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GENETIC DISEASES, GENETIC COUNSELLING AND SELECTIVE BREEDING

SECTION A: TYPICAL EXAM QUESTIONS

Question 1: 8 minutes

HINT:

When you do multiple choice questions, cover the answers options. Read through the question, then think of the correct answer, then look at which answer option is closest to the answer that you thought of. If you don't know the answer, then look at the options. Place a tick next to the answer that appears correct and a cross next to the answer option that appears to be wrong. If you have ticked two, then re-read the question and try to find the answer that is the most correct.

Example:

A person uses their eyes to ...

- A see objects that are far away ✓
- B smell objects that are close by ✓
- C hear children playing far away ✓
- D taste a glass of orange juice ✓

1.1. 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

1.2. Which of the following is not a genetic disease?

- A Cystic fibrosis
- B Albinism
- C Anorexia
- D Haemophilia

1.3. _____ is a symptom of Huntington's Chorea.

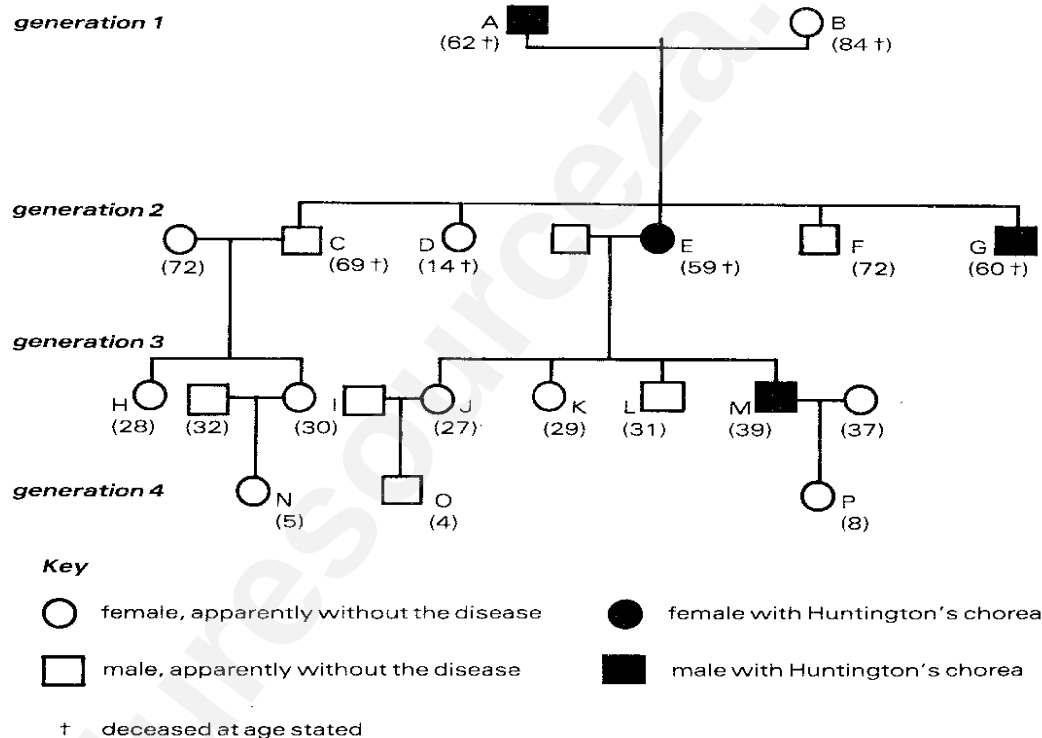
- A The pancreas develops fibrous cysts
- B The physical structure of the brain shrinks between 20 and 30 per cent of its original size
- C Poor blood supply to the muscles due to swelling causing muscle atrophy and gangrene
- D Violent muscle spasms.

- 1.4. 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%
- 1.5. 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
- 1.6. 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
- 1.7. 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
- 1.8. 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
- 1.9. Haemophilia is when there is extreme blood thickening throughout your body causing blood clots.
- A True
 - B False
- 1.10. Cystic Fibrosis is a common genetic disease in white people and it is caused by an autosomal gene which is hereditary:
- A True
 - B False

(10)

Question 2: 25 minutes**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



(With kind thanks and acknowledgement from Data response exercises in Biology, P.W. Ayerst et al: Shuter & Shooter 1989.)

(HINT: Read the passage twice. Then look at the pedigree chart of the family tree. Then move onto the questions.)

