What is increased nuchal translucency or large NT?
During an ultrasound, measurements are taken of many parts of the developing baby. Nuchal translucency (NT) measures an area of fluid at the back of a baby’s neck. This is done between 11 and 14 weeks in pregnancy. NT fluid is seen in all babies. When there is more fluid than usual, it is called “increased nuchal translucency” or “large NT”. The NT is considered large when it is 3.0 mm (about 1/8 inch) or more.

Does a large NT mean the baby has a problem?
Usually not. Most babies with a large NT will be healthy at birth. However, a large NT size means a higher chance for problems.

What type of problems are found in babies with a large NT?
Although most babies with a large NT will not have any problems, the following conditions are more likely to be found in babies with a large NT:

- **Chromosome problem** – This is a difference in the genetic make-up of the baby that can cause intellectual disability and birth defects. The most common chromosome problems seen with a large NT are Down syndrome (an extra chromosome 21), trisomy 18 (an extra chromosome 18), and Turner syndrome (a missing X chromosome).

- **Heart defect** – A change in the way the heart has formed. Some heart defects do not need any treatment, while others may require surgery or cause heart failure.

- **Noonan syndrome** – A genetic condition that affects many areas of the body. This condition includes a higher chance for heart problems at birth, shorter height, and a common facial appearance. Some people with Noonan syndrome have learning problems or delays in development. Many people with Noonan syndrome do not need any special care. Special genetic testing is needed to diagnose Noonan syndrome.

- **Pregnancy problems** – There is a higher chance for miscarriage and stillbirth, especially when the NT is very large, or fluid is seen in other parts of the baby’s body. If there are other concerns about the baby, you will get information from the counselors and doctors involved in your care.

Will a large NT go away?
The extra fluid seen with a large NT usually goes away after the 14th week in pregnancy. A follow-up ultrasound is routinely scheduled at 18 to 21 weeks in pregnancy to provide more information about the baby.

This information is not intended to diagnose health problems or to take the place of medical advice or care you receive from your physician or other health care professional.

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What can follow-up testing tell me about the baby?
Follow-up testing can tell you if the baby has certain health problems. You can decide if you want to learn more about the baby’s health during pregnancy.

NIPT (noninvasive prenatal testing) – A blood test that screens for some common chromosome problems. However, NIPT does not identify all chromosome problems. This test can also raise false alarms in a healthy pregnancy. If testing shows a higher chance for a chromosome problem, more testing is needed to be sure. NIPT does not test for Noonan syndrome or heart defects.

CVS or Amniocentesis (diagnostic testing) – Tests that accurately identify chromosome problems in a developing baby by taking a sample directly from the pregnancy. The sample from CVS or amniocentesis may also be tested for Noonan syndrome. There is a small risk for miscarriage associated with these procedures (less than 1 in 500 risk).

Ultrasound – An ultrasound is routinely done between 18 to 21 weeks to check the developing baby and the placenta. This procedure can find some physical birth defects; however, not all birth defects can be found by ultrasound. If there is concern about a heart defect, a special ultrasound of the baby’s heart, called a fetal echocardiogram, may be done. There is no known risk to the mother or baby from ultrasound.

What if all of the follow-up tests are reassuring?
Prenatal testing can find many types of problems during pregnancy, but there is no way to be absolutely certain about a baby’s health before birth. The good news is that with reassuring test results, you will most likely have a healthy baby.

Where can I get more information?
You can speak with your doctor, nurse practitioner, nurse midwife, or a genetic counselor if you have additional questions about this ultrasound finding.

Kaiser Genetics Departments: Website: http://genetics.kp.org/
Fresno….(559) 324-5330   Oakland……..(510) 752-6298   San Francisco..(415) 833-2998
Modesto..(916) 614-4075   Sacramento..(916) 614-4075   San Jose……..(408) 972-3300

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