

# Genetic Diseases Notes

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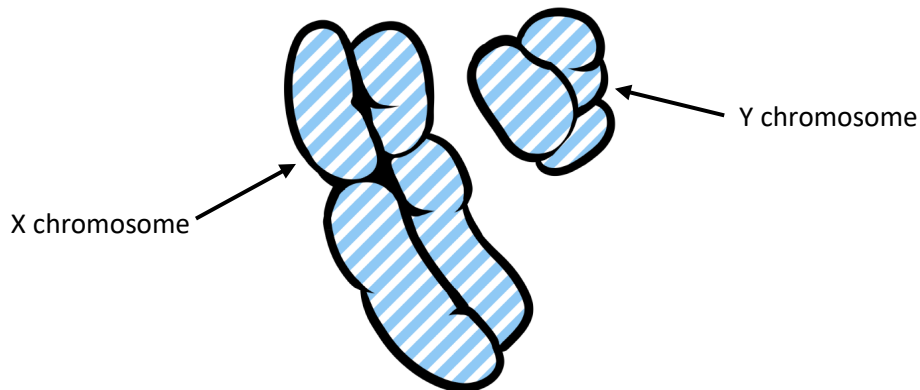
An individual can inherit a genetic disease from one or both of their parents depending on the gene involved. These work differently from diseases caused by microorganisms; they are not contagious. They are inherited, not caught.

The **two** genetic diseases covered in Edexcel Biology are both **sex linked**. What we mean by this is that the genes that cause issues are found on the 23<sup>rd</sup> pair of chromosomes; the X and Y chromosomes.

So, what's the going on with the sex chromosomes?

**The X chromosome is longer than the Y chromosome.**

There are more genes on the X chromosome, for example the genes for the **ability to see red and green** and the **ability to clot your blood** when you have a cut so that you don't bleed to death.



As you can see in the diagram above the X chromosome is indeed longer than the Y chromosome.

So, if there is a faulty gene on the lower part of the X chromosome than males **do not have a second copy**. This means they will get the disease if the gene is present.

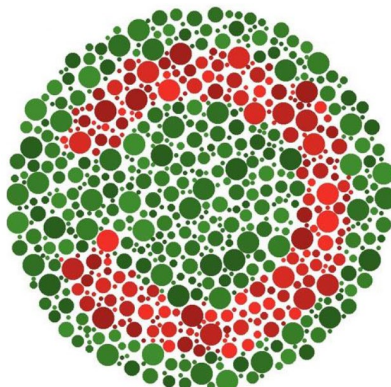
Females will have a second gene which may be dominant. This will protect them from the disease. They will just carry the gene, but are not affected. This is why both red green colour blindness and Haemophilia are both very rare in females.

That's the background, now let's look at both red green colour blindness and Haemophilia in detail.

**Red green colour blindness:** individuals who have this condition are unable to tell the difference between the colours red and green.

**Red green colour blindness is caused by a recessive allele on the X chromosome.**

Tests for this are quite easy to do, you may or may not have seen a diagram like the one below. Diagrams like this are used to determine if an individual has red green colour blindness.



The most common type of questions involving red green colour blindness are:

**Type 1:** Asking you to state what form of allele causes red green colour blindness.

**Type 2:** Stating on which chromosome the allele for red green colour blindness is found.

**Type 3:** A Punnet square or genetic diagram. Where you can be asked to draw either one and calculate the probability of one of the offspring from the cross having the condition.

The punnet square for sex linked conditions look a little different to the standard punnet squares. As the genes are on the X chromosome, we write the sex chromosomes as the gametes.

We show the allele on the X chromosome as a power.

See the diagram below for the cross between an unaffected male and a heterozygous carrier female

|          |       | Parent 1  |           |
|----------|-------|-----------|-----------|
| Parent 2 |       | $X^R$     | $X^r$     |
|          | $X^R$ | $X^R X^R$ | $X^R X^r$ |
|          | Y     | $X^R Y$   | $X^r Y$   |

As you can see, there is a 50 : 50 chance of a male having red green colour blindness.

## Haemophilia

**Haemophilia:** people with this condition are **unable to clot their blood resulting in excessive bleeding**, even from small cuts or bruises, this can be fatal.

**Haemophilia is caused by a recessive allele on the X chromosome. Haemophilia is more common in males.**

The most common type of questions involving Haemophilia are:

**Type 1:** Asking you to state what form of allele causes Haemophilia.

**Type 2:** Stating on which chromosome the allele for Haemophilia is found.

**Type 3:** A Punnet square or genetic diagram. Where you can be asked to draw either one and calculate the probability of one of the offspring from the cross having the condition.

