ECE 4923

Topic : Cystic Fibrosis

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What is Cystic Fibrosis?

-> Cystic fibrosis (CF) is the most common fatal genetic disease.

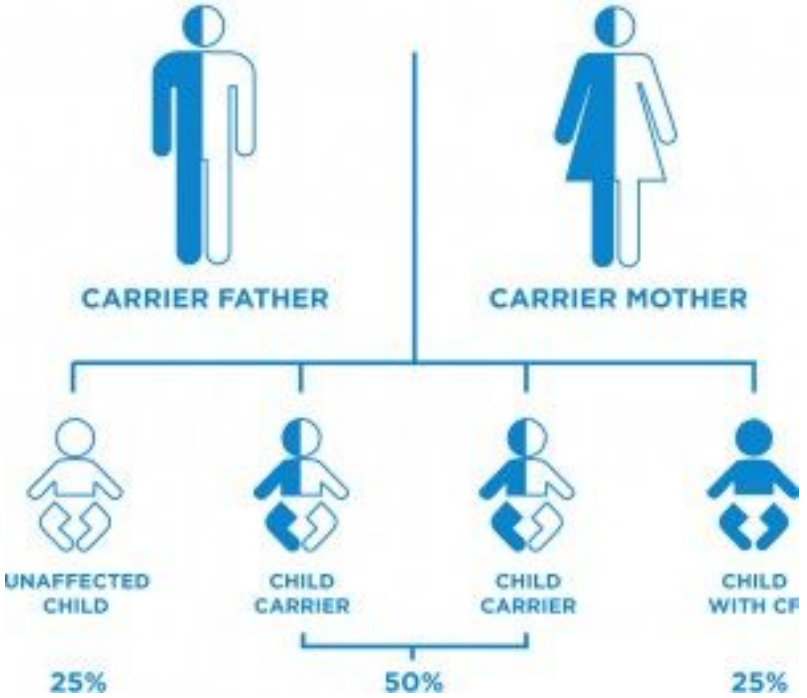
- Mainly affects the digestive system and lungs.
- Infection of lungs -> destruction of lungs -> death.

Reference : Cystic Fibrosis Canada

How do we get it?

- When two CF carriers have a child:
  - 25 % chance that the child will be born with CF.
  - also a 50 % chance that the child will be a carrier.
  - 25 % chance neither will happen.

- Sweat test may be administered for diagnosis
  - Amount of salt content in the sweat measured
    - If the result positive:
      - there's more salt in the sweat than usual



Reference : Cystic Fibrosis Canada

Signs and Symptoms:

Babies with CF usually no signs of CF at birth.

Common problems in babies with CF can include:

- Problems gaining weight
- Meconium ileus or bowel disturbances.
- Salty tasting sweat

Problems that can occur after the newborn period:

- Breathing problems (such as persistent cough)
- Problems conceiving children

Reference : Newborn Screening Ontario

Relation to genetics?

- CF is caused by mutations in a single gene located on chromosome 7,termed the cystic fibrosis transmembrane regulator (CFTR) gene.

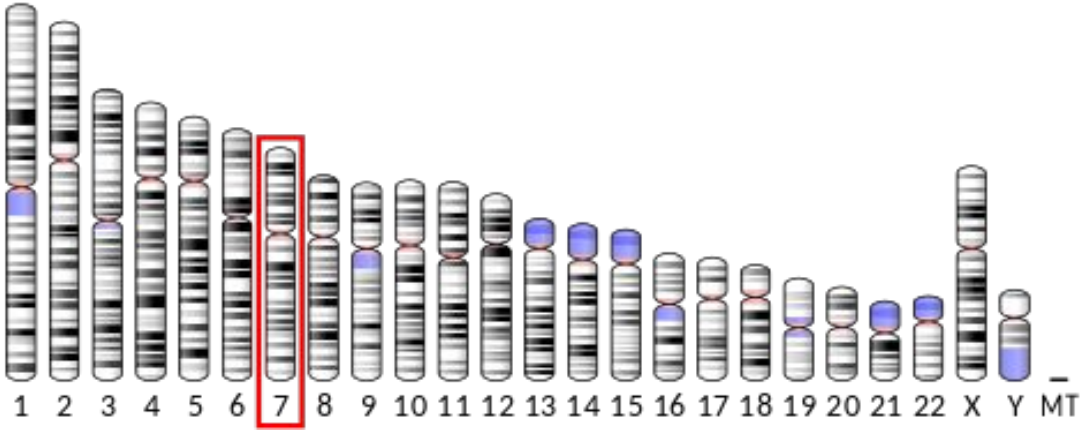


Image taken from : Wikipedia

Relation to genetics?

