



Prenatal Testing for Birth Defects: Helping You Decide

Most pregnancies end with the birth of a healthy baby. However, every pregnant woman has a small chance of having a baby with a birth defect. Prenatal testing can find some of these birth defects before a baby is born.

There are 2 types of prenatal tests: **Screening tests and diagnostic procedures.** Either type of prenatal test is available to you, regardless of your age. The tests are optional, so the decision about whether or not to have testing, or which test to choose, is up to you.

What are prenatal screening tests?

Prenatal screening tests can help you find out if you have a higher or lower chance of having a baby with certain birth defects. Screening tests do not look for all types of birth defects, but they can help detect:

- **Down syndrome** - a chromosome abnormality that causes intellectual disability and some physical birth defects.
- **Trisomy 18** - a chromosome abnormality that causes severe intellectual disability and physical birth defects.
- **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 rare genetic condition that causes intellectual disability and physical birth defects.

Screening tests do not increase the chance of miscarriage.

What screening tests are available?

Serum Integrated Screening

Serum Integrated Screening involves two blood tests. For each blood test you need a special lab form filled out by your obstetrician's office. Once you have the form, testing can be done at any Kaiser Permanente laboratory. The first blood test is done between 10 weeks and 13 weeks 6 days of pregnancy. The second blood test is done between 15 and 20 weeks of pregnancy. The test results are available about 2 weeks after the second blood test. Serum integrated screening tests for all of the conditions listed above.

Sequential Integrated Screening

Sequential Integrated Screening uses the same two blood tests as Serum Integrated Screening. It also includes a special ultrasound, called a nuchal translucency (NT) ultrasound. The NT ultrasound is done between 11 weeks 2 days and 14 weeks 2 days of pregnancy. The NT ultrasound is available at a limited number of Kaiser Permanente medical offices, so you may have to travel to have the ultrasound done. A preliminary result for Down syndrome and trisomy 18 is available after the ultrasound and first blood test. Final results are available about 2 weeks after the second blood test. Sequential Integrated Screening tests for all of the above conditions and has a slightly better detection rate for Down syndrome and trisomy 18.

Quad Screening

If you started prenatal care at 14 weeks or later, you can still have a screening

test. Quad screening involves one blood test done between 15 to 20 weeks of pregnancy. Results are available in about 2 weeks. Quad screening tests for all of the conditions listed above.

What the screening results mean

Most women who have a screening test receive "screen negative" results. This means that the chance the baby has one of the birth defects the test screens for is low. The chance is low enough that further testing is not routinely offered. A screen negative result does not guarantee that your baby has no health problems or birth defects.

Some women who have a screening test receive a "screen positive" result. This means the chance for a birth defect is high enough to consider more testing. Women with screen positive results are contacted by the Genetics Department. A genetic counselor will discuss the choices for diagnostic testing, such as amniocentesis or chorionic villus sampling (CVS). You can choose to have further testing to help find out why your test was screen positive. The chance of having a screen positive result is higher as women get older. Most women with screen positive results go on to have healthy babies.

What are prenatal diagnostic procedures?

Prenatal diagnostic procedures can tell for certain if specific birth defects are present in your baby. Diagnostic procedures can find more than 99 percent of all babies with a chromosome abnormality. The procedures do not test for all birth defects or all types of intellectual disability.

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What prenatal diagnostic procedures are available?

Chorionic villus sampling (CVS)

CVS is usually done between 10 and 13 weeks of pregnancy. It is done by putting a thin flexible tube through your vagina and cervix or by using a thin needle through your lower abdomen (belly). The procedure involves taking a small sample of the early placenta (the organ that nourishes the developing baby). CVS checks for chromosome abnormalities in the growing baby. The risk for miscarriage after CVS is less than 1 in 300.

Amniocentesis

Amniocentesis is usually done between 15 and 20 weeks of pregnancy. It is done by taking a sample of the fluid surrounding the baby using a thin needle that passes through your abdomen (belly). This fluid is used to detect any chromosome abnormalities in the growing baby.

Amniocentesis can also find some other birth defects like spina bifida. The risk for miscarriage after amniocentesis is less than 1 in 300. (When amniocentesis is done before 15 weeks of pregnancy, the miscarriage risk is a little higher.)

Special note: You will have a prenatal ultrasound in the second trimester (usually between 17 to 20 weeks). Ultrasound is a good test to diagnose neural tube defects, abdominal wall defects, and some other physical birth defects. Some ultrasound findings may also increase the concern about a chromosome abnormality in the baby. However, only CVS or amniocentesis can accurately diagnose a chromosome abnormality during pregnancy.

What the diagnostic results mean

Most women who have a diagnostic procedure will get normal results. Normal prenatal diagnosis results can provide reassurance that your baby does not

have Down syndrome or another chromosome problem. However, no test can completely guarantee that your baby has no health problems or birth defects.

Once in a while, a prenatal diagnosis result reveals that the developing baby has a chromosome abnormality. If this happens, you will be offered genetic counseling and consultation with one or more specialists. These discussions will include the options available for your pregnancy based on your specific situation.

How much does each test cost?

The actual amount varies depending on your Kaiser Permanente coverage plan. Kaiser members are responsible for any co-pays or fees related to each test. For a screening test, this may include laboratory costs, as well as an additional cost for the NT ultrasound. If you choose to have a diagnostic test, there may be a cost for the procedure. Contact Member Services at (800) 464-4000 if you have questions about your coverage.

How can I decide?

Deciding whether to have a screening test, a diagnostic procedure, or no testing for birth defects during your pregnancy is very personal. It can be helpful to weigh the pros and cons. For example, ask yourself: "What are the reasons to choose this test?" and "What would I do with the results?" List your responses. If you find that you need more information or extra support, consider what additional resources might be helpful to you. You may want to talk further with your provider, review other educational materials, talk with your partner or a close friend, or ask to see a genetic counselor.

Other resources

For more information on prenatal testing, you can look at the Genetics web site: genetics.kaiser.org.

The following decision tree can help you decide which test, if any, is right for you.

Would knowing that my baby has a birth defect help me prepare for having a baby with special needs? Would I consider ending my pregnancy if the baby had a birth defect?

If you answered **NO** to both questions, you may decide not to have any prenatal testing for birth defects.

If you answered **YES** to either question, you may want to consider having some type of prenatal testing.

Do I want to know whether my pregnancy is at higher risk for certain birth defects?
Do I need more information before deciding to have a diagnostic procedure?
Am I willing to accept a test that might miss these birth defects?

Or

Do I want to know for certain if my baby has a chromosome abnormality?
Am I willing to accept a test that has a small risk for miscarriage?

If you answered **YES** to these questions, you may want to consider having Serum Integrated Screening or Sequential Integrated Screening.

If you answered **YES** to these questions, you may want to consider having CVS or Amniocentesis.

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.