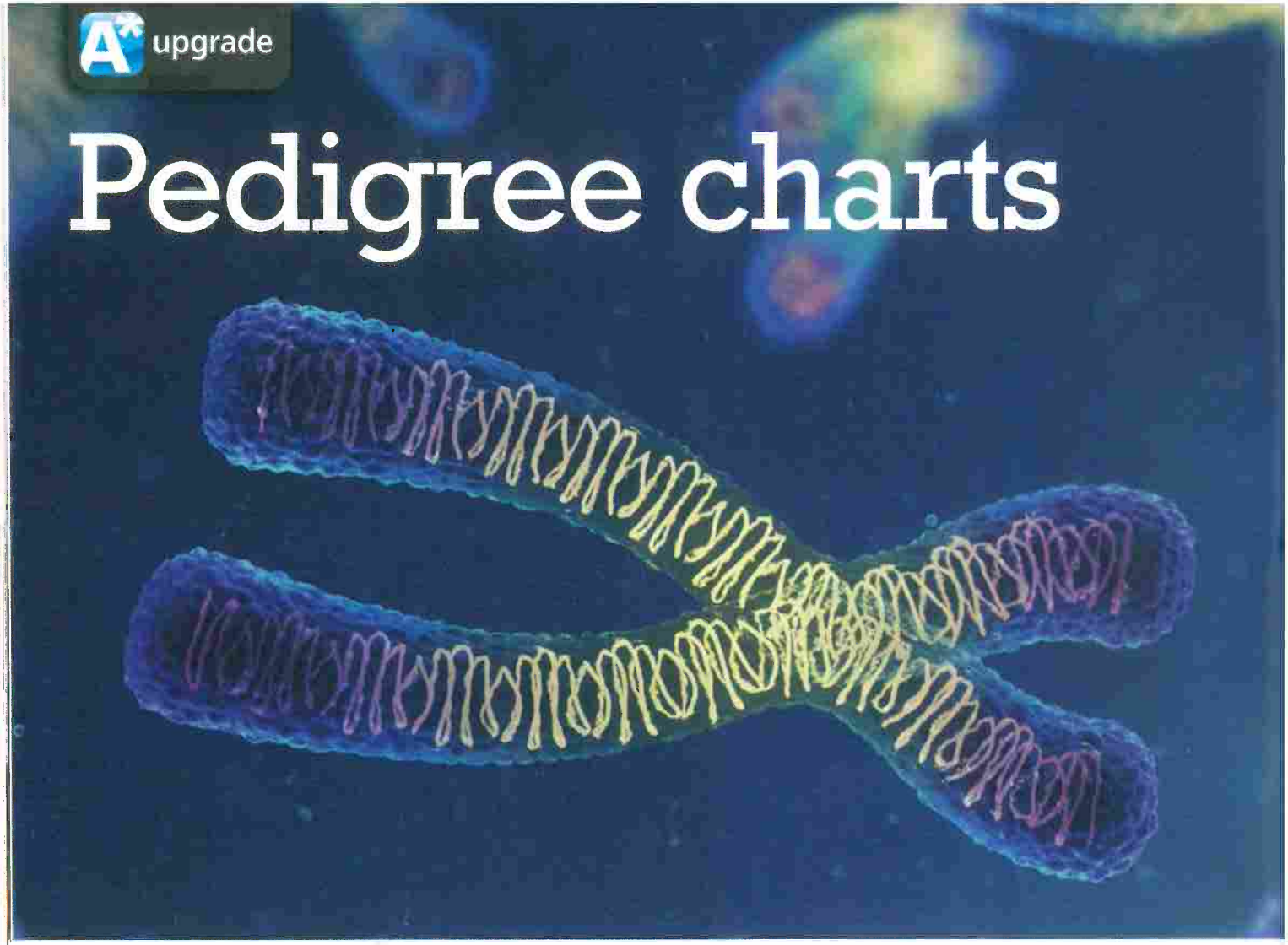
upgrade

Pedigree charts



A-level teachers Kim Stafford and Suzanne Foulds help to unravel the mysteries behind the pedigree chart — a topic often misunderstood by A-level students

Key words

Inheritance
Pedigree chart
Autosomal
Sex-linked

A diagram that shows the occurrence of a particular **phenotype** in a family is called a pedigree chart. It indicates the pattern of inheritance of the alleles of the gene that code for the phenotype from one generation to the next. Exam questions using pedigree charts have proved tricky for students in the past with, on average, less than 20% of students being awarded full marks. In this *Upgrade*, we look at pedigree charts showing the inheritance of different phenotypes and indicate how you can use the information in these charts to answer exam questions.

