

Name of the Student: _____

Max. Marks : 23 Marks

Time : 23 Minutes

Q1.

Read the following passage.

Complete achromatopsia is a form of complete colour blindness. It is caused by having only rods and no functional cone cells. People with complete achromatopsia have difficulty in seeing detail. Complete achromatopsia is caused by an autosomal recessive allele and is usually very rare in populations with only one in 40 000 being affected. However on the Pacific island of Pingelap ten percent of the population are affected. 5

One form of red-green colour blindness is caused by a sex-linked recessive allele which affects more men than women. People with this red-green colour blindness are unable to distinguish between red and green, and also between other colours. They have green-sensitive cones but the photoreceptive pigment they contain does not function. 10

Scientists investigated the use of gene therapy to correct red-green colour blindness in monkeys. They injected viruses containing the gene for the green-sensitive pigment directly into the eyes of the monkeys. Although the monkeys maintained two years of colour vision, there is debate on whether this form of gene therapy is worthwhile. No clinical trials of this procedure have been carried out on humans. Current research into the treatment of red-green colour blindness involves the use of induced pluripotent stem cells (iPS cells). The use of iPS cells could have advantages over the use of gene therapy. 15 20

Use the information in the passage and your own knowledge to answer the following questions.

- (a) People with complete achromatopsia have difficulty in seeing detail (lines 2–3).

Explain why.

(3)

- (b) Ten percent of the population on the Pacific island of Pingelap are affected by complete achromatopsia (lines 3–6).

Use the Hardy-Weinberg equation to calculate the percentage of this population who are heterozygous for this disorder.
Show your working.

Answer = _____ %

(2)

- (c) Red-green colour blindness affects more men than women (lines 7–8).

Explain why.

(2)

- (d) People with red-green colour blindness are unable to distinguish between red and green, and also between other colours (lines 8–10).

Explain why.

(3)

- Suggest how iPS cells could correct red-green colour blindness.

(2)

- Using the information from the passage, suggest and explain reasons why.

(3)

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Q2.

Short tandem repeats (STRs) are short sequences of DNA, usually 2 to 7 base pairs. STRs are repeated a number of times, one after another. For example, the STR D5S818 is made up of AGAT repeated 7 to 16 times.

STRs are found throughout the whole genome.

The repeated sequences in STRs are common to all humans. Due to variation in the number of repeats, STRs can be used in genetic fingerprinting.

- (a) Describe how STRs could be removed from a sample of DNA.

(2)

- (b) Genetic fingerprinting using STRs requires amplification of the STRs using the polymerase chain reaction (PCR). The short base sequences either side of a specific STR are known.

Explain the importance of knowing these base sequences in PCR.

(2)

- (c) A single STR molecule consisting of a 12 **base pair** sequence of AGAT was amplified 50 times using PCR.

Calculate the total number of base pairs in all the STR molecules after 50 cycles of PCR.

Show your working.

(d) During genetic fingerprinting, the different STRs are separated by gel electrophoresis.

Give **two** features of STRs which enable them to be separated by gel electrophoresis.

1 _____

2 _____

(2)

(Total 8 marks)