

- 4 Nail patella syndrome is a rare genetic disease, which causes deformity or absence of some or all of the nails and the absence of the patella (knee-cap).

Fig. 4.1 shows a pedigree from an affected family. The ABO blood groups of the members of the family are also shown.

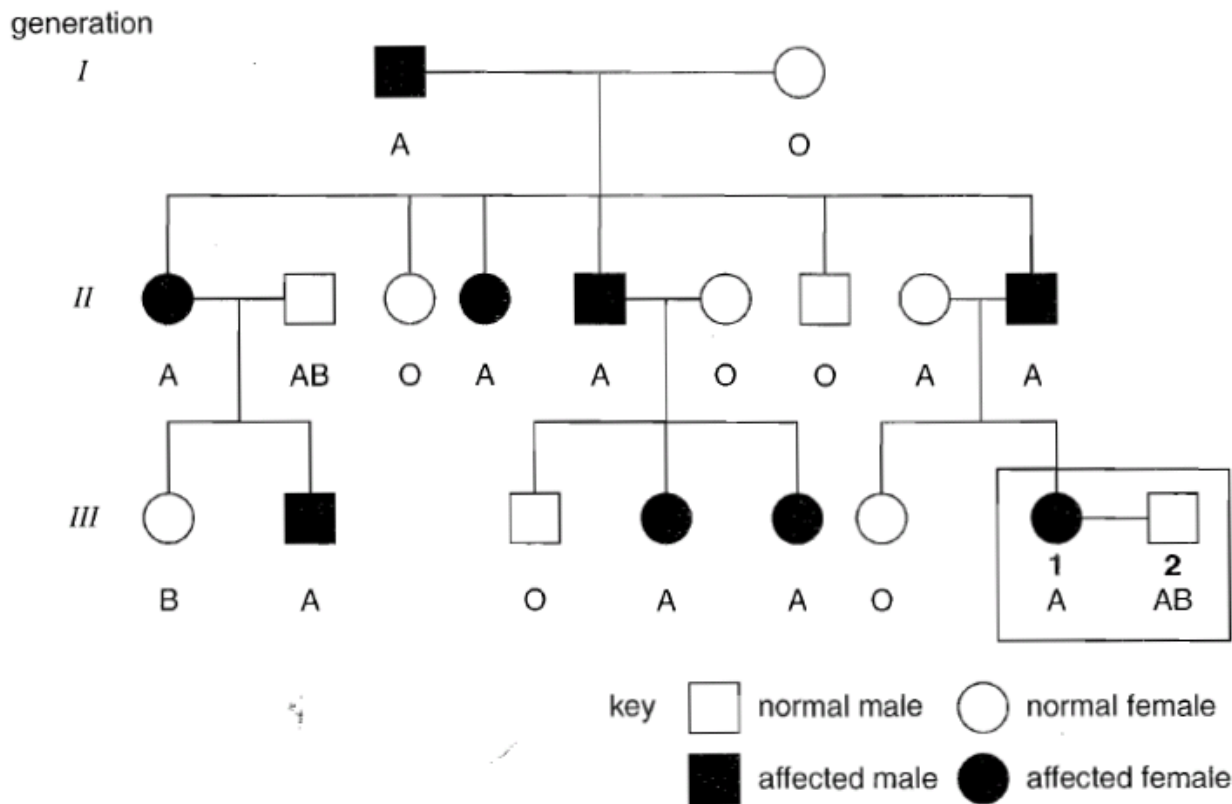


Fig. 4.1

- (a) (i) Using the symbols  $N/n$  for the nail patella locus and  $I^A / I^B / I^O$  for the ABO blood group locus, state the genotype of the parents in **generation I**.

male .....

female .....

[4]

- (ii) State **and** explain the type of inheritance shown by nail patella syndrome.

.....  
 .....  
 .....  
 .....  
 ..... [3]

5 The gene that causes nail patella syndrome is linked to the **ABO** blood group gene. This is one of the few examples of autosomal linkage in humans that is well understood.

**As known examples of autosomal linkage in humans are very rare, an imaginary example has been used in the following questions.**

It has been proposed that individuals who have a long index finger (locus **D/d**) also have a short big toe (locus **T/t**).

An individual with a long index finger and a short big toe has five children with a partner who has a normal length index finger and a normal length big toe.

All their children have a normal length index finger and a normal length big toe.

(b) State whether the allele for a long index finger is dominant or recessive. Give a reason for your answer.

allele .....

reason .....

.....

.....[2]

(c) Using the symbols **D/d** for the index finger length locus and **T/t** for the toe length locus,

(i) state the genotype of the children;

.....[2]

(ii) state the possible genotypes of the parent with a normal length index finger and a normal length big toe.

.....[2]

- (d) Researchers investigated a large sample of people with the heterozygous genotype for both index finger and big toe length. Each person had children with a partner who had a long index finger and a short big toe.

The number of children with each phenotype is shown below.

- long index finger and a short big toe 83
- long index finger and a normal big toe 21
- normal index finger and a short big toe 20
- normal index finger and a normal big toe 80

The ratio of phenotypes for unlinked loci is predicted to be 1 : 1 : 1 : 1

The chi-squared ( $\chi^2$ ) test was conducted on these data, giving a value for  $\chi^2$  of 73.06.

Table 5.1

degrees of freedom	probability (p)				
	0.10	0.05	0.02	0.01	0.001
1	2.71	3.84	5.41	6.64	10.83
2	4.61	5.99	7.82	9.21	13.82
3	6.25	7.82	9.84	11.35	16.27
4	7.78	9.49	11.67	13.28	18.47

- (i) Use the value of  $\chi^2$  and Table 5.1 to find the probability of these results differing from the expected ratio by chance.

.....[1]

- (ii) State the conclusions that may be drawn from the probability found in (d)(i).

.....  
 .....  
 .....  
 .....[3]

- (iii) The difference between the observed and expected results in this investigation occurs because the loci for index finger length and big toe length are on the same chromosome (linked).

Explain by means of a genetic diagram, how the actual results of these crosses occurred.

