Pedigree possibilities

Cystic fibrosis is a genetic disease caused by a range of **recessive alleles** that are estimated to be carried by one in 25 people in Australia. Sufferers of cystic fibrosis have thickened body fluids, experience difficulty in breathing, and have poor digestion. In families with a known history of the disease, genetic counselling can offer advice on the chance of passing the condition on to children.

What are the chances?

If two healthy people have a child who is affected by cystic fibrosis, both parents must be **carriers** of the recessive alleles (call them c), which cause the disease. They must also have a normal **allele**, C, because they do not have the disease themselves, so their **genotypes** are both Cc. Knowing the genotype of both parents makes it possible to determine the possible genotypes and **phenotypes** of the offspring.

When the mother produces ova and the father produces sperm, each could contain either the c or C allele. This is best shown in a table called a **punnett square**.

Mother's genotype: Cc
Father's genotype: Cc

		Mother	
	Possible gametes	C	c
Father	C	CC normal	Cc normal
	c	Cc normal	cc cystic fibrosis

A punnett square is used to predict the outcomes of a cross. In this one, the top row has the possible genotypes of **gametes** from one parent, and in the left column the possible gametes from the other. *Remember, each gamete has only one set of* ***chromosomes*** *so it can only contain one allele.* In the body of the table, the genotype that could be formed from each egg and sperm combination is written, and underneath is the resulting phenotype. In this case the parents have, for each birth, a one in four chance of having an affected child.

If a female with blood type AB has children with an O blood-group male, what blood groups could the children have?

Possible egg genotypes

Possible sperm genotypes	Eggs / Sperm	IA	IB
	i	IAi A blood	IBi B blood
	i	IAi A blood	IBi B blood

Each child has a 50/50 chance of having A-type blood or B-type blood.

	X^c	X
X	XX^c	XX
Y	X^cY	XY

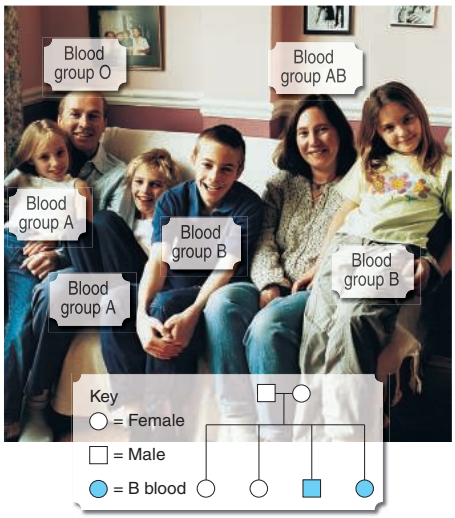
The possible children of a woman carrying the allele for colour-blindness and a normal male are shown in this punnet square for a sex-linked recessive condition. Note that sex-linked alleles are denoted as X with a superscript c. Her sons have a 50% chance of inheriting the condition; her daughters will all have a normal phenotype, but half could be carriers of the condition.

For some diseases it is no longer necessary to rely on probabilities. Genetic tests can be used to screen for the alleles involved in certain genetic diseases, so that people at risk can find out whether they have inherited them. In some cases people can then take steps to reduce the risk of developing the disease, and can be aware of the chance of passing the condition on to their children.

Pedigrees

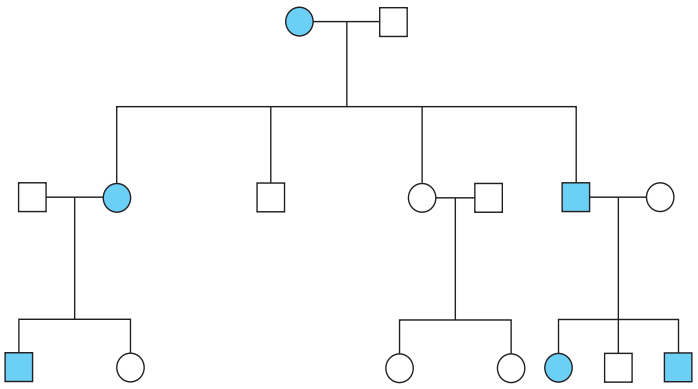
Pedigrees are family tree diagrams that chart whether a characteristic is present. A pedigree can sometimes also help determine if a characteristic is dominant or recessive.

