

## Information about prenatal testing

Congratulations on your pregnancy !

At this stage it is important to inform you that there are tests which are available to you – to help identify that your baby is healthy and chromosomally normal.

Before discussing these tests, it is important to know some basic information.

1. There are chromosomal risks related to both maternal and paternal age. By far the most significant is maternal age. Advanced maternal age (AMA) is defined as age > 35 years old at the time of delivery. In women older than 35 there are increased genetic risks in pregnancy. These risks are primarily related to the fact that the eggs from which the pregnancy was conceived have been present from the time the mother was an embryo. The aging process of the egg involves mechanisms of cellular division. Defects in the cellular division process can lead to abnormal chromosome numbers in the fetus. Abnormal chromosome numbers lead to defects such as Down syndrome (extra chromosome 21 or trisomy 21), trisomy 18, and trisomy 13. Many of these abnormalities in chromosome numbers will lead to a higher miscarriage. As a woman gets older, there is a higher chance that she may have a baby with a chromosomal abnormality. As a woman gets older – there is also a higher risk of miscarriage. Most miscarriages occur because nature has identified that there is a problem with the baby.

The table below indicate the risks of chromosomal problems and miscarriage rates associated with age.

Age	Down Syndrome risk	Any Chromosomal Abnormality	Miscarriage Risk
20	1 in 1,667	1 in 526	10%
30	1 in 952	1 in 386	12%
35	1 in 378	1 in 192	18%
40	1 in 106	1 in 66	34%
45	1 in 30	1 in 21	53%

2. Every baby is at risk for having a birth defect. This is irrespective of a woman's age. Common birth defects include such things as spina bifida, cleft lip, cleft palate, club foot, hernias etc etc. The average risk is about 3 – 5 %. These abnormalities are not necessarily associated with chromosomal

abnormalities so would not be detected by chromosomal testing such as amniocentesis. Most major abnormalities however can usually be picked up by a detailed fetal ultrasound which is usually recommended to all pregnant women at 18 weeks gestation. In Vitro fertilization and ICSI may be associated with a slightly increased risk of such abnormalities.

## Available prenatal Screening

### **What is a screening test?**

It is very important to remember what a screening test is before getting one performed. This will help alleviate some of the anxiety that can accompany test results. Screening tests do not look only at results from the blood test. They compare a number of different factors (including age, ethnicity, results from blood tests, etc...) and then estimate what a person's chances are of having an abnormality. These tests DO NOT diagnose a problem; they only signal that further testing should be done.

The tests that are available include the following.

### Nuchal translucency screening

Nuchal translucency (NT) is an ultrasound measurement of fluid normally found under the skin behind the baby's neck. It is used as a screening test to determine if your baby is at increased (or decreased) risk of certain abnormalities. The NT is measured between 11-14 weeks of pregnancy by ultrasound examination. All babies have some amount of fluid collected behind the neck. The amount of fluid increases as the baby grows so it is important to measure how big the baby is when measuring the NT. The NT measurement is a very precise measurement, which should only be performed by a specially trained certified sonographer.

The NT will be increased in most but not all pregnancies affected with Down syndrome. In other words, most babies with Down syndrome will have more fluid present behind their neck than is expected. Because of this, the NT measurement along with the maternal serum screening blood test results can be used to estimate the chance of Down syndrome in a pregnancy. It is important to understand that an increased NT measurement does not mean the baby has Down syndrome.

A markedly increased NT measurement can also be seen in pregnancies affected with other chromosome problems such as Trisomy 18 and in babies with heart defects. Overall, most babies with an increased NT will be healthy.

Nuchal translucency testing and Integrated prenatal screening ( which includes a blood test at 12 weeks PLUS the Nuchal screen) is not readily available in British Columbia, however efforts are being made to try and improve availability. Preference is given to women over the age of 38 years and to twin pregnancies.

### Bio chemical screening ( blood tests) - SIPS

There are certain hormones and proteins produced by your pregnancy. ( baby/babies) .These can be measured in your blood at various times in your pregnancy. At this time these screening blood tests are done first at between 10 – 14 weeks gestation, and then secondly at between 15 – 20 weeks. The blood tests will only be interpreted once the second sample has been taken.

This screening program in BC is referred to as the SIPS test ( this stands for Serum Integrated Prenatal Screen)

### How is the SIPS test performed?

As mentioned above this test is done in 2 stages. The first blood sample was drawn between 10 weeks and 14 weeks, and the second sample between 15 and 20 weeks. Once the laboratory has both samples then the testes run. The laboratory will then calculate the risk of the baby having a problem based on maternal age, ethnicity, maternal weight, and if possible will integrate into this calculation the results of a nuchal translucency ultrasound. There is information on nuchal translucency ultrasound elsewhere in this handout.

### What are the risks and side effects to the mother or baby?

Except for the discomfort of drawing blood, there are no known risks or side effects associated with these blood tests

### Who will be offered this test ?

All pregnant women should be offered the SIPS , but it is especially recommended for women who:

- Have a family history of birth defects
- Are 35 years or older
- Used possible harmful medications or drugs during pregnancy
- Have diabetes and use insulin
- Had a viral infection during pregnancy
- Have been exposed to high levels of radiation

### What does the SIPS look for?

The screen is measuring high and low levels of AFP ( alpha-fetoprotein) , PAPP A ( pregnancy associated plasma protein A) and abnormal levels of hCG( human chorionic gonadotropin) and estriol ( a form of estrogen) . The results are combined with the mother's age, weight, ethnicity and gestation of pregnancy in order to assess probabilities of potential genetic disorders.

High levels of AFP may suggest that the developing baby has a neural tube defect such as spina bifida or anencephaly. However, the most common reason for elevated AFP levels is inaccurate dating of the pregnancy.

Low levels of AFP and abnormal levels of hCG and estriol may indicate that the developing baby has Trisomy 21( Down syndrome), Trisomy 18 (Edwards Syndrome) or another type of chromosome abnormality.

Abnormal levels of PAPP A may be associated with a higher risk of Down syndrome however may also be a mock for other pregnancy related complications

Although the primary reason for conducting the test is to screen for genetic