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 2 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 (death) 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 offers standard screening testing to all pregnant women in California. Tests screen for: Down syndrome, trisomy 18, neural tube defects, abdominal wall defects, and Smith-Lemli-Opitz syndrome. There are 2 tests you may take, depending on how far your pregnancy has progressed.

Integrated screening (serum or sequential)

If you're before 14 weeks of pregnancy, State Screening includes 2 steps:

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

Blood testing can be done at any Kaiser Permanente lab, 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 to 20 weeks

If you're more than 14 weeks pregnant, quad screening provides a result from a second-trimester blood test alone.

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.

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. A genetic counselor will discuss options with you for follow-up testing, which may include ultrasound, noninvasive prenatal testing (NIPT), and diagnostic procedures.

Noninvasive Prenatal Testing (NIPT)

NIPT is a screening test available to women who:

- Are 35 or older.
- Have a screen positive result from the State Screening tests.
- Are 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 of pregnancy that measures fragments of the fetal DNA found in the mother's blood. This screening test targets specific chromosome problems, such as 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 1 to 2 weeks after the blood test, but not everyone gets a result. About 3 to 5 percent of NIPT tests come back with NO result.

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Possible NIPT results

Low risk: It is very unlikely that the baby has any of the screened chromosome problems. 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. 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 a 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, and other chromosome problems. Each test has a very small risk for miscarriage (1 in 500).

Diagnostic Procedures

Chorionic villus sampling (CVS): 10 to 13 weeks

CVS is done by passing a thin needle through your lower abdomen 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 to 3 percent of samples require follow-up to obtain a final result.

Amniocentesis: 15 to 20 weeks

Amniocentesis is done by taking a sample of the fluid from around the baby, by using a thin needle that passes through your abdomen. 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

If CVS or amniocentesis shows that the developing baby has a chromosome problem, you are offered genetic counseling and consultation with one or more specialists.

Test Costs

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

How to 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 of miscarriage to get the most information about my pregnancy?

For more information about any of these tests, talk with your Ob/Gyn doctor.

Additional resources:

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

This information is not intended to diagnose or to take the place of medical advice or care you receive from your physician or other health care professional. If you have persistent health problems, or if you have additional questions, please consult with your doctor.

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