# GENETICS

1. **A 35 year old man presents with an inherited neurological disorder. His father developed the disease in his 60s and his daughter was born 2 years ago with a severe form of the condition. His mother, sister, wife and other child, a son, are unaffected. What is the mode of inheritance?**
   a. autosomal inheritance
   b. Mitochondrial inheritance
   c. Polygenic inheritance
   d. Trinucleotide repeat disease
   e. Xlinked inheritance

2. **Which of the following conditions may be detectable by growth monitoring?**
   a. Hyperthyroidism
   b. Hypothyroidism
   c. Pseudohypoparathyroidism
   d. XYY Syndrome
   e. Insulin dependent diabetes mellitus

3. **You are asked advice by a young professional couple, Mr and Mrs X. Mrs X is 9 weeks pregnant. Mr X's brother and his partner had a child with cystic fibrosis. As a result, Mr X was screened and found to carry the DF508 mutation for cystic fibrosis. Mrs X declines to be tested. What are the chances of Mr and Mrs X's child having cystic fibrosis, given that the gene frequency for this mutation in the general population is 1/20.**
   a. 1/4
   b. 1/20
   c. 1/40
   d. 1/80
   e. 1/160

4. **In meiosis which of the following is true?**
   a. DNA replication occurs during meiosis 1.
   b. At the beginning of meiosis 2, each cell contains 23 single chromosomes.
   c. Anaphase lag results in one of the 2 daughter cells receiving an extra part of one chromosome.
   d. Nondisjunction at mitosis (meisois 2) results in mosaicism.
   e. The incidence of Down's Syndrome due to translocation increases with increasing maternal age.

5. **Which ONE of the following have their own self replicating DNA?**
   a. Golgi body
   b. Lysosomes
   c. mitochondria
   d. Peroxisome
   e. Rough Endoplasmic Reticulum

6. **A 35 year old male presents with oral and genital mucocutaneous ulcerations associated with polyarthritis affecting the lower limbs. He is currently on warfarin for an recent episode of pulmonary embolism. Which of the genetic association is most commonly associated with his condition:**
   a. HLA A3
   b. HLA B5
   c. HLA B27
   d. HLA DR3
   e. HLA DR2

7. **A 62 year old man has experienced substernal chest pain upon exertion with increasing frequency over the past 1 year. An electrocardiogram shows T wave inversion in the anterolateral leads at rest. He has a total serum cholesterol of 7.0 mmol/l. On angiography, he has an 85% narrowing of the left anterior descending artery. Which of the following events is most likely to occur in this patient?**
   a. A systemic artery embolus from thrombosis in a peripheral vein.
   b. A systemic artery embolus from a left atrial mural thrombus.
   c. Pulmonary embolism from a left ventricular mural thrombus
   d. A systemic artery embolus from a left ventricular mural thrombus.
   e. Pulmonary embolism from thrombosis in a peripheral vein.

8. **With respect to lipoprotein transport and metabolism in the body, the following statements are correct EXCEPT:**
   a. Chylomicrons are synthesized in the liver.
   b. HDL is assembled in the extracellular space.
   c. Arterial walls contain cells with LDL receptors.
   d. VLDL transformation to LDL occurs in adipose tissue.
   e. Cholesterol is required for the formation of red blood cell membranes.

9. **Which of the following is a feature of hereditary haemorrhagic telangiectasia?**
   a. a good response to oestrogen therapy
   b. cerebral arteriovenous malformations
   c. GI haemorrhage as the usual presenting feature
   d. telangiectasia of the mucous membranes, but not the skin
   e. tendency of lesions to become less obvious with age

10. **Genetic anticipation occurs characteristically in all the conditions except**
   a. myotonia dystrophica
   b. spinocerebellar ataxia type 1
   c. Marfan's syndrome
   d. Huntingdon's disease
   e. Fragile X syndrome

11. **Transcription RNA (tRNA) has three bases specific for a particular amino acid with which it binds tomessenger RNA (mRNA). This specific area of tRNA is called the**
   a. anticodon
   b. codon
   c. exon
   d. intron
   e. transposon

