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
- BM issues
  - Fatty diarrhea
- 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=player_embedded](http://www.youtube.com/watch?v=8UCWoz6gUp8&feature=player_embedded)
• 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
Works Cited


Image Credits

• Title - http://www.listen2yourgut.com/blog/blog/blogimages/2008/07/nebulizer.jpg
• Modern Diagnosis - http://thednafiles.files.wordpress.com/2008/04/cfchannel.jpg
Thank You! 😊