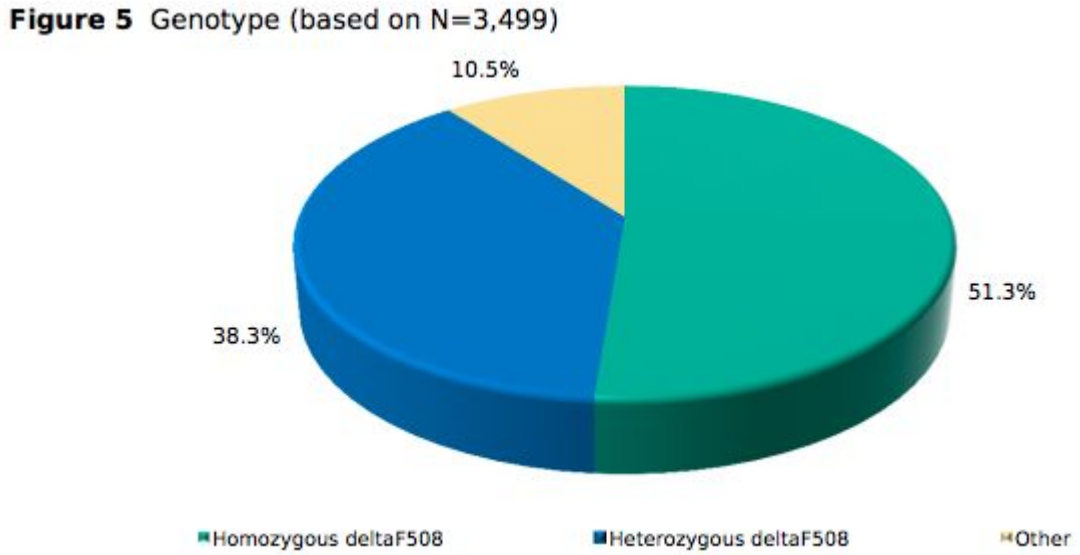
- The CFTR gene codes for a protein called the transmembrane conductance regulator which functions as a chloride channel and is involved in many cellular functions.

- The most common mutation worldwide is a three base-pair deletion resulting in the deletion of the phenylalanine residue at amino acid position 508, commonly referred to as deltaF508.

Reference: Canadian CF Patient Data Registry – 2010 Report

* Phenlylalanine : an aromatic essential amino acid; a component of proteins

Relation to genetics?



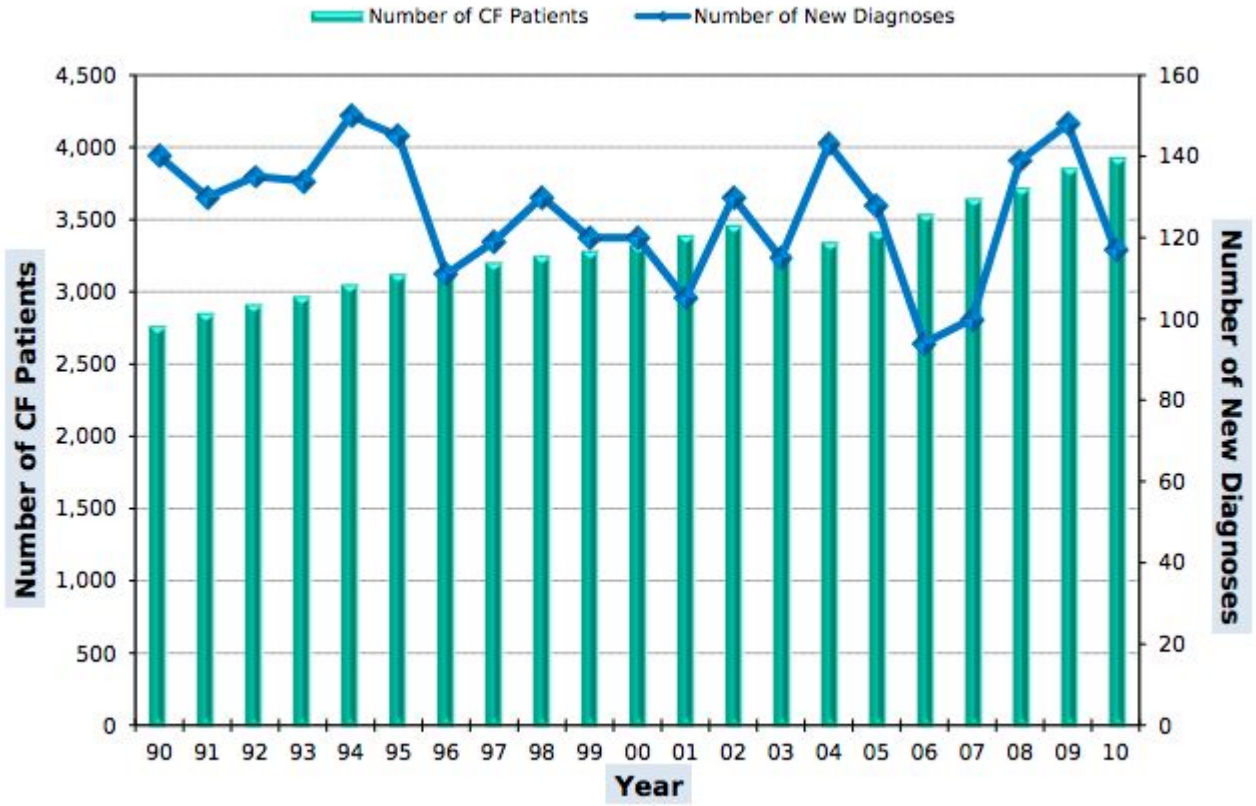
Reference: Canadian CF Patient Data Registry –2010 Report

