

GENETICS

1. A 26 year old man with history of hereditary haemorrhagic telangiectasia is planning to start a family. What is the mode of inheritance?

- A. Autosomal dominant with incomplete penetrance
- B. Autosomal co-dominant
- C. Autosomal recessive with incomplete penetrance
- D. Autosomal dominant
- E. Autosomal recessive

Ossler-Weber ^{inf}
anomaly

D

2. A 16 year old girl with amenorrhoea, normal secondary sexual character, broad spaced breast and webbed neck. What is the single most diagnosis?

- A. Klinefelter's syndrome
- B. Turner's syndrome
- C. Down's syndrome
- D. Hypothyroidism
- E. Tay-sachs disease

B

3. An otherwise healthy 13-year-old boy presents with recurrent episodes of facial and tongue swelling and abdominal pain. His father has had similar episodes. What is the single most likely diagnosis?

- A. C1 esterase deficiency
- B. HIV disease
- C. Mumps
- D. Sarcoidosis
- E. Sjogren's syndrome

(Hereditary angioedema)

A

4. A 59-year-old man has shown a change in his mood and personality over a 9-month period. He has subsequently developed difficulty with memory and concentration, and then progressive fidgety movements of his limbs and facial musculature. By the time of medical assessment he has frank choreiform movements and a Mini-Mental State Examination of 21/30. Other examination is normal. He was adopted and therefore no information on his family history is available. He has three adult children (aged 27, 30 and 33 years) of whom the two youngest are asymptomatic. However, the oldest son has recently been investigated by the Neurology Department for slightly erratic behaviour and fidgety restless movements of both legs. Based on the likely clinical diagnosis, which one of the following genetic patterns of this condition is most likely?

- A. Autosomal-dominant inheritance with anticipation
- B. Autosomal-dominant with variable penetrance
- C. Autosomal-recessive inheritance
- D. X-linked inheritance
- E. Mitochondrial disorder

A

4. This is the measure of depression of a set of values. Choose the **single term** from the list of options given.

- A. Standard deviation (root mean square deviation)
- B. Specificity
- C. Sensitivity
- D. Probability
- E. Prevalence

A

5. This is the likelihood of a test reporting positive when the condition is being tested in actually present. Choose the **single term** from the list of options given.

- A. Standard deviation (root mean square deviation)
- B. Specificity
- C. Sensitivity
- D. Probability
- E. Prevalence

C

6. At the time that a condition is studied this is the number of patients in the whole population with the condition. Choose the **single term** from the list of options given.

- A. Prevalence
- B. Specificity
- C. Sensitivity
- D. Probability
- E. Prevalence

A

Single Best Answer

7. A town has a population of 500,000. In a five year period 100 people presents with cancer Y. During the same period 1250 cases of cancer Y are registered by death certification. The one year survival rate is zero percent. What is the **annual prevalence** of cancer Y per million in this period?

- A. 200
- B. 400
- C. 500
- D. 800
- E. 1250

B

8. The comparison of the occurrence of the putative cause between two groups of subjects one with a disease and the other without.

- A. Case-control study.
- B. Student test.
- C. Period prevalence.
- D. Probability.
- E. Cohort study.

A

9. The incidence of the disease is compared between two groups of subjects, one is exposed to a putative casual factor and the other is not.

- A. Case-control study.
- B. Student test.
- C. Period prevalence.FVH
- D. Probability.
- E. Cohort study.

E

QUESTIONS

5. A 26 year old man with history of hereditary haemorrhagic telangiectasia is planning to start a family. What is the mode of inheritance?

- A. Autosomal dominant with incomplete penetrance
- B. Autosomal codominant
- C. Autosomal recessive with incomplete penetrance
- D. Autosomal dominant
- E. Autosomal recessive

6. You are performing the postnatal check on a 3 day old girl. She has swollen hands and feet, and you find it difficult to palpate her femoral pulses. Select the most likely karyotype.

- A. 45XO
- B. 47XXX
- C. 47XXY
- D. 47XY(13)
- E. 47XY(18)

7. You are asked to see a 1 hour old baby. He is dysmorphic, with a small chin and low-set ears. He also has a small head and rocker-bottom feet. Select the most likely karyotype.

