The California Prenatal Screening Program

The result of your screening test is:

"SCREEN POSITIVE due to a Large Nuchal Translucency"

This means there is an increased risk that the fetus may have a birth defect.

Positive Lg NT

As part of your prenatal care, you had a screening test either in the First Trimester or Second Trimester of pregnancy. This included:

- a blood test and
- a nuchal translucency ultrasound

The screening result tells you the ***risk***, or chance, that the fetus (unborn baby) has a certain birth defect. For example, a risk could be 1 in 40, or 1 in 5,000.

The result of your screening was:
"Screen Positive - Large Nuchal Translucency (NT)".
A nuchal translucency is a special ultrasound that measures the thickness at the back of the fetus' neck. A Large NT means there is a high risk of a chromosome abnormality or a heart defect in the fetus.

When a Large NT is seen in a fetus, there is a greater than 1 in 5 chance of a chromosome abnormality.

Which abnormalities are associated with Large NT?

Chromosomes are packages of genetic information found in every cell of the body. Birth defects can occur when there are too few or too many chromosomes.

Common chromosome abnormalities are:

Turner syndrome: This condition causes short body, short neck, heart problems and reproductive problems. This affects female fetuses and is caused by a missing X chromosome.

Down syndrome: This condition causes mental disability and some serious health problems. Down syndrome is caused by an extra chromosome #21.

Trisomy 18: This condition causes severe mental disability and very serious health problems. Trisomy 18 is caused by an extra chromosome #18.

Because the fetal NT was large...

Your doctor will refer you for genetic counseling and diagnostic testing at a Prenatal Diagnosis Center (PDC). Specific tests will tell you if there are abnormalities such as Turner syndrome, Down syndrome, or Trisomy 18.

A large NT in the fetus **also means there is an increased risk for heart defects**. This could involve blood vessels of the heart or the heart itself.

Fetal Echocardiogram is a special ultrasound of the fetal heart. It can detect many heart defects. It is often done between 18 weeks and 24 weeks.

This special ultrasound is **not covered** by the California Prenatal Screening Program. Your doctor may recommend this test which would be billed to your insurance.

These follow-up services **are available** at a State Approved Prenatal Diagnosis Centers at no additional charge:

Follow-up Services	Time of Service
Genetic Counseling	At first appointment
Chorionic Villus Sampling (CVS)	1st Trimester
Ultrasound	2nd Trimester
Amniocentesis	2nd Trimester

You may accept or decline a referral to a Prenatal Diagnosis Center. After genetic counseling, you may accept or decline further follow-up services.

Genetic counseling is the first service

A genetic counselor will discuss your screening results and what they mean. Your family's health history is also reviewed. The counselor will give you information about the follow-up testing available to you. **You may decline any services or tests at any time.** Be sure to ask the counselor any questions you may have.

The counselor will explain that you have several testing options to choose from: CVS, ultrasound, and amniocentesis discussed on the next pages.

CVS (Chorionic Villus Sampling)

This diagnostic test can only be done early in pregnancy, **between 10 and 14 weeks.** This test removes a few cells from the placenta (in the uterus), using a very thin needle or tube. The fetus is not touched. The cells from the placenta contain the same chromosomes as the fetus. The chromosomes are counted and examined.

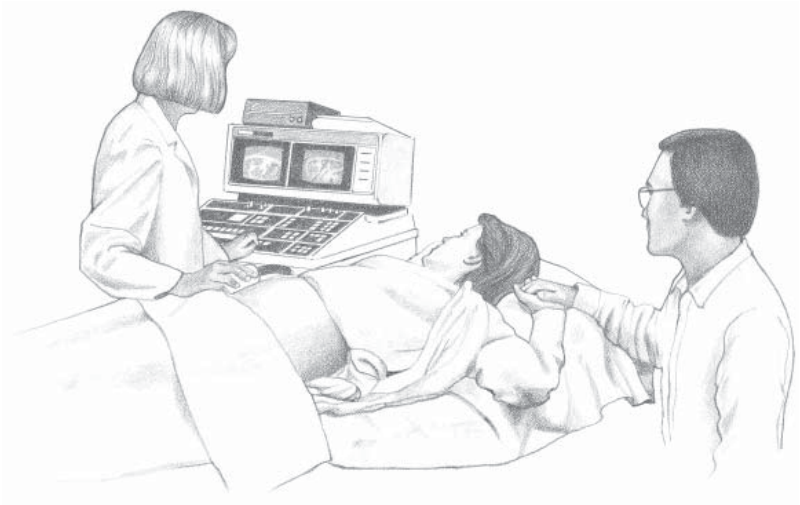
CVS can tell you if your fetus has a chromosome abnormality such as Turner syndrome, Down syndrome or Trisomy 18. It is considered a safe test when performed by the medical experts at a State-approved Prenatal Diagnosis Center. The risk of miscarriage due to CVS is small – less than 1 in 100.

The results of CVS are ready in about two weeks.

Ultrasound at a State-approved Prenatal Diagnosis Center

This test is also called a sonogram. This high level ultrasound is done in the second trimester. It is a very detailed picture of the entire fetus done by doctors with special training. It may identify some birth defects.

You can have this high level ultrasound even if you decline CVS, or amniocentesis.



Amniocentesis at 15 – 24 weeks

This diagnostic test involves removing a small amount of the fluid around the fetus. A thin needle is used to remove a small quantity of the fluid. The fetus is not touched. This fluid contains cells from the fetus. The chromosomes in these cells are counted and examined.

Amniocentesis can tell if the fetus has a chromosome abnormality such as Turner syndrome, Down syndrome or Trisomy 18. Amniocentesis is considered a safe test when done by the medical experts at a State-approved Prenatal Diagnosis Center. The risk of miscarriage due to amniocentesis is small – less than 1 in 100 .

The results of the amniocentesis are ready in about 2 weeks.

If my blood was drawn in the first trimester, should I have another blood specimen drawn?

A patient or her doctor might decide to have another blood specimen drawn in the 2nd trimester of pregnancy instead of having diagnostic testing. Because of the large NT measurement, the screening result will still be positive. The pregnancy will still be considered at high risk for chromosome abnormalities.

What if a chromosome abnormality is found?

A doctor or genetic counselor would give you information about the fetus' condition. Sometimes the birth defect is very mild. Sometimes the fetus is born with mental or physical disabilities. Sometimes the birth defect is severe and the fetus may die. Counselors and special programs are available throughout California to support parents and help children achieve their full potential.

Patient Options

Options for continuing or ending the pregnancy will be discussed during genetic counseling. The decision is entirely up to you.

The California Prenatal Screening Program does not pay for any other medical services after authorized follow-up tests and genetic counseling. Referrals for medical care and support services are available for parents and families through the Prenatal Diagnosis Center.

Please remember:
A "Screen Positive Result" does not always mean there is a birth defect. Sometimes all the diagnostic tests are normal and the fetus is healthy.

The California Prenatal Screening Program

California Department of Public Health
Genetic Disease Screening Program
850 Marina Bay Parkway, F175
Richmond, CA 94804
866-718-7915 (Toll Free)

For more information see our website:
www.cdph.ca.gov/programs/pns

11/2013