12. **A 44 year old man has had no major medical problems throughout his life, except for arthritis pain involving all extremities for the past couple of years. He has had worsening orthopnoea and ankle oedema in the past six months. He is afebrile. There is no chest pain. A chest Xray shows cardiomegaly with both enlarged left and right heart borders, along with pulmonary oedema. Laboratory test findings include sodium 139 mmol/L, potassium 4.3 mmol/L, urea 7 mmol/L creatinine 95 μmol/L, and glucose 8.6 mmol/L. Which of the following additional laboratory test findings is he most likely to have?**
   a. Anticentromere antibody titer of 1:320
   b. Erythrocyte sedimentation rate of 79 mm/Hr

c. Haemoglobin of 10.7 g/dL with MCV of 72 fL
d. Serum ferritin of 3400 pmol/L
e. Spherocytes in his peripheral blood smear

**13. Which ONE of the following is associated with Marfan's syndrome?**
a. Autosomal recessive inheritance
b. increased upper : lower body ratio
c. Mental retardation
d. Pulmonary stenosis
e. Retinal detachment

**14. Autosomal recessive disorders include**
a. Achondroplasia
b. Congenital Adrenal Hyperplasia
c. Familial hypercholesterolaemia
d. Hereditary Haemorrhagic Telangiectasia
e. Huntington's disease

**15. In Down syndrome, which is the commonest congenital heart defect?**
a. Atrial septal defect
b. Atrioventricular septal defect
c. Patent ductus arteriosus
d. Tetralogy of Fallot
e. Ventricular septal defect

**16. Lipoprotein lipase deficiency is associated with:**
a. Abetalipoproteinaemia
b. Combined hyperlipidaemia
c. Familial combined hyperlipidaemia
d. Familial Hypercholesterolaemia
e. Marked Hypertriglyceridaemia

**17. Concerning Neurofibromatosis Type 1 (NF1), which one of the following statements is true?**
a. Bilateral acoustic neuromas are common
b. Clinical severity in individuals is similar in a given family
c. New mutations occur rarely
d. Pigmented spots on the iris are a characteristic feature
e. The diagnosis is likely if two caféaulait patches are present

**18. The following are features of pseudohypoparathyroidism:**
a. Increased urinary phosphate and cAMP with PTH infusion
b. Low serum PTH
c. Low serum calcium and low serum phosphate
d. Low serum calcium and high serum phosphate
e. Shortened 2nd and 3rd metacarpals

**19. Reverse transcriptasePCR is used to ampify:**
a. Antibodies
b. DNA
c. RNA
d. Protein
e. Plasmids

**20. Which of the following is a characteristic feature of familial hypercholesterolaemia?**
a. Autosomal dominant inheritance
b. elevated chylomicrons
c. hypertriglyceridaemia
d. increased expression of LDL receptors
e. Palmar xanthomas

**21. The parents of a child with cystic fibrosis consult you wishing to know what is the risk of their next child being a carrier of the condition. Which ONE of the following percentages is the correct risk?**
a. 0%
b. 25%
c. 50%
d. 75%
e. 100%

**22. Which of the following is NOT true regarding the polymerase chain reaction:**
a. It is used to amplify DNA but not RNA
b. The amount of DNA required makes it unsuitable for early prenatal diagnosis
c. Synthetic short DNA primers which flank the sequence of interest are required to initiate the amplification
d. It utilizes the thermostable properties of Taq DNA polymerase
e. It can be used to detect the presence of viral DNA in human disease

**23. The Polymerase Chain Reaction (PCR) is used to amplify small amounts of DNA for further analysis. First the DNA double helix must be split into two strands. This is achieved by**
a. alkali solution
b. centrifugation
c. DNA polymerase
d. heating to nearly 100°C
e. viral reverse transcriptase

**24. Protein synthesis occurs within cells. A particular molecule that is produced in the nucleus initiates protein synthesis. This molecule matures in the cytoplasm and binds to the ribosome. This molecule is**
a. messenger RNA
b. ribosomal RNA
c. RNA nucleotide
d. RNA polymerase
e. transfer RNA

