What is Down Syndrome?

Humans normally have 23 pairs of chromosomes (46 total) and these are the genetic blueprints that make us who we are. Abnormalities can affect the number and structure of chromosomes. Because chromosome abnormalities affect every cell and every organ of the body, they may cause a variety of birth defects. Down syndrome occurs when a baby has an extra copy of the 21st chromosome, resulting in a total of 47. It is the most frequent serious chromosome abnormality that is present in newborns. It is associated with mental retardation, distinctive features, and an increased risk for structural birth defects, such as heart malformations, intestinal problems, and brain abnormalities.

FACTS

1. The vast majority of babies are healthy at birth.
2. All pregnancies have a small chance (3%) for physical or mental birth defects.
3. The most common cause of mental disability is Down syndrome and trisomy 18.

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Dr. Gulinson's

Nuchal Translucency Testing

First trimester screening is performed early in the pregnancy – usually between 10 ½ and 13 ½ weeks of pregnancy. The screening combines measurements from both a blood test and an ultrasound examination.

A small sample of the mother's blood, drawn from her arm, is required for the screening. The sample is then analyzed for two pregnancy hormones (usually free beta or intact hCG and PAPP-A). Both substances are normally produced during pregnancy.

The ultrasound examination involves measuring the amount of fluid accumulated under the skin at the back of the baby's neck. This normal accumulation of fluid is known as the nuchal translucency (or NT) measurement and it is often increased when a developing baby has Down syndrome. It is critical that this measurement be accurate, therefore it should always be performed by a physician or sonographer that has proper certification.

The ultrasound and blood results are then combined with maternal factors such as age and weight to calculate the chance of Down syndrome in the current pregnancy.

How long do the results take?
You can usually expect your test results to come back in approximately one week or less from the time the blood sample is obtained.

How will the results be reported?
You will receive an individual assessment of the chance for Down syndrome in *your* pregnancy (for example 1 in 800). While each patient will interpret their personal risk somewhat differently, in general, laboratories will choose a cut-off and any risk that is higher than the cut-off will be reported as "screen positive". All values lower than the cut-off are reported as "screen negative".

How accurate are the results?
Your individual risk based on the ultrasound examination and blood test is a very accurate reflection of the chance for your developing baby to have Down syndrome. Ninety percent of pregnancies with Down syndrome will have results in the screen positive range. This means that roughly 9 out of 10 of the pregnancies with Down syndrome will come back as 'screen positive'. However, a 'screen positive' result does not mean that the baby has Down syndrome. It simply means that the risk is high enough that you should consider further evaluation of the pregnancy.

What else can the first trimester screening tell me?
First trimester screening can also determine whether the pregnancy has an increased chance for trisomy 18. Trisomy 18 is a chromosome change that results in severe mental retardation and multiple birth defects. First trimester screening detects approximately 90% of pregnancies with trisomy 18.

What does it mean if my first trimester screening test result is positive for Down syndrome?
When a laboratory calculates the chance for a developing baby to have Down syndrome the results can range from 1 in 5 (20%) to 1 in 10,000 or less (0.01%). Each laboratory establishes a cut-off risk and any risk that is over that cut-off is considered to be *screen positive*. Being told your result is screen positive does not mean that the developing baby definitely has Down syndrome; it simply means that the chance is high enough for you to be offered further testing. In fact, the majority of women with 'screen positive' results do *not* go on to have a baby with Down syndrome.
Screening tests are not diagnostic and do not provide definitive answers. Screening tests identify a high-risk group.