Education Services Ltd

EB Education Revision Guide

How to work with Genetics: Part 2

...helping everybody achieve Even Better

Genetic Diseases

Cystic Fibrosis

Cystic fibrosis is a genetic disease which is caused by a recessive allele (represented by **f**).

It creates a thick, sticky mucus and mainly affects the lungs and pancreas.

Someone who is homozygous recessive (**ff**) for the disease will develop cystic fibrosis and suffer symptoms of this disease.

Someone who is homozygous dominant (**FF**) or heterozygous (**Ff**) will not develop cystic fibrosis.

This means that in order to inherit the disease, the child must inherit a recessive allele from both parents.

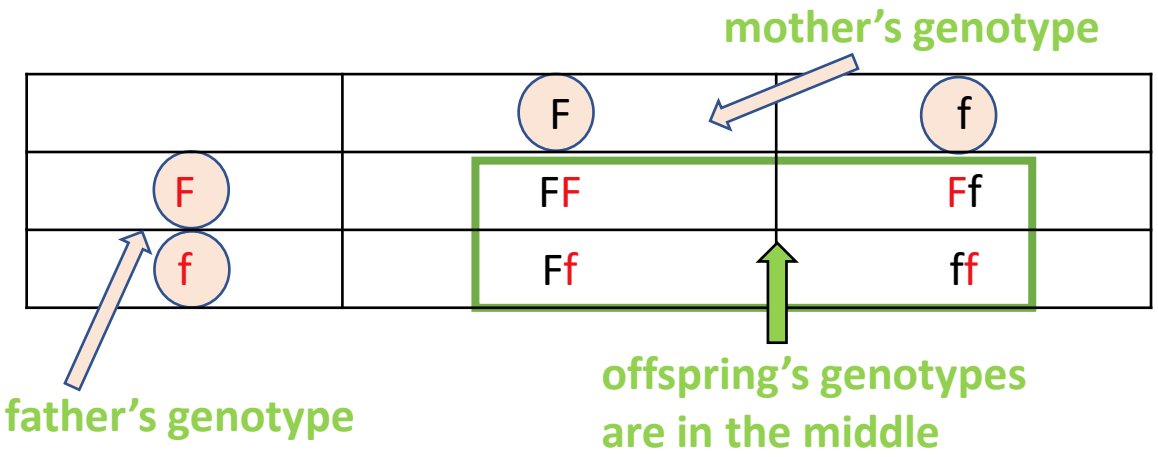
People who are heterozygous (**Ff**) are known as **carriers** – this is because they carry the recessive allele and can pass it on, but do not develop the disease.

The Punnett square below shows that if both parents are heterozygous, and therefore carriers for cystic fibrosis, there is:

25% chance of a child having cystic fibrosis

50% chance of a child being a carrier

25% chance of a child being unaffected and not a carrier



Education Services Ltd

Family Pedigrees

What is it?