**25. A 17yearold girl is short in stature for her age. She has not shown any changes of puberty. She has a webbed neck. Her vital signs include Temperature 36.6°C Respiratory rate 18/min Pulse 75 bpm and BP 165/85 mmHg. On physical examination, she has a continuous murmur heard over both the front of the chest as well as her back. Her lower extremities are cool with poor capillary filling. A chest radiograph reveals a prominent left heart border, no oedema or effusions, and rib notching. Which of the following pathologic lesions best explains these findings?**
a. Constriction of the aorta past the ductus arteriosus
b. Lack of development of the spiral septum and partial absence of conus musculature
c. Shortening and thickening of chordae tendineae of the mitral valve
d. Single large atrioventricular valve
e. Supravalvular narrowing in the aortic root

**26. Autosomal dominant conditions include**
a. Betathalassaemia
b. Cystic fibrosis
c. Marfan syndrome
d. Wilson's disease
e. Xeroderma Pigmentosa

**27. A 19 year old woman is found to have a cardiac murmur characterized by a midsystolic click. An echocardiogram reveals mitral insufficiency with upward displacement of one leaflet. There is also aortic root dilation to 4 cm. She has a dislocated right ocular crystalline lens. She dies suddenly and unexpectedly. The medical examiner finds a prolapsed mitral valve with elongation, thinning, and rupture of chordae tendineae. A mutation involving which of the following genes is most likely have be present in this patient?**

a. Betamyosin
b. CFTR
c. FGFR
d. Fibrillin
e. Spectrin

**28. A Plasmid is best described as**

a. a recombinant section of DNA
b. a small viral particle
c. bacterial DNA separate from the chromosome
d. consist of multiple copies of a single gene
e. having multiple origins of replication

**29. A 59 year old woman has had insulin dependent diabetes mellitus for over two decades. The degree of control of her disease is characterized by the laboratory finding of a HbA1c of 10.1%. She complains of repeated episodes of abdominal pain following meals. These episodes have become more frequent and last for longer periods over the last couple of months. On physical examination, there are no abdominal masses andno organomegaly of the abdomen, and she has no tenderness to palpation. Which of the following findings is most likely to be present:**

a. Ruptured aortic aneurysm
b. Hepatic infarction
c. Mesenteric artery occlusion
d. Acute pancreatitis
e. Chronic renal failure

**30. A 51yearold healthy man is found to have bilateral breast enlargement. He says that this is normal for him and that he has not noted any change in years. Which of the following is most likely to be present?**

a. 47, XXY karyotype
b. History of antidepressant drug therapy
c. Increased risk for breast carcinoma
d. Increased testosterone levels
e. Seminoma of the testis

**31. Two strains of Escherichia coli are isolated and both are resistant to ampicillin. Strain A retains its resistance to amplicillin when grown form multiple generations in the absence of ampicillin. However strain B loses its resistance when grown in the absence of ampicillin. Which of the following best explains the loss of antibiotic resistance in strain B?**

a. Changes in the bacterial DNA gyrase
b. Downregulation of the resistance gene
c. Loss of a plasmid containing the resistance gene
d. Mutations in the resistance gene
e. Transposition of another sequence into the resistance gene

**32. Which of the following is true regarding chromosomes?**

a. Down's syndrome is most commonly due to an extra copy of chromosome 21 inherited from the father.
b. A Fetus with triploidy will have 47 chromosomes
c. Heterochromatin is mostly composed of active genes
d. The normal human karyotype consists of 22 pairs of autosomes
e. Telomeres provide the point of attachment to the mitotic spindle

**33. A 36 year old man attends clinic with his wife after failing to conceive after 10 years of marriage. Examination reveals that he his tall, thin and has bilateral gynaecomastia. Investigations show high levels of urinary gonadotrophins. What is the most likely diagnosis?**

a. Andropause
b. Gaucher's disease
c. Kleinfelter's syndrome
d. Marfan syndrome
e. Noonan's syndrome

**34. Which one of the following statements is correct?**

a. adult polycystic renal disease is inherited as an autosomal recessive trait
b. reflux nephropathy is inherited as an autosomal recessive trait
c. nephrogenic diabetes insipidus is inherited as an autosomal dominant trait
d. Alport's syndrome affects females more severely than males
e. medullary sponge kidney is typically not inherited but is a congenital condition.