Pedigree charts are usually used to show inheritance of human diseases. A pedigree chart represents a male by a square and a female by a circle. The symbols are either shaded (has the phenotype) or not (lacks the phenotype in question) — for example, whether an individual suffers from a genetic disease or not. Relationships in pedigree charts are shown by a series of lines (see Figure 1).

In exam questions you might be required to identify and explain the evidence that shows whether the gene involved is **autosomal** or **sex-linked**, and which alleles of the gene are **dominant** and **recessive**. You might also be asked to predict the **genotype** of individuals or calculate the probability of the next offspring having a particular phenotype.

Autosomal recessive condition

Tay-Sachs disease is an inherited human disorder that causes progressive damage to the nervous system. The absence of an enzyme leads to the build-up of molecules called gangliosides, which stops the cells in the brain from functioning. Sufferers usually die during childhood. The allele for Tay-Sachs disease, **t**, is recessive to allele T, present in unaffected individuals. Figure 2 shows the inheritance of Tay-Sachs in one family.

In an exam question, you might be asked to explain the evidence to show that the allele causing Tay-Sachs is recessive and not dominant. There are two ways to tackle this problem.

- Look for two unaffected parents who have an affected child. In this example, neither parent 3 nor 4 is affected by Tay-Sachs, but one of their sons (individual 9) and one of their daughters (individual 11) both suffer Tay-Sachs. This can only be explained if the allele causing Tay-Sachs is recessive.
- Turn the question around and look for evidence that it cannot be dominant. In this example, assume the Tay-Sachs allele is dominant, in which case all unaffected people

would be **homozygous** recessive. If parents 3 and 4 were homozygous recessive, all their children would also be homozygous recessive, so would not have the condition. The fact that individuals 9 and 11 do have Tay-Sachs now provides evidence that your assumption was wrong and that the allele for Tay-Sachs is recessive.

Whichever method you use, you must give the evidence that you have found in your answer, i.e. parents 3 and 4 have children 9 and 11. You must then explain how this evidence supports the fact that the allele in question is recessive, i.e. both parents must be **heterozygous** and each have passed on a recessive allele to individuals 9 and 11.

In the same question, you might also be asked to provide evidence that the gene is not sex-linked. So using the second technique above, let's assume it is sex-linked (but still recessive). Since he does not show the condition, the genotype of individual 3 would then be X^TY and, since she inherited her father's X chromosome, the genotype of individual 4 would be X^TX^t. Using a Punnett square, you could then find the genotypes of the children these parents could have.

	X^T	X^t
X^T	X^TX^T	X^TX^t
Y	X^TY	X^tY

None of the female children could have the condition

From this Punnett square, you can see that the only possible female genotypes would be X^TX^T or X^TX^t, i.e. neither would suffer Tay-Sachs. However, individual 11 is a girl with Tay-Sachs, which shows that the gene cannot be sex-linked. This would be your evidence, but how would you explain this in your answer? If it was sex-linked, father 3 would pass on the dominant healthy allele on the X chromosome, so none of the daughters would be affected. This is not the case since daughter 11 has the condition.

Terms explained

Autosomal A gene carried on any chromosome that is not a sex chromosome.

Dihybrid inheritance The inheritance of two characteristics coded for by genes that are located on different pairs of homologous chromosomes.

Dominant allele An allele that is always expressed in the phenotype, usually represented using an upper case letter.

Genotype The alleles of the gene(s) controlling a particular feature.

Heterozygous A genotype with two different alleles of a single gene.

Homozygous A genotype with the same alleles of a gene.

Phenotype Measurable characteristic shown by an individual, resulting from an interaction between the genotype and environmental factors.

Recessive allele An allele that is expressed in the phenotype only in the absence of the dominant allele, usually represented using a lower case letter.

Sex-linked gene A gene carried on a sex chromosome.

