Genetic Disorders

Some disorders can be inherited genetically. They can be transmitted from the parents to the offspring via the inheritance of different alleles. Here are some examples:

1. **Cystic Fibrosis**
2. **Polydactyly**

1. Cystic fibrosis

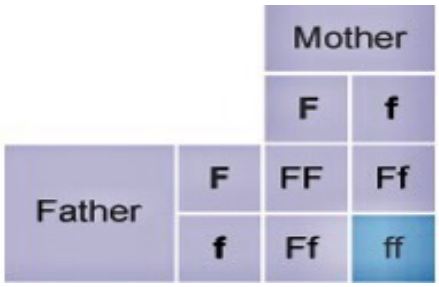
- Cystic fibrosis is an inherited disorder of cell membranes that mainly affects the **lungs and digestive system.** They can become **clogged** with lots of **thick, sticky mucus** as too much is produced. Over many years, the lungs become increasingly damaged and may eventually stop working properly.
- **Treatment:** A number of treatments are available to help reduce the problems caused by the condition, but unfortunately average life expectancy is reduced for people who have it.
- **Causes of the Disease:** It is caused by a faulty recessive allele on **chromosome 7.** To be born with cystic fibrosis, a child has to inherit two copies of this faulty gene - one from each of their parents. Their parents will not usually have the condition themselves, because they will only carry one faulty gene and one that works normally.
- **Cystic Fibrosis allels:**

  - the recessive allele (lower case), which can be shown as f
  - the dominant allele (capital letter), which can be shown as F

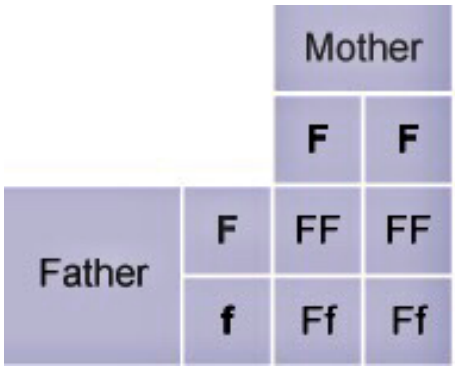
- **Probability of having a disease**

1. An individual who is **homozygous (ff)** with the recessive allele will develop cystic fibrosis.
2. Someone who is **heterozygous (Ff)** will be a carrier of the recessive allele, but will not develop cystic fibrosis and have no symptoms.
3. Someone who is **homozygous with the dominant allele (FF)** will not develop cystic fibrosis.

1. If both parents are **heterozygous, Ff.** The chance of them producing a child with cystic fibrosis is 1 **in 4, or 25%**. As in shown in diagram.

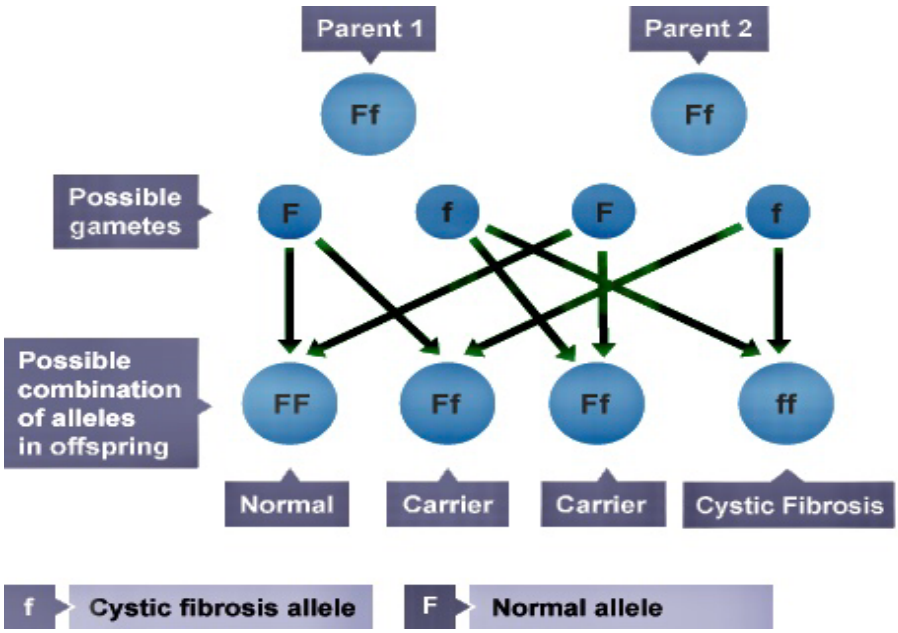


2. Only one parent (**the father) has a copy of the recessive allele (Ff)** and **mother has (FF).** There is no chance of them producing a child with cystic fibrosis. There is a ration of **FF and Ff is 1 : 1 or 50%.** Means out of 4 there is **2 child has a carrier** Allele as shown in punnet square diagram.

