

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.

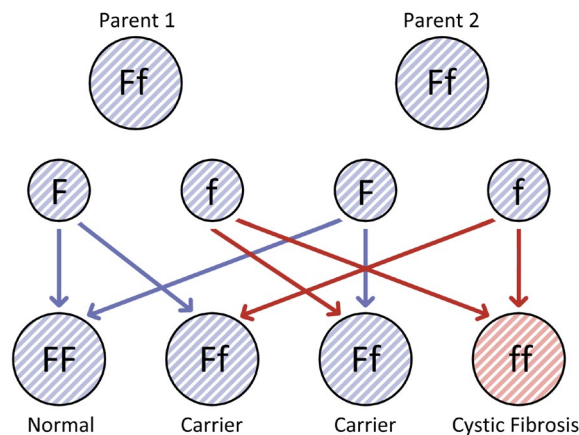
For the AQA GCSE specification we need to learn about two genetic diseases, these are **Cystic Fibrosis** and **Polydactyly**.

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?

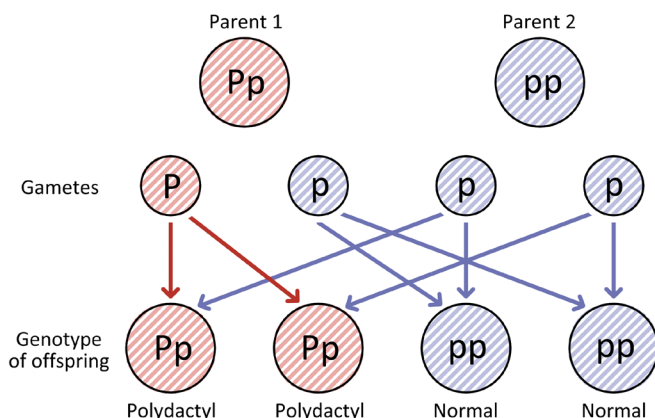
1. An individual might have difficulty breathing, as the **excess mucus produced can block airways**. It can also increase the diffusion distance, making diffusion of gases take longer.
2. A male might have difficulty reproducing as the **excess mucus can block the vas deferens** (sperm duct) this can 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.