**35. A 22 year old lady is affected by an inherited disorder. She has two brothers who are unaffected. She has two sisters both are affected. Her father is affected but not her mother. What is the mode of inheritance?**

a. Autosomal Dominant
b. Autosomal Recessive
c. Mitochondrial
d. Xlinked Dominant
e. Xlinked Recessive

**36. A 60 year old Chinese man has been started on quinine for leg cramps by his General Practitioner. He presents, a week later, with 5 days of darkened urine and 2 days of increasing breathlessness, back pain and fatigue. Investigations show a haemoglobin of 7.0 g/dl and raised reticulocyte count. Which of the following best explain this drug reaction?**

a. autoimmune haemolytic anaemia
b. glucose6phosphate dehydrogenase deficiency
c. hereditary spherocytosis
d. pyruvate kinase deficiency
e. sickle cell disease

**37. Benign Essential Tremor:**

a. Is present characteristically at rest
b. Occur with lesion in sub thalamus
c. Occur in liver disease
d. Alchohol improves the tremor
e. Is autosomal recessive in inheritance

**38. In which of the following is mental retardation an expected finding?**

a. Alkaptonuria
b. Cystinuria
c. Glycogen storage disease
d. Lactose intolerance
e. Maple syrup urine disease

**39. In X linked recessive inheritance, which of the following is true?**

a. The male to female ratio is 2:1.
b. Each son of a female carrier has a 1:4 risk of being affected.
c. Each daughter of a female carrier has a 1:4 risk of being a carrier.
d. Daughters of affected males will all be carriers.
e. The family history is often positive since new mutations are rare.

**40. The level of cellular telomerase activity will affect:**

a. The rate of cell growth
b. Cell death
c. The number of cell divisions a cell is capable of undergoing
d. Cell survival
e. RNA synthesis

**41. Which of the following haematological disorders is inherited as an autosomal recessive condition?**

a. Antithrombin III deficiency
b. Protein C deficiency
c. Glucose6phosphate dehydrogenase deficiency
d. Pyruvate kinase deficiency
e. Acute intermittent porphyria

**42. A 20 year old female patient is refered with primary amenorrhoea. Investigations reveal a 46 XY karyotype. Which of the following concerning the condition is true?**

a. It is likely that her mother received stilboestrel in pregnancy
b. It is likely that her mother received Carbimazole for thyrotoxicosis during pregnancy
c. Low testosterone and oestradiol concentrations would be expected
d. The diagnosis is likely to be testicular feminisation syndrome
e. The diagnosis is Noonan's syndrome

**43. Which ONE of the following is a recognised feature of achondroplasia?**

a. Autosomal recessive inheritance
b. May be diagnosed radiologically at birth
c. Increased liability to pathological fractures
d. Shortened spine
e. Subfertility

**44. Restriction enzymes**

a. Cut DNA
b. Join two pieces of DNA together
c. Synthesize DNA
d. Degrade DNA
e. Are involved in cell cycle arrest

**45. The following is true about Cystic Fibrosis:**

a. Is an autosomal dominant condition.
b. Is due to mutation of CFTR gene on chromosome 17
c. Skin test may be positive for aspergillus
d. Median survival rate is 10 to 15 years.
e. Is a cause of mental retardation.

**46. The incidence of Down syndrome in children born to women aged less than 30 years is**

a. 1:600
b. 1:800
c. 1: 1000
d. 1:1200
e. 1:1400

**47. A routine ultrasound at 18 weeks gestation in a diabetic mother reveals a male foetus with an endocardial cushion defect. Other abnormalities include increased nuchal thickening and a "double bubble" sign. Which of the following conditions is most likely to have contributed to this set of findings:**

a. Maternal use of ACE inhibitor
b. Marfan syndrome
c. Maternal folate deficiency
d. Trisomy 21
e. Congenital syphilis

**48. A 40 year old male is diagnosed with Dystrophia myotonica. Which one of the following features would be expected in this patient?**

a. Autosomal recessive inheritance
b. Cataracts
c. Fasiculations would predominate
d. Progressive external ophthalmoplegia
e. Preserved tendon reflexes despite muscle wasting

**49. Which of the following is true regarding chromosomes?**

a. Down's syndrome is most commonly due to an extra copy of chromosome 21 inherited from the father.
b. A Fetus with triploidy will have 47 chromosomes
c. Heterochromatin is mostly composed of active genes
d. The normal human karyotype consists of 22 pairs of autosomes
e. Telomeres provide the point of attachment to the mitotic spindle