Treatment :

- Medications to help fight infection
- Airway clearance and physiotherapy
- Pancreatic enzyme supplements to help digest food
- Vitamins and supplements
- Other treatment as needed

Reference : Newborn screening Ontario

Stats:



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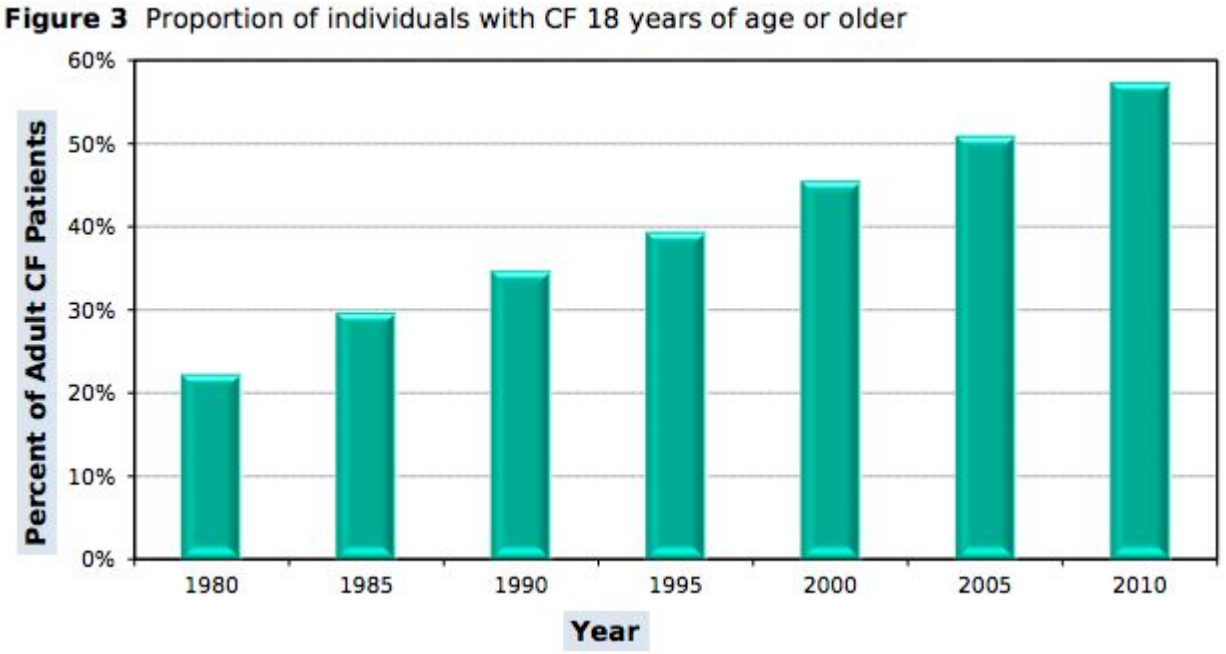

