2 - 57)
A. Structural chromosomal defect.
B. Numerical chromosomal defect.
C. Aminoacidopathy.
D. Disorder of carbohydrate metabolism.
E. Disorder of lipid metabolism.
Answer: C

56. Screening of congenital hypothyroidism: (End Round 2 - 57)
A. It is done in the first day after birth.
B. Hypothyroidism is diagnosed when serum T4 is more than 7 ug/dL.
C. Hypothyroidism is diagnosed when serum T3 is less than 300 ng/dL.
D. Hypothyroidism is diagnosed when serum TSH is more than 40 uIU/ml.
E. It is done by taking 2 ml of blood in test tube.
Answer: D

57. In neonatal screening TSH was 30 uIU/ml, what should be done: (End Round 3 - 57)
A. This test result is suspicious for diagnosis of hypothyroidism and the serum TSH should be repeated after 7 days
Answer: A

58. Screening by blood in newborn: (End Round 1 - 58)
A. Phenylketonuria
Answer: A (Phenylketonuria)

59. Most common treatable cause of mental retardation: (End Round 1 - 58)
A. Hypothyroidism
Answer: A (Hypothyroidism)

60. Cause of phenylketonuria? (End Round 2 - 58, End Round 1 - 59)
A. Phenylalanine hydroxylase deficiency
Answer: A (Phenylalanine hydroxylase deficiency)

61. If TSH is above 50 uIU/ml on newborn screening? (End Round 2 - 58)
A. Confirmation of hypothyroidism by serum T3 and T4
Answer: A (Confirmation of hypothyroidism by serum T3 and T4)

62. Newborn screening method in Egypt (for hypothyroidism)? (End Round 2 - 58, End Round 1 - 59, Midterm MNU1)
A. Heel prick
Answer: A (Heel prick)

63. Which of the newborn metabolic screening tests is done at 3-5 days of age: (Final 58)
A. Phenylketonuria
Answer: A (Phenylketonuria)

64. TSH = 25 uIU/ml on newborn screening: (End Round 1 - 59)
A. Suspicious and re-evaluate TSH
Answer: A (Suspicious and re-evaluate TSH)

65. How is newborn screening performed in Egypt? (End Round 2 - 59, Midterm 60, Midterm 61)
A. Urine sample
B. Arterial blood sample
C. Heel prick test
D. Venous blood sample
E. Cord blood sample
Answer: C (Heel prick test)

66. In classic Phenylketonuria, which enzyme is defective? (End Round 2 - 59, Midterm 60, Midterm 61, Midterm MNU1)
A. Dihydrobiopterin synthetase
B. Phenylalanine synthetase
C. Phenylalanine hydroxylase
D. Dihydrobiopterin reductase
E. Tyrosine reductase
Answer: C (Phenylalanine hydroxylase)

67. A newborn had a serum TSH concentration of 50 uIU/ml during newborn screening, what is the most appropriate course of action? (End Round 2 - 59)
A. This infant has hypothyroidism and a confirmatory test by assessment of serum T3, T4 should be done
B. The test result is normal, parents should be reassured
C. This test result is suspicious for diagnosis of hypothyroidism and the serum TSH should be reevaluated after 30 days
D. This test result is suspicious for diagnosis of hypothyroidism and the serum TSH should be reevaluated after 60 days
E. This infant has hypothyroidism and no further tests are required
Answer: A

68. At what age should the blood sample for phenylketonuria (PKU) screening be obtained? (Midterm 60, Midterm 61)
A. 12 hours after birth.
B. Immediately after birth.
C. 2 weeks after birth.
D. 3-7 days after birth.
E. 2 hours after birth.
Answer: D (3-7 days after birth)

69. A newborn had a serum TSH concentration of 25 uIU/mL during newborn screening. What is the most appropriate course of action? (Midterm 61, Midterm MNU1)

A. This test result is suspicious for diagnosis of hypothyroidism and the serum TSH should be reevaluated
B. The test result is normal, parents should be reassured
C. This infant has hypothyroidism and no further tests are required
D. The test is suspicious for diagnosis of hypothyroidism and thyroid ultrasound should be done immediately
E. This infant has hypothyroidism and a confirmatory test by assessment of serum T3, T4 should be done

Answer: A

70. Congenital hypothyroidism is diagnosed if serum TSH level on neonatal screening is? (Final 61)

A. > 20 uIU/ml
B. > 30 uIU/ml
C. > 40 uIU/ml
D. > 25 uIU/ml
E. > 35 uIU/ml

Answer: C

5. Teratogenicity

71. Dose of Vitamin A causing teratogenic defect: (End Round 1 - 57)

A. > 15,000 IU/day

Answer: A (> 15,000 IU/day)

72. Mother with SLE, child is affected by: (End Round 1 - 57)

A. Congenital heart block

Answer: A (Congenital heart block)

73. The following statement is correct as regard teratogenic agents causing neural tube defect: (End Round 2 - 57)

A. Anticonvulsant, carbamazepine and trimethoprim.
B. Sodium valproate, trimethoprim and maternal hyperthermia.
C. Maternal hyperthermia, tetracycline and HIV.
D. Tobacco, phenobarbitone and trimethoprim.
E. Maternal diabetes, maternal hyperthermia and sodium valproate.