- 2.1. Is the gene causing this disease dominant or recessive? Provide a reason for your answer from the family tree provided. (2)
- 2.2. Is Huntington's chorea a sex-linked disease. Provide a reason for your answer from the family tree provided. (2)

- 2.3. 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)
- 2.4. Provide the most probable genotype for each of the following individuals: A, B, C and E. (4)
- 2.5. What are the probabilities that individual N, O and P will suffer from this disease? Explain your answer in each case. (6)
- 2.6. 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)
- 2.7. 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)

"The science of eugenics is aimed at improving the inborn qualities of future generations of mankind. One way it hoped to do this was by sterilizing people with genetic diseases so that they were unable to pass on their defective genes to the next generation"

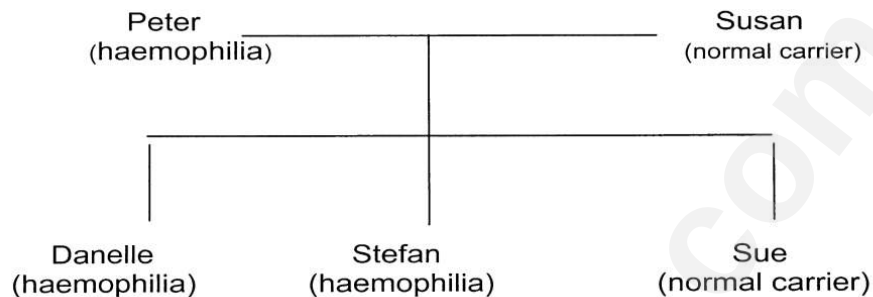
8. Imagine that a gene X, which causes a defective trait, is dominant and that it is present in 10% of the population. Also assume that everyone in the world suffering from the genetic disease caused by X were to be sterilized before they were able to reproduce. What would the frequency be of the gene in the next generation? Explain your answer. (3)
9. Explain why the system of eugenics by sterilization would not work for a disease like Huntington's chorea. (2)
10. Briefly discuss your opinion of eugenics and whether this process should be legislated in South Africa. . (3)

(Reminder: when you are asked for your opinion – it must be based on facts and not just a feeling. Also remember that you need to choose a view point – you are either for or against the process. You MUST NOT discuss both for and against in your answer as only the first response will be marked)

Question 3: 7 minutes (Taken from Gauteng Prelim 2009 Paper 1)

Haemophilia is a sex-linked hereditary disease that occurs as a result of a recessive allele on the X-chromosome. Study the family tree below and answer the questions that follow:

(Use the symbols **H** for normal and **h** for haemophilia above the sex chromosomes, for example.: $X^H X^h$)



- 3.1. Write down the genotype of Stephan. (2)
- 3.2. Peter and Susan would like to have a fourth child. Calculate the percentage probability of this child having haemophilia. (6)

SECTION B: SOLUTIONS AND HINTS**Question 1:**

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

Question 2:

1. Dominant ✓. The presence of the gene in heterogynous state still causes the disease in individuals. ✓
2. No. Both males and females are equally affected. ✓✓
3. She died at the age of 14 ✓before the onset of the disease. ✓If B is homozygous recessive, then the probability is that D is possibly homozygous recessive and therefore will not carry the gene. ✓
4. A = Hh ✓
B = hh ✓
C = hh ✓
E = Hh ✓

5. N – no chance since the 2nd generation lived to 72 and 69 years. Highly probable that birth parents of N are homozygous recessive (hh) ✓✓
O – Possible since E died of Huntington's chorea and must be Hh. J could therefore be carrying the dominant gene. ✓✓
P – Highly probable since M must be Hh the chances are 50% that P is carrying the dominant gene. ✓✓
6. The ages of the individuals in generations 3 and 4 are between 4 and 39. ✓ Only M has been diagnosed (aged 39). ✓ None of the other individuals have reached the age where the symptoms can be recognized. ✓
7. c) low. ✓ If 1 parent is hh and one is Hh the chance for homozygous HH is zero. If both parents are Hh then the chance for homozygous HH is only 25%. ✓
8. Zero ✓ since none of the people carrying the X gene would be able to reproduce since they had been sterilized. No X gene would be transferred to the next generation. ✓ This will also be based on the assumption that the genetic disease caused by gene X could be identified before reproductive age of the individual is reached. ✓
9. The genetic disease is only identified many years after reproductive age is reached. ✓ In most cases, infected individuals have already reproduced thereby passing the gene on to the next generation. ✓
10. Learners respond with either 'for' or 'against'. ✓
For: discuss enforced protection of possible offspring/ financial burden on the parents and state should defect be passed on/emotional stress placed on family should defective offspring be produced/quality of life of defective offspring etc. ✓✓
Against: discuss that this is against the constitutional rights of people living in South Africa. Sterilization causes problems with hormonal balances and health. Individuals should be provided with options through Genetic counselling so that they are able to make their own educated decisions regarding sterilization. People must retain their rights etc. ✓✓

Question 3:

3.1. XhY ✓✓ (2)

3.2 $P1: XhY \times XHXh$ ✓

Gametes: $Xh \ Y \ XH \ Xh$

$F1: XHXh \ \checkmark \ XhXh \ \checkmark \ XHY \ \checkmark \ XhY \ \checkmark$

50 % ✓ probability of having haemophilia (6)

SECTION C: ADDITIONAL CONTENT NOTES**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.

