Genetic technology – 2021 Biology A2 9700

1. Nov/2021/Paper_41/No.4

(a) In 1984, the geneticist Alec Jeffreys invented a DNA testing technique, known as DNA profiling, that produces a DNA banding pattern on a gel. The DNA banding pattern (profile) is unique to each individual.

DNA profiling can be used in police forensic work to catch criminals.

Since 1987, police in many countries have collected and stored DNA from crime scenes to create DNA profiles, which they try to match with the DNA profiles of criminal suspects.

(i) DNA at a crime scene may be obtained from hairs and traces of blood, semen and saliva.

Explain why PCR may be needed before DNA from a crime scene can be profiled.

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(ii) Explain why electrophoresis produces a DNA banding pattern on a gel.

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(b) GEDmatch is described as 'an open data personal genomics website'. It can be used by people who want to upload their DNA data to trace their ancestors and other relatives.

In 2018, police in the USA solved a large number of serious crimes. Some of these crimes had been unsolved for over thirty years. The police used GEDmatch to profile DNA taken from crime scenes and to look for matching DNA profiles. In many cases the police found partial matches to the relatives of criminals. This allowed the criminals to be identified and then charged on the basis of a complete DNA profile match.

(i) Suggest why the police strategy of comparing crime scene DNA with the GEDmatch database was so successful.

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(ii) Explain why GEDmatch is an example of bioinformatics.

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(iii) The first successful conviction resulting from the use of GEDmatch by the police was widely reported.

Some journalists and broadcasters thought that the GEDmatch website should not have been used by the police in this way.

In the days following the news, the number of citizens choosing to upload their DNA data to GEDmatch increased from 1500 to 5000 a day.

Comment on the social **and** ethical issues raised by this first successful conviction.

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[Total: 10]

2. Nov/2021/Paper_42/No.4

(a) Genome-wide association studies find links between single nucleotide polymorphisms (SNPs) and phenotypic features such as human diseases. SNPs are points on the DNA that vary in the population because of DNA base substitutions.

A genome-wide association study investigates the effect of genetic variation on a disease. A large number of people with the disease and a large number of healthy control individuals provide DNA. Microarray chips are used to identify the genotype of each individual at many SNPs.

The Wellcome Trust Case Control Consortium (WTCCC) study was an important genome-wide association study.

- The study used a microarray chip that identified each person's genotype at 500 000 different SNPs.
- The study looked for links between SNPs and 7 different diseases.
- For each disease, 2000 people with the disease were tested.
- Their results were compared with the results of 3000 healthy control individuals.

(i) Outline how microarrays are used in the analysis of genomes.

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(ii) Explain why bioinformatics was important to the WTCCC study.

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(b) Fig. 4.1 summarises results for three diseases in the WTCCC study. The 22 human autosomes and the X chromosome (chromosome 23) are shown.

Chromosome locations with SNPs that are associated with a disease at a statistically significant level (greater than 5 arbitrary units) are shown in black.

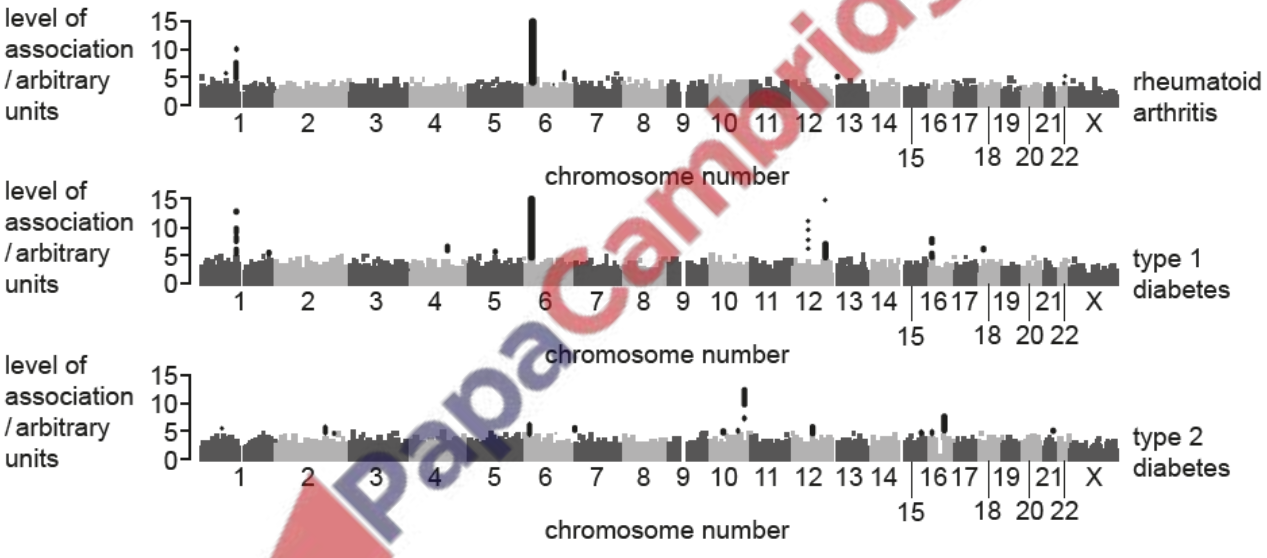

Fig. 4.1

(i) Identify the chromosomes that contain SNPs that have a high level of association with **both** rheumatoid arthritis and Type 1 diabetes.

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(ii) With reference to Fig. 4.1, compare the genetic basis of the three diseases.

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(c) Individuals can choose to have their DNA analysed on a microarray chip to predict their risk of developing different diseases.

Outline the social **and** ethical considerations of this type of DNA analysis.

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3. Nov/2021/Paper_42/No.7

(a) The Labrador is a variety of domestic dog. Labradors have fur that can be brown, black or yellow.

In Labradors, *TYRP1* is one gene that codes for fur colour. This gene has two alleles, **B** and **b**.

- The dominant allele, **B**, codes for the enzyme tyrosinase that functions in the pathway to produce melanin, leading to black fur.
- The production of melanin in Labradors is very similar to the production of melanin in humans.
- The recessive allele, **b**, codes for an enzyme that results in the production of a brown form of melanin, leading to brown fur.

Outline how melanin may be produced in Labradors to produce black fur.

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(b) Another gene, *MC1R*, interacts with *TYRP1*.

MC1R has two alleles, **E** and **e**.

- The dominant allele, **E**, allows the alleles of *TYRP1* to be expressed.
- The recessive allele, **e**, prevents the alleles of *TYRP1* from being expressed.
- When no form of melanin is produced the Labrador will have yellow fur.

(i) Construct a genetic diagram to show the ratio of possible offspring from a cross between a black male Labrador, heterozygous for both genes, and a yellow female Labrador, heterozygous for *TYRP1*.

parental phenotype	black	×	yellow
parental genotype			
gametes			
offspring genotypes			
offspring phenotypes			

ratio .....

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(ii) State the term used to describe a protein that is involved in the control of gene expression in eukaryotes.

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