




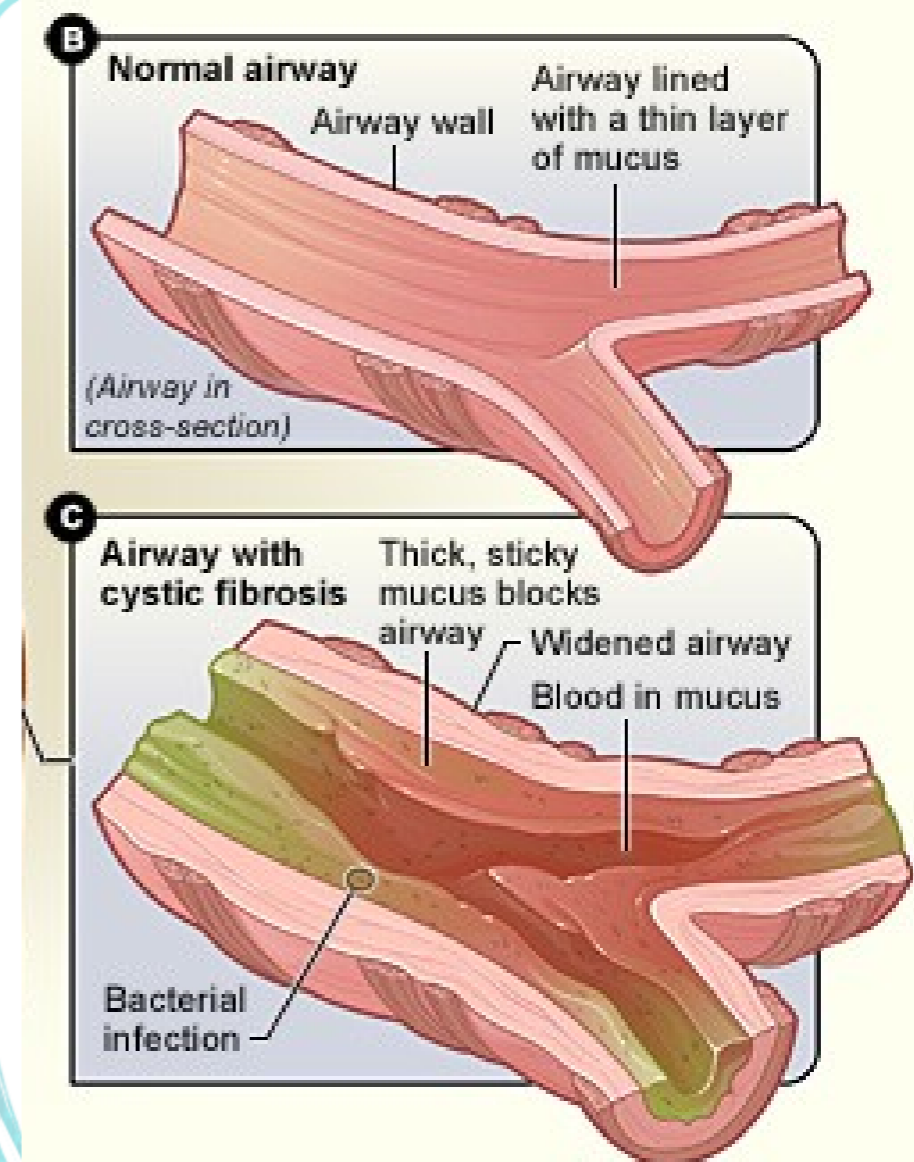
Cystic Fibrosis

Carolyn Sinow
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About Cystic Fibrosis (CF)

- * Considered a common genetic disorder (1:3500)
- * Most common in Caucasians
- * Inherited disease of the mucus glands
- * Body produces abnormally thick and sticky mucus



Symptoms

- * Damage to respiratory system
 - Breathing problems
 - Bacterial infections in the lungs
 - Coughing/ wheezing
 - Inflammation
- * Digestive problems
 - Diarrhea
 - Malnutrition
- * Salty sweat
 - Dehydration
- * Infertility

Diagnosis

- * Newborn screening: genetic or blood tests
- * “Salty baby”
- * Sweat test is the most useful diagnostic test
- * Chest X-ray, lung function test, sputum culture
- * Prenatal testing: amniocentesis or chorionic villus sampling (CVS)
- * Genetic tests for carriers

Treatment

- * No cure
- * Until 1980's, most CF patients died in childhood or teenage years; now most live to over 35 years old.
- * Goals:
 - Prevent and control lung infections
 - Remove mucus from lungs
 - Provide nutrition
 - Prevent dehydration