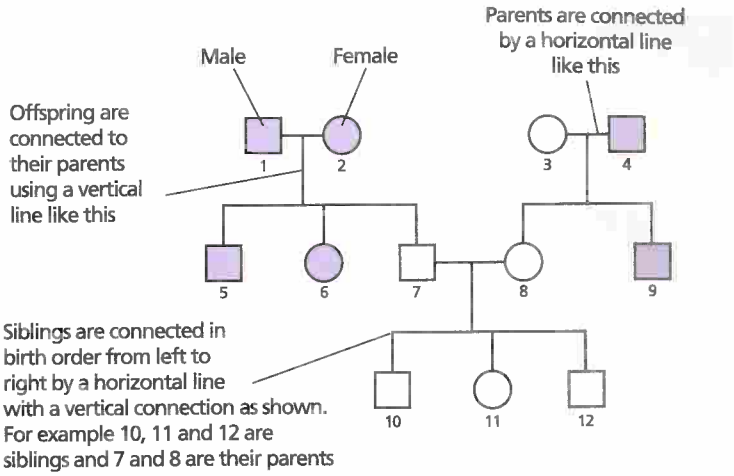


Figure 1 A pedigree chart showing the common denotations used to display the inheritance of a specific phenotype

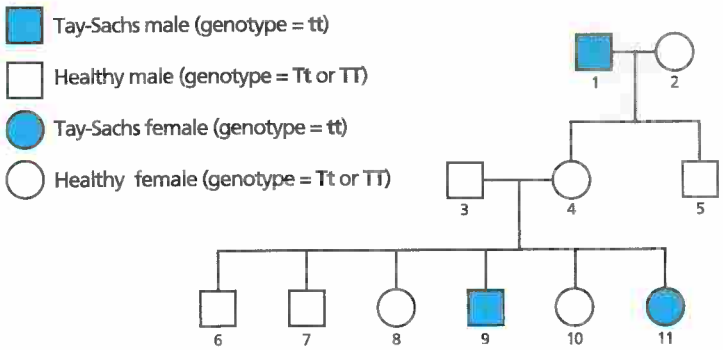


Figure 2 A pedigree chart to show the inheritance of Tay-Sachs disease in a family

Autosomal dominant condition

The gene for the Rhesus blood group (for further information see Biological Sciences Review, Vol. 28, No. 2, pp. 6–9) has two alleles. The allele for Rhesus positive, R, is dominant and that for Rhesus negative, r, is recessive. The pedigree chart in Figure 3 shows the inheritance of the Rhesus blood group in one family.

Our first question asks for evidence that the allele for Rhesus positive is dominant. The evidence for this is quite easy to spot, but if you are unsure then follow one of the techniques we have been through. Let's assume that the allele for Rhesus positive is recessive. If so, this would

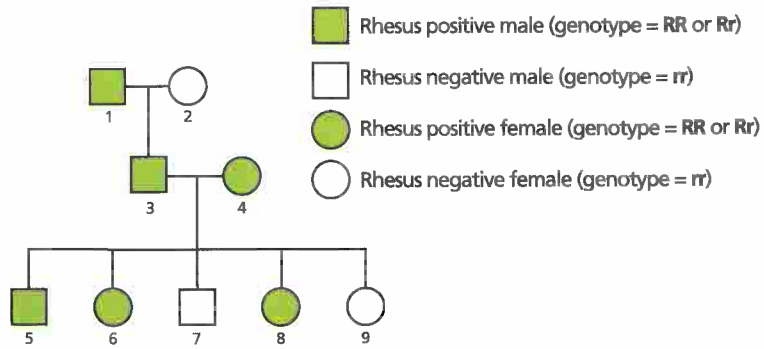


Figure 3 A pedigree chart showing the inheritance of blood groups

Further reading

O'Dell, K. (2015) 'Grandma's secret: a genetics puzzle', Biological Sciences Review, Vol. 27, No. 3, pp. 31–33.

mean that individuals 3 and 4 would both have the genotypes **rr**. This means all of their offspring would have the genotype **rr**, so all their children would be Rhesus positive. .Since individuals 7 and 9 are Rhesus negative, the assumption cannot be true and the allele for Rhesus positive must be dominant. So the explanation would be that the parents must be heterozygous and so must both pass on the recessive allele for Rhesus negative to individuals 7 and 9.

The question might also ask for evidence that the gene for the Rhesus blood group is not sex-linked. Again, using our method, let's use parents 3 and 4 and assume that the gene is on the X chromosome. If so, female 4 would have the genotype X^RX^r or X^RX^R and male 3 would have the genotype X^RY. Let's use a Punnett square to find the genotypes of the offspring these parents could have.