- A. 45XO
- B. 47XXX
- C. 47XXY
- D. 47XY(13)
- E. 47XY(18)

8. A 15 year old boy presents to the general pediatrics clinic. He and his family are concerned as he has not entered puberty yet. He is tall for his age and has small, firm testis. He is a shy boy and has some behavioral problems at school. Select the most likely karyotype.

- A. 45XO
- B. 47XXX
- C. 69XXY
- D. 47XY(21)
- E. Klinefelter syndrome
- F. 47XXY

9. You are asked to see a 1 hour-old baby. He is dysmorphic with a round face. On examination you notice that he is very floppy. Select the most likely karyotype.

- A. 47xy
- B. 47XXX
- C. 47XXY
- D. 47XY(13)
- E. 47XY(18)

10. A 5 year old boy presents to the outpatient clinic. He has learning difficulties, a large head and large testicles. He has quite a characteristic facial appearance, with a prominent forehead and large ears. Select the most likely karyotype.

- A. 47XY(18)
- B. 47XY(13)
- C. 47XY(21)
- D. 69XXY
- E. Fragile X syndrome

11. 8 months old boy with incontinence and has E.coli positive in urine. , select the most likely karyotype.

- A. 45XO
- B. 47XXY
- C. 47XY(18)
- D. 47XY(13)
- E. 47XY(21)

12. A 7 year old boy wets the bed every day. , select the most likely karyotype.

- A. 47XXY
- B. 47XY(18)
- C. 47XY(13)
- D. 47XY(21)
- E. 69XXY

The scientific basis of prenatal medicine

13. Prenatal screening is recommended if ultrasound scan at 16 weeks confirms that the foetus is male and the mother has had an affected son previously. Choose the single most likely condition

- A. Duchene muscular dystrophy
- B. Cystic fibrosis
- C. Spina bifida
- D. Down's syndrome
- E. Spinal muscular atrophy

14. Detailed prenatal ultrasound scan is recommended if maternal serum at 16 weeks show a significantly increased level of alpha fetoprotein. Choose the single most likely condition

- A. Duchene muscular dystrophy
- B. Cystic fibrosis
- C. Spina bifida
- D. Down's syndrome
- E. Spinal muscular atrophy

QUESTIONS

- D. Fetal cardiocentesis
E. Genotyping fetal cell in maternal circulation
20. The mother of a child with Duchene muscular dystrophy plans to become pregnant in the near future. She enquires about the earliest test to identify if the fetus of a future pregnancy would be affected. Choose the single most likely condition.
- A. Amniocentesis
B. Pre-implantation genetic diagram
C. Chorionic villus sampling (CVS)
D. Fetal cardiocentesis
E. Genotyping fetal cell in Maternal circulation
21. A 20 year old woman is 17 weeks pregnant. She is worried about the possibility of her baby with spina bifida as she did not take folic acid supplement before conception and in early pregnancy. Choose the single most likely condition.
- A. Pre-implantation genetic diagram
B. Amniocentesis
C. Chorionic villus sampling (CVS)
D. Fetal cardiocentesis
E. Genotyping fetal cell in Maternal circulation
F. Ultrasound scan at 18-20 week gestation
22. A 29 year old woman reports that her partner's father has recently been diagnosed as Huntington chorea. She wants to know what diagnostic test would be certain to avoid problem in a future child. Choose the single most likely condition.
- A. Amniocentesis
B. Pre-implantation genetic diagram
C. Chorionic villus sampling (CVS)
D. Fetal cardiocentesis
E. Ultrasound scan at 18-20 weeks gestation
F. Ultrasound scan at 12 weeks gestation

- C. Stellate ganglion
D. Deep inguinal ring
E. Transpyloric plane
26. Removal of a glioma. Which is the SINGLE most appropriate anatomical structure involved?
A. Cricoid cartilage
B. Rectus sheath muscle
C. Duramater
D. Conjoined tendon
E. Intercostal muscles
- 27.. Midpoint between the supra-sternal notch and pubic symphysis. What is the Single most appropriate land mark?
A. Fundus of the Gall bladder
B. McBurney's point
C. Stellate ganglion
D. Deep inguinal ring
28. A patient with vesicles in the maxillary division of trigeminal nerve. Which area of mucus membrane will be involved?
A. Palate
B. Cheek
C. Cornea
D. Conjunctiva
17. Inserting a drain in the mid axillary line. What is the SINGLE most appropriate anatomical structure?
a. External iliac muscle
b. Linea Alba
c. Rectus sheath muscle
d. Conjoined tendon
B. Intercostal musc
C. . Transpyloric plane
29. Fifth intercostal space in the anterior axillary line. What is the Single most appropriate land mark?

