

The Heart of the Matter

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Lee and Nancy are having a baby! The couple has been trying to conceive for quite a while and a pregnancy has been long anticipated. A specialized ultrasound is done at their obstetrician's office to look at their baby's anatomy. Blood is also drawn to measure two chemicals, freeBeta and PAPP-A, for which high and low levels may be seen in babies with certain birth defects. During the ultrasound the OB measures the amount of fluid in the baby's neck, called a nuchal translucency measurement (NT). The larger the NT is, the more likely the baby may have health problems.

At their appointment, the couple receives the news that their baby's NT is larger than expected. A genetic counselor meets with Lee and Nancy to explain the potential meanings of this finding, including the possibility of chromosome and heart defects. The counselor tells them that although it is possible their baby will be healthy, it appears more likely that there could be a problem. The blood chemistry shows normal levels of freeBeta and PAPP-A, reducing the likelihood of a chromosome abnormality, but exactly what is causing the problem is not yet known. The couple is understandably concerned by this unexpected turn of events.

At 13 weeks of pregnancy, when this ultrasound was performed, the baby is about the size of a peanut. It is difficult at this stage of development to see the tiny fetal heart well enough to diagnose most heart defects, so we are unsure of the prognosis at this point. We do know when a heart defect is present, the NT is often enlarged because fluid accumulates in the baby's neck.

A few anxious weeks later Nancy has an amniocentesis. The results show the baby has normal female chromosomes and the couple, though somewhat relieved, is still worried. Nancy and Lee are referred to a heart specialist. This specialist performs echocardiography at around 18 weeks. The fetal echo shows all four chambers of the baby's heart beating in amazing detail. During this test the cardiologist discovers the problem—the baby has a hole in her heart.

Though Lee and Nancy are concerned, they feel some relief by learning what is happening to them. About one in a hundred babies is born with some type of heart defect, making it one of the most common health concerns. By learning of their baby's condition before birth, the couple is able to change their plans for delivery. Instead of delivering at their local community hospital, Nancy makes plans to go to a specialized medical center at a local university where neonatal heart specialists will be on hand to care for the baby.

When baby Chloe is born, a pediatric surgeon repairs her heart. The hole is closed thanks to the skillful hands of the surgeon and the miraculous technology that allowed the physicians to diagnose and correct the baby's serious condition. Lee and Nancy spend a few nights in the hospital getting very little sleep after their daughter's surgery. Chloe does recover and Lee and Nancy finally bring their tiny daughter home. While in the past Chloe might have died from her condition, her life is an example of the miracles we often take for granted from modern medicine.

GeneCare Medical Genetics Center, in Chapel Hill, and is a partner with the Fetal Medicine Foundation to provide of education and training in the measurement of nuchal translucency. For more information please call (919) 942-0021 or 1-800-277-4363 or visit our web site: www.genecare.com.