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 want 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 is the mother 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
- Women over the age of 35 years have an opportunity to reduce the risk of having a Down's syndrome baby.



Learner Note: 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. You must include genetic counselling and screening in your answer and back the answers up with reasons that are acceptable within the norms of society. Take careful note of the mark allocation.

SECTION D: HOMEWORK



Learner Note: As you attempt the homework you need to ensure that you are able to answer the questions in the allocated time frames. If you get stuck, refer to either the additional notes or your class teacher.

Cystic fibrosis is a common genetic disease caused by an autosomal gene, which is recessive. The gene must therefore be present in both alleles and be homozygous, to cause the disease. It is estimated that one in 25 people carry the disease and one in 2000 suffer from the disease. Life expectancy is from one year to about 30 years. Since this genetic disease is common, geneticists looked for an advantageous reason and have found that this condition gives an increased resistance to cholera.

1. What is the difference between sex-linked genetic disorders and autosomal genetic disorders? Use examples to explain the difference. (4)
2. You have been told that the baby you are expecting has cystic fibrosis. The genetic counsellor has discussed the symptoms of the disorder and how the disease progresses. You have to make a decision: Do you terminate the pregnancy or are you going to have this child? Support your answer with reasons. (4)
3. A group of Grade 12 learners did a survey about the frequency of genetic disorders in a small human population. The results are shown in the table below. The number of people in the population is 1 200.

RESULTS OF THE SURVEY ON THE FREQUENCY OF GENETIC DISORDERS IN A POPULATION

GENETIC DISORDERS	FREQUENCY IN POPULATION (%)
Albinism	7
Down's syndrome	x
Sickle cell anaemia	10
Without any disorder	78

- 3.1. According to the table, which disorder is most common in this community? (1)
- 3.2. Calculate the number of people with Down's syndrome. Show ALL working. (3)
- 3.3. Give ONE reason why genetic counselling would be important for parents who are expecting children that may be sufferers of a disorder such as Down's syndrome. (1)

SECTION E: SOLUTIONS TO SESSION 17 HOMEWORK

- 1.1. May have fewer side effects ✓

May not be contaminated ✓/will be in its natural form

No problem from a religious perspective ✓

Can be mass produced ✓/produced faster

Avoids killing animals ✓ (3)

(Mark first THREE answers only in learner's script)

- 1.2. Against:

- risk to human health ✓

- risk to the environment ✓

- risk to the health and well-being of other organisms ✓

- interference with nature ✓/God's creation

- cultural sensitivity ✓ e.g. objection to the use of pigs and cows (2)

(Mark first TWO answers only in learner's script)

- 2.1 a sudden/rapid ✓
unexpected/random/unpredictable ✓
change/mistake ✓
in DNA ✓ (2)
(Mark any 2)
- 2.2 it affects chromosome 1/no indication it is sex-linked or affects gonosomes ✓ (1)
- 2.3 genetic counsellor/geneticist ✓ (1)
- 3.1 May 7 2003 (full date) ✓ (1)
- 3.2 Futhi means replica/repeat ✓
A replica is an exact ✓ copy ✓
The calf is a genetically identical/a clone of/has the same DNA as the parent ✓ (3)
- 3.3 (a) sexual reproduction (or explanation thereof) ✓ (1)
(b) **Any 2 of** ... crossing over random fusion of gametes; ✓ independent assortment/random alignment during meiosis ✓ (2)
- 3.4 Protective covering of the egg left on in normal cloning ✓ (1)
- 3.5 uterus ✓ (1)
- 3.6 Many (clones) can be made of highly productive livestock ✓ (1)
- 3.7 Producing the clone, i.e. viable embryo itself ✓
OR
When the egg is inserted into the recipient ✓ (1)
- 3.8 Isolation phase: Any 1 of ... Remove gene for high yield from high yielding donor ✓
Engineer phase: Any 1 of ... Genetically engineer a bacterium/plasmid to contain this gene ✓
Insertion phase: Any 1 of ... Infect the cow with the bacterium ✓ (3)
OR any other reasonable 3 step process