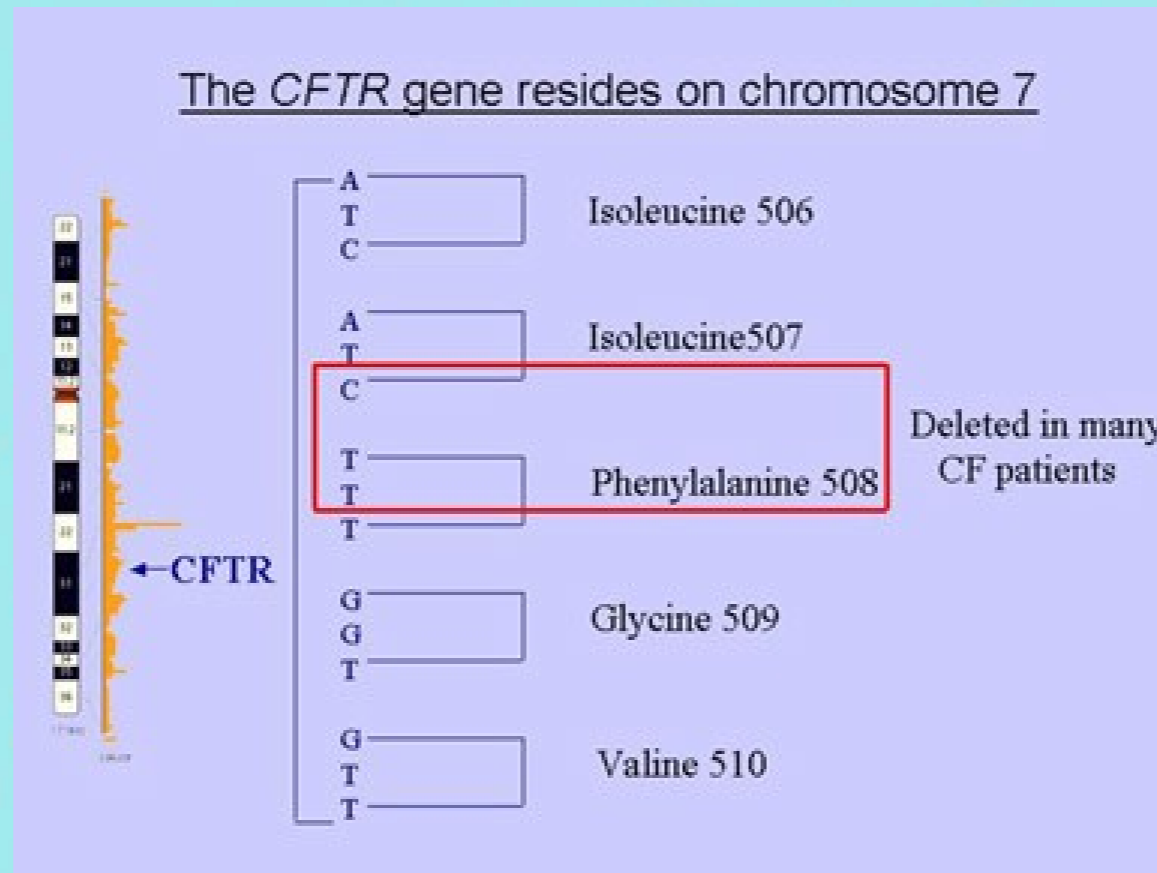
Treatment Continued

- * Chest physical therapy (CPT)
- * Aerobic exercise
- * Medicines: antibiotics, anti-inflammatory, bronchodilator, mucus thinners
- * Diet rich in salt, calories, fat, protein
- * Check-ups every 3 months

Molecular Genetics

- * Autosomal recessive
- * Caused by mutations on CFTR gene (cystic fibrosis transmembrane conductance regulator gene)
- * Can't regulate flow across chloride channels
- * Causes thick, sticky mucus which clogs airways and glands
- * Other unknown genes may affect severity of CF

Affect of Knowing Genetics



<http://www.mcgill.ca/hostres/diseases/cystic/>

- * Has helped diagnostics (genetic tests of CFTR gene)
- * Has not helped development of treatment

References

- * <http://www.nlm.nih.gov/medlineplus/cysticfibrosis.html>
- * http://www.nhlbi.nih.gov/health/dci/Diseases/cf/cf_what.htm
- * <http://ghr.nlm.nih.gov/condition/cystic-fibrosis>
- * <http://www.ncbi.nlm.nih.gov/omim/219700>
- * <http://www.ncbi.nlm.nih.gov/omim/602421>