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TESTING OPTIONS FOR CHROMOSOMAL ABNORMALITIES AND BIRTH DEFECTS

Introduction

Almost all children in the United States are born healthy. Out of 100 newborns, only two or three have major birth defects. A birth defect is a physical problem that is present at birth. It also is called a congenital disorder or malformation. For approximately 70% of babies born with birth defects, the cause is unknown. In other cases, birth defects are inherited through genes or chromosomes.

Birth defects are caused by an error in the way the baby develops. A birth defect may affect how the body looks, works, or both. Many birth defects are mild, but some can be severe. Babies with birth defects may need surgery or medical treatment. They might be mentally disabled, physically disabled, and/or have a shortened life span. Most birth defects occur at conception or during the first 3 months of pregnancy. Some birth defects can be found before birth with special tests. Others might not be diagnosed until birth or later in life.

We will be offering you different tests at specific times during the pregnancy which will detect many birth defects. Even if you have all the tests outlined below, there can be no guarantee that your baby will not have a birth defect; however, if the tests show your baby to be at "low risk", the chance of the baby having a birth defect is very small.

Below are the different options available at this time for testing for birth defects.

Anatomical Fetal Ultrasound

At about 18-21 weeks we strongly encourage every patient to have a high level ultrasound by a specialist to make sure that the baby's organs are developing properly. The scan will look at many organs including the brain, kidneys, heart, bladder, spine, and stomach. At this time, if you want to find out the sex of the baby, you can. If the baby is found to have an abnormality of one or more organs, further tests might be recommended. If the baby has a serious abnormality, the doctor will explain how this may affect your child's health and functioning. For some birth defects, we may recommend specific management such as delivery at a specialized hospital, closer surveillance of the baby during the pregnancy or other treatments to improve the outcome for your baby. If the birth defect has very serious implications, you might be given the option of terminating the pregnancy, if that would be the right choice for you.
Screening for Chromosomal Abnormalities

All patients will be given the option of testing for chromosomal abnormalities. As a woman ages, there is a slowly increasing chance that the baby will have an abnormal number of chromosomes. If we calculate your chance of delivering a baby with an abnormal number of chromosomes to be less than 1 chance in 200 (0.5%), we will offer you noninvasive testing (an ultrasound and blood tests) that will recalculate your risks based on your age and the result of those tests. This test is called **Sequential Screen**. This is a sonogram of your baby at a very specific time in the pregnancy – between 11 and 13 ½ weeks, a blood test on you at the same time, and then a second blood test on you at about 16 weeks. In this sonogram, the radiology tech is doing a measurement called **Nuchal Translucency** where the amount of fluid under the skin of the baby's neck is measured. Babies who have an abnormal number of chromosomes tend to have an increased amount of fluid in this area at that gestational age. If these tests show that your risk is increased, we will refer you for further testing as described below for women who are at increased risk, to determine if there is an abnormality. The **Sequential Screen** will detect about 90% of chromosomal abnormalities with a 3.5% false positive rate (3.5% of women who are tested will be referred for further testing and it will be determined with those tests that the baby is normal). The **Sequential Screen** will also detect 80% of open neural tube defects (incomplete closure of the fetal

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spine which can cause spina bifida or anencephaly). If it is too late to do the testing at 11 – 13 ½ weeks, the second part of the test, which is a blood test at around 16 weeks can be done alone (**MSS4**). This will detect 75-80% of chromosomal abnormalities and 80% of open neural tube defects. The results for each of these tests are usually available within two weeks.

Testing for Women at Increased Risk for a Baby with Chromosomal Abnormalities

If we calculate your risk of having a baby with a chromosomal abnormality to be increased (that is, if it is greater than about 0.5% or 1 chance in 200), we will refer you to a genetic counselor so they can go over your specific risks and discuss testing options. Women who would be considered at increased risk include: maternal age greater than 35 at delivery or paternal age greater than 55, a prior child or pregnancy loss with a chromosomal abnormality, or an abnormal Sequential screen or MSS4. Most women elect to start out with noninvasive testing which will include a blood test called **Noninvasive prenatal testing (NIPT)**. Blood is drawn from your arm like any other blood test. This test is able to pick up your fetus' "free DNA" and from that detect some of the most common chromosomal abnormalities including Trisomy 21 (Down's syndrome), Trisomy 18, Trisomy 13 and sex chromosome abnormalities. This test is 80-99% accurate depending on the specific abnormality they are looking for. The result of this test is available within about two weeks. They are also able to determine the sex of the baby. A specialized sonogram along with this blood test will pick up most chromosomal abnormalities. If the above tests indicate that it is highly likely that your baby has an abnormality, or if you are at increased risk and request it, you can have a test called **Amniocentesis**. In this test, a needle is passed through your abdomen to collect some of the amniotic fluid surrounding the baby. Fetal cells that have been sloughed off into the fluid are collected and "grown" to evaluate the baby's chromosomes. It usually takes 2-3 weeks for the final results, although preliminary results might be available within 10 days. Because this procedure is invasive, it carries a small chance of complications (0.3-1%) such as infection, ruptured membranes, miscarriage or failure of the cells to grow.

See our video on this subject: https://www.youtube.com/watch?v=HoW-rRtd9cQ

Cystic Fibrosis Screening

Cystic Fibrosis is a recessive genetic disorder that affects about 1 in 3,500 births. A person who has cystic fibrosis has TWO abnormal genes – one abnormal gene from their mother and one from their father. Most people who carry this gene have no family history of cystic fibrosis. A person who has cystic fibrosis (has two abnormal genes) has abnormally thick mucus and can have respiratory and gastrointestinal problems. Sometimes these problems are very severe and can require hospitalizations, extensive home care or a shortened life span.

The chance of carrying the gene with no family history varies depending on your ethnic origin. Caucasians and people of Eastern European Jewish descent (Ashkenazi) have the highest carrier rate – about 1 in 25 compared to Hispanic with a 1 in 58 chance, African American with 1 in 61 chance and Asians with a 1 in 94 chance.

We will give you the option of being tested to see if you are a carrier. If you are a carrier, we will check the baby's father. If you are both carriers, there is a 25% of having an affected child and you could elect to have amniocentesis to see if your fetus has two abnormal genes. There is no way to improve or change the outcome once you are pregnant, however, if you desired, you could terminate a pregnancy if it was determined that the fetus had two abnormal genes and would have cystic fibrosis. If you would like to be tested to see if you are a carrier, let us know.

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Chromosomal Abnormalities at Term, Live-born Newborns

Maternal Age	Risk of Abnormalities
20	1/526
21	1/526
22	1/526
23	1/500
24	1/476
25	1/476
26	1/476
27	1/455
28	1/435
29	1/417
30	1/385
31	1/385
32	1/322
33	1/286
34	1/238
35	1/192
36	1/156
37	1/127
38	1/102
39	1/83
40	1/66
41	1/53
42	1/42
43	1/33
44	1/26
45	1/21
46	1/16
47	1/13
48	1/10
49	1/8