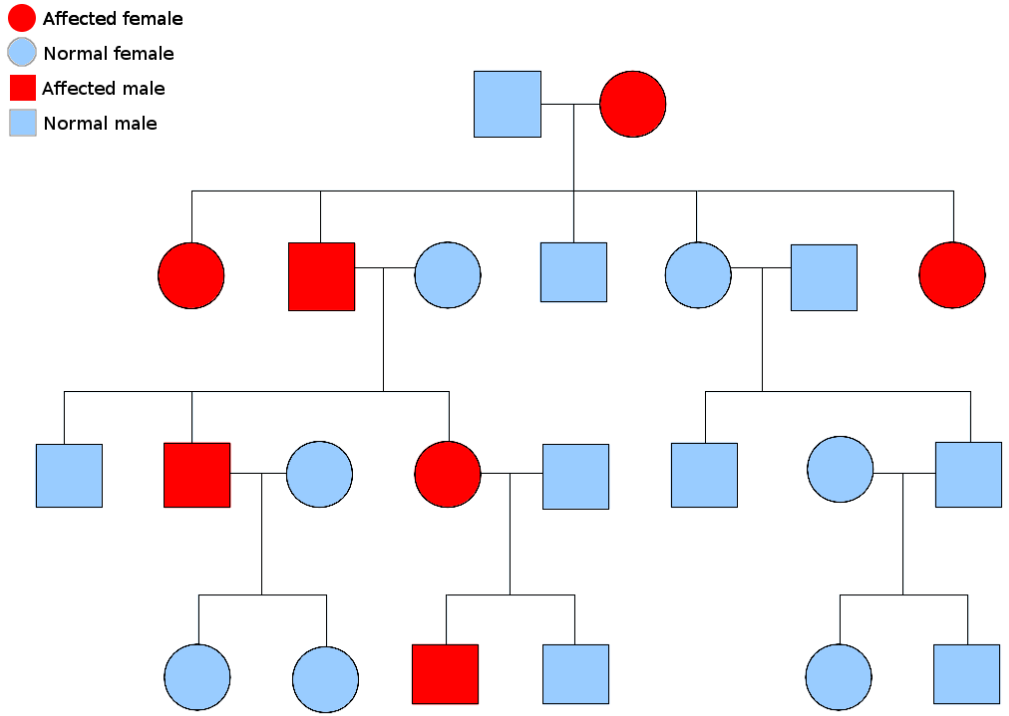
A family pedigree diagram can be used to demonstrate how alleles are passed through generations of a family.

Horizontal lines link parents to each other and vertical lines link parents to their children.

Females are represented with circles and males with squares.

You can use the key to identify who is affected and who is not.

Example 1:



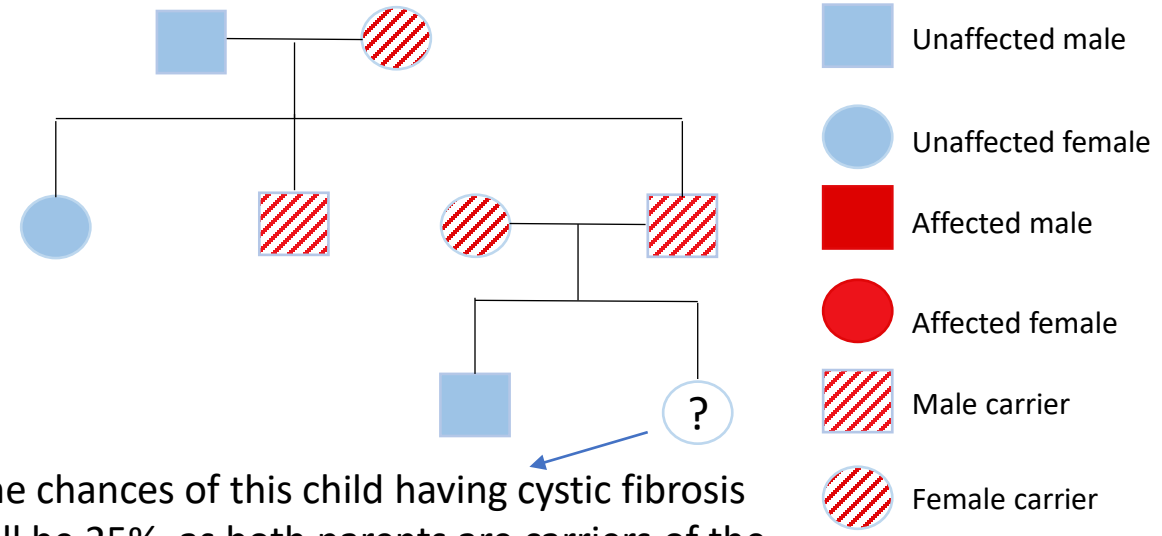
Family Pedigrees

What is it?

This example shows a family pedigree for a family with members who are carriers of cystic fibrosis.

This chart shows that cystic fibrosis is a recessive disorder – as individuals can carry the allele but do not have the disorder.

Example 2:



The chances of this child having cystic fibrosis will be 25%, as both parents are carriers of the allele, and are therefore heterozygous (**Ff**). Possible genotypes of the offspring are **FF, Ff, Ff, ff**.

