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Integrated Prenatal Screening

What is an integrated screen?

An integrated screen is a test done during pregnancy that tells you the chance of your baby having Down syndrome, trisomy 18, or spina bifida. The integrated screen does not diagnose these conditions.

How is an integrated screen done?

An integrated screen has 3 parts – An ultrasound done between 11 and 14 weeks of pregnancy and 2 blood draws, done at different times, from the mother's arm.

Ultrasound

During the ultrasound, the baby's length is measured to confirm the due date. Ultrasound is also used to measure the small space under the skin behind your baby's neck, called the nuchal translucency (NT). This space is often larger than normal when the baby has Down syndrome or trisomy 18.

Blood Draws

The first blood draw is done in the first trimester, usually the same day as the NT ultrasound. The second blood draw is done in the second trimester, between 15 and 20 weeks of pregnancy. The blood tests measure certain proteins and hormones made by the baby and placenta, which are found in every pregnant woman's blood. The amounts of these proteins and hormones are often different when the baby has Down syndrome, trisomy 18, or spina bifida.

What do I learn from an integrated screen?

An integrated screen has 3 results, which are numbers such as "1 in 4,000 (0.025%)" or "1 in 75 (1.3%)". These numbers are the odds or chances that your baby has Down syndrome, trisomy 18, or spina bifida. The results are usually available within a week after the second blood draw.

The integrated Screen can detect 9 out of 10 cases (90%) of Down syndrome and trisomy 18, and 8 out of 10 cases (80%) of spina bifida. But, it will not detect all cases of these birth defects, and it does not test for any other health problems. If you are having a multiple birth, an integrated screen is less accurate but may still be helpful.

What are the benefits of an integrated screen?

An integrated screen provides you with information about your baby's health without any risk to your pregnancy.

What are the risks of an integrated screen?

Most people are worried when they get an abnormal integrated screen result. It can cause stress for the rest of your pregnancy, especially if you decide not to have all the follow up testing.

What does a "positive" result mean?

A few women (about 5%, or 1 out of 20) have a "positive" integrated screen. This does not mean their baby has a birth defect. It only means that the risk of their baby having 1 of these 3 conditions is higher than a certain level. Most women who have a positive integrated screen result will have a healthy baby.

If you have a positive integrated screen result, your health care provider will offer you follow-up, such as a visit with a genetic counselor, more specific blood testing and an ultrasound to carefully examine the baby's anatomy. You may choose to have an amniocentesis. Amniocentesis is a test that will tell you whether to not your baby has Down syndrome, trisomy 18, or spina bifida.

What does a "negative" result mean?

Most women (95%, or 19 out of 20) will have a "negative" integrated screen. This does not mean their baby is completely healthy. It only means that the chance of the baby having 1 of these 3 conditions is lower than a certain level. A few women who have a negative integrated screen result will have a baby with Down syndrome, trisomy 18 or spina bifida.

This handout gives information to help you decide if you want to have an integrated screen. Having this test is up to you. Talk with your health care provider to learn more.

Information provided and adapted from University of Washington Medical Center patient education.

WHAT IS CYSTIC FIBROSIS?

Cystic Fibrosis (CF) is a genetic condition that affects one out of every 2,500 Caucasian people in the United States.

- CF causes problems in how the lungs and digestive system work.
- People with CF get many lung infections, like pneumonia.
- Frequent coughing, diarrhea, poor growth, and infertility can also occur.
- Most people with CF will have symptoms before their first birthday.
- A few people have mild problems and do not have symptoms until they become adults.
- CF does not cause developmental delay or a difference in how a person looks.
- There is no cure for CF.
- CF usually shortens the length of time a person lives. Most people with CF live into their 30's.