- A. Apex beat
- B. Chest-drain insertion
- C. Stellate ganglion
- D. Transpyloric plane

E. Vena cava opening in to the diaphragm

30. A 45 year old lady presents with history of double vision and facial numbness. Which anatomical site is most likely to be affected?

- A. Cerebral cortex
- B. Trigeminal Nerve
- C. Oculomotor Nerve
- D. Brain Stem

E. Basal Ganglia

31. The artery that supplies the anterior right ventricular wall. What is the SINGLE most appropriate option?

- A. Acute marginal branch
- B. Left anterior descending artery
- C. Coronary sinus
- D. Circumflex artery
- E. Right coronary artery

- A. Sodium valproate
- B. Diethylstilbestrol DES
- C. Alcohol
- D. Phenytoin
- E. Retinoic acid

33. Hearing defects, absent ears, ocular anomalies, renal anomalies, phocomelia (severe limb malformations). Choose the **single most likely causative factor** from the list of options given

- A. Glucocorticoids
- B. Phenytoin
- C. Thalidomide
- D. Alcohol
- E. Indometacin

34. Craniofacial alterations, cleft palate, neural tube defects, cardiovascular malformations, absent or defective ears, small jaw, kidney anomalies and low IQ. Choose the **single most likely causative factor** from the list of options given

- A. Sodium valproate
- B. Warfarin
- C. Alcohol
- D. Phenytoin
- E. Retinoic acid

35. Premature closure of the ductus arteriosus. choose the **single most likely causative factor** from the list of options given

- A. Glucocorticoids
- B. Phenytoin
- C. Thalidomide
- D. Alcohol
- E. Indometacin

36. Lumbosacral spina bifida with meningocoele, often accompanied by midfacial hypoplasia, deficient orbital ridge, prominent forehead, congenital heart disease and decreased postnatal growth.

- A. Phenytoin
- B. Thalidomide
- C. Tetracycline
- D. Sodium valproate
- E. Retinoic acid

37. Cleft lip, cardiac anomalies and skeletal defects choose the **single most likely causative factor** from the list of options given.

- A. Thalidomide
- B. Alcohol
- C. Indometacin
- D. Norethisterone
- E. Tetracycline

- C. Phenytoin
- D. Nifedipine
- E. Cyclosporin

45. A lady on treatment for epilepsy develops ataxia and gum hypertrophy. What is the most likely causative agent?

- A. Sodium Valproate
- B. Carbamazepine
- C. Phenytoin
- D. Nifedipine
- E. Cyclosporin

46. Severe mucositis is seen with

- A. Radiotherapy
- B. Cyclosporin
- C. Digoxin
- D. Carbamazepine
- E. Nifedipine

47. A boy on treatment for generalized tonic-clonic epilepsy develops features of hepatic failure in few weeks of starting treatment. What is the predisposing factor?

- A. Sodium valproate (wt. gain, reversible hair loss)
- B. Carbamazepine
- C. Phenytoin
- D. Alcohol
- E. Verapamil

48. A 46-year-old lady on long term treatment for psychiatric problem developed polyuria and polydipsia. Osmolality results revealed nephrogenic diabetes insipidus. There was also chronic renal impairment, a raised TSH level and leukocytosis. Which one of the following is the most likely agent which might be responsible?

- | | |
|--------------|----------------|
| A. Clonidine | D. Risperidone |
| B. Clozapine | E. Trazodone |
| C. Lithium. | |

49. A 42 year old patient with a history of schizophrenia for which he is taking antipsychotic agent. He becomes physically unwell and is found to have unstable blood pressure, high body temperature, sweating, raised white blood cell count and raised CPK. Choose the single most likely causative drug.

- A. Lithium
- B. Chlorpromazine
- C. Imipramine
- D. Clozapine
- E. Fluoxetine

Neuroleptic syndrome

50. A 38 year old patient with a history of schizophrenia for which he is taking antipsychotic agent. He presents with extrapyramidal effects, reduced sexual libido and erectile impotence. His prolactin is raised. Choose the single most likely causative drug.

- | | |
|-------------------|---------------|
| A. Lithium | E. Fluoxetine |
| B. Chlorpromazine | |
| C. Imipramine | |
| D. Clozapine | |