Cystic Fibrosis

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Biochem 118

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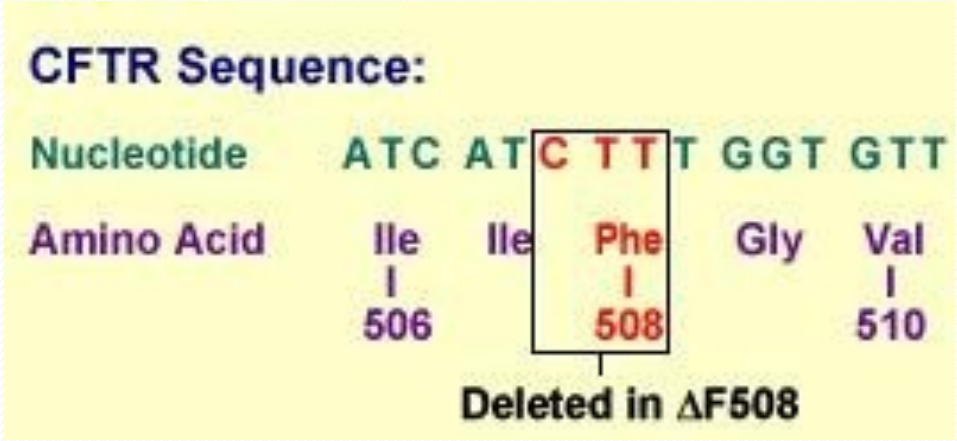
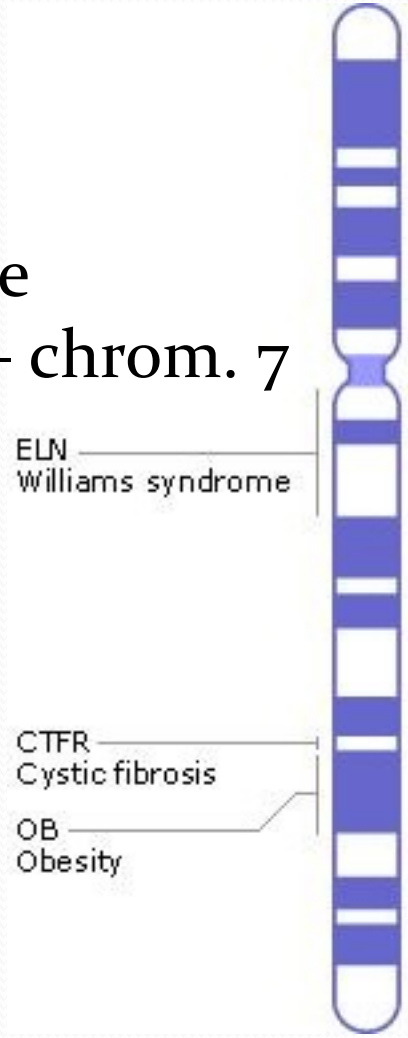
What is Cystic Fibrosis (CF)?

- Autosomal recessive disorder – 25%
- 1 in 4,000 children are born with CF in US
- Cystic Fibrosis Transmembrane Conductance Regulator (CFTR) gene mutation(deletion) – chrom. 7
- CFTR – chloride ion transport
- Thick mucus lining – pulmonary disease



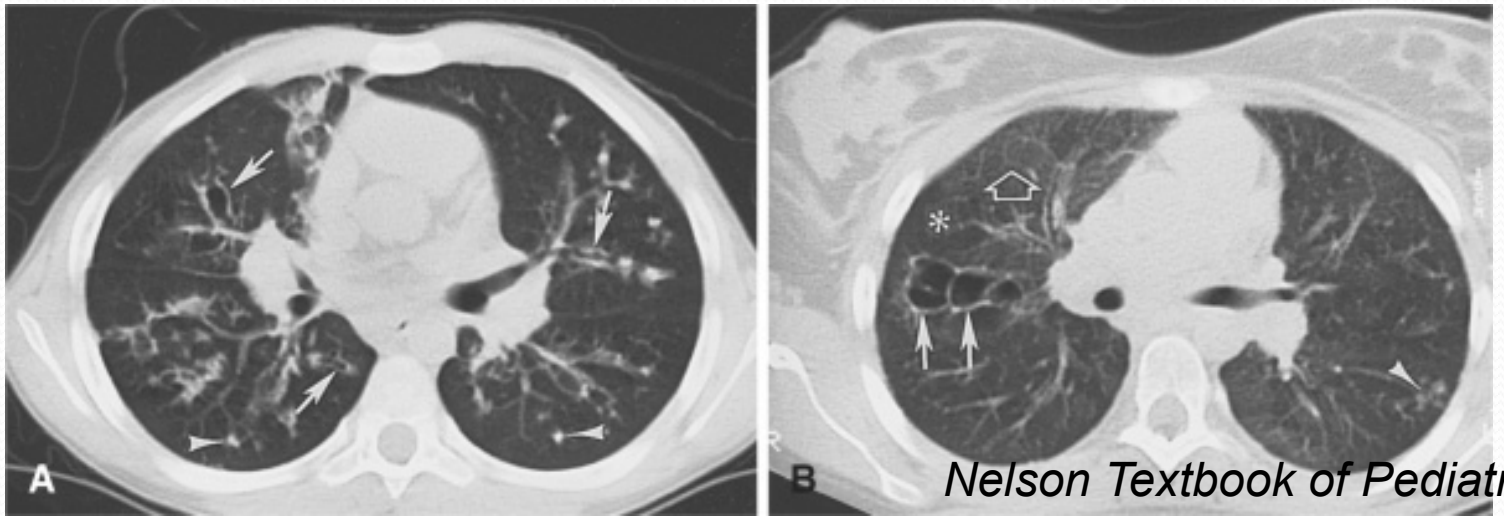


Classical Diagnostic Methods

- Chloride (sweat) test
- Blood tests – assess infection, involvement of certain organs,
- Chest x-rays - mucus buildup
- Pulmonary function tests – ability to exchange O_2 and CO_2
- Sputum cultures – *Pseudomonas aeruginosa*
- Pancreatic function tests - ducts in the pancreas, preventing digestive enzymes from reaching the intestines
- Stool – fat absorption
- Sterile males

