

Cystic Fibrosis (CF)



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BioC 118Q – Brutlag

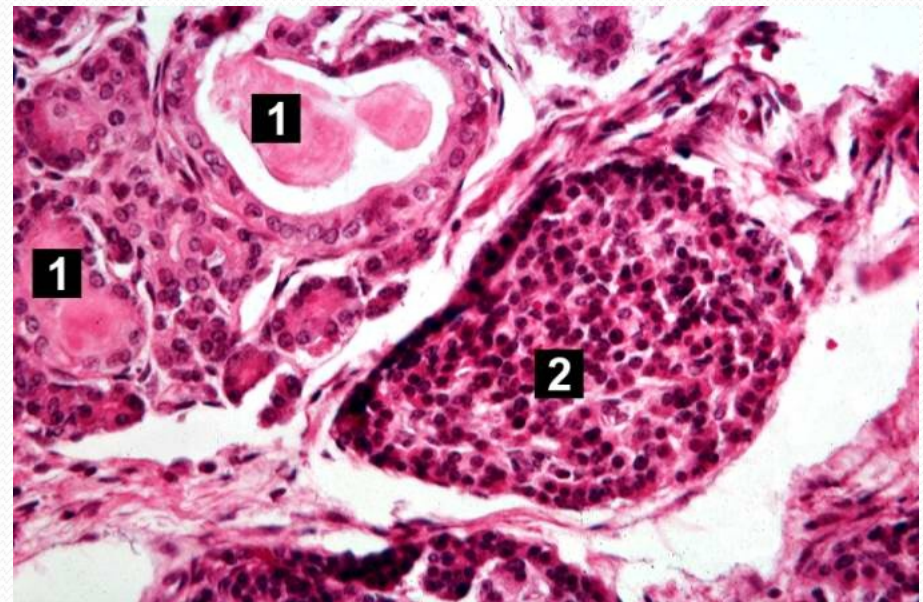
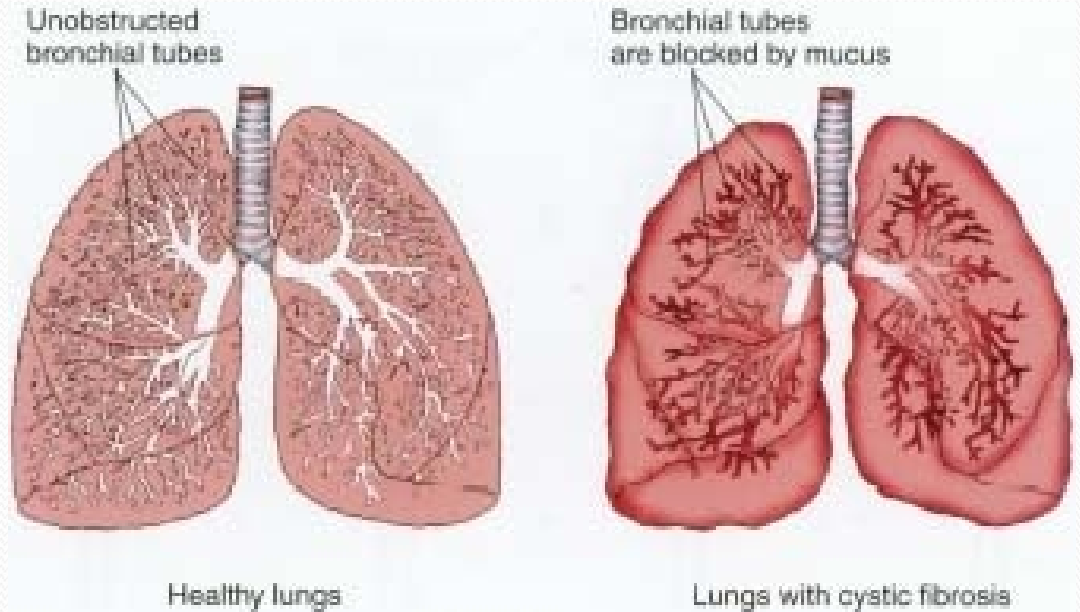
History & Statistics

- 1938 – Dorothy Hansine Andersen
- 30,000 US (most common fatal)
- 70,000 world
- 1,000 new annually
- 70% diagnosed by age 2
- 45% live past age 18
- 1/25 Northern Europeans carry
- < 1/500 Asians carry