	X^R	X^r
X^R	X^RX^R	X^RX^r
Y	X^RY	X^rY

All the female children will be Rhesus positive

You can see from the Punnett square that all the females would be Rhesus positive. So if the gene was sex-linked, parents 3 and 4 could not have a daughter with the genotype of individual 9. This would be your evidence and the explanation would be that the gene could not be sex-linked or the father 3 would pass on the dominant allele to his daughters, so they would all be Rhesus positive.

Question 1: There is another piece of evidence showing that the allele for Rhesus positive is not sex-linked. Can you find this evidence and explain it?

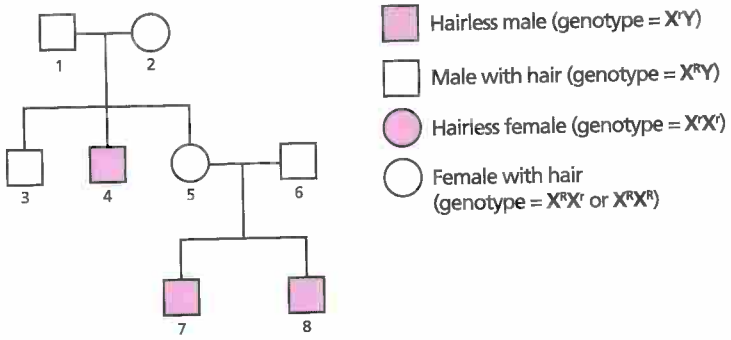


Figure 4 A pedigree chart showing the inheritance of hairlessness in beef cattle

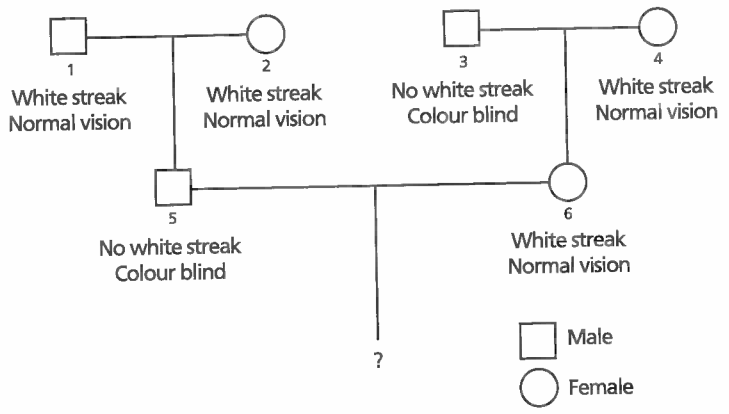


Figure 5 A pedigree chart showing the inheritance of colour blindness and the presence of a white streak in human hair

Sex-linkage

A single gene controls the presence of hair on beef cattle. The gene is carried on the X chromosome. Its dominant allele, R, causes hair to be present and its recessive allele, r, causes hairlessness. The pedigree chart in Figure 4 shows the pattern of inheritance of these alleles in a group of cattle.

We can use this pedigree chart to show that the allele for hairlessness is recessive. Let's assume that the allele for hairlessness is dominant, in which case parents 1 and 2 would have the genotypes X^rY and X^rX^r, respectively. As a result, all the offspring would have hair; as you can see individual 4 does not. This is the evidence, and the explanation would be that the mother must be heterozygous (remember, with sex-linkage in mammals, males cannot be heterozygous) and have passed on the recessive allele to individual 4.

Question 2: Explain the other piece of evidence from the chart that shows hairlessness is controlled by a recessive allele.

Pedigree charts involving dihybrid inheritance

Dihybrid inheritance charts are much more challenging because they take into account two different phenotypes. Colour blindness in humans is controlled by a gene on the X chromosome. The allele for colour blindness, X^b, is recessive to the allele for normal colour vision, X^B. Another gene controlling the presence of a white streak in the hair is not sex-linked, with the allele for the presence of a white streak, H, being dominant to the allele for the absence of a white streak, h. The pedigree chart in Figure 5 shows the inheritance of these phenotypes.

You should be able to analyse and interpret this chart as you would with the others. Just take each phenotype on its own and follow through the technique we have used above.

Question 3: Explain the evidence that shows the allele controlling the absence of a white streak in the hair is recessive.

Question 4: What are the genotypes of individuals 5 and 6?

Question 5: What is the probability that the first child of individuals 5 and 6 will be a colour-blind boy with a white streak in his hair?