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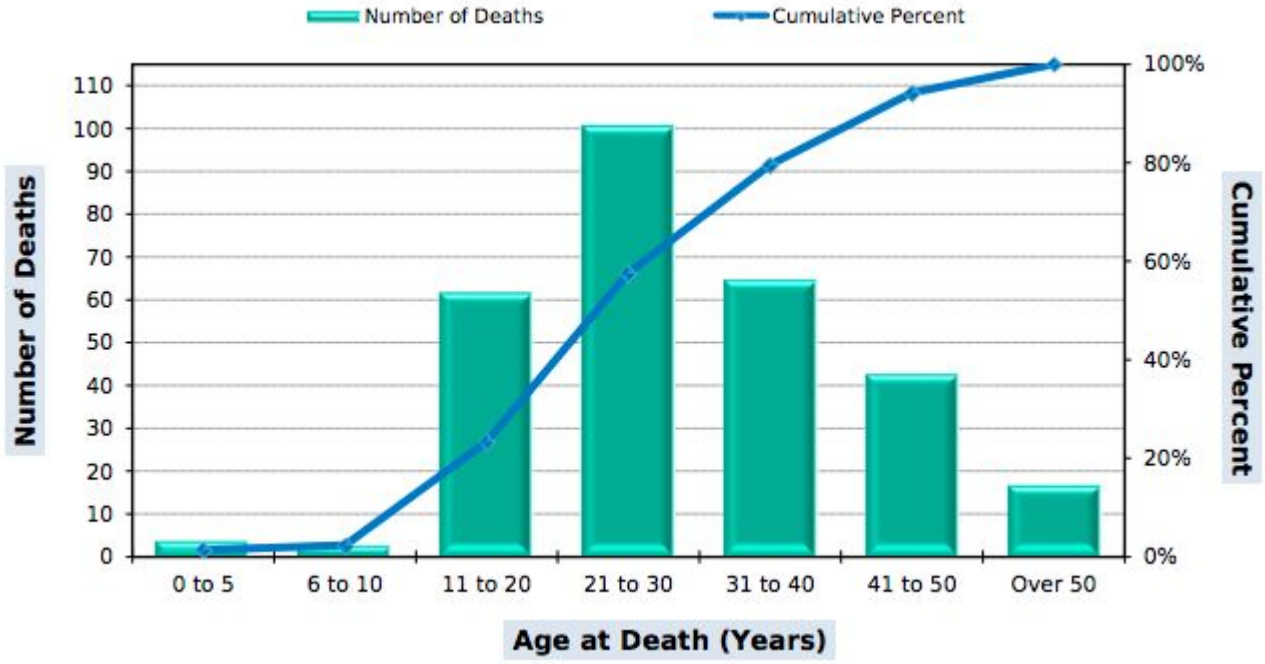


Reference: Canadian CF Patient Data Registry – 2010 Report

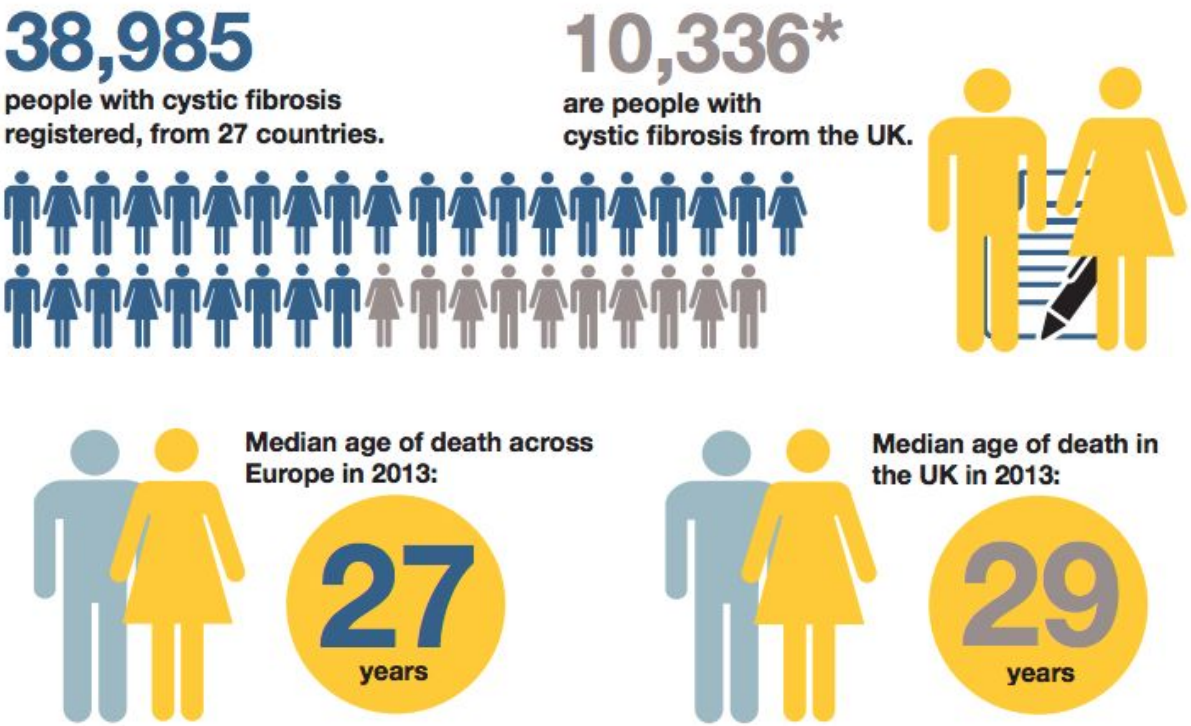
Stats:

In the US:

- More than 30,000 people are living with cystic fibrosis.
- Approximately 1,000 new cases of CF are diagnosed each year.
- More than 75 percent of people with CF are diagnosed by age 2.
- More than half of the CF population is age 18 or older.

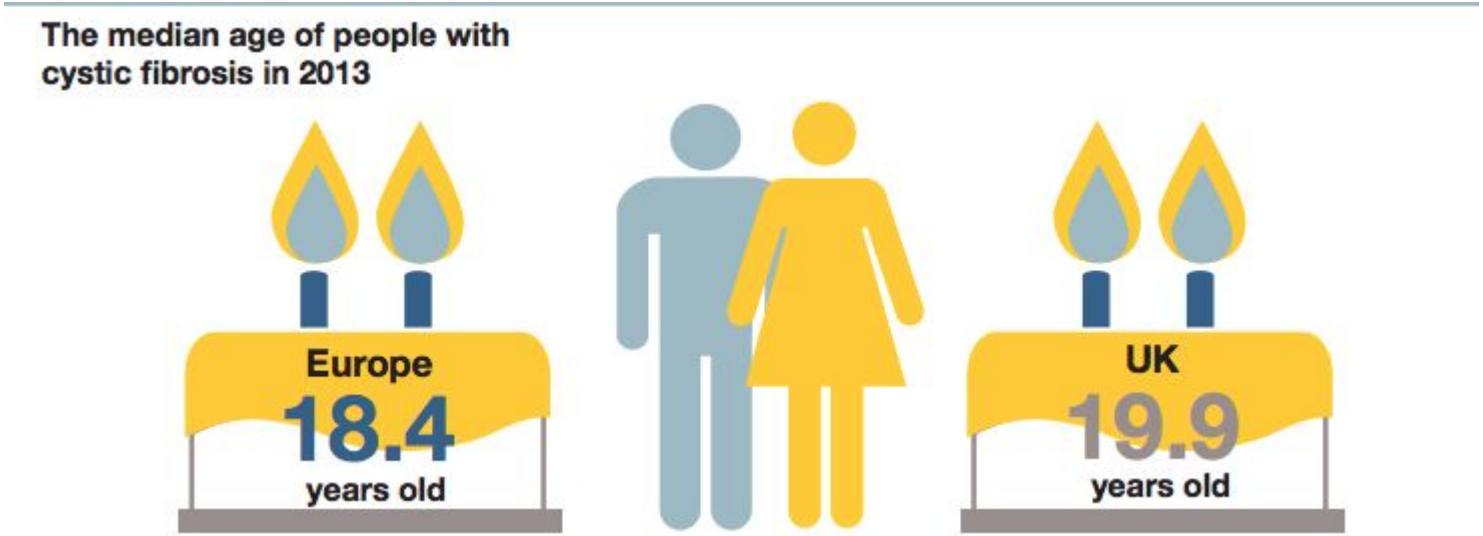
Reference: Cystic Fibrosis Foundation Patient Registry (USA)

Stats:



Reference: European Cystic Fibrosis Society Annual Data Report 2013

Stats:



Reference: European Cystic Fibrosis Society Annual Data Report 2013

What can you do?

- CF Newborn Screening for early diagnosing
  - Except QC,all provinces and territories have implemented newborn screening.
- Educate the society
- Volunteer for Cystic Fibrosis Canada

Nearest Clinics:

1. IWK Health Centre, Halifax
2. QEII Health Sciences Centre, Halifax
3. Saint John Regional Hospital, Saint John

Reference : Cystic Fibrosis Canada

Questions?