So yes, quite similar to the style of questions to red green colour blindness.

See the diagram below for the cross between a homozygous dominant individual and a heterozygous carrier.

|          |       | Parent 1  |           |
|----------|-------|-----------|-----------|
| Parent 2 |       | $X^H$     | $X^h$     |
|          | $X^H$ | $X^H X^H$ | $X^H X^h$ |
|          | Y     | $X^H Y$   | $X^h Y$   |

As you can see from the Punnet Square:

1. There is a 0% chance of a female having Haemophilia if the father is not affected and the mother is a carrier of the allele.
2. There is a 50% chance of a male child being born with Haemophilia if the father is not affected and the mother is a carrier of the allele.
3. The father would have to have Haemophilia and the mother would have to be a carrier to produce females with Haemophilia.

In the section below we have included notes on the genetic conditions of Cystic fibrosis and Polydactyly.

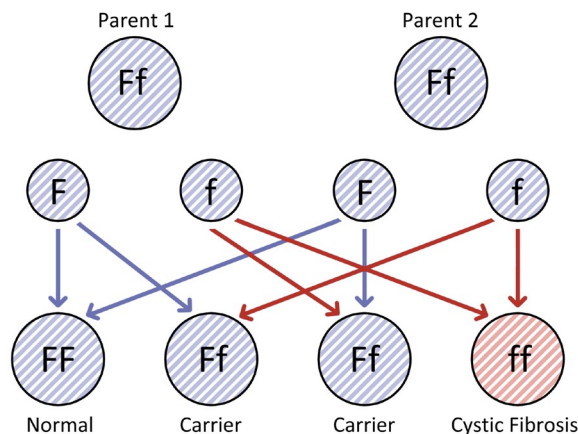
For the Edexcel specification we don't need to memorise these, but we should know how the inheritance works if they give it to us in a question so we have included them here for additional understanding and knowledge 😊

## Cystic Fibrosis

**What is cystic fibrosis?** It is a genetic disease that causes some membranes of the body to produce excess mucus. It affects mucus producing tissues like the respiration system, the digestive system and the reproductive system.

Cystic fibrosis is a recessive condition meaning that two recessive alleles are needed to have the condition. So, the individual would need to be homozygous recessive.

**How is it transmitted?** It is an inherited condition so one allele is inherited from each parent.



The figure to the left shows the genetic diagram showing the inheritance of cystic fibrosis from two heterozygous (carrier) parents.

The cross produces one **homozygous dominant** (normal) individual, two **heterozygous** (carriers) individuals, and one **homozygous recessive** (cystic fibrosis) individual.

**What are the effects of cystic fibrosis on an individual?**

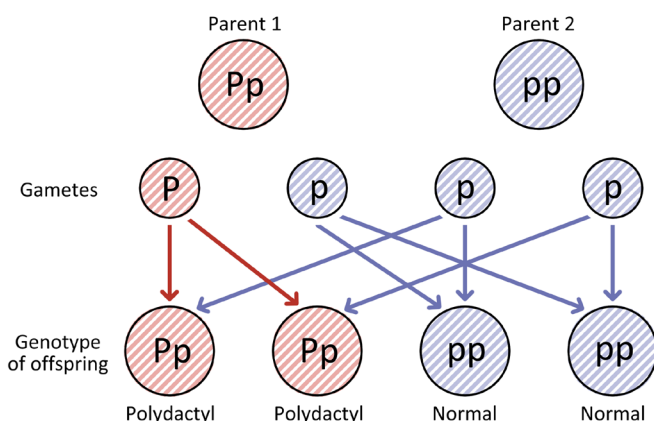
1. An individual might have difficulty breathing, as the **excess mucus** produced **can block airways**. It can also increase the diffusion distance, making diffusion of gases take longer.
2. A male might have difficulty reproducing as the **excess mucus can block the vas deferens** (sperm duct) this can prevent the sperm from being released thereby **preventing them from reaching the fallopian tube**.
3. A female might have difficulty reproducing as the **excess mucus can block the fallopian tubes**, this can **prevent sperm from reaching the Ovum (egg) to fertilise it**.

## Polydactyly

**Polydactyly:** an inherited disorder where an individual has extra digits, so six fingers or six toes.

**Polydactyly is caused by a dominant allele.**

As it is caused by a dominant allele the individual that has the condition will be either **homozygous dominant (PP)** or **heterozygous (Pp)**.



The figure shows the inheritance of polydactyly.