X-Sheet 7

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Terminology & definitions

**Albinism**: inherited condition where melanocytes do not synthesise (make) melanin in the skin, so the skin lacks pigment colour.

**Dominant allele**: an allele that masks or omits over the allele partner of a chromosome pair and the dominant characteristic is seen in the homozygous and heterozygous state.

**Gene mutation**: a change of one or more bases in the nuclear DNA of an organism.

**Gene pool**: the sum of all the genes present in one population.

**Gene therapy**: a process where a mutant gene is masked or replaced.

**Heritable variation**: variation that is passed on from parents to offspring under normal conditions.

**Genome**: the genetic make-up of an individual, representing genetic instructions from the egg and also the sperm.

**Haemophilia**: hereditary, sex-linked disease that is recessive in humans and carried by females but affects males and causes the lack of blood clotting ability in the individual.

**Melanin**: a protein pigment that gives colour to the skin, hair and eyes.

**Predisposed/predisposition**: when there is a chance that the person could suffer from a disease we say they have a predisposition to that disease.

**Sickle-cell anaemia**: a hereditary disease caused by defective haemoglobin and characterized by a sickle-shaped red blood cell.

Key Concepts / Diagrams/ Explanations

**Gene mutations**: Gene mutations may result from internal changes in the nucleus during **meiosis and mitosis**. External factors may change the genetic structure like:

- Unhealthy diet and alcohol abuse
- Environment substances like smoke, specific types of paint, some dyes and colourants, tar and benzene, pollution
- Nuclear radiation, ultraviolet radiation and X-rays
- Viruses

**Identifying carriers**: Genetic testing is conducted to detect and identify genetic disorders. Genetic testing will include:

- Identification of blood proteins
- Analysis of the individual’s DNA
- Checking of the chromosomes and gene sequencing

**Genetic traits are transferred in three ways**:  
- Autosomal dominant alleles (e.g. Huntington’s Chorea)  
- Autosomal recessive alleles (e.g. Albinism, Sickle-cell anaemia, Cystic fibrosis)  
- Sex-linked recessive alleles (e.g. Haemophilia and colour-blindness)

Colour blindness is caused by a sex-linked gene where only males are affected. A person who is colour-blind has difficulty with the perception of red and green.
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Genetic counselling:
Genetic counsellors are doctors who have specialized in sympathetic counselling techniques and are able to assist people to understand genetic disorder and therefore prevent possible gene transfer. The history of the family is studied by the Genetic counsellor to determine a possible pattern of inheritance when:
- A person has been diagnosed with a genetic disorder
- The children with a genetic disorder
- A person wants to know the possibility of passing a disorder to their children based on family history
Counselling takes place before, during and after procedures and analysis of results.

Genetic screening:
Genetic screening is a process to detect the presence of mutant genes. A sample of muscle tissue is used to expose chromosomes to identify mutant genes. Genetic screening will be done for:
- **prenatal diagnosis** - to detect disorders in unborn babies if the mother is concerned about alcohol or drug abuse during pregnancy, injury, family history or age. Counselling about the quality of life of the child should the parents choose to have the baby or whether they choose to terminate the pregnancy
- **carrier diagnosis** - to identify whether one or both parents carry mutant genes. The genetic counsellor will discuss the implications of having children. The informed parents then choose to have children or to adopt.
- **predictive diagnosis** - to predict future diseases that the person may be likely to suffer from. With Huntington’s chorea, symptoms only show after the age of 40, so if a parent suffers from the disease, the children stand a high chance of carrying the dominant gene.

Techniques used for genetic screening:
- **Chorionic villus sampling**: sex-linked mutations can be identified, the sex of the foetus can be determined and conditions like cystic fibrosis and Huntington’s disease can be identified. Termination of the pregnancy is less traumatic physically and emotionally.
- **Amniocentesis**: this procedure takes place at 15 to 16 weeks of pregnancy. The pregnancy will have progressed to six months by the time results are available. Termination of the foetus at 6 months can be physically dangerous for the mother and also emotionally very traumatic.
- **Gene probes and DNA analysis**: analysis of DNA obtained from cells removed by amniocentesis, chorionic villus sampling, blood or squamous epithelial cells from the mouth. Restriction enzymes break the DNA and fragments are separated according to size. A probe is used to identify any mutant genes.

Benefits of genetic screening:
- Reduces risks and suffering of individual effected with a genetic disease as carriers are identified before offspring are produced.
- Parents are afforded the opportunity to make an informed, educated decision, whether to keep the embryo or terminate the pregnancy.
- If predisposed genetic conditions are identified, an individual can change their lifestyle to reduce the possibility of the disease.
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- Women over the age of 35 years have an opportunity to reduce the risk of having a Down’s syndrome baby.

