GENETICS MRCOG 1 Quizz February 2020

SBA 1
A routine ultrasound at 18 weeks' gestation in a diabetic mother reveals a male fetus 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 Congenital syphilis
B Marfan's syndrome
C Maternal folate deficiency
D Maternal use of ACE inhibitor
E Trisomy 21

SBA 2
In which of the following malignancies are tumour suppressor genes MLH1 and MSH2 affected?

A Familial adenomatous polyposis
B Hereditary non-polyposis colonic carcinoma (HNPCC)
C Retinoblastoma
D Wilms' tumour
E Xeroderma pigmentosa

SBA 3
Which one of the following conditions is inherited in an autosomal dominant manner and is associated with multiple colonic polyps?

A Familial adenomatous polyposis
B Hereditary non-polyposis colonic carcinoma (HNPCC)
C Retinoblastoma
D Wilms' tumour
E Xeroderma pigmentosa

SBA 4
Which of the following statements best describes the action of reverse transcriptase?
A Bacterial enzyme which cleaves foreign DNA at a specific recognition site
B Enzyme producing cDNA
C Enzyme responsible for DNA replication
D Enzyme responsible for DNA transcription
E Enzyme used in the polymerase chain reaction

SBA 5
Regarding the human chromosomes, which of the following statements is correct?
A Barr body is caused by the presence of an inactive X chromosome
B Barr body is only found in people who are phenotypically female
C Cells containing YO chromosome are compatible with life
D The Y chromosome is larger than the X chromosome
E There are 23 pairs of autosomal chromosomes

SBA 6
Which of the following is not a tumour suppressor gene?
A APC
B BRAC1
C c-myc
D p53
E VHL

SBA 7
A 38-year-old woman with a strong familial history of breast cancer has been diagnosed with a breast malignancy.
Which oncogene is associated with the development of breast cancer?
A BCL-2
B BRAC3
C HER-2
D N-MYC
E RET

SBA 8
A 41-year-old man undergoing an elective inguinal hernia repair develops a marked pyrexia intraoperatively.
Malignant hyperthermia is a recognised complication with the use of which drug?
A Dantrolene
B Diclofenac
C Lidocaine
D Propofol
E Suxamethonium

SBA 9
Increased numbers of chromosomes occur in which of the following?
A Fragile-X syndrome
B Jacobsen syndrome
C Klinefelter's syndrome
D Turner's syndrome
E Retinoblastoma

SBA 10

Which of the following conditions is characterised by abnormal chromosome patterns?

A Cleft lip palate
B Congenital renal agenesis
C Duchenne muscular dystrophy
D Haemophilia
E Patau syndrome

SBA 11
In which syndrome would a single Barr body be found?

A Females of genotype XXX
B Klinefelter's syndrome
C Males with Down's syndrome
D Most cases of Turner's syndrome
E Subjects with testicular feminisation syndrome

SBA 12
Which organelle has its own self-replicating DNA?
A Golgi body
B Lysosomes
C Mitochondria
D Nucleolus
E Rough ER

SBA 13
A 46 XY karyotype associated with a female phenotype is found in which of the following?
A A child whose mother has been treated with stilboestrol
B A patient with testicular feminisation syndrome (androgen insensitivity)
C An infant whose mother has thyrotoxicosis
D An infant whose mother is treated with prednisolone
E Noonan's syndrome

SBA-14 -15-16-17-18

A woman presents to the labour ward at approximately 40 weeks' gestation in spontaneous labour. She is unbooked and has received no antenatal care. On abdominal palpation, she feels large of her reported gestational age. A small mal baby is born by emergency caesarean section due to fetal distress. He is found to have microcephaly, a prominent occiput, a cleft lip and palate, clenched hands and polydactyl. Soon after birth, he has apnoeic episodes and has difficulty feeding.

What chromosomal abnormality is the most likely cause of this baby's presentation?

A Microdeletion of chromosome 15
B Microdeletion of chromosome 22
C Trisomy 13
D Trisomy 18
E Trisomy 21

A 32-year-old woman is 15 weeks pregnant in her second pregnancy. She opts to have antenatal screening and has blood taken as part of the quadruple test. The result shows reduced levels of α-fetoprotein and unconjugated oestriol with elevated β-human chorionic gonadotrophin.

Which of the following is the most likely explanation for the screening results?

A Down's syndrome
B Edwards' syndrome
C Multiple pregnancy
D Neural tube defect
E Normal pregnancy

A 37-year-old primiparous woman is 14 weeks pregnant. Following serum screening, the pregnancy is found to have an increased risk of trisomy 21. She wishes to have further testing to confirm whether the fetus is affected.

In view of her current gestation, what is the most appropriate diagnostic test?

A Amniocentesis
B Cell-free fetal DNA sampling
C Chorionic villus sampling
D Cordocentesis
E Nuchal translucency imaging

A 29-year-old woman seeks genetic counselling as she has a number of her female relatives who have had either breast or ovarian cancer. Both her mother and her sister have been diagnosed with breast cancer. DNA sequencing subsequently shows that she carries a mutated form of the BRCA1 gene.