Sex-Linked Genetic Disorders

What are they?

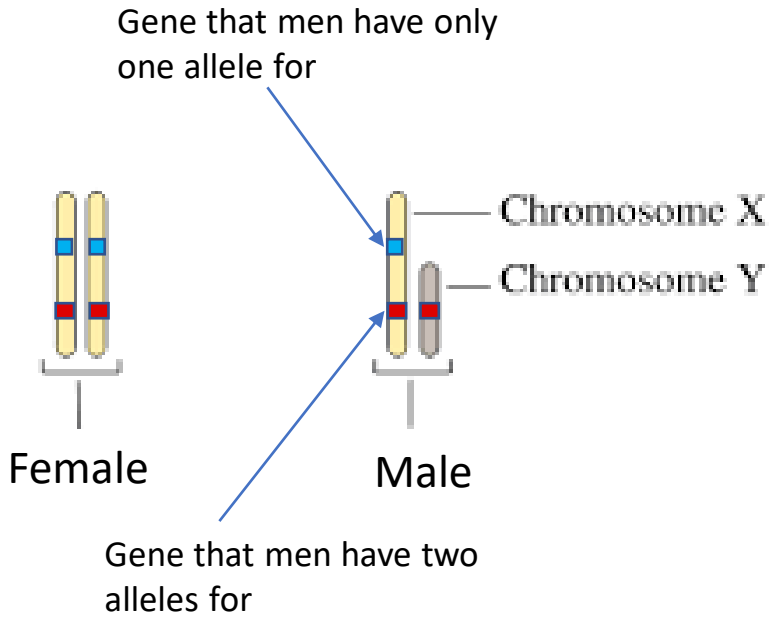
Males are more likely to inherit some genetic disorders. These are known as sex-linked disorders.

If an allele coding for a characteristic is found on a sex chromosome (X or Y), it is sex-linked.

Most genes on the sex chromosomes are found on the X-chromosome, because the Y chromosome is much smaller, and so does not have as many genes.

As the genotype of men is XY – they only have one X chromosome. This means they often only have one allele (on the one X chromosome) for sex-linked genes.

This makes it more likely for males to express this characteristic if it is recessive.



Sex-Linked Genetic Disorders

Colour blindness

A faulty allele found on the X chromosome causes colour blindness. The Y chromosome does not have an allele for colour vision.

When completing a Punnett square for sex-linked disorders, both the chromosome and the allele are included.

X and Y represent the chromosomes, N is the normal colour vision allele, n is the faulty colour vision allele.

