Genetic Screening for Cystic Fibrosis

A New Choice for You and Your pregnancy
Goals for this Discussion

• Present information about a new screening test for a common genetic disease
• Discuss the nature of this disease, cystic fibrosis
• Discuss how testing can be accomplished
• Discuss what the test will tell you
• Discuss what the test won’t tell you
• Discuss the risks of genetic testing
• Discuss how the test results can be managed
What is Cystic Fibrosis (CF)?

• Cystic fibrosis is an inherited disease which may lead to the development of very thick, easily dried out mucous in nose, lungs, and intestines
• Disease may be mild or very severe
  – Generally it is a chronic life-long illness requiring some kind of lifetime treatment
  – No present cure
Areas affected by CF

• Lungs
  – Thick clogging mucous
  – Frequent coughing and wheezing
  – Frequent pneumonia and bronchitis
• Chronic sinus infections
• Digestion problems
• Altered reproduction
  – Men may be infertile; pregnancy may be risky for affected women
Healthcare for CF patients

- Daily breathing treatments, chest therapy, and medications
- Daily vitamins, enzymes for digestion, and careful diet
- Treatment or preventative treatment for infections
- Treatment for development of diabetes
Outcome of Effective Treatment for CF

- Normal or near normal growth
- Normal intellectual development
- Increasing lifespan
  - About 50% of people with CF live to age 30
  - Babies born now may have average length of life to over 50
Diagnosing CF

- About 1 in 2500 to 1 in 3000 Caucasian babies have CF
- In a child, the diagnosis is made by measuring the amount of salt in the sweat
- Also made by the clinical appearance of the condition
- DNA testing may determine the genetic cause of the disease
What causes CF?

• CF is an inherited (genetic) condition caused by a pair of genes which are not working properly
  – Genes are genetic material passed from parents to children
  – Genes determine how and what proteins are made in the body
  – Genes are made up of DNA
  – Changes in DNA which change the protein the gene makes are called **mutations**
Genetics of CF

- Genes are inherited in pairs, one from the mother, one from the father
  - If the parents carry one mutated gene and one normal gene, they are called carriers
    - **Carriers of one mutated gene are completely normal**
Genetics of CF

• Cystic fibrosis occurs when **both** genes for the CF protein are changed (mutated)
  – Only if the baby **gets an abnormal gene from mother** AND **father** will it have cystic fibrosis
What is your chance of being a CF carrier?

• The chance for a person to be a carrier of CF depends largely on their ethnic background
  – Highest carrier rates in people of Caucasian and Ashkenazi Jewish background
    • Chance is 1 in 29 that people in those groups carry CF
    • Includes people whose background is from England, Scotland, Wales, Scandinavia, Europe
### Chance of Being a CF Carrier by Ethnic Background

<table>
<thead>
<tr>
<th>Ethnic group</th>
<th>Affected child</th>
<th>Carrier rate</th>
<th>Ability to Detect mutation</th>
</tr>
</thead>
<tbody>
<tr>
<td>Europ. Cauc.</td>
<td>1/3000</td>
<td>1/29</td>
<td>80%</td>
</tr>
<tr>
<td>Ash. Jewish</td>
<td>1/3000</td>
<td>1/29</td>
<td>97%</td>
</tr>
<tr>
<td>Hispanic Am</td>
<td>1/9200</td>
<td>1/46</td>
<td>57%</td>
</tr>
<tr>
<td>African Am</td>
<td>1/15,000</td>
<td>1/65</td>
<td>69%</td>
</tr>
<tr>
<td>Asian Am</td>
<td>No data</td>
<td>1/90</td>
<td>unknown</td>
</tr>
</tbody>
</table>
Other Effects of Ethnic Background

• The likelihood that testing can detect a mutation depends on the ethnic group
  – Non-Caucasian or non-Jewish groups are already at low risk for having children with CF
  – Testing may not add much additional information about these groups
CF Carrier Screening

- Testing is available to provide information about your risk for being a CF carrier
- This is termed “screening” testing because it is testing people who do not have the condition
What if there is a family history of CF?