Tips for pedigree charts

We have shown you some simple techniques to use so that you can work through examples on your own, to work out whether a gene is autosomal or sex-linked and which of its alleles is dominant or recessive. Showing your working on the exam paper is perfectly acceptable and will help you t

Table 1

Type of genetic inheritance	To show...	Evidence	Explanation
Autosomal recessive	The allele is recessive	Two unaffected parents have an affected child	The parents must be heterozygous (carriers)
	The allele is not sex linked	Two unaffected parents have an affected daughter	If the allele was sex-linked, the unaffected father would pass on the dominant allele on his X chromosome, so none of the daughters would be affected
Autosomal dominant	The allele is dominant	Two affected parents have an unaffected child	Both parents must be heterozygous and pass on their recessive alleles
	The allele is not sex linked	Two affected parents have an unaffected daughter	If the allele was sex-linked, the affected father would pass on the dominant allele on his X chromosome, so all daughters would be affected

identify the evidence. If you are still struggling, the summary in Table 1 might help.

In some pedigree charts there will be evidence to show whether a gene is autosomal or sex-linked and its alleles dominant or recessive; in others there might be none. This depends on the size of the family tree and the phenotypes present. Having read this *Upgrade*, you should now be able to identify and explain evidence and work out genotypes of individuals displayed on a pedigree chart, an important technique in genetic research today and a tool often used in genetic counselling.

BiologicalSciencesReview*Extras*

Go online for the answers to the questions in this *Upgrade*: www.hoddereducation.co.uk/biologicalsciencesreviewextras

Kim Stafford and **Suzanne Foulds** are experienced A-level examiners. They are A-level biology teachers based at Xaverian College and lecture on The University of Manchester Life Science Foundation Year programme.

Disruptive selection

Imagine you are an eagle, soaring over your northern Canadian homeland. The snow beneath you is peppered by patches of dark brown scrubland. Rabbits are your favourite meal, and there are plenty. They vary in colour from brilliant white to a dark brown, and every shade between.

You rarely get to taste the white ones though — they are so well camouflaged on top of the snow that you hardly ever see them. Nor do you get to enjoy the dark brown ones — invisible amid the bushes. But the rabbits that are intermediate in colour? You will have eaten many of these. They simply cannot hide from you — they are too dark to hide on the snow, and too light to be hidden among the bushes.

How do you think natural selection might affect this population of rabbits? Clearly the white and dark brown rabbits — the extreme phenotypes — have a better chance of avoiding being eaten by eagles. They should, therefore, be selected over the intermediate phenotypes, which we would expect to become less common. This would be disruptive, or diversifying, selection, because it causes the population to split into two (or sometimes more) distinct phenotypes (see Figure 1), both of which have a better chance of survival than any phenotype in between. This is very different from stabilising selection (see p. 5 this issue), which selects for just one phenotype — the optimum for survival.

Although disruptive selection is rare, it is one of the main causes of sympatric speciation (see BIOLOGICAL SCIENCES REVIEW, Vol. 28, No. 3, p. 19). The two or more phenotypes may start to occupy different ecological niches (in this case some in snow-covered habitat, others in scrubland). Even though they are in the same geographical area, they may not come into contact with each other again to breed. They would then be said to be reproductively isolated, and could evolve into separate species.

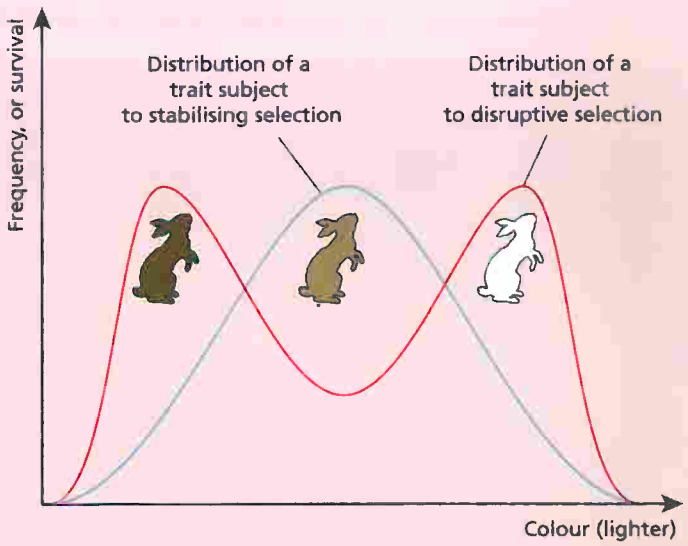


Figure 1

Jonny Miller