To the Learner:

- Please make sure that you know the various Genetic diseases, the carrier possibilities and how the genetic trait causing disease is transferred.
- This unit is often used to evaluate your values so make sure that you know about the counselling process and the importance of giving a person the opportunity to make an informed and educated decision.
- You may be asked about your view on whether or not you would terminate a pregnancy if you were told that you are carrying a Down Syndrome baby or a similar situation. Include genetic counselling and screening in your answer and back your answers up with reasons that are acceptable within the norms of society. Take careful note of the mark allocation.

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X-ample Questions

Question 1:

Huntington’s chorea

Huntington’s chorea is a rare hereditary disease that is only noticeable at late middle age with the first signs after the age of thirty. During the course of the disease, the brain tissue is damaged causing a person to become restless, moody and depressed. Later, severe muscle spasms develop all over the body and the person becomes insane and then dies. The disease cannot be cured and there is no known way of slowing down its progression. The family tree shows the pedigree chart of the disease in one family. Since the disease can be observed from middle age only, the age of each person is provided in brackets. A cross indicates that the person died at that age. The factor causing Huntington’s chorea is very rare and is known to occur in less than one person in a million. Assume that person B is homozygous.

1.1. Is the gene causing this disease dominant or recessive? Provide a reason for your answer from the family tree provided. (2)

1.2. Is Huntington’s chorea a sex-linked disease. Provide a reason for your answer from the family tree provided. (2)
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1.3. Although individual D did not apparently suffer from the disease, it was not known whether she carried the gene for Huntington’s chorea. Explain the genetic basis for this statement. (3)

1.4. Provide the most probable genotype for each of the following individuals: A, B, C and E. (4)

1.5. What are the probabilities that individual N, O and P will suffer from this disease? Explain your answer in each case. (6)

1.6. Use the evidence in question 1.5 to explain why there are so few people known to be sufferers of Huntington’s chorea in generations 3 and 4. (3)

1.7. What is the likelihood of a person being homozygous for Huntington’s chorea? Choose from:
   a) very high
   b) fairly high
   c) low or
   d) extremely low.
   Explain your answer. (2)

Question 2:
(Taken from NSC Examination Paper 1, Exemplar 2008 – question 1.5)
Cystic fibrosis is an inherited disorder of the human body caused by a recessive gene. This disorder affects mucus production causing blockage of tiny air passages in the lungs. Study the pedigree diagrams below and answer the questions that follow.

2.1. Name the genotypes represented by 1 and 3 in the diagrams respectively. (2)

2.2. What is Thandi’s genotype? (1)

2.3. Does Thandi suffer from cystic fibrosis? (1)

2.4. Thandi and Sipho intend getting married. Show, using a pedigree diagram and the key above, ALL the possible genotypes of any sons they might have. (4)

2.5. Explain what Thandi and Sipho should consider before deciding whether to have children or not. (3)
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X-ercise

1. The external factors that do not play a role in the changes in the gene mutation are:
   A. viruses
   B. environmental toxins
   C. healthy diet with minimal alcohol intake
   D. nuclear radiation, ultraviolet radiation and X-rays

2. Which of the following is not a genetic disease?
   A. Cystic fibrosis
   B. Albinism
   C. Anorexia
   D. Haemophilia

3. ________ is a symptom of Huntington’s Chorea.
   A. The pancreas develops fibrous cysts
   B. The physical structure of the brain shrinks between 20 and 30 per cent of its original size.
   C. Poor blood supply to the muscles due to swelling causing muscle atrophy and gangrene
   D. Violent muscle spasms.

4. Approximately what percentage of all birth defects have genetic defects that are apparent at birth:
   A. 0.5%
   B. 2%
   C. 1%
   D. 1.5%

5. The benefits of genetic screening are...
   A. reduce the risks of genetic diseases
   B. parents are able to make an informal decision to keep the embryo or terminate the pregnancy
   C. Carriers are then identified before offspring are produced
   D. all of the above

6. A person with the genetic disease Albinism, the following will occur...
   (i) they will have white hair on their head, body and eyebrows
   (ii) when exposed to sunlight they will get a slight tan before peeling leaving darker spots that resemble freckles
   (iii) eyes look red because they are photosensitive
   (iv) they have a lack of melanin in their skin

   A. i, ii, iv
   B. ii, iii, iv
   C. i, iii, iv
   D. i, ii, iii
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7 Environmental toxins such as cigarettes, tobacco, alcohol abuse, specific types of paint, some dyes and colourants, tar, benzene and exhaust fumes cause genetic mutations.
   A  True
   B  False

8 Down’s syndrome, Klinefelter’s syndrome and Turner’s syndrome are as a result of:
   A  hereditary diseases
   B  bad health and diet
   C  chromosomal mutation
   D  genetic mix-up

9 Haemophilia is when there is extreme blood thickening throughout your body, causing blood clots.
   A  True
   B  False

10 Cystic Fibrosis is a common genetic disease in white people and it is caused by an autosomal gene that is hereditary.
   A  True
   B  False

Answers to the X-ercise questions:

1. C
2. C
3. B
4. C
5. D
6. C
7. B
8. D
9. B
10. B