Prenatal Testing for Birth Defects:
Testing Options for Twin Pregnancies

Most twin pregnancies end with the birth of two healthy babies. 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 two types of tests that can give you information about birth defects during pregnancy:

- **Screening tests**
- **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. There is no risk for miscarriage from a screening test. Screening tests do not look for all types of birth defects, but in a twin pregnancy they can estimate your chance for:

- **Down syndrome** - a chromosome abnormality that causes intellectual disability and may cause physical birth defects.
- **Trisomy 18** - a chromosome abnormality that results in severe intellectual disability and often causes 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).

What screening tests are available?

The California Prenatal Screening Program offers a choice of screening tests. If you are *less than 14 weeks* pregnant you have the option of having either Serum Integrated Screening or Sequential Integrated Screening.

Serum Integrated screening uses two blood tests to estimate your risk for birth defects. The first blood test is done between 10 weeks and 13 weeks 6 days of your pregnancy. The second test is done between 15 weeks and 20 weeks of pregnancy. Your results are completed about one to two weeks after the second blood test. In a twin pregnancy, Serum Integrated screening tests for Down syndrome, neural tube defects, and abdominal wall defects. Serum Integrated screening does ***not*** test for trisomy 18 in a twin pregnancy.

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. This ultrasound is done between 11 weeks 2 days and 14 weeks 2 days of pregnancy. NT ultrasound is offered at limited locations in Northern California, so you may need to travel to have this ultrasound. A preliminary result for Down syndrome and trisomy 18 is available after the ultrasound and first blood test. Final results are available about one to two weeks after your second blood test. In a twin pregnancy, Sequential Integrated screening tests for Down syndrome, trisomy 18, neural tube defects, and abdominal wall defects.

Quad Marker screening is a single blood test done between 15 weeks and 20 weeks of pregnancy. Results are completed about one to two weeks later. The Quad Marker test makes screening possible for those who start their prenatal care after 14 weeks of pregnancy. In a twin pregnancy, Quad Marker screening tests for Down syndrome, neural tube defects, and abdominal wall defects. It does ***not*** screen for trisomy 18 in twin pregnancies.

What do the screening results mean?

Most women who have a screening test receive a "screen negative" result. This means the chance is low for either one of the babies to have any of the birth defects being screened for.

Your test result is considered "screen positive" if the chance for one of these problems is higher than a certain cutoff. A screening test cannot tell for sure if the baby has a birth defect. If your result is screen positive, a Genetic Counselor will talk with you about the result and the option of more testing, including diagnostic testing.

When a baby has one of the specific birth defects, the test will usually be screen positive, prompting follow-up. However, every screening test misses some babies with these conditions. Screening tests on a twin pregnancy are more likely to miss a birth defect than testing on a single baby. In addition, some pregnancies without birth defects have a screen positive result, even though the babies are fine.

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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.

What prenatal diagnostic procedures are available?

Chorionic villus sampling (CVS)

CVS is usually done between 10 weeks and 13 weeks 6 days 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). In a twin pregnancy, the procedure takes a small sample from each separate placenta (the organ that nourishes the developing baby). Attempts to take a sample from each placenta are not always successful with a CVS procedure. In 25% to 50% of twin pregnancies, the CVS procedure cannot be done. The risk for miscarriage after CVS in a twin pregnancy is somewhat higher than the 1 in 300 risk with a singleton pregnancy.

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). After a sample from the first baby is taken, a safe, temporary dye is sometimes added into the fluid. Then a sample is taken from the second baby. The risk for miscarriage after amniocentesis in a twin pregnancy is somewhat higher than the 1 in 300 to 1 in 500 risk with a singleton pregnancy.

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 do the diagnostic results mean?

Most women who have a diagnostic procedure will get normal results. Normal prenatal diagnosis results can provide reassurance that your babies do not have Down syndrome or another common chromosome problem. However, no test can completely guarantee that your babies have no health problems or birth defects.

Once in a while, a prenatal diagnosis result reveals that one or both of the developing babies has a chromosome abnormality. If this happens, you will be offered genetic counseling and consultation with specialists. The discussion will include a review of your options based on your pregnancy history and your unique situation.

How much does each test cost?

The actual cost depends 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?

You should consider several questions before deciding which test, if any, is right for you.

- Would knowing that one or both of my babies have a birth defect help me prepare for having a baby with special needs?
- Would I consider ending my pregnancy if both of the babies have a birth defect?
- Would I consider having a selective reduction (terminating one fetus) if only one baby has a birth defect?

If you answered no to all of these questions, you may decide not to have any testing for birth defects during your pregnancy.

If you answered yes to any of these questions, you may want to consider having some type of prenatal test.

Screening tests can provide information about your risks for certain birth defects and may help you make a decision about having a diagnostic procedure. Screening tests do not have any risk for miscarriage; but they do not identify all babies with the birth defects included in the screening test.

Some women prefer to go directly to a diagnostic procedure such as CVS or amniocentesis in order to know for sure about the birth defects these procedures can identify. But, both diagnostic procedures have a small risk for miscarriage.

Prenatal testing for birth defects is optional. It is up to you to decide whether to have a screening test, a diagnostic procedure, or no testing for birth defects during your pregnancy.

Other resources

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

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