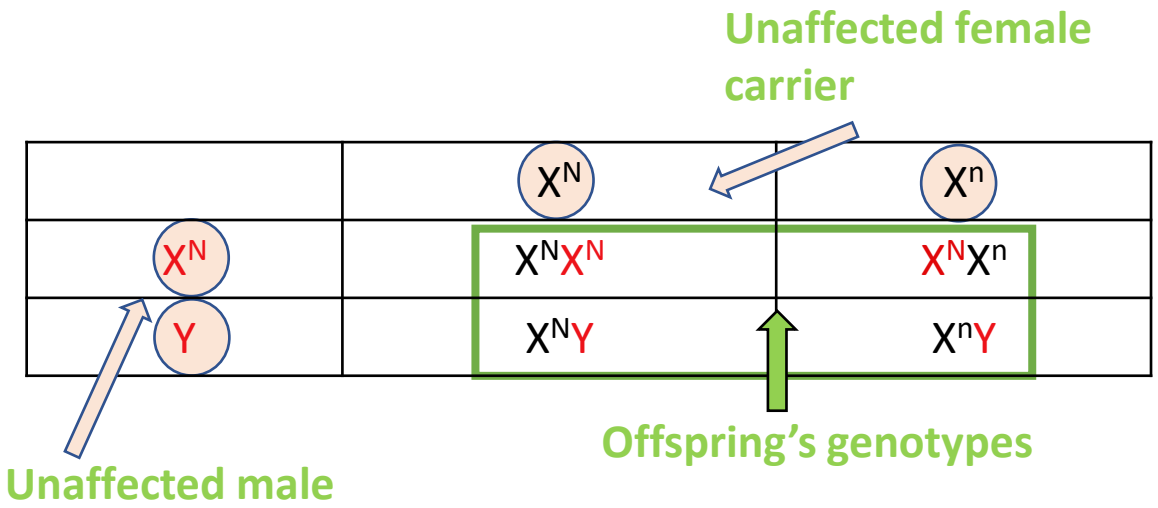
X^N = X chromosome with normal allele

X^n = X chromosome with faulty allele

Haemophilia is also a sex-linked disorder. This is a disease where blood does not clot properly, so sufferers of the disorder bleed for longer than usual if they cut themselves.

Diagram

25% of the offspring will be colour blind, a 3: 1 ratio. These will all be male.



Blood Groups

How do you inherit them?

Multiple alleles determine which blood group you inherit.

There are four different blood types in humans:

O A B AB

There are three different alleles for the gene determining blood type.

I^O I^A I^B

I^A and I^B are what is known as co-dominant. This means that neither of these alleles are dominant over the other. If a person has the genotype I^AI^B – they will express both alleles, and have the blood group AB.

I^O is a recessive allele. This means that to get blood type O – you need to have two recessive alleles, I^OI^O.

I^AI^O – this means you will be blood group A, as A is dominant over O.

I^BI^O – this means you will be blood group B, as B is dominant over O.

Mother's genotype:
Heterozygous
Blood group A

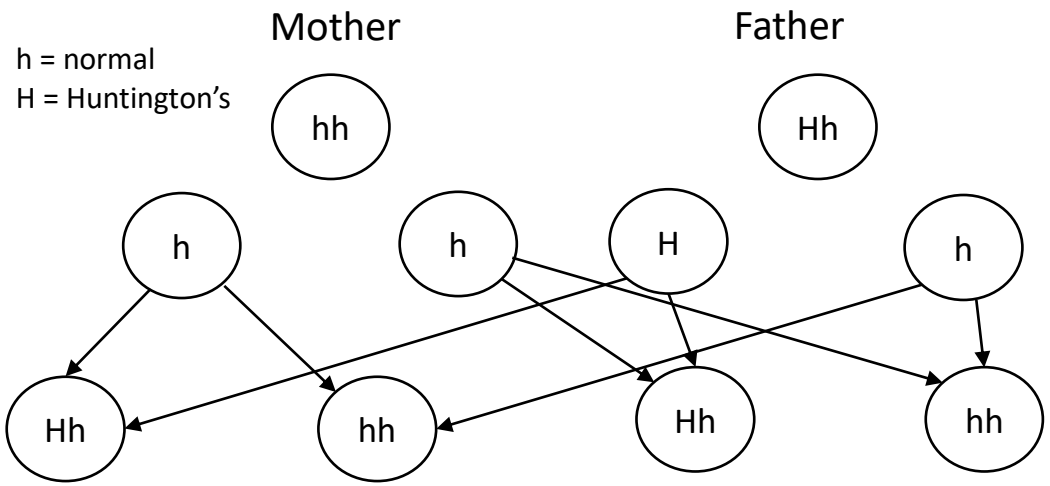
	I^A	I^O
I^B	I^AI^B	I^BI^O
I^O	I^AI^O	I^OI^O

Father's genotype:
Heterozygous
Blood group B

There is a 25% chance of a child having any of the four blood groups

Your turn:

1. Huntington's disease is an inherited disease.
The diagram below shows how it can be inherited.



a) Complete the following sentences.
Huntington's disease is caused by a …… allele.
Individuals suffering from Huntington's disease can have the …… HH or ……