Answer: B & E

74. Regarding FDA drug risk classification, antiepileptics are considered category: (End Round 2 - 57)

A. Category A
B. Category B
C. Category C
D. Category D
E. Category X

Answer: D

75. Mother got pregnant while taking oral contraceptive (combined progesterone and estrogen), what are possible teratogenic effect(s): (End Round 2 - 57)
A. 2 to 4 fold increase in VATER association
B. Sirenomelia
C. Neural tube defect
D. 2 to 4 fold increase in VACTER association
E. Intracranial calcification
Answer: A

76. Teratogenic factors are responsible for: (End Round 3 - 57)
A. 3% of congenital anomalies.
B. 10% of congenital anomalies.
C. 13% of congenital anomalies.
D. 15% of congenital anomalies.
E. 20% of congenital anomalies.
Answer: B

77. Teratogenic agent(s) that can cause VATER association: (End Round 3 - 57)
A. Carbamazepine.
B. Maternal diabetes, tetracycline and HIV.
C. Maternal diabetes and oral contraceptive.
D. Oral contraceptive, trimethoprim and maternal hyperthermia.
E. Trimethoprim and maternal diabetes.
Answer: C

78. Alcohol teratogenic effect: (End Round 1 - 58)
A. Hypoplastic (smooth) philtrum
Answer: A (Hypoplastic philtrum)

79. Tetracycline teratogenic effect: (End Round 1 - 58)
A. Enamel dysplasia
Answer: A (Enamel dysplasia)

80. Cytomegalovirus congenital infection: (End Round 1 - 58)
A. Periventricular calcification
Answer: A (Periventricular calcification)

81. Warfarin causes which anomaly? (End Round 2 - 58)
A. Stippled epiphyses
Answer: A (Stippled epiphyses)

82. Congenital rubella syndrome? (End Round 2 - 58)
A. Cataract
Answer: A (Cataract)

83. Ebstein anomaly is caused by? (End Round 2 - 58)
A. Lithium
Answer: A (Lithium)

84. Birth defects attributed to teratogen: (End Round 1 - 59)
A. 10%
Answer: A (10%)

85. Valproic acid teratogenic effect: (End Round 1 - 59)
A. Neural tube defect
Answer: A (Neural tube defect)

86. Congenital toxoplasmosis: (End Round 1 - 59, Midterm MNU1)
A. Chorioretinitis
Answer: A (Chorioretinitis)

87. Which of the following is a teratogenic effect of warfarin? (End Round 2 - 59, Midterm 60, Midterm 61)
A. Neural tube defect
B. Renal failure
C. Micrognathia
D. Teeth enamel dysplasia
E. Stippling of the epiphysis of lumbosacral area
Answer: E (Stippling of the epiphysis of lumbosacral area)

88. Which of the following teratogenic agents is associated with Ebstein's anomaly? (End Round 2 - 59)
A. Lithium
B. Valproate
C. Vitamin A
D. Trimethoprim
E. Lead
Answer: A

89. Which of the following is characteristic for congenital rubella syndrome? (End Round 2 - 59, Midterm 60, Midterm 61)
A. Cataract
B. VSD
C. Periventricular cerebral calcifications
D. Corneal opacities
E. Obstructive hydrocephalus
Answer: A (Cataract)

90. What is the percentage of birth defects attributed to teratogens? (Midterm 60, Midterm 61, Midterm MNU1)

A. 20%
B. 5%
C. 15%
D. 25%
E. 10%

Answer: E (10%)

91. A 25 years old woman with epilepsy, receiving valproate therapy. She is planning to get married and asked your advice regarding the possible teratogenic effects of this drug. Which of the following is a teratogenic effect of valproate? (Midterm 60, Midterm 61, Midterm MNU1)

A. Obstructive hydrocephalus
B. Neural tube defects
C. Tooth enamel dysplasia
D. Optic atrophy
E. VATER association

Answer: B (Neural tube defects).

92. A 26 years old female, attending our antenatal outpatient's clinic, was found positive for toxoplasmosis, during the 13th week of pregnancy. Which of the following is a feature of congenital toxoplasmosis? (Midterm 60, Midterm 61)

A. Intracranial calcifications
B. Microcephaly
C. Corneal opacities
D. Cleft lip/palate
E. Limb abnormalities

Answer: A (Intracranial calcifications)

93. Which of the following is a teratogenic effect of tetracycline? (Midterm 60, Midterm 61)

A. Ear deformities
B. Congenital heart disease
C. Renal failure
D. Neural tube defect
E. Teeth enamel dysplasia

Answer: E

94. Which of the following is characteristic for congenital cytomegalovirus infection? (Midterm 60)

A. Pulmonary stenosis
B. Cataract
C. Corneal opacities
D. Periventricular cerebral calcifications
E. Obstructive hydrocephalus

Answer: D

6. Overlap Questions

95. XXY is a karyotyping of: (End Round 2 - 57)
A. Klinefelter syndrome
Answer: A (Klinefelter syndrome)
Note: Not in the current Genetics lecture curriculum.

96. Clenched fists is found in: (End Round 3 - 57)
A. Edward syndrome
B. Patau syndrome
C. Down syndrome
D. Klinefelter
E. Turner
Answer: A
Note: Not in the current Genetics lecture curriculum. Edward's Syndrome is Trisomy 18

97. Autosomal Dominant disorder: (End Round 1 - 58)
A. Hereditary spherocytosis
Answer: A (Hereditary spherocytosis)
Note: Tests Autosomal Dominant inheritance, though Hereditary Spherocytosis is not listed among the AD examples in the current lecture curriculum.

98. A 24-months girl came to outpatient clinic with her parents because of short stature. X-ray of her hands revealed metacarpal bone shortening. Which of the following is the most likely diagnosis? (End Round 2 - 58, End Round 2 - 59, Final MNU1)
A. Celiac disease
B. Turner syndrome
C. Hypothyroidism
D. Familial short stature
E. Growth hormone deficiency
Answer: B (Turner syndrome)
Note: Not in the current Genetics lecture curriculum (covered under Short Stature in Endocrinology). Appeared in End Round 2 - 58 and Final MNU1, and as a full clinical case in End Round 2 - 59.