Classical Treatment

- Antibiotics - lung and sinus infections.
- Inhaled medicines - open the airways
- Bronchial drainage – 1-4 times a day
- Chest PT
- Pancreatic enzyme replacement
- Oxygen therapy
- Lung transplant is an option in some cases



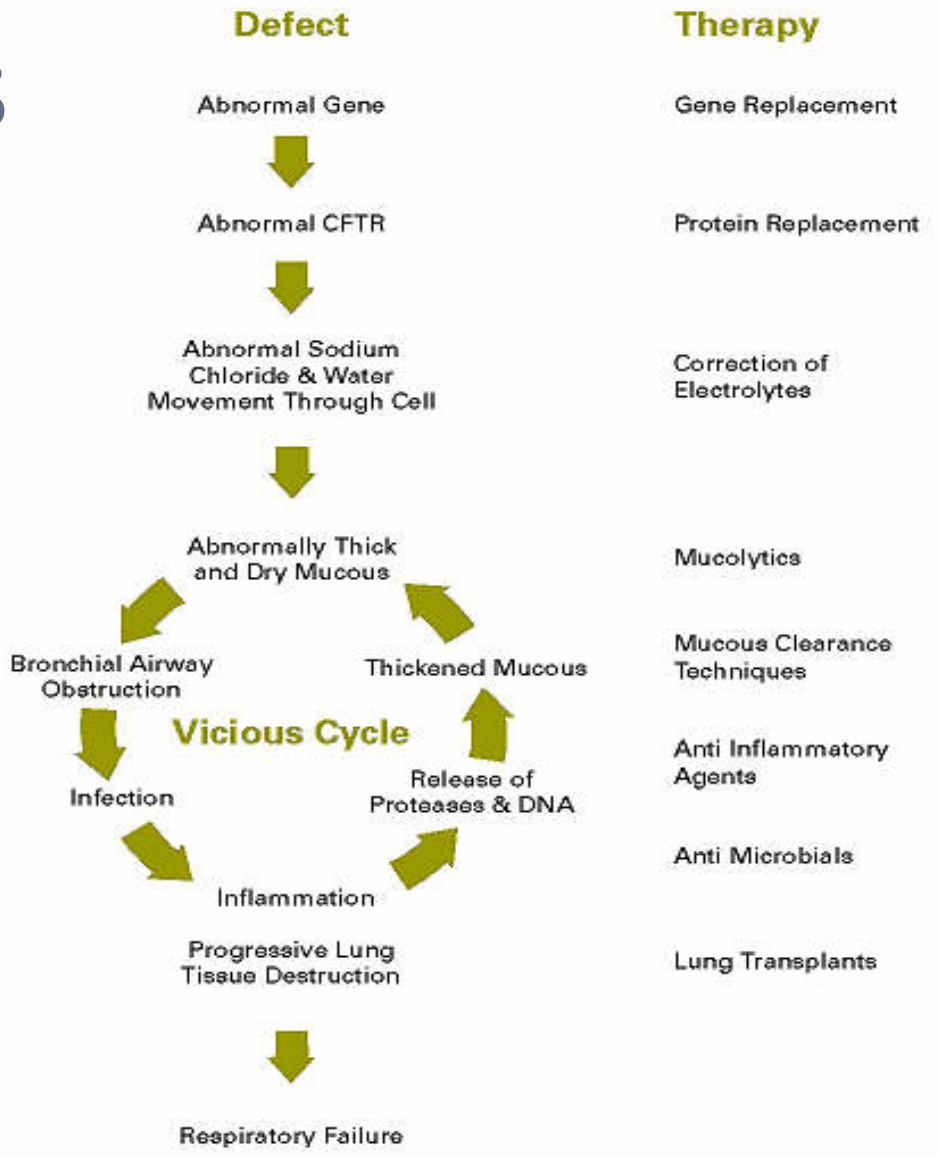
Nelson Textbook of Pediatrics, 18th ed

Novel Diagnostics after knowing causative gene

- Discovery of gene in 1989
- Family history
- Gene test – detect CFTR gene mutation
- Detection of CF in a fetus is now possible through genetic testing
- Immunoreactive trypsinogen (IRT) test – newborns
  - Not diagnostic, frequent false-positives
- All states will screen for CF by end of 2009
- Heterozygotes (carriers) – typhoid protection

Novel therapies

- Ideal goal: replace the faulty gene
- CF good candidate, 1 yr
- External CFPR
- Chemical chaperone
- Traditional therapies with gene specificity



References

- http://www.ncbi.nlm.nih.gov/books/bv.fcgi?call=bv.View..ShowSection&rid=gnd.section.279
- http://www.ncbi.nlm.nih.gov/entrez/dispomim.cgi?id=219700
- http://ghr.nlm.nih.gov/condition=cysticfibrosis
- http://www.ornl.gov/sci/techresources/Human_Genome/posters/chromosome/cftr.shtml
- http://www.lifespan.org/adam/healthillustratedencyclopedia/1/000107.html
- http://www.news.harvard.edu/gazette/1998/07.09/CysticFibrosisG.html
- http://www.childsdoc.org/fall97/cf/cf.asp