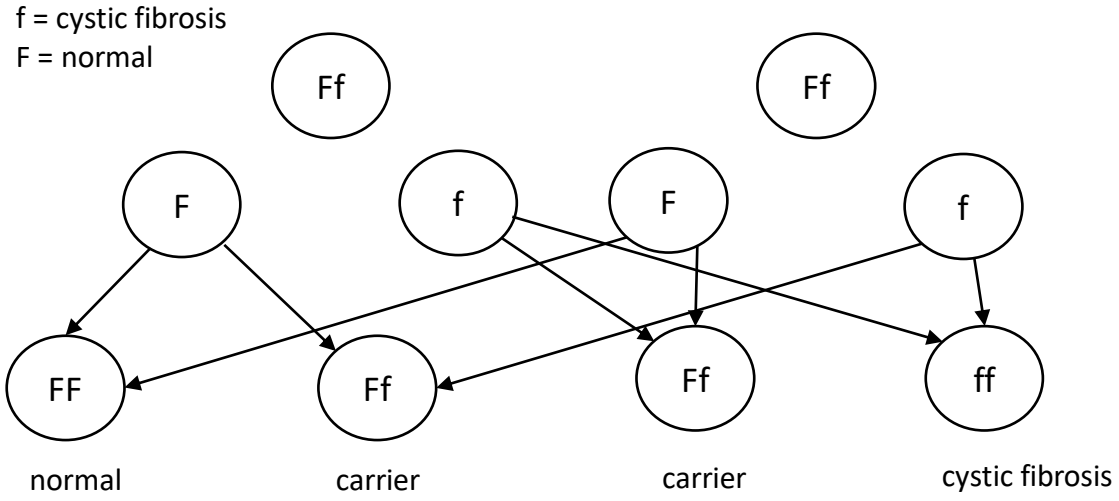
b) Complete the Punnett square below for two heterozygous parents.

c) Using the Punnett square calculate the probability of the offspring of these parents developing Huntington's disease.

……

Your turn:

d) Cystic fibrosis is also an inherited disorder. The diagram below shows how it is inherited.



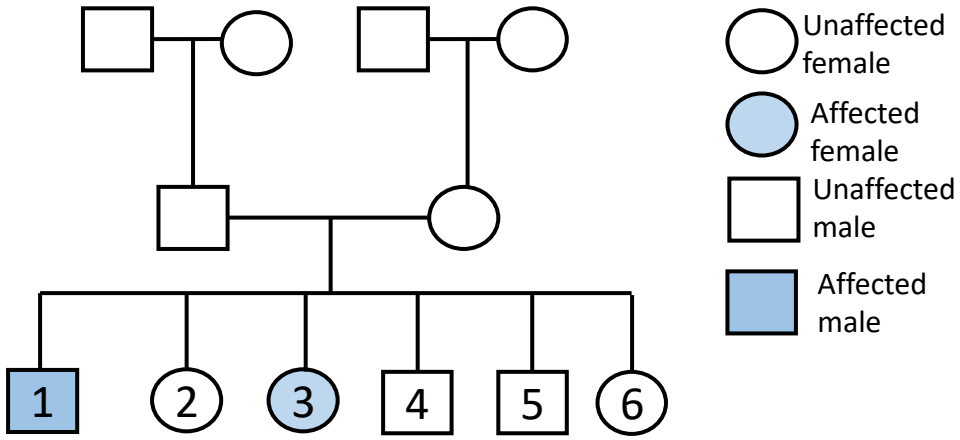
Even though both sets of parents are heterozygous, explain why the chance of inheriting Huntington's disease is greater than the chance of inheriting cystic fibrosis.

……

……

……

2. The family pedigree diagram below shows the inheritance of cystic fibrosis in a family.
Cystic fibrosis is caused by recessive alleles.



a) How many offspring in the 3rd generation have cystic fibrosis?

……

Your turn:

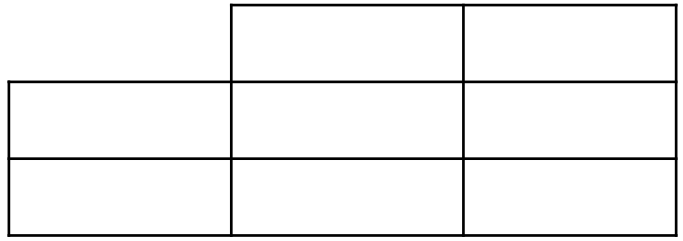
b) Which of the below is the correct description for individual 3 in the 3rd generation

i) Heterozygous for cystic fibrosis
ii) A carrier of the cystic fibrosis allele
iii) Homozygous recessive for cystic fibrosis
iv) Homozygous dominant for cystic fibrosis

c) Explain how offspring in the 3rd generation can inherit cystic fibrosis when their parents and grandparents do not suffer from the disorder.