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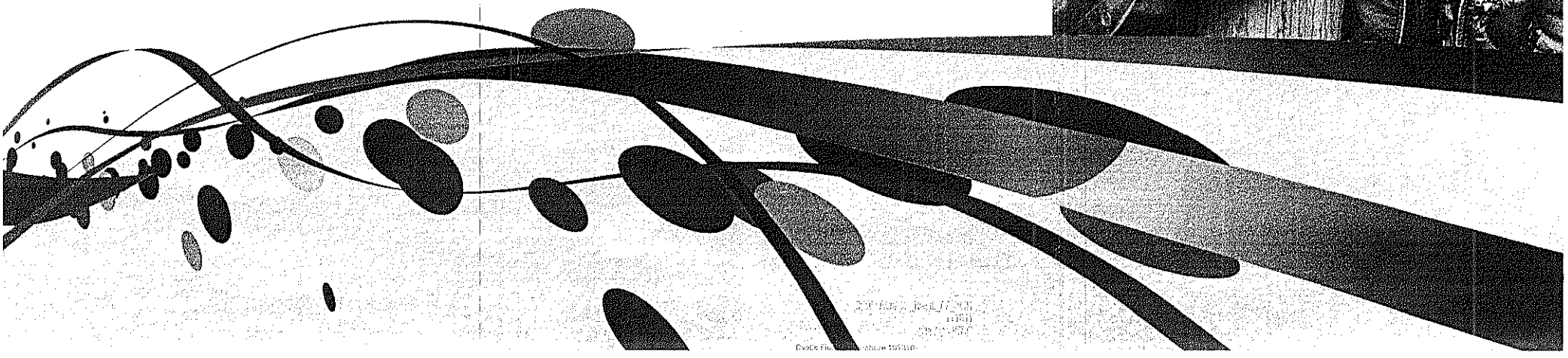
Cystic Fibrosis

Patient Information



WHAT CAUSES CYSTIC FIBROSIS (CF)?

- CF is caused by a change in a gene inherited from both parents.
- Genes are the instruction booklets in our bodies that determine how we grow and develop.
- Each person has two copies of every gene.
- In people with CF, both copies of the same gene do not work.

WHAT DOES IT MEAN TO BE A CARRIER?

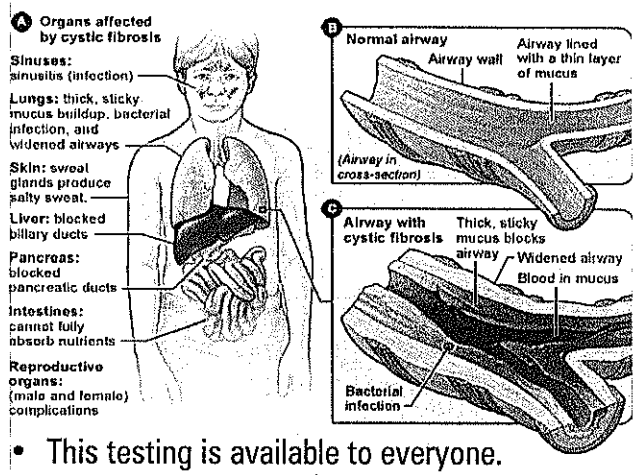
- A person who is a carrier for CF has one copy of the gene that works normally and one copy of the gene that does not work.
- Carriers of CF do not have symptoms.

WHAT ARE MY CHANCES OF BEING A CARRIER?

- Your chance of being a carrier depends on your family history and ancestry (ethnic background).
- People of Caucasian (white) and Ashkenazi Jewish ancestry have a higher chance of being a carrier than people of other ethnic backgrounds.
- If you are Caucasian, you have a 1 in 25 chance (4%) of being a carrier.
- People of Hispanic, African American, Native American and Asian ancestry have a lower chance of being a carrier.
- You may be a carrier even if nobody in your family has CF.
- Your chance of being a carrier is higher if someone in your family has CF.

WHAT IS CYSTIC FIBROSIS CARRIER SCREENING?

- This testing will help determine if you are a carrier for CF and your chance to have a child with CF.
- The testing requires a blood sample.



- This testing is available to everyone. This testing is optional and is not required.
- A positive result means you are a carrier. If you are a carrier and are pregnant or considering pregnancy, carrier testing should be offered to your partner as well.
- A negative result reduces the chance that you are a carrier but does not eliminate it.

WHAT IF MY PARTNER AND I ARE BOTH CARRIERS?

- If both parents are carriers, they have a 1 in 4 chance (25%) to have a child with CF with each pregnancy.
- Diagnostic testing during pregnancy can tell you if the baby has CF.
- If you are both carriers, the next step would be to talk about diagnostic testing with a pregnancy care provider or genetic counselor.