• You and your husband will need genetic counseling about the family history
• Genetic testing for CF will be offered if desired
• You do not fall into the general screening program for CF because you are already a higher risk
Is there a benefit to me to have Genetic Screening for CF?

• Genetic screening can identify if you are at higher risk than you thought for having a baby with cystic fibrosis.

• Genetic screening may be reassuring to you that you have a lower chance of having a baby with this serious problem.
How do I find out if I am a carrier for CF?

• You must sign a consent form stating that you understand what testing means and that you want to be tested

• A blood specimen is needed
  – May be part of New Mothers’ labs if not yet drawn
  – If those labs already drawn, another tube must be drawn

• If you need to think about testing and want to wait, fresh blood may need to be drawn later
Genetic Testing Process

• IF the mother gives consent (checks “Yes” on the consent form), then the blood is tested
  – DNA testing is done on the blood
  – Results come back in approximately 3 weeks

• IF the mother does NOT give consent (checks “No” on the consent form), then no blood is drawn and no testing for CF is done
The results show: a mutation is not found

- If no mutation is found, the risk is reduced and no further testing is done
  - No further CF testing is needed in any other pregnancy unless the father changes and has a family history of CF
What does it really mean: No mutation?

- No mutation means that no abnormality was found when your DNA was studied to see if it showed multiple different mutations

- **HOWEVER**, testing cannot say 100% you are not a carrier; it just reduces your chance of being a carrier and having an affected baby
  - A woman of European Caucasian background without family history of CF is tested for CF. No mutations are found
    - Her risk of being a carrier changes from 1 in 29 to 1 in 140 with a very low chance for an affected baby
The results show: a mutation is found

- If the test finds a mutation (it is a positive test), then **you are a carrier** of a specific known change in the DNA for CF.

- The next step is to find out if your partner is also a carrier.

- **Only if both parents are carriers can you have a baby with CF.**
What if my partner is also a carrier?

- If both parents are found to be carriers of mutations in CF, then they need further information:
  - Detailed genetic counseling about their 1 in 4 risk of having an affected baby
  - Prenatal diagnosis including amniocentesis or other testing to identify if the baby is affected may be of interest to some couples
  - The baby can also have genetic testing at birth
Benefits of Prenatal Diagnosis for CF

• Family can prepare for the birth of a baby with special needs
• Baby will benefit from early treatment from birth to improve health
• Some couples may not wish to continue a pregnancy if they know their baby is affected with cystic fibrosis
New advances in CF

• Even if mutations are found in both parents, it does not always predict how severely affected a child will be
  – Medical advances have improved the lifetime care for CF patients and will continue to do so
What if my baby’s father is unavailable or unknown?

- If the father is not available because he is stationed somewhere else, blood can be drawn from him and mailed back for testing.
- If the father is unknown, the risk to the baby is based only on the mother’s carrier risk.
  - It is possible that an affected baby may be missed if the father is not tested.
Should I get Genetic screening for CF?

• The choice is up to you!
  – The greatest value is if you belong to an ethnic group which has a higher risk of having CF
    • Includes European Caucasian and Ashkenazi Jewish
  – Other ethnic groups have such a low risk of CF that testing may add very little to their knowledge
    • Includes African Americans, Hispanic Americans, and Asian Americans
Do I have to have Genetic Screening for CF?

• No!
• If you do not want genetic screening for CF, mark “NO” on the consent form
  – Your blood will not be tested unless you consent
• If you decide later you want testing, you can give your consent and a new specimen of blood will be drawn
• If you don’t want to be tested, you will still get the same good prenatal care that you would get if you did get tested
  – There’s no penalty for not testing!
Reminder about testing

• All genetic tests are specific—they only look for one particular condition
  – If you do not have a mutation for CF, it does not mean that you could not have a mutation for some other genetic condition entirely

• There is no testing available for all genetic disorders

• If you have a family history of some other genetic problem, you may benefit from genetic counseling to discuss what is your risk of being a carrier of that problem