……
……
……
……
……

c) Complete the Punnett square below to show the inheritance of cystic fibrosis in the 3rd generation.



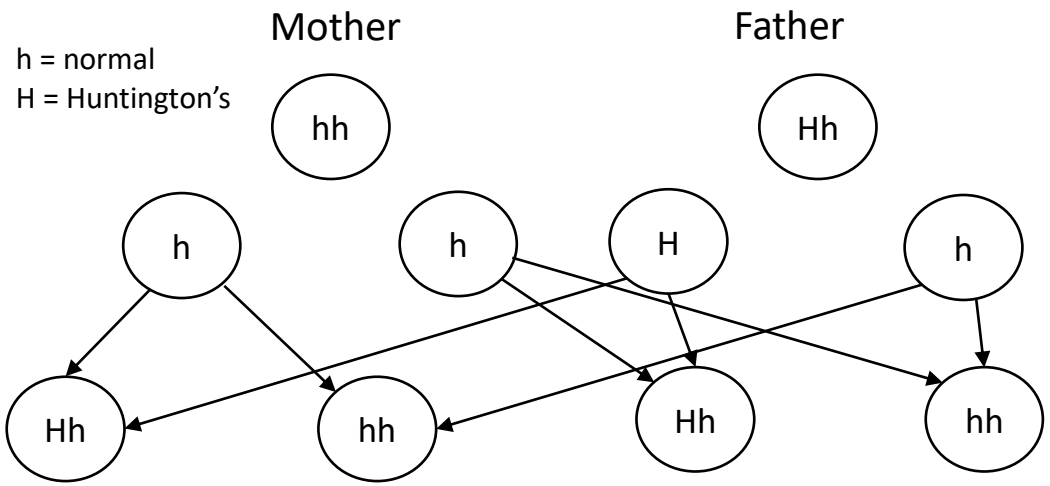
d) Explain why unaffected individuals in the 3rd generation would find a pedigree analysis useful.

……
……
……

Education Services Ltd

Answers:

1. Huntington's disease is an inherited disease.
The diagram below shows how it can be inherited.



a) Complete the following sentences.
Huntington's disease is caused by a dominant allele.
Individuals suffering from Huntington's disease can have the genotype HH or Hh

b) Complete the Punnett square below for two heterozygous parents.

