Prenatal Testing for Birth Defects: Helping You Decide

Most babies are healthy at birth. However, every woman has a small chance of having a baby with a birth defect. Prenatal testing can help find some birth defects before a baby is born. These tests are optional, so the decision about whether or not to have testing, or which test to choose, is up to you.

There are two types of tests that look for birth defects: **screening tests and diagnostic procedures.**

What Screening Tests Tell You

Screening tests estimate your personal risk for birth defects without any risk to the pregnancy. Screening tests do not look for all types of birth defects, but they can help determine your risk for some conditions, such as:

- Down syndrome - a chromosome problem that causes intellectual disabilities and can include physical birth defects.
- Trisomy 18 or trisomy 13 - two types of chromosome problems that cause severe developmental issues, physical birth defects, and a high mortality rate in infancy.
- Open neural tube defects - openings in the baby's spine or skull.
- Abdominal wall defects - openings in the baby's abdomen (belly).
- Smith-Lemli-Opitz syndrome – a very rare developmental disorder that affects many parts of the body.

Most babies with one of these birth defects will have a screening result that prompts more follow-up; however, not all babies with these conditions are identified by a screening test.

State Screening Tests

The California Prenatal Screening Program is the standard prenatal screening test and is routinely offered to pregnant women in California. This test screens for: Down syndrome, trisomy 18, neural tube defects, abdominal wall defects, and Smith-Lemli-Opitz syndrome. State Screening is currently recommended for women who choose to start with a screening test. There are two tests you may take, depending on how far your pregnancy has progressed.

Integrated Screening (Serum or Sequential)

If you are before 14 weeks in pregnancy, State Screening includes two steps:

- **First trimester screen:**
  Blood test between 10 weeks and 13 weeks 6 days of pregnancy.
  Optional - Nuchal translucency (NT) ultrasound between 11 to 14 weeks
- **Second trimester screen:**
  Blood test between 15 and 20 weeks of pregnancy.

Blood testing can be done at any Kaiser Permanente laboratory, but NT ultrasounds are only available at select Kaiser Permanente sites. NT ultrasound is not required to get a State Screening result, but it can give you an early risk result. NT ultrasound may also provide early detection of some physical birth defects.

Quad Screening – 15-20 weeks

If you are after 14 weeks in pregnancy, quad screening provides a result from a second trimester blood test alone.

State Screening Results

Your final State Screening result is sent to your provider about 1 to 2 weeks after the second blood test. A preliminary result for Down syndrome and trisomy 18 is available for women who have a NT ultrasound and the first blood test.

Possible State Screening results

"Screen negative": The chance is low that the baby has any of the screened birth defects and no further testing is offered. You are notified by mail.

"Screen positive": The chance for a birth defect is high enough to consider more testing, although most women with screen positive results have healthy babies. You are contacted by a genetic counselor. The genetic counselor discusses options for follow-up testing, which may include ultrasound, non-invasive prenatal testing (NIPT), and diagnostic procedures.

Non-invasive Prenatal Testing (NIPT)

NIPT is a screening test available to women 35 or older, women with a screen positive result from the State Screening test, and women at high risk for certain chromosome problems. NIPT is not currently offered to low-risk women. NIPT is a blood test done after 10 weeks in pregnancy that measures fragments of the fetal DNA (called cell-free DNA) found in the mother's blood. This screening test targets select chromosome problems: Down syndrome, trisomy 18, trisomy 13, and sex chromosome problems. It does not provide information about other birth defects.

NIPT Results

Final results are available about one to two weeks after the blood test, but not everyone gets a result. About 3 to 5% of NIPT tests come back with NO result.

Possible NIPT results -

"Low risk": It is very unlikely that the baby has any of the screened chromosome problems. You are notified by secure message or by phone. Additional testing is needed to screen for neural tube defects and abdominal wall defects.

"High risk": The baby is likely to have a chromosome problem. You are contacted by a genetic counselor. Diagnostic testing is needed to verify a chromosome problem.

What Diagnostic Procedures Tell You

Diagnostic procedures accurately diagnose chromosome problems during pregnancy. Testing is done directly on fetal cells, which are examined under the microscope. The procedures do not test for all birth defects or all causes of intellectual disability, but will identify Down syndrome, trisomy 18, trisomy 13, as well as other chromosome problems. Each test has a small risk for miscarriage (less than 1 in 300).

What diagnostic procedures are available?

Chorionic villus sampling (CVS) - 10-13 weeks

CVS is done by passing a thin needle through your lower abdomen (belly) or by putting a flexible tube through your vagina and cervix. The doctor takes a small sample of the early placenta (the organ that nourishes the developing baby). CVS cannot be done for all pregnancies and about 2-3% of samples require follow-up to obtain a final result.

Amniocentesis - 15-20 weeks

Amniocentesis is done by taking a sample of the fluid from around the baby, using a thin needle that passes through your abdomen (belly). Amniocentesis also tests for other birth defects, like spina bifida.

Special note: Ultrasound is routinely done in the second trimester to examine the baby's anatomy. This procedure can diagnose some physical birth defects, including neural tube defects and abdominal wall defects. Sometimes ultrasound shows findings that raise concern about a possible chromosome problem in the baby. However, CVS or amniocentesis is needed to accurately diagnose a chromosome problem before birth.

Diagnostic Testing Results

You are called with results about two weeks after your procedure. Most women learn that the baby has typical chromosomes. This provides reassurance that your baby does not have a chromosome problem. If you want, you can also find out the baby's sex.

If CVS or amniocentesis reveals that the developing baby has a chromosome problem, you are offered genetic counseling and consultation with one or more specialists. These discussions cover the options available for your pregnancy based on your specific situation.

How much does each test cost?

The costs of prenatal tests are not always fully covered by your Kaiser Permanente insurance. The actual cost varies depending on your coverage plan. Check with the benefits office to find out what your policy covers.

How can I decide?

Deciding whether or not to have prenatal testing for birth defects is very personal. Consider your answers to the following questions:

- Do I want to learn about birth defects during my pregnancy?
- What would I do with the results?
- Do I want more information about my pregnancy before I decide to have a diagnostic procedure?
- Do I want to know "for sure" about chromosome problems?
- Am I willing to accept a small risk for miscarriage to get the most information about my pregnancy?

If you are having testing in your pregnancy, you only need ONE test to start with.

If you choose to start with a screening test, State Screening is the standard screening test. Both State Screening and NIPT offer information about common chromosome problems, but State Screening has the added benefit of identifying other birth defects and potential pregnancy problems. **However, the chance for a screen positive result from State Screening is higher as your age increases.**

To get more information about any of these tests talk with your OB provider.

Additional resources:

- Kaiser Permanente Genetics website: **www.genetics.kaiser.org**
- Member Services at 1-800-464-4000 for questions about coverage
- California Prenatal Screening website: **http://www.cdph.ca.gov/programs/PNS**

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