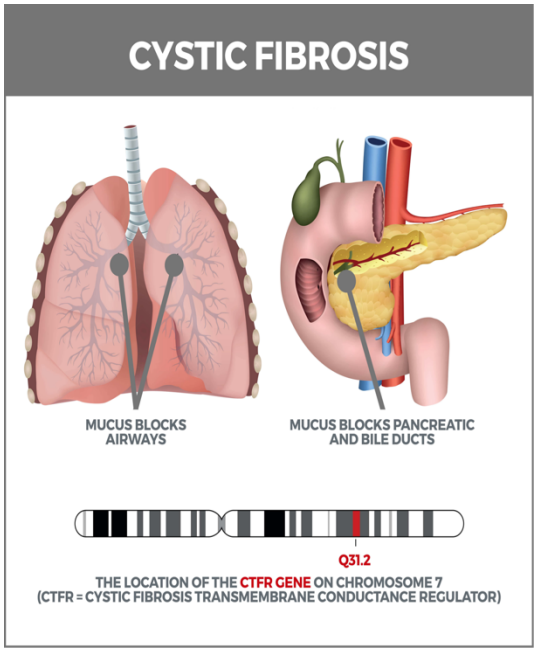


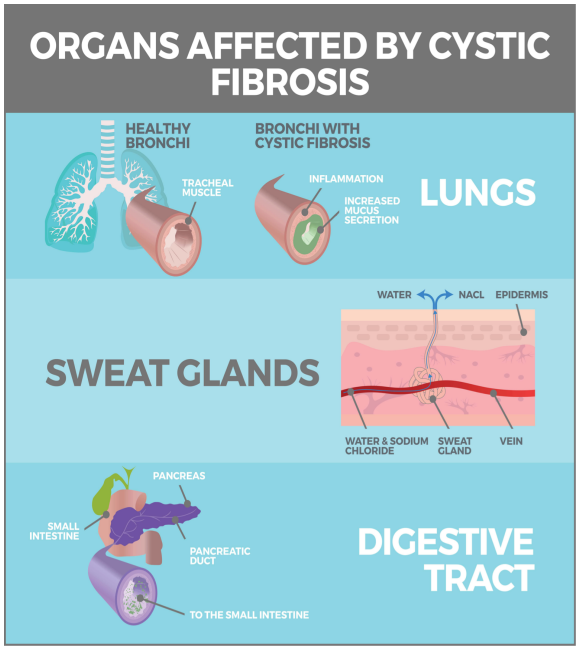
➢ Genetic Diagram of heterozygous parents

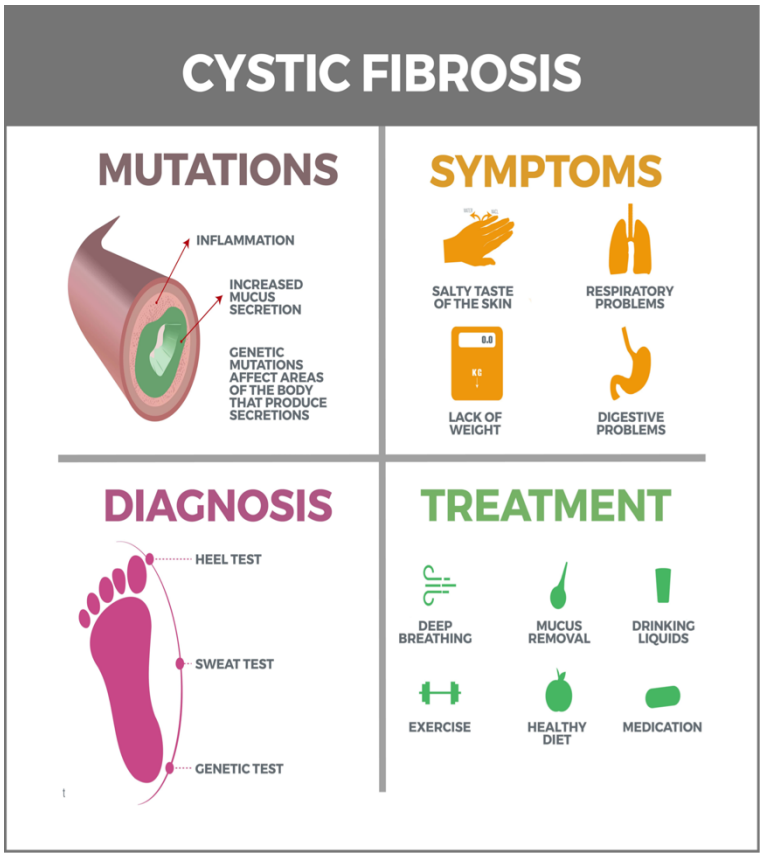
If we cross the Ff father with Ff mother then the result are shown in diagram.



