A level Biology A

H420/03 Unified biology

Question Set 11

1 Agammaglobulinemia and Vici syndrome are both genetic diseases.

(a) Agammaglobulinemia results in a lack of mature B lymphocytes in a person's blood.

(i) Suggest and explain one symptom of agammaglobulinemia. **[2]**

greater susceptibility to pathogens as fewer antibodies are produced

(ii) Fig. 4 shows the inheritance pattern of agammaglobulinemia in a family.

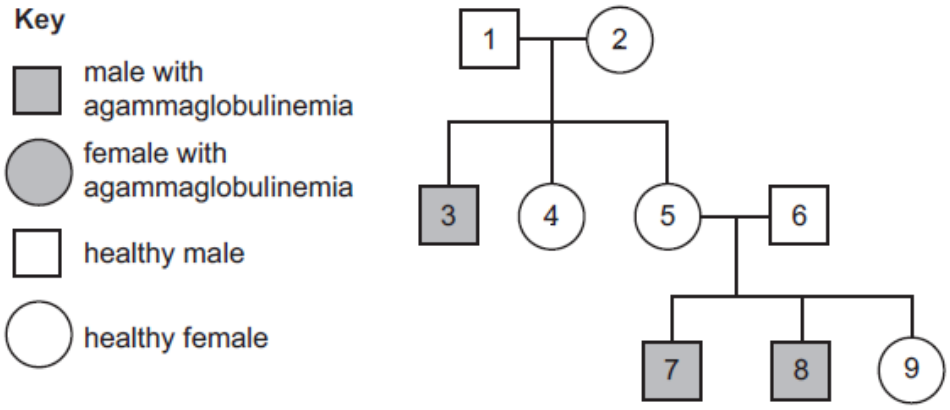

Fig. 4

What conclusions can you draw about the location and nature of the allele responsible for causing agammaglobulinemia? Explain your conclusions. **[4]**

- recessive allele: healthy parents 1&2, 5&6 produce children with disease
- 2&5, mothers are heterozygous } as on X chromosome and only males have disease
- likely to be sex-linked

(b) Vici syndrome is a genetic disease that shows a recessive inheritance pattern. The allele responsible for Vici syndrome is found on chromosome 18.

(i) Two carriers of Vici syndrome have six children.

Calculate how many of the six children you would expect to:

- have Vici syndrome
- be carriers of Vici syndrome.

	D	r
D	DD	Dr
r	Dr	rr

Vici syndrome ..... 2 .....

Carriers ..... 3 .....

Vici (rr): $6 \times \frac{1}{4} = \frac{3}{2} = 1.5$ so 1 or 2 **[1]**

Carrier (Dr): $6 \times \frac{2}{4} = 3$

* **(ii)** A daughter of these parents and a male carrier of Vici syndrome have a child.

Calculate the probability of the child having Vici syndrome.

Answer = ... 0.25 ..... **[1]**

daughter: DD : 0.25, Dr : 0.5, rr : 0.25 } Dr : 0.5

male: DD × Dr = X = 0; Dr × Dr = 0.5 × 0.25 = 0.125; rr × Dr = 0.25 × 0.5 = 0.125 } 0.25

(c) DNA profiling can be used to analyse the risk of inheriting conditions such as agammaglobulinemia and Vici syndrome.

(i) To produce a DNA profile, DNA first needs to be purified.

Explain why a protease enzyme is added to the mixture during the DNA purification process. **[1]**

protease digests proteins associated with DNA

(ii) DNA samples can be amplified using the polymerase chain reaction (PCR).

In theory, how many fragments of DNA might be present after 12 cycles of PCR?

Assume one DNA fragment was present at the beginning of the PCR process. Represent your answer as a $\log_{10}$ value.

$2^{12} = 4096$

$\log_{10}(4096) = 3.612$

..... $10^{3.612}$ ..... fragments **[2]**

(iii) Suggest why the figure you calculated in **(ii)** may not be achieved in practice. **[1]**

lack of primers & free nucleotides

(iv) State the name of the enzyme used in PCR to synthesise new DNA strands. **[1]**

Taq DNA polymerase

(v) DNA fragments are separated to produce a DNA profile using electrophoresis.

A student wrote the following description of the electrophoresis procedure:

> We will set up an agarose gel plate and place the DNA samples in the wells at the cathode. Voltage will be passed through the gel for one minute. The gel will then be placed in purified water and we will be able to see the banding pattern of each DNA sample.

Describe **two** changes you would make to the student's procedure and explain how these changes would improve electrophoresis. **[2]**

- use alkaline solution ⇒ solution carries charge
- use radioactive / fluorescent probes ⇒ to visualise bands

Total Marks for Question Set 11: 15