**50. A 28 year old lady presents with multiple caféaulait spots. A diagnosis of neurofibromatosis type 1 is made. What is true of the NF1 gene?**

a. Inherited in a recessive fashion
b. Inherited in an Xlinked fashion
c. On chromosome 17
d. On mitochondrial genome
e. Related to NF2 gene

# Genetics - Answers



1. d
2. b
3. d
4. d
5. c
6. b
7. d
8. a: Chylomicrons are formed in the gut from exogenous triacylglycerols and cholesterol. They are released into the lymph and thereby enter the blood.They are not formed in the liver.
9. b: In hereditary haemorrhagic telangiectasia there may also be pulmonary AV malformations.Epistaxis, not GI haemorrhage, is the usual presenting feature. Lesions become more obvious with age and affect mucuous membranes as well as skin. Oestrogen therapy is sometimes advocated but the effect, if any, is small.
10. c: Anticipation means increased severity/earlier age of onset of disease with successive generations. Other conditions with anticipation include spinocerebellar ataxia type 1 and dentatorubral pallidoluysian atrophy.
11. a: mRNA has codons which are bound by the anticodons on tRNA during translation of protein synthesis. Exons are noncoding sequences in the mRNA and introns are areas of unknown function. Transposons are genetic sequences that have been transposed from one part of DNA to another.
12. d
13. e
14. b: All the others are autosomal dominant of course.
15. b: 50% of Down syndrome births have congenital heart disease. Defects in order of decreasing frequency are: B, E, C, D and A.
16. e: Lipoprotein lipase deficiency is autosomal recessive and associated with increased chylomicrons and marked hypertriglyceridaemia.
17. d
18. d
19. c
20. a
21. c: As both parents are carriers for the CF gene then the chances of another child being affected (homozygote) is 1 in 4 (25%). The chances of their child being free from the CF gene is also 1 in 4 (25%) and the chances of a child being a carrier (heterozygote) is 1in 2 (50%).
22. b: RtPCR is used to amplify RNA rather than PCR specifically. Preimplantation diagnosis uses IVF and genetic analysis of 3 day old embryos, before selective transfer of unaffected embryos to uterus.
23. d
24. a
25. a: She has coarctation of the aorta, and the constriction is postductal, allowing prolonged survival. Her physical characteristics also suggest Turner syndrome (monosomy X).
26. c: All the others are autosomal recessive of course.
27. d
28. c
29. c
30. a
31. c
32. d
33. c: Gaucher's and Marfan syndrome do not present with infertility. Noonan's is associated with short stature. Kleinfelter's is a sex chromosome disorder affecting 1:400 1: 600 male births typically with 47 XXY, XXXYY or XXYY. Andropause is the term for the gradual decrease in serum testosterone concentration with age, but does not occur, usually, until after the age of 50.
34. e: PKD is usually autosomal dominant although the infantile form is autosomal recessive. Nephrogenic DI is usually Xlinked. Features of Alport syndrome (hereditary nepritis, haematuria, progressive renal failure and highfrequencynerve deafness)are usually more marked in in males. Neither reflux nephropathynor medullary sponge kidneys are hereditary conditions.
35. d
36. b
37. d: There is no tremor at rest, but a rhythmic oscillation develops when the patient holds the arms outstretched. A positive family history is obtained in over half of such patients and the pattern of inheritance in such families indicates an autosomal dominant trait. Alcohol suppresses essential tremor, but the mechanism responsible is unknown
38. e
39. d
40. 3: The telomere is a DNA sequence at the end of each chromosome which becomes progressively shorter with each division the cell undergoes. When it is reduced to a critical length the cell is not capable of dividing, the enzyme telomerase is able to lengthen the telomere thus preventing this occurring.
41. d
42. d
43. b
44. a: Restriction enzymes cut DNA at sequences specific for each restriction enzyme, they are vital tools for molecular biology and molecular genetic research.
45. c: Cystic fibrosis is an autosomal recessive condition and is due to mutation of CFTR gene on chromosome 7. 20% develop bronchopulmonary aspergillosis. Median survival rate is 25 to 35 years and is currently improving.
46. d: Maternal age also affects incidence of hydrocephalus, anencephaly and achondroplasia.
47. d
48. b
49. d
50. c