- Out of 4 3 offspring are normal but out of 3 only one is **normal other 2 having a carrier allele.**
- Out of **4 1 offspring are cystic fibrosis patient**

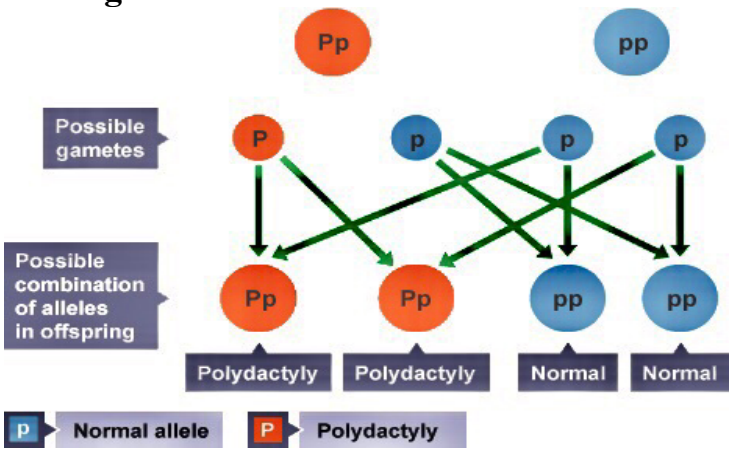

CYSTIC FIBROSIS
MUCUS BLOCKS AIRWAYS
MUCUS BLOCKS PANCREATIC AND BILE DUCTS
Q31.2
THE LOCATION OF THE CTFR GENE ON CHROMOSOME 7
(CTFR = CYSTIC FIBROSIS TRANSMEMBRANE CONDUCTANCE REGULATOR)


ORGANS AFFECTED BY CYSTIC FIBROSIS
HEALTHY BRONCHI
TRACHEAL MUSCLE
BRONCHI WITH CYSTIC FIBROSIS
INFLAMMATION
INCREASED MUCUS SECRETION
LUNGS
SWEAT GLANDS
WATER
NACL
EPIDERMIS
WATER & SODIUM CHLORIDE
SWEAT GLAND
VEIN
PANCREAS
SMALL INTESTINE
PANCREATIC DUCT
TO THE SMALL INTESTINE
DIGESTIVE TRACT