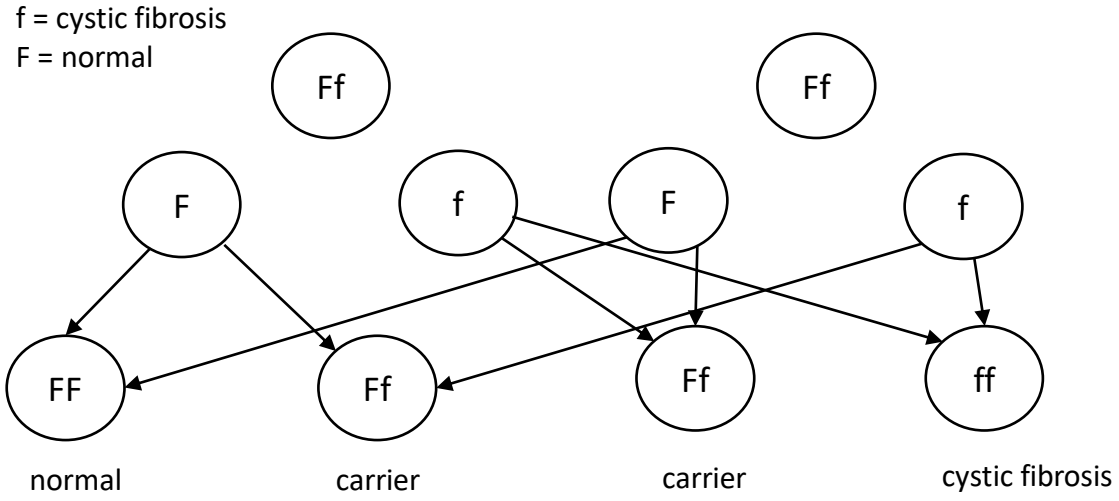
	H	h
H	HH	Hh
h	Hh	hh

c) Using the Punnett square calculate the probability of the offspring of these parents developing Huntington's disease.

75% chance

Answers:

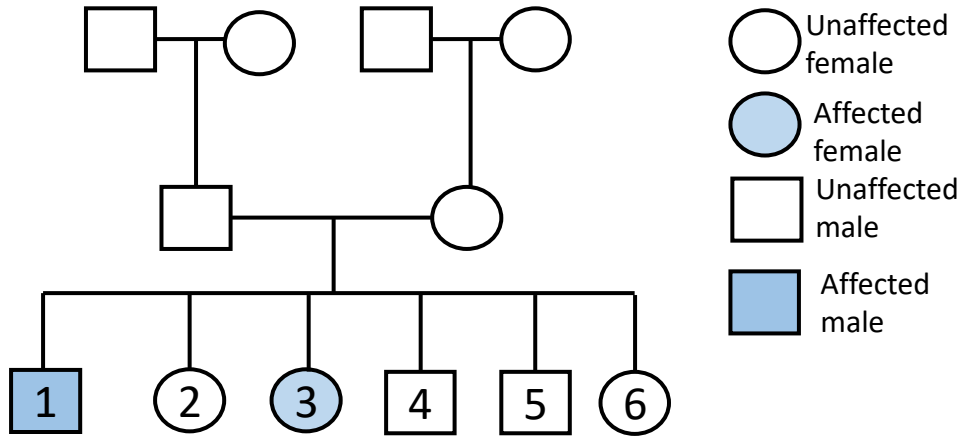
d) Cystic fibrosis is also an inherited disorder. The diagram below shows how it is inherited.



Even though both sets of parents are heterozygous, explain why the chance of inheriting Huntington's disease is greater than the chance of inheriting cystic fibrosis.

Huntington's is caused by a dominant allele, and cystic fibrosis by a recessive allele, therefore only 1 allele needs to be inherited to have Huntington's, whereas 2 alleles need to be inherited to get CF

2. The family pedigree diagram below shows the inheritance of cystic fibrosis in a family.
Cystic fibrosis is caused by recessive alleles.



a) How many offspring in the 3rd generation have cystic fibrosis?

Two

Education Services Ltd

Answers:

b) Which of the below is the correct description for individual 3 in the 3rd generation

i) Heterozygous for cystic fibrosis
ii) A carrier of the cystic fibrosis allele
iii) Homozygous recessive for cystic fibrosis
iv) Homozygous dominant for cystic fibrosis

c) Explain how offspring in the 3rd generation can inherit cystic fibrosis when their parents and grandparents do not suffer from the disorder.

Both parents in the 2nd generation must carry the recessive allele and be carriers. The offspring in the 3rd generation who inherit cystic fibrosis must have inherited a recessive allele from each parent to be homozygous recessive.

c) Complete the Punnett square below to show the inheritance of cystic fibrosis in the 3rd generation.

	F	f
F	FF	Ff
f	Ff	ff

d) Explain why unaffected individuals in the 3rd generation would find a pedigree analysis useful.

It can determine how likely it is that their offspring could inherit the CF allele. If they are carriers (heterozygous) there is a 50% chance they will pass on the gene. If both are heterozygous there is a 25% chance of offspring inheriting two recessive alleles.

Education Services Ltd

For more help and resources, or to work with us as a tutor, please contact us

www.ebeducationservices.co.uk

contact@ebeducationservices.co.uk

0161 442 5270

...helping everybody achieve Even Better