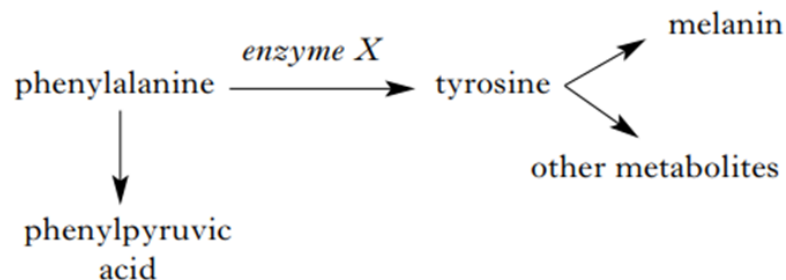


Antenatal & Postnatal Screening

1. During antenatal care, which **two** techniques can be used to obtain cells for production of a karyotype?

- A Chorionic villus sampling (CVS) and amniocentesis
- B Ultrasound imaging and chorionic villus sampling (CVS)
- C Amniocentesis and pre-implantation genetic diagnosis (PGD)
- D Pre-implantation genetic diagnosis (PGD) and ultrasound imaging

2. Phenylketonuria (PKU) is a metabolic disorder which can be lethal in childhood. It is caused by an inability to make *enzyme X*, shown in the metabolic pathway below.



Which substance would have to be removed from the diet for someone who has this disorder?

- A Phenylalanine
- B Enzyme X
- C Tyrosine
- D Melanin

3. The following are three forms of antenatal screening.

- 1 Anomaly scan
- 2 Amniocentesis
- 3 Chorionic villus sampling

Which of these involve the production of a karyotype?

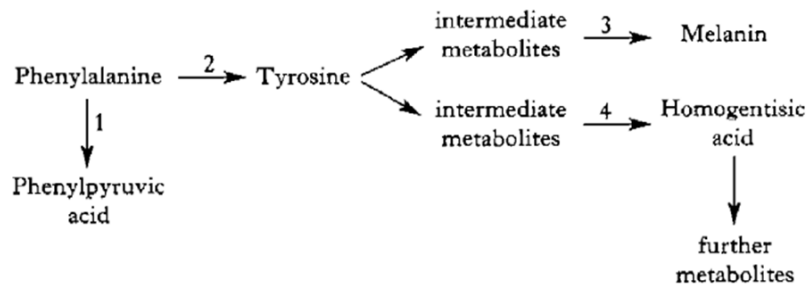
- A 1 only
- B 2 only
- C 2 and 3 only
- D 1,2 and 3

4. A feature of phenylketonuria in humans is

- A the synthesis of excess phenylalanine
- B an inability to synthesise phenylalanine
- C the synthesis of excess tyrosine from phenylalanine
- D an inability to synthesise tyrosine from phenylalanine.

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- 5 The diagram below shows part of a metabolic pathway. Each stage is controlled by an enzyme.



Phenylketonuria (PKU) is caused by a mutation of the gene required to make enzyme

- A 1
- B 2
- C 3
- D 4.

- 6 In the condition phenylketonuria (PKU), the human body is unable to

- A synthesise phenylalanine from tyrosine
- B secrete phenylalanine from cells
- C absorb phenylalanine into the bloodstream
- D convert phenylalanine to tyrosine.

- 1 Adenosine deaminase (ADA) deficiency is an autosomal recessive disorder that affects the immune system. It is caused by a mutation in the gene that codes for this enzyme.

- (a) Describe the difference between an autosomal disorder and a sex-linked disorder.

1

- b) Amniocentesis and chorionic villus sampling (CVS) are examples of antenatal tests that can be carried out.

Describe an advantage and a disadvantage of using amniocentesis rather than chorionic villus sampling (CVS).

2

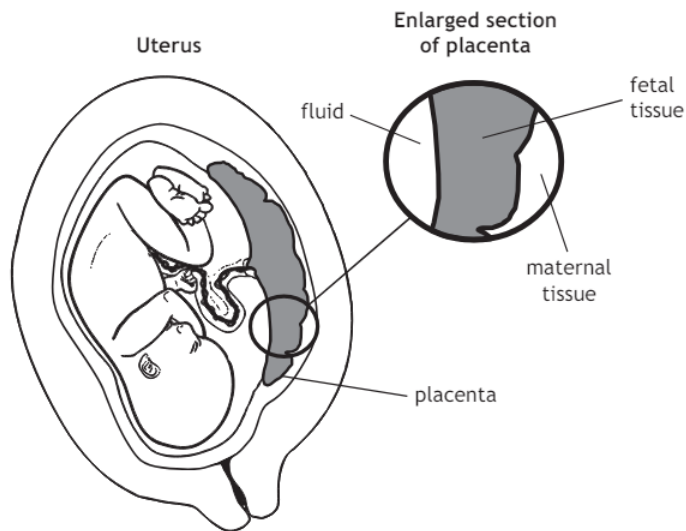
Advantage _____

Disadvantage _____

Antenatal & Postnatal Screening

2 Chorionic villus sampling (CVS) is a technique which can be used during antenatal screening. The cells obtained from CVS are used to prepare a karyotype.

- (a) The diagram below shows the uterus of a pregnant woman with a section of the placenta enlarged.



- (i) Place a cross (X) on the diagram of the **enlarged section of placenta** to indicate the area from which cells are removed during CVS.

1

- (ii) Describe the process by which a karyotype is produced from cells removed during CVS.

2

- (iii) Suggest an advantage of using CVS rather than amniocentesis during antenatal screening.

1

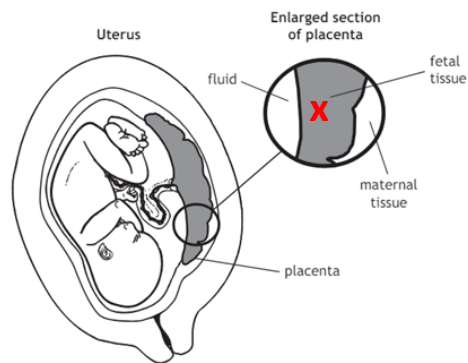
Antenatal & Postnatal Screening

1. A
2. A
3. C
4. D
5. B
6. D

1a) An autosomal disorder is located on chromosomes 1 to 22 while a sex linked disorder is located on chromosome 23.

b) Advantage – amniocentesis has a lower risk of miscarriage. Disadvantage – amniocentesis is carried out later in the pregnancy.

2(i)



(ii) Cells are cultured/allowed to divide 1 mark

Karyotypes then show the chromosomes 1 mark

(iii) CVS can be carried out earlier in the pregnancy