Via which mode of inheritance is the BRCA1 gene mutation transmitted?

A Autosomal dominant inheritance
B Autosomal recessive inheritance
C Mitochondrial inheritance
D X-linked dominant inheritance
E X-linked recessive inheritance

A 34-year-old woman delivers a male baby with Down's syndrome. Chromosomal analysis following his birth is suggestive of familial Down's syndrome.

What chromosomal event best describes the aetiology of familial Down's syndrome?

A Microdeletion
B Nonsense mutation
C Reciprocal translocation
D Robertsonian translocation
E Triplet repeat expansion

Both individuals in a couple are both known to carry the trait for a haemoglobinopathy. They decline any invasive testing when they conceive their first pregnancy. At an anomaly scan at 20 weeks' gestation the fetus is found to have severe hydrops fetalis. In utero death occurs at 22 weeks' gestation.

What is the most likely cause of the fetal demise?

A Alpha-thalassaemia with deletion of 4 α-globin genes
B Beta-thalassaemia major
C Glucose-6-dehydrogenase deficiency
D Haemoglobin H disease
E Sickle cell disease

A 40 year old woman presents at 14 weeks of gestation. She opts to have antenatal screening. Her blood tests show an increase in βhCG and low PAPPA. At her dating scan there is a raised nuchal translucency noted. What is the most likely explanation for these results?
A Down syndrome
B Edward's syndrome
C Normal pregnancy
D Patau's syndrome
E Twin pregnancy

What cells in the spermatogenesis process can undergo mitotic division?
A Primary spermatocytes
B Secondary spermatocytes
C Spermatids
D Spermatogonia
E Spermatozoa

Male infertility in a patient with cystic fibrosis is likely to be due to which condition?
A Testicular failure
B Oligospermia
C Hypothalamic failure
D Congenital absence of vas deference
E Congenital absence of testes

Which error in meiosis is the most common cause of Down syndrome?
A Translocation
B Nondisjunction
C Mosaicism
D Inversions
E Aneuploidy

Which chromosomal abnormality has a known association with severely impaired semen quality?
A Paracentric inversion of the Y chromosome
B Microdeletions of the Y chromosome
C Microdeletions of the X chromosome
D Formation of ring chromosomes
E Formation of isochromosomes of the X chromosome

A couple have presented to a clinic with suspected infertility. Diagnostic tests reveal that the man has azoospermia, testicular atrophy and gynaecomastia. Which karyotype result would be most likely in this case?

A 47 XY+21
B 47 XYY
C 47 XXY
D 46 XY
E 46 XX

A teenager presents to a gynaecology clinic with primary amenorrhoea and delayed puberty. Further investigations reveal a karyotype as 45 XO. What condition is the teenager likely to have?
A Turner syndrome
B Mayer-Rockitansky-Kuster-Hauser syndrome
C Kallmann syndrome
D Down syndrome
E Congenital adrenal hyperplasia

If both parents carry the trait for beta thalassaemia, what is the chance of a female baby having beta thalassaemia?
A 75%
B 50%
C 25%
D 15%
E 10%

The following clinical features are compatible with what genetic syndrome: renal abnormalities, omphalocele, holoprosencephaly and polydactyly?
A Patau syndrome
B Gardner syndrome
C Fetal alcohol syndrome
D Edwards syndrome
E Down syndrome

A 15 year old boy presents with primary infertility (azoospermia). He is relatively tall for his family with mild gynaecomastia and soft, small testicles. What it is the potential genetic cause?
A Trisomy 15
B 47 XYY
C 47 XXY
D 47 XXX
E 45 XO

A woman has been diagnosed with carrying the BRCA1 gene. What is her lifetime risk of ovarian cancer?
A 70–90%
B 60–70%
C 40–60%
D 30–40%
E 10–30%

A woman has been diagnosed with carrying the BRCA1 gene. What is her lifetime risk of breast cancer?
A 60–90%
B 50–80%
C 40–60%
D 30–40%
E 10–30%

Chorionic villus sampling (CVS) is associated with fetal limb reduction if performed too early in gestation. What is the earliest appropriate gestational age to perform CVS?
A 12 weeks
B 11 weeks
C 10 weeks
D 9 weeks
E 8 weeks

What effect does Down syndrome have on the quadruple test?
A - Increased alphafetoprotein, decreased human chorionic gonadotrophin, decreased unconjugated estradiol and increased inhibin
B - Decreased alphafetoprotein, increased human chorionic gonadotrophin, increased unconjugated estriol and increased inhibin
C -Decreased alphafetoprotein, increased human chorionic gonadotrophin, decreased
D -Decreased alphafetoprotein, increased human chorionic gonadotrophin, decreased unconjugated estriol and increased inhibin
E- Decreased alphafetoprotein, decreased human chorionic gonadotrophin, decreased unconjugated estradiol and increased inhibin