



## First Trimester Screening & Nuchal Translucency

First Trimester Screening is a screening test that evaluates a woman's risk of having a child with Down syndrome or Trisomy 18. Maternal blood tests & nuchal translucency ultrasound are two available screening tools that are used in combination to provide a numerical risk estimate of your pregnancy being affected by one of these conditions.

### What is Down syndrome?

Down syndrome happens when a baby has an extra chromosome 21 (three copies instead of two). People with Down syndrome have mild to moderate intellectual disabilities and other potential health problems. There is no way to predict how serious the problems will be or how mildly or severely the child will be affected.

### What is trisomy 18?

Trisomy 18 happens when a baby has an extra chromosome 18 (three copies instead of two). Babies born with Trisomy 18 have severe malformations commonly affecting the heart and the brain. Many are miscarried or stillborn. All babies with Trisomy 18 have severe developmental delays. The chance of having a baby with Trisomy 18 increases with age.

### What is a nuchal translucency?

A nuchal translucency (NT) is an ultrasound measurement of the fluid at the back of a baby's neck between 11 and 13<sup>+6 days</sup> weeks of pregnancy (best timeframe between 12 and 13<sup>+3 days</sup> weeks). It is a screening test to determine if your baby is at an increased or decreased risk of certain abnormalities. All babies have some fluid collection behind the neck, but babies with Down syndrome or Trisomy 18, tend to have an unusually high amount. Therefore, an NT provides valuable insight specific to your pregnancy.

### Who performs nuchal translucency at The Kelowna Regional Fertility Centre?

NT's are performed by a Fetal Medicine Foundation (FMF) certified ultrasonographer under the supervision of Dr. Kathy Wise, M.D., F.R.C.S. Dr. Wise is an Obstetrician Gynecologist who has completed the Nuchal Translucency Theory Course and Quality Assurance Program and oversees our FMF certified ultrasonographer.

### What options are available at The Kelowna Regional Fertility Centre for prenatal genetic screening?

There are two options available for women who wish to have a nuchal translucency ultrasound as part of their prenatal genetic screening. Screening tests combine the measurements from the ultrasound with results from at least one blood test. The two options are:

1. First Trimester Screening (FTS)
2. Integrated Prenatal Screening (IPS)



### **First Trimester Screening (FTS)**

FTS consists of a nuchal translucency ultrasound measurement combined with a blood test (PAPP-A and free beta-hCG) performed between 11 and 13<sup>+6 days</sup> weeks gestation. The results will provide a numerical risk estimate of the chance of Down syndrome and trisomy 18.

#### Advantages:

The main advantage of FTS is the speed of the results which are available within 7-10 days of your screen. FTS can provide women with reassurance regarding their baby's health early in the pregnancy. FTS has the ability to detect 85-90 % of babies with Down syndrome or Trisomy 18. The detection rate increases to 95% with nasal bone measurements.

#### Disadvantages:

Approximately 5% of patients will have a positive screen when their baby is in fact normal and healthy. This is called a false positive and may lead to an unnecessary amniocentesis. In addition, this test does not screen for open neural tube defects (spina bifida) which requires another blood test (AFP) in the 2<sup>nd</sup> trimester of your pregnancy.

### **Integrated Prenatal Screening (IPS)**

This screening test combines measurements from a nuchal translucency ultrasound with two separate provincially funded blood tests to estimate the chances of a developing baby having Down syndrome, trisomy 18 or an open neural tube defect. The first blood test is done between 10 and 13<sup>+6 days</sup> weeks, the NT ultrasound between 11 and 13<sup>+6 days</sup> weeks and the second blood test between 15 and 20<sup>+6 days</sup> weeks gestation. The bloodwork is arranged by your primary obstetrical caregiver and is done at an outside lab. Only the NT is done here at The Kelowna Regional Fertility Centre.

#### Advantages:

Results from an IPS offer a slightly higher detection rate (up to 95%), and slightly lower false positive rate (<5%) than a FTS assessment, meaning that fewer women will be offered an unnecessary amniocentesis. In addition, IPS also includes a screen for open neural tube defect as part of the second blood test.

#### Disadvantages:

The main disadvantage of IPS is the delay in obtaining results. IPS results are not available until 10 days after the second blood test (16 to 17 weeks). This can delay decision making about further testing options and thus diagnosis.

### **How do I get my results?**

Your FTS results will be available within 7-10 days and we will contact you directly. These results will also be sent to the family physician, midwife, or obstetrician that will be following you

through your pregnancy. Please note that IPS results will only be available to the ordering physician and you will be contacted by that office and not our clinic.

**What is a “Screen Positive” result?**

If your result is reported as “screen positive” for Down syndrome or Trisomy 18, it means that your test result has shown a higher risk than the test cut off. You will be offered further counselling to help you better understand this result as well as definitive testing with amniocentesis. It is important to know that most patients with a “screen positive” result actually have a normal healthy baby.

**What is a “Screen Negative” result?**

If your result is reported as “screen negative” for either Down syndrome or Trisomy 18, it means that your test result shows a risk that is lower than the test cut off. No further testing is required. There is a small chance that your screen is falsely negative and your child does have Down syndrome or Trisomy 18.

**What is the cost?**

Please refer to the Kelowna Regional Fertility Centre’s current fee schedule.