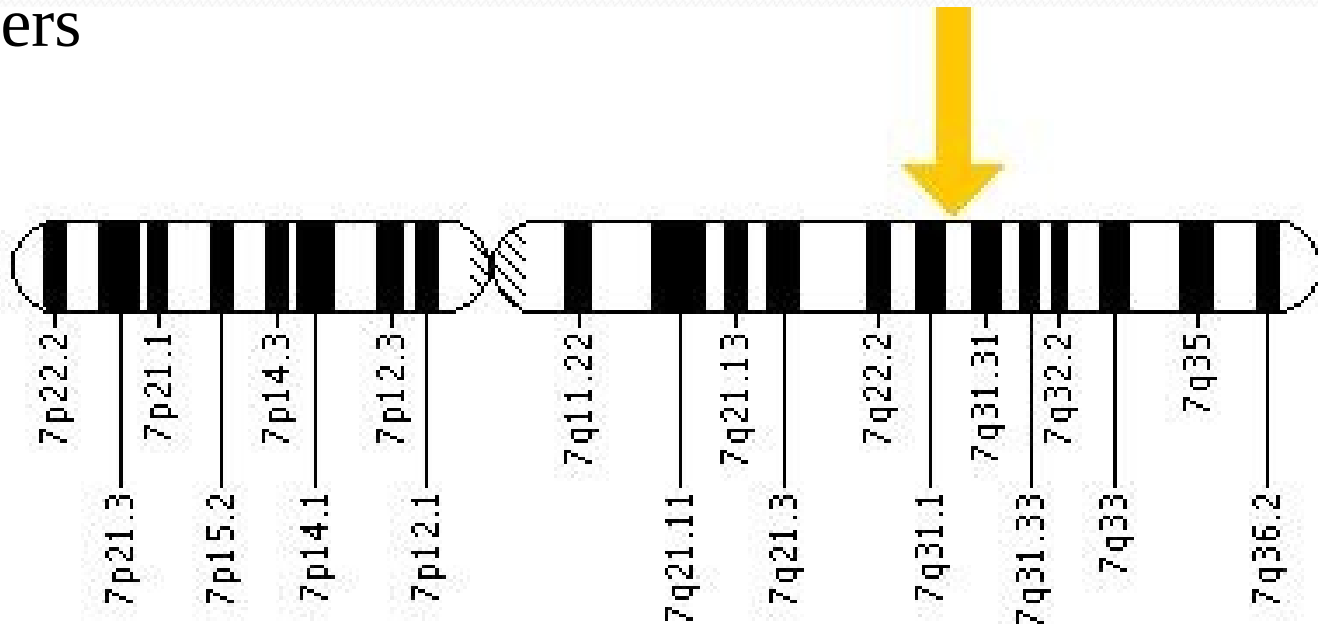
Symptoms

- Salty sweat
- Lung infections
- Shortness of breath
- Poor nutrient absorption
 - Fatty diarrhea
- BM issues
- Delayed puberty
- Infertility



Genetic Facts

- Autosomal recessive
- Chromosome 7, q, 31
- Carriers



Classic Diagnosis

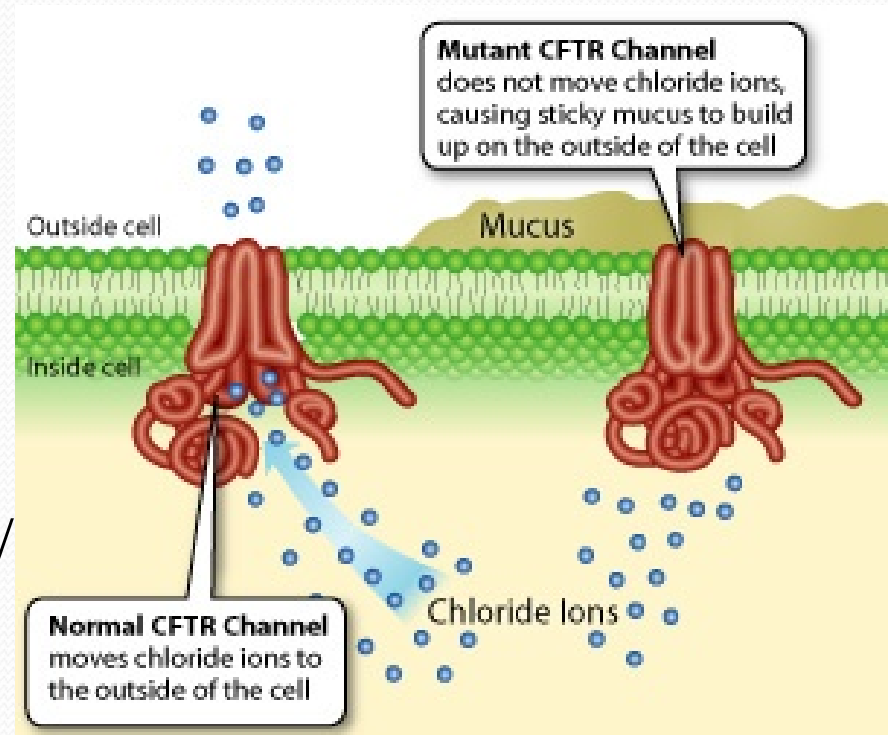
- Sweat test – 1950s
 - <http://www.youtube.com/watch?v=8UCWoz6gUp8&feature=play>
- Often confused with upper respiratory issues
- Thought to be linked to celiac disease
- Also mistaken for Vitamin A deficiency

Classic Treatment

- Most children did not live to adolescence
- If discovered early, nutrition

Modern Diagnosis

- CFTR – delta-F508 (1989)
- Pre-birth tests
 - CVS
 - Amniocentesis
- Neonatal screening
 - <http://genes-r-us.uthscsa.edu/>
- Sweat test



Modern Treatment

- Airway clearance
 - Nebulizers
 - Inhalers
- Implanted ports
- Extra nutrition
 - Tube feeding



Novel Genetic Methods

- Genetic counseling and family planning
 - Carrier testing
- Targeted mutation analysis
- Gene therapy in the works (1993)
 - <http://www.cff.org/research/DrugDevelopmentPipeline/>

Real-Life Experience

- Median age 37
- Pills, inhaler – antibiotics
- 2-3 hour daily treatment for mucus removal from lungs

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Thank You! 😊