Cystic Fibrosis

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Cystic Fibrosis: A Hereditary Disease

- Comes from the “cysts” that often cover the pancreas and apparent fibrosis
- Caused by a mutation of the **Cystic Fibrosis Trans-membrane conductance regulator**
- Also known as:
  - Fibrocytic disease of the pancreas
  - Mucoviscidosis
  - Mucoviscidosis of the pancreas
  - Pancreas fibrocytic disease
  - Pancreatic cystic fibrosis
The Genetic Basis

- The CFTR mutation occurs at the indicated spot on Chromosome 7.

- An [Autosomal] Recessive Trait so TWO alleles of the mutated CFTR are necessary for the disease to be represented. Essentially, at least one of the homologous alleles on chromosome 7 must be functional. As such many people are carriers of Cystic Fibrosis, and do not actually exhibit the disease.

- 66% of Cystic Fibrosis cases involve a deletion of 3 nucleotides that code for the amino acid phenylalanine.

- Essentially the mucus glands of several organs are not formed properly and are unable to function adequately. An example would be the mucus lining in the lungs that due to CF become inflamed with thick mucus constricting breathing and may lead to death.
Racial Comparison of Chances for CF

The Chance of Being a CF Carrier Depending on Race/Ethnicity

<table>
<thead>
<tr>
<th>Ethnicity/Race</th>
<th>Chance of Being a CF Carrier</th>
<th>Chance Both Partners Are CF Carriers</th>
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<tbody>
<tr>
<td>European Caucasian, Ashkenazi Jewish</td>
<td>1 in 29</td>
<td>1 in 841</td>
</tr>
<tr>
<td>Hispanic American</td>
<td>1 in 46</td>
<td>1 in 2,116</td>
</tr>
<tr>
<td>African American</td>
<td>1 in 65</td>
<td>1 in 4,225</td>
</tr>
<tr>
<td>Asian American</td>
<td>1 in 90</td>
<td>1 in 8,100</td>
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</tbody>
</table>

REMEMBER: Both parents must be carriers for the baby to develop CF.
Common Symptoms and Facts about Cystic Fibrosis

Some Statistics:

- About 1,000 new cases of cystic fibrosis are diagnosed each year.
- More than 70% of patients are diagnosed by age two.
- More than 40% of the CF patient population is age 18 or older.
- The predicted median age of survival for a person with CF is more than 37 years.

Usual Symptoms:

- very salty-tasting skin
- persistent coughing, at times with phlegm
- frequent lung infections
- wheezing or shortness of breath
- poor growth/weight gain in spite of a good appetite; and
- frequent greasy, bulky stools or difficulty in bowel movements
- Dehydration
- Infertility (common in men)

http://www.cff.org/AboutCF/
Symptomatic Diseases
- Possible diseases from having cystic fibrosis

<table>
<thead>
<tr>
<th>Disease</th>
<th>Explanation</th>
<th>Example</th>
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<tbody>
<tr>
<td>Lung/Sinus Disease</td>
<td>Inflammation, infection in respiratory system, clogged lungs and possible lung/heart failure</td>
<td></td>
</tr>
<tr>
<td>Gastrointestinal, liver, and pancreatic disease</td>
<td>Difficulty digesting foods and extracting nutrients from these foods. Linings of the organs in the systems are covered with a thick mucus.</td>
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<tr>
<td>Endocrine disease and growth</td>
<td>Because CF affects the pancreas poor digestion and illnesses like clubbing (enlargement of fingers and toes) and Type I &amp; II diabetes. Also CF hinders Vitamin D uptake which can harm bone density</td>
<td></td>
</tr>
</tbody>
</table>
What will your child look like?

Father:
- $bb =$ two recessive genes for blond hair
- $Cc =$ one dominant and one recessive allele for cystic fibrosis (carrier)

Mother:
- $BB =$ two dominant alleles for brown hair
- $Cc =$ one dominant and one recessive allele for cystic fibrosis (carrier)
Punnett Squares & Results

- **Bb, Bb, Bb, Bb**: 100% Chance of Brown Hair in Child
- **Cc, Cc, Cc, Cc**: 25% Cystic Fibrosis in Child
- **Cc, Cc, Cc, Cc**: 50% Chance of CARRIER
- **Cc, Cc, Cc, Cc**: 25% Chance of Nothing
Cystic Fibrosis: Prognosis

- There is currently no cure for cystic fibrosis; however, do not worry, new treatments have extended life expectancy for the afflicted.

<table>
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<th>Treatment</th>
<th>Explanation</th>
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| Antibiotics                | Help fight lung infections  
- Aerosolized antibiotics are available  
- Drawback: Bacteria may eventually develop a resistance to the antibiotics |
| 2. Mucus-Thinning Drugs:   | Aerosolized drugs that provide an enzyme that helps to breakdown the mucus  
- Drawback: May cause irritation of the airway and sore throat |
| 4. Bronchial Airway drainage: | Physical removal of the mucus in the lungs by electric chest clappers and other  
> mechanical aids |
| 5. Good nutrition:         | Causes malnourishment because pancreatic enzymes do not reach small intestine  
- High calorie nutrition with vitamins and enteric-coated pancreatic enzymes to  
> maintain weight |
| 6. Lung Transplantation:   | A major surgery that requires the replacement of both lungs  
- High-risk procedure |
| 8. Exercise:               | Helps to improve lung function and loosens up mucus                         |
Prenatal and Post-natal Testing

Prenatal:
- **Amniocentesis**: “In amniocentesis, your doctor will insert a hollow needle through your abdominal wall into your uterus to obtain cells from the fluid (amniotic fluid) around the baby. The fluid is then tested to see if both of the baby’s cystic fibrosis transmembrane conductance regulator (CFTR) genes are normal.”

- **Chorionic Villus Biopsy**: “In a chorionic villus biopsy, your doctor will use an ultrasound to guide a thin tube through your vagina and cervix into your uterus and remove a tiny piece of the placenta to biopsy. The cells of the placenta are then tested to see if the baby has CF.”

Post-Natal:
- **Genetic Testing**: “The Food and Drug Administration (FDA) has approved the first DNA-based blood test to help detect cystic fibrosis (CF). It is called the Tag-It™ Cystic Fibrosis Kit. This test analyzes human DNA to find genetic variations that may indicate the disease. The test will be used to help diagnose cystic fibrosis in children and to identify adults who are carriers of the gene variations. This test identifies a group of variations in the cystic fibrosis transmembrane conductance regulator (CFTR) gene, which is the cause of cystic fibrosis. The FDA approved this test based on a study of hundreds of DNA samples. These samples were able to show that the test identifies the CFTR gene variations with a high degree of certainty.”
If your child acquires CF here is some supplementary information

- Ongoing medical care is important. You should seek treatment from a team of doctors, nurses, and respiratory therapists who specialize in CF. These specialists are often located at CF Foundation Centers in major medical centers.

- Good self-care includes:
  - Eating a healthy diet
  - Avoiding tobacco smoke
  - Washing your hands often to reduce your chances of infection
  - Exercising frequently
  - Drinking lots of fluids
  - Doing chest physical therapy every day
  - Having annual flu and other appropriate vaccinations
  - Taking your medicines as prescribed

- Courtesy of the National Heart and Lung Institute
Works Cited

- http://upload.wikimedia.org/wikipedia/commons/d/d9/