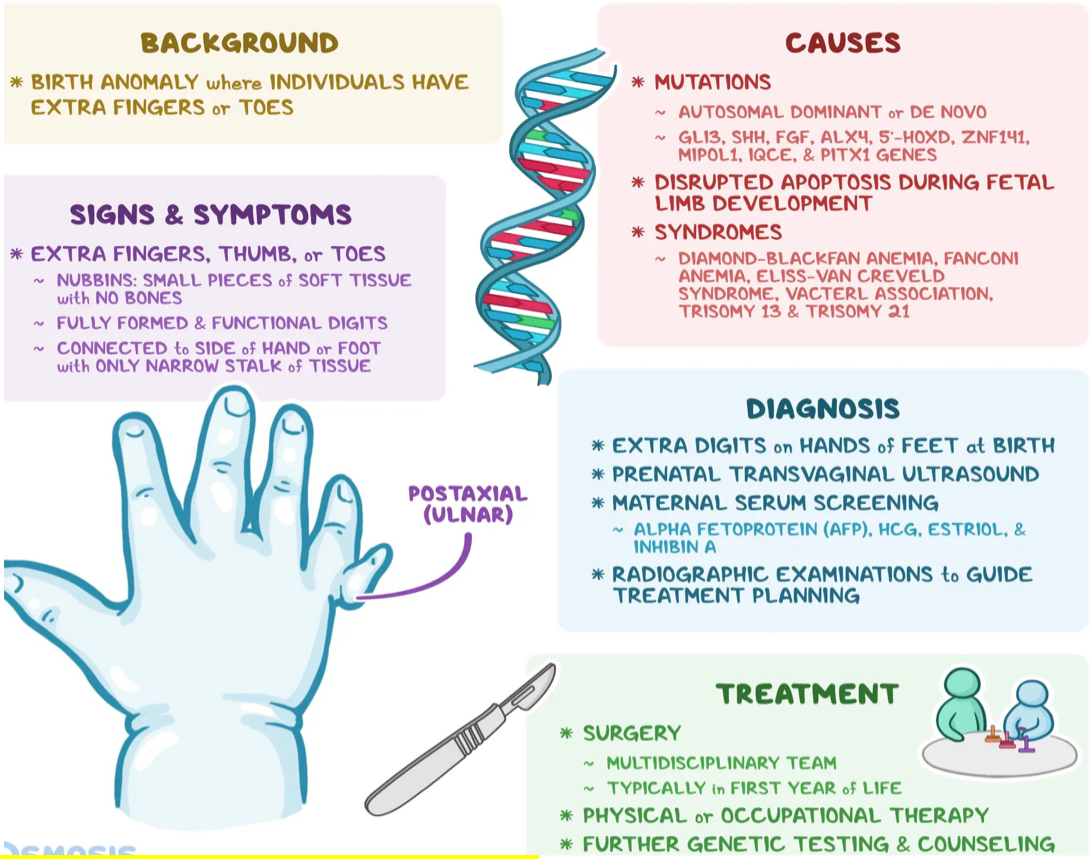

CYSTIC FIBROSIS
MUTATIONS
INFLAMMATION
INCREASED MUCUS SECRETION
GENETIC MUTATIONS AFFECT AREAS OF THE BODY THAT PRODUCE SECRETIONS
SYMPTOMS
SALTY TASTE OF THE SKIN
RESPIRATORY PROBLEMS
LACK OF WEIGHT
DIGESTIVE PROBLEMS
DIAGNOSIS
HEEL TEST
SWEAT TEST
GENETIC TEST
TREATMENT
DEEP BREATHING
MUCUS REMOVAL
DRINKING LIQUIDS
EXERCISE
HEALTHY DIET
MEDICATION

2. Polydactyly

- Polydactyly is an inherited condition in which a person has **extra fingers or toes.** It is caused by a **dominant** allele of a gene. This means it can be passed on by just one allele from one parent if they have the disorder.
- Someone who is homozygous (PP) or heterozygous (Pp) for the dominant allele will develop polydactyly.
- **Genetic Diagram**



- Offspring need to carry just one dominant allele from their parents to inherit the polydactyly condition.
- The **probability** of the offspring having polydactyly is **50% (2 of the 4)** and 50% not having it (normal). This can be expressed as a ratio, **2:2** which can be simplified to 1:1.



Genetic tests or screening

➤ Genetic testing involves analysis of a person's DNA to see if they carry alleles that cause genetic disorders. It can be done at any stage in a person's life.

1. **Antenatal testing: It** is used to analyse an individual's DNA or chromosomes before they are born. This testing is offered to couples who may have an increased risk of producing a baby with an inherited disorder, but it can't detect all the risks of inherited disorders.
2. **Neonatal testing:** It is known as the new born blood spot test involves analysing a sample of blood that is taken from pricking a baby's heel. It detects genetic disorders in order to treat them early.
3. **Pre-implantation genetic diagnosis (PGD) or IVF: It** is used on embryos before implantation. Fertility drugs stimulate the release of several eggs. The eggs are collected and fertilised in a Petri dish. This is known as in vitro fertilisation (IVF). Once the embryos have reached the eight-cell stage, one cell is removed.
4. The cells are tested for the disorder-causing alleles. Embryos that don't contain the disorder allele are implanted into the uterus.

Issues with screening:

1. **Ethics in Normal Pregnancy** Once the parent has their results, there is an ethical quagmire. They will be in the knowledge of a portion of the health of the child. This can lead to an element of choice in some cases. Parents may get the option of terminating the pregnancy. Some therefore, believe that this testing is unethical, as it

could ruin the opportunity for an embryo to live a life. Others may not terminate the pregnancy, but use the result to prepare for pregnancy and the life of the child.

2- Ethics in IVF : During IVF, if an embryo is found to have a genetic disorder, it is not implanted. There are questions regarding the ethics of this, as the embryo is then destroyed.

3- Cost : One thing to remember is that all of this testing is very expensive, however the cost of the healthcare for the child who suffers from a disorder may be much greater.

4- False results : If the results are a false positive or a false negative, they could lead to great trauma to a family.