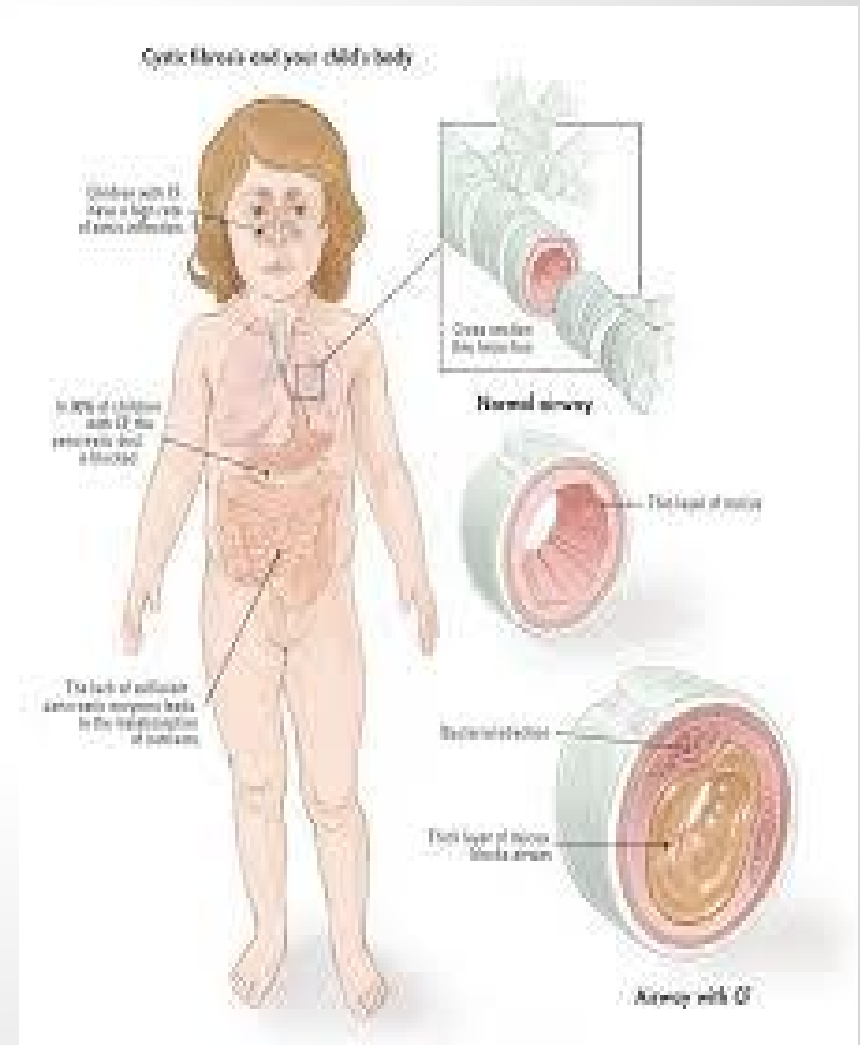


Cystic Fibrosis

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What is Cystic Fibrosis?

Cystic Fibrosis is an autosomal recessive genetic disorder that affects most critically the lungs, and also the pancreas, liver, and intestine. It is characterized by abnormal transport of chloride and sodium across an epithelium, leading to thick, viscous secretions.



History of Cystic Fibrosis

- Cystic Fibrosis has been around for thousands of years but had not been documented until the 20th century when a few people in Germany reported cases of disfiguration and some of the symptoms of Cystic Fibrosis in young children.
- There were some descriptions that had been documented before that in stories and journals, but had not been confirmed by any doctors.
- A larger resurgence of genetic mutations linked to Cystic Fibrosis were discovered in the 80's

Symptoms

- Depends on severity
- Symptoms may appear after birth or in adulthood
- Can be diagnosed in first month of birth or later
- Affected in digestive and respiratory system
- Wheezing, lung infections, coughs, thick mucus, inflamed nasal passage, etc.
- Constipation, bad smelling stool, lack of weight/growth.
- Go to doctors after seeing these symptoms

Influence In Science

- been around for thousands of years
- not documented until the 20th century
- many cases reported in germany
- large resurgence in the 80's

Treatment

- There is no cure for cystic fibrosis, but treatment can ease symptoms and reduce complications.
- Close monitoring and early, aggressive intervention is recommended.
- Managing cystic fibrosis is complex, so you should obtain treatment at a center that specializes in cystic fibrosis.

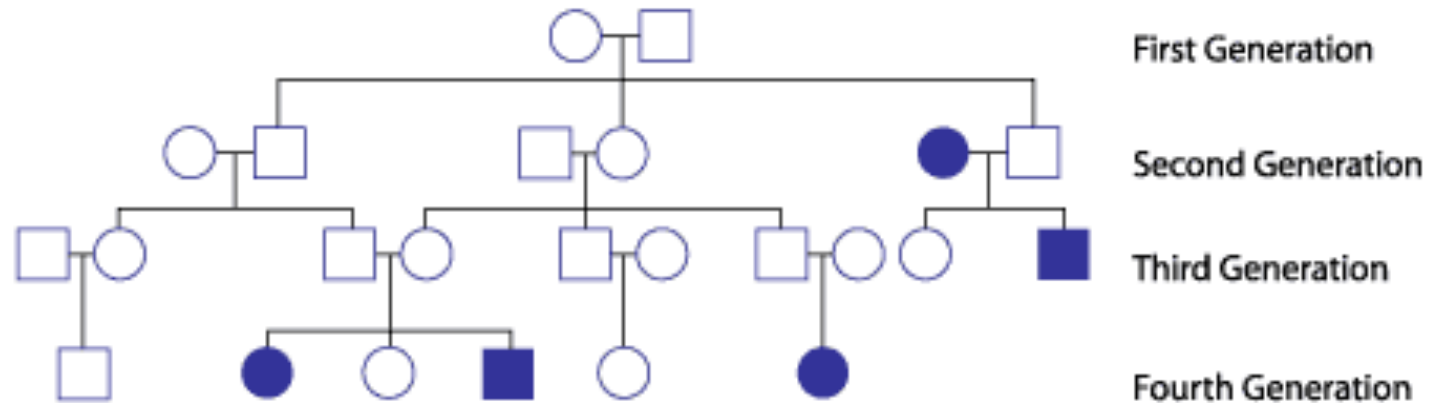
Genetics of Cystic Fibrosis

- Cystic Fibrosis is a dormant gene that lies harmless in most people, only altering other genes when cells go through meiosis with another rare mutated cystic fibrosis cell.
- It is hard to tell when this is going to happen in offspring since the dormant mutation is so rare and usually unnoticeable.

Diagnosis of Cystic Fibrosis

- Blood test. The test looks for variation in a gene to know the cause of the disease
- Immunoreactive trypsinogen
- sweat chloride test
- chest X-ray
- lung function tests
- measurement of pancreas function
- secretin stimulation test
- trypsin and chymotrypsin in stool

Inheritance Patterns



- Female
- Male
- ■ Person with cystic fibrosis
- Linking parents
- | Linking parents to children

Punnett Square

	C	c
C	CC	Cc
c	Cc	cc

25% Homozygous
Dominant

25% Homozygous
Recessive

50% Heterozygous

Thank you

For listening and watching

Citations

- <http://www.mayoclinic.com/health/cystic-fibrosis/DS00287/DSECTION=symptoms>
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