GeneCare

The Nose Knows

By Philip Buchanan, PhD, FACMG and Jeff Taska, MS, CGC

Roberta and Charles are having a baby! The couple has been trying to conceive for quite a while and a pregnancy has been long anticipated. After a home pregnancy test for confirmation, Roberta calls her obstetrician to schedule an appointment. At the OB=s office the couple is told about a new method of screening for birth defects through a combination of a specialized ultrasound and blood test. This screening helps identify pregnancies at increased risk for chromosomal conditions and certain other medical disorders.

Charles has a niece with Down syndrome and so is familiar with the condition. His young niece, Allison, was born with a hole in her heart and has significant learning delays, but spends most of her time in regular classroom environment with other children her age. Charles has noticed that although his niece has her mother=s blue eyes, she is also has some features which are unlike other family members and are instead characteristic of Down syndrome. For instance, Allison has a small button nose, small ears, and her neck appears shorter than other children. Charles wonders if his child will be born with Down syndrome. One way to address this question early is to look for characteristics of Down syndrome before a baby is born.

Roberta=s blood is drawn and her OB does an ultrasound at around the 11th week of her pregnancy. Her OB is specially trained and certified to measure the Aneck size@ and Anose size@ of a baby with ultrasound and can use these measurements to help estimate the chances the baby has Down syndrome. The Aneck size@ of a baby is called a nuchal translucency (NT), and the larger an NT measurement is, the more likely a baby has Down syndrome or other health problems.

Interestingly, the nasal bone is either absent or small in most fetuses with Down syndrome, just as children with Down syndrome tend to have smaller noses. A baby whose nasal bone is absent on ultrasound is much more likely to have Down syndrome than the average baby. Conversely, if a nasal bone is seen on ultrasound in the first trimester it reduces the chances the baby has Down syndrome. Nasal bone and NT are the best prenatal ultrasound predictors for Down syndrome available.

A few days after the ultrasound is performed, the results of Roberta=s blood test are complete. The chemicals in Roberta=s blood, the NT measurement from the ultrasound, and information such as Roberta=s age and Charles= family history are all factored into giving the couple their final result, which is that they are not at increased risk for their to have Down syndrome. Charles and Roberta=s baby=s nasal bone is seen at their ultrasound and a photo of their baby=s profile is made as a keepsake.

(Nasal bone measurement is not currently included in the standard risk calculation for Down syndrome screening, but eventually may be formally included once further studies are performed).

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 nasal bone and nuchal translucency. For more information please call (919) 942-0021 or 1-800-277-4363 or visit our web site: www.genecare.com.