- A carrier shape may be half-shaded or have a little 'c' written in it.
- A horizontal line between a circle and a square represents a mating pair.
- The vertical lines represent the offspring, also called *progeny*.

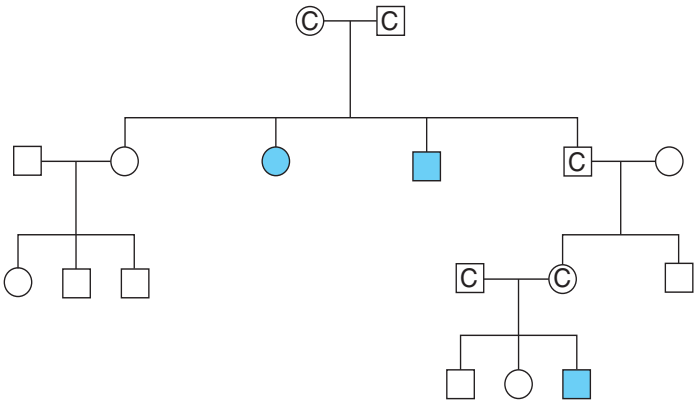


Sex-linkage in pedigrees

Look at the pedigree of Queen Victoria on the previous page. Her sons each had a 50% chance of inheriting haemophilia from her, while her daughters had a 50% chance of being carriers. The disease is able to skip a generation and males are more commonly affected. This is a typical pedigree for a recessive **sex-linked condition**.



The above diagram shows a possible pedigree pattern for the inheritance of Huntington's disease, a dominant condition. Each child of an affected parent has a 50% chance of being affected; so approximately half will be affected and half won't.



The above diagram shows a possible pedigree pattern for a recessive gene such as cystic fibrosis. Two unaffected parents can produce affected children. The parents in this case are carriers.

Activities

REMEMBER

1 Copy the table below. Using an arrow, link each of these with their correct representation in a pedigree:

Male	marriage
Female	shaded
Affected	small c or half-shaded
Carrier	square
Not affected	offspring
Horizontal line	circle
Vertical line	left unshaded

CALCULATE

2 Mendel had pure-breeding yellow seed (a dominant character Y) and pure-breeding green seed (recessive y) pea strains.

(a) Draw up a punnett square representing a cross between heterozygous yellow seed peas.

(b) What are the ratios of the genotypes and phenotypes of the offspring?

THINK

3 Copy and complete the missing labels in the following punnett square:

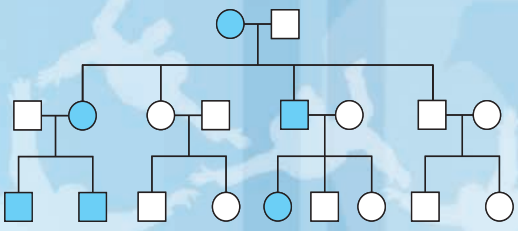
Genotypes of parents: father _____, mother _____

	Mother's _____	
	i	IA
IA		
IB		

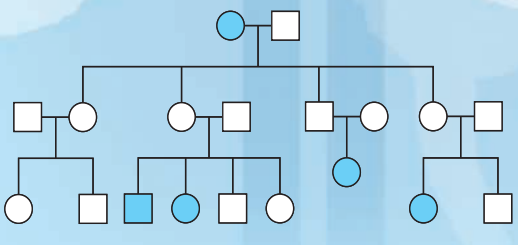
Blood types of the children:
50% are _____, 25% are _____, 25% are _____

4 Are the traits shown in the pedigrees below dominant or recessive? Explain your reasoning.

(a)



(b)



(c) Mark any carriers on each family tree.

5 If Bill with O-type blood married Jill with B-type blood, what are all the possible genotypes and phenotypes of their children? (*Hint:* Jill could have two different genotypes, so use two different punnet squares to work it out.)

6 A dominant red-flowered plant with genotype RR is crossed with a recessive white-flowered plant with genotype rr.

(a) What are the genotypes and phenotypes of the plants raised from their seeds?

(b) Use a punnet square to perform a cross between two heterozygous red plants with genotype (Rr). What are the ratios of the different phenotypes?

learning

I CAN:

- ☐ fill in a punnet square
- ☐ use a punnet square to predict phenotypes
- ☐ produce a pedigree chart for three generations of a family
- ☐ understand how sex-linked conditions are passed on.