UPMC
LIFE CHANGING MEDICINE

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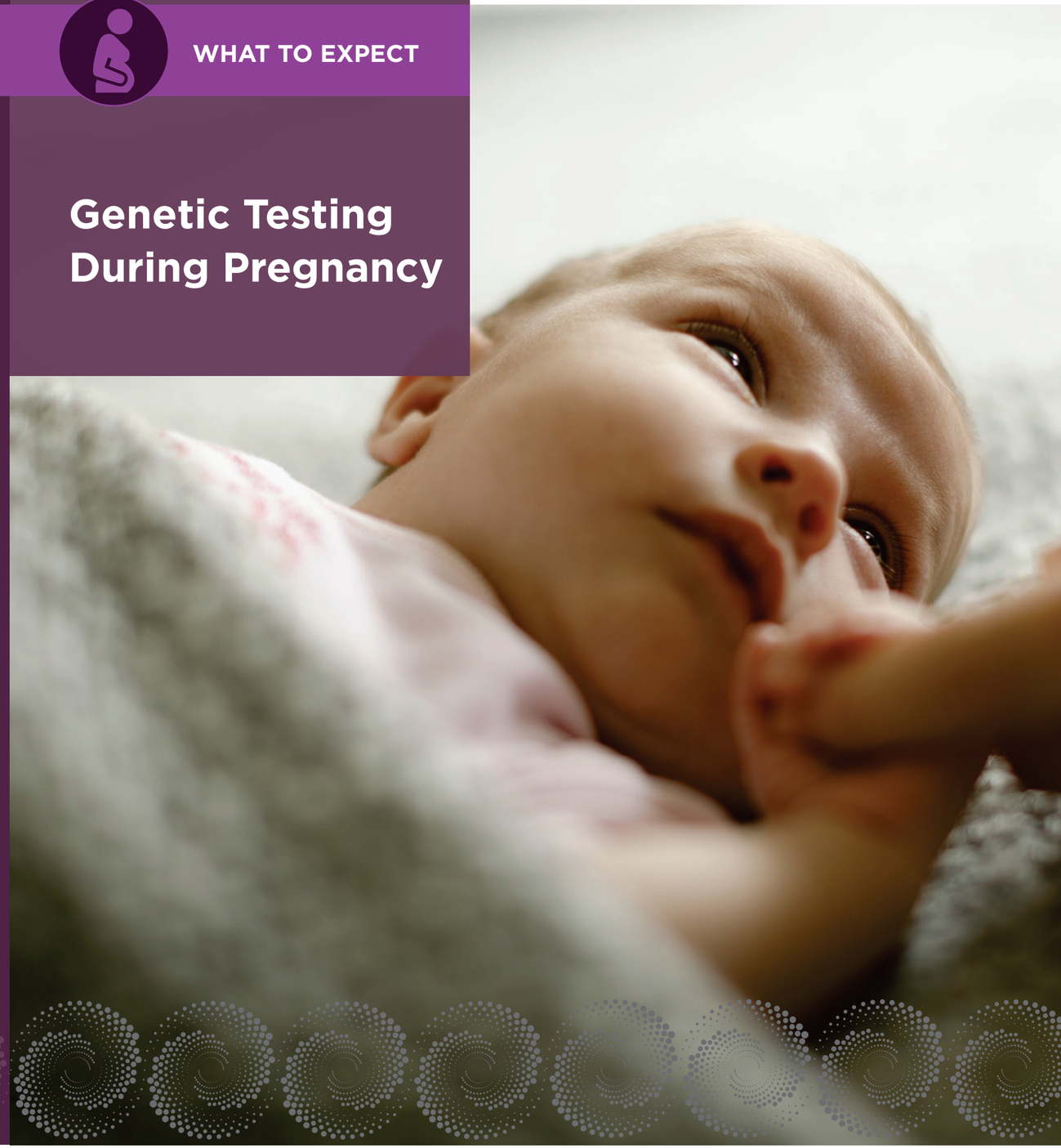
WHAT TO EXPECT

Genetic Testing During Pregnancy



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UPMC | MAGEE-WOMENS

Genetic Testing During Pregnancy

Is genetic counseling for me?

The health of a child can be affected by many different things. A genetic counselor can provide balanced information about all of your options and allows you to use your own values to make choices about your care. These tests are offered to all women during pregnancy. You may want to consider genetic counseling or prenatal genetic testing if you:

- Want to better understand how genetics can affect your baby
- Are 35 years or older and pregnant or thinking of becoming pregnant
- Have a family member with a genetic disease or birth defects
- Have a health problem such as diabetes, high blood pressure, or depression
- Have lost 3 or more pregnancies
- Are concerned about medicines, chemicals, or infections during pregnancy
- Have been told something unexpected was found on an ultrasound of the baby
- Have other risk factors such as alcohol, smoking, or drug use during pregnancy
- Are from an ethnic background with a high risk for specific genetic conditions

Family history is important. Ask your family and your partner's family about possible health conditions and talk with your doctor or genetic counselor about them.

Will insurance cover testing?

- Most insurance plans will cover some kind of genetic screening during pregnancy.
- The cost of testing can be different for people on different insurance plans.
- Options may be available to help lessen the cost of testing.
- A genetic counselor will help you understand how much testing may cost you.

Genetic testing is never required. It is always your choice.

Testing Options

You can talk to your doctor or genetic counselor about what testing options are right for you. There are many to consider.

Screening tests can be done without risk to your pregnancy but are unable to provide a sure yes or no answer to whether your baby has the condition in question.

Before 14 Weeks — 1st Trimester Ultrasound ☐

Checks the general health of the pregnancy and tells you a due date

After 10 Weeks — Non-Invasive Prenatal Screening (NIPS) ☐

Screens for chromosome conditions like Down Syndrome, Trisomy 18, Trisomy 13, and X and Y chromosome disorders

11 to 14 Weeks — 1st Trimester Screening (FTS) ☐

Screens for Down syndrome, Trisomy 18 and evaluates the nuchal translucency and nasal bone by ultrasound

15 to 20 Weeks — Quad Screen (Quad) ☐

Screens for Down syndrome, Trisomy 18, and spina bifida and allows the opportunity to identify risks related to plasma protein levels

18 to 20 Weeks — Anatomy Scan Ultrasound ☐

Checks the baby from head to toe for problems in the way your baby is growing

Family history can be important! Talk to family members about health conditions that may be important to a pregnancy.

Diagnostic tests can tell you if your baby has a genetic disease or not, but come with risks for early delivery or miscarriage of the pregnancy.

11 to 14 Weeks — Chorionic Villus Sampling (CVS) ☐

Tests cells from the placenta to determine if they baby has a genetic or chromosomal condition;
Risk: 1/300

After 16 Weeks — Amniocentesis (Amnio) ☐

Takes cells from the fluid around the baby to determine if the baby has a genetic or chromosomal condition;
Risk: 1/500

At Delivery — Postnatal Testing ☐

Allows for diagnostic genetic testing after delivery using cord blood, tissue, or other sources of DNA.

Carrier screening can test for a genetic disease. Typically if both parents carry a change in the same gene, your baby may be at risk. Common conditions for carrier screening include:

For Anyone ☐

- Cystic Fibrosis (CF)
- Spinal Muscular Atrophy (SMA)

Other Tests to Consider ☐

- Fragile-X Syndrome
- Sickle Cell Anemia
- Thalassemia

Testing for multiple conditions at once ☐

- Core panel
- Jewish panel
- Expanded carrier screening
- Custom panel for you

Results

Screening tests will say low-risk or high-risk. If a screening test gives a high risk result, diagnostic testing is recommended for more information or a definitive yes or no answer.

Diagnostic tests will give 1 of 4 results:

- **Negative** - this result means no disease-causing changes were found for the disease or diseases being looked at.
- **Positive** - this result means a change was found which is expected to affect the baby's health.
- **Uncertain** - this result means a change was found, but it is not known if this change may affect the baby's health.
- **Unexpected** - this result means a change was found that is not related to the reason the test was done but may still affect the baby or parents in some way.

Getting a positive or uncertain result may mean you can consider additional testing. Talk to your genetic counselor or doctor about your options. Diagnostic tests can offer yes or no answers, but also come with a small procedure-related complication risk, which includes miscarriage or early pregnancy loss.

Carrier screening will usually be positive or negative. Positive results mean you are a carrier for a genetic condition. Testing your partner may help you understand risks to the pregnancy. Negative means you did not carry the conditions the test looked for.

Resources

- March of Dimes Birth Defect Foundation
  www.marchofdimes.com
- Family Voices
  www.familyvoices.com
- Genetic Alliance
  www.geneticalliance.org
- MedLinePlus Genetics
  www.medlineplus.gov/genetics
- National Healthy Mothers Health Babies Coalition
  www.hmhb.org
- Family History Tool
  www.hhs.gov/familyhistory