

Cecil Andrews College

**Year 11**

**ATAR**

**Human Biology**

**Test 5**

**Variation, Genetics and Inheritance**

Syllabus Points:

* crossing over, non-disjunction and random assortment of chromosomes during meiosis will produce gametes with different genetic content
* variations in the genotypes of offspring, including gender, arise as a result of the processes of meiosis and fertilisation
* probable frequencies of genotype and phenotype of offspring can be predicted using Punnett squares and by taking into consideration patterns of inheritance, including the effects of dominance,   
  co-dominance, autosomal or sex-linked alleles, and multiple alleles: Huntington's disease, phenylketonuria (PKU), ABO blood groups, red–green colour blindness/haemophilia show different inheritance patterns
* pedigree charts can be constructed for families with a particular genetic disorder and can be used to reveal patterns of inheritance and assist in determining the probability of inheriting the condition in future generations
* DNA profiling identifies the unique genetic make-up of individuals and can be used in determining parentage

**Name: \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_**

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| **Section** | **Available Marks** | **Achieved Marks** |
| **Multiple Choice** | **20** |  |
| **Short Answer** | **58** |  |
| **Extended Response** | **10** |  |
| **Total** | **88** |  |

**Section One: Multiple-choice (20 Marks)**

This section has **20** questions. Answer **all** questions on the separate Multiple-choice Answer Sheet provided. For each question shade the box to indicate your answer. Use only a blue or black pen to shade the boxes. If you make a mistake, place a cross through that square, do not erase or use correction fluid, and shade your new answer. Marks will not be deducted for incorrect answers. No marks will be given if more than one answer is completed for any question.

1. The best description of a person in terms of his/her observable features is called their:
   1. profile.
   2. genotype.
   3. phenotype.
   4. genome.
2. A person who has two identical genes for a particular trait is termed a:
   1. hybrid.
   2. homozygote.
   3. heterozygote.
   4. hemizygote.
3. A couple have a family of four daughters. Which of the following statements about the sex chromosomes of the family are true?
   * 1. All the sex chromosomes of the parents are X.
     2. All the sex chromosomes of the husband are X.
     3. All the sex chromosomes of the wife are X.
     4. All the sex chromosomes of the daughter are X.
     5. Half the sex chromosomes of the husband are Y and half are X.
     6. Half the sex chromosomes of the wife are X and half are Y.
4. I, II and III.
5. II, III and IV.
6. III, IV and V.
7. IV, V and VI.
8. The sex of an offspring is determined by the presence or absence of the Y chromosome. If the offspring is male, it can be concluded that:
   1. the mother’s gamete contained an X and a Y chromosome.
   2. the mother’s gamete contained a Y chromosome.
   3. the father’s gamete contained an X and a Y chromosome.
   4. the father’s gamete contained a Y chromosome.
9. In humans, polydactyly (presence of an extra digit) is due to an autosomal recessive gene. A man with polydactyly is to marry a normal woman whose father has an extra digit and they want to know the probability of having children with polydactyly. A genetic counsellor would advise them that the probability of having children with polydactyly would be:
10. 0.
11. 50%.
12. 100%.
13. 25%.
14. Sex-linked diseases are not usually found in females because the sex-linked genes are usually recessive and:
    1. occur on the homologous portions of both X and Y chromosomes.
    2. occur on the Y chromosome.
    3. occur on only one of the female’s X chromosomes.
    4. provide immunity for the female.
15. The allele for curly hair is autosomal dominant to the allele for straight hair. If a heterozygous curly-haired woman and a homozygous straight-haired man have a daughter, what is the probability that she will have curly hair?
    1. 0%.
    2. 25%.
    3. 50%.
    4. 100%.
16. At which of the following stages is the sex of the individual determined?
    1. When the ovum is formed.
    2. When the sperm is formed.
    3. When the zygote is formed.
    4. When the embryo is 16 weeks old.
17. A good example of co-dominance occurs in the ABO blood grouping system. This occurs between the following alleles:
    1. IA and IB.
    2. IA and i.
    3. IB and i.
    4. IA,, IB and i
18. Colour-blindness is an X-linked recessive condition. What is the probability that a man who is colour-blind and married to a woman who is a carrier, could have a child who is colour-blind?
    1. 0.25
    2. 0.50
    3. 0.75
    4. 1.00

THE NEXT FOUR QUESTIONS REFER TO THE FOLLOWING INFORMATION

The diagram below shows the family tree for the inheritance of albinism through three generations. (Mode of inheritance for albinism is autosomal recessive).

**4**

**2**

**9**

1. Individual 2 has the alleles:
   1. aa.
   2. Aa.
   3. AA.
   4. either AA or Aa.
2. Individual 4 has the alleles:
   1. aa.
   2. Aa.
   3. AA.
   4. either AA or Aa.
3. Individual 9 obtained the alleles for albinism from his:
   1. mother only.
   2. father only.
   3. mother and father.
   4. grandparents 2 and 4.
4. If individual 7 is heterozygous for albinism and marries a man who does not carry the albinism gene, what is the probability that they will have a child with the condition?
   1. 0%.
   2. 25%
   3. 50%.
   4. 100%
5. Alternative forms of a gene are termed:
   1. chromatids.
   2. homologues,
   3. karyotypes.
   4. alleles.
6. A child with blood group O has a mother with blood group A. The father must therefore have:
   1. AB blood group.
   2. IAIA genotype.
   3. Genotypes IAi, IBi or ii.
   4. Genotype ii only.
7. In a monohybrid cross:
   1. several characteristics are studies.
   2. one pair of characteristics is studied.
   3. two genotypes are studied.
   4. two phenotypes are studied.
8. Mitochondrial DNA is inherited from the:
   1. father.
   2. mother.
   3. grandparent.
   4. offspring.
9. A pair of homologous chromosomes:
   1. are identical in their gene content.
   2. carry one pair of genes concerned with the same physical characteristic.
   3. carry dominant genes only on one chromosome and recessive genes only on the other.
   4. carry genes derived from one parent only.
10. Crossing over occurs in which stage of meiosis:
    1. prophase I
    2. anaphase I
    3. metaphase I
    4. metaphase II

**Section Two: Short answer (58 Marks)**

This section has **nine (9)** questions. Answer **all** questions. Write your answers in the spaces provided.

Supplementary pages for the use of planning/continuing your answer to a question have been provided at the end of this Question/Answer booklet. If you use these pages to continue an answer, indicate the original answer where the answer is continued, i.e. give the page number.

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1. Cystic Fibrosis (CF) is an autosomal recessive disorder. A couple with a history of CF in their families undergo genetic counselling before trying to conceive a baby.
   1. It is found that the man is a carrier for CF and the woman is homozygous normal. Predict the possible genotypes and phenotypes of their future children. (5 marks)
   2. The genetic counsellor drew a pedigree to show the couple how CF has been inherited in their families. In the space provided, construct the pedigree diagram the counsellor would have drawn. Include:

* three labelled generations
* the couple’s parents
* the couple and their siblings. The man has an older sister; his sister was affected with CF.   
  The woman is the oldest child, with a younger sister and an even younger brother who are unaffected
* the prediction of three future children, who are all boys. (8 marks)

1. The pedigree below shows the inheritance of an X-linked disease.
   1. Label the generations and individuals. (1 Mark)
   2. Is the gene for this disease dominant or recessive? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ (1 Mark)
   3. Explain your answer\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(2 Marks)

* 1. What are the genotypes of the following individuals shown in the pedigree?
     1. I 2 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
     2. III 2 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
     3. III 3 \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ (3 Marks)
  2. Female III 1 is pregnant. Assume this disease is red-green colour-blindness and the father of the child has normal vision. What is the probability that the child will be a red-green colour-blind son? Show your working out.

(2 Marks)

1. If a woman with blood group AB marries a man who is homozygous for blood group B, what are the possible genotypes and phenotypes they can produce if they have offspring?

Show all workings.

(3 Marks)

1. Draw the following pedigree in the space provided.

Bill marries Sue. They have four children, Bob, Sally, Rosie and Tom.

Bob marries Cathy, and Rosie marries Jim. The other two children do not marry.

Bob and Cathy have five children, Bert, Ernie, Harry, Charlie and Pete.

Rosie and Jim have only one child whom they name Jane.

(3 Marks)

Jane marries Edmund and they have identical twin girls. Draw a diagram to show this.

(2 Marks)

1. Sickle-cell anaemia is a genetic disease which in the homozygote causes the red blood cells to become sickle shaped and lose their oxygen-carrying capacity. In the heterozygote, both types of red blood cells are formed. The person is then said to have sickle-cell trait (not anaemia). People who have sickle-cell trait are quite healthy but they can suffer a shortage of oxygen at high altitude or under extreme physical exertion.
   1. What is this disease’s mode of inheritance? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(2 Marks)

* 1. If a couple who both have the sickle-cell trait have children, what is the probability that their first child will have sickle-cell trait?

(3 Marks)

* 1. If a person who has sickle-cell trait marries a normal homozygote, what is the probability that any one of their children will have sickle-cell anaemia?

(3 Marks)

1. Assume that the gene for brown eyes is dominant B, and the gene for blue eyes is recessive b. A brown-eyed woman whose mother had blue eyes marries a man with brown eyes. They produce four children all of whom are brown eyed.
   1. What is the genotype of the mother? \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(1 Mark)

* 1. What are the possible genotypes of her husband? Explain. \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(2 Marks)

* 1. If their fifth child is blue-eyed, what is the husband’s genotype? Explain your answer.

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(3 Marks)

1. Red-green colour blindness is an X-linked recessive condition. Alison has normal vision, as do her parents, Dick and Dora, but Alison’s brother Bob has colour- blindness.
   1. What are the genotypes of this family?
      1. Alison \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
      2. Dick \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
      3. Dora \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_
      4. Bob \_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_ (4 Marks)
   2. How could you be sure of Alison’s genotype?

\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_\_

(2 Marks)

1. Why do couples who are first cousins have a slightly higher risk of having a child with an inherited disorder than unrelated couples?

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(3 Marks)

1. Haemophilia is a recessive, sex-linked trait. A person with haemophilia is lacking certain proteins that are necessary for normal blood clotting. A woman who is heterozygous for haemophilia marries a normal man.
2. What are the genotypes of the parents? (1 Mark)
3. What are the probabilities their children will inherit the condition? (2 Marks)

1. What is the probability that a male offspring will have haemophilia? (1 Mark)

1. What is the probability of having a haemophiliac female offspring? (1 Mark)

**Section Three: Extended answer (10 Marks)**

This section contains **two (2)** questions. You must answer **one (1)** question. Write your answers in the lined pages provided.

Supplementary pages for the use of planning/continuing your answer to a question have been provided at the end of this Question/Answer booklet. If you use these pages to continue an answer, indicate at the original answer where the answer is continued, i.e. give the page number.

Responses could include clearly labelled diagrams with explanatory notes; lists of points with linking sentences; clearly labelled tables and graphs; and annotated flow diagrams with introductory notes.

Suggested working time: 50 minutes.

1. All species show differences between individuals. The differences between members of a species are called variation. Describe the processes that cause and maintain this variation in humans.
2. Families with a known condition are often given access to the services of a genetic counsellor. Describe how genetic counsellors use pedigrees and genetic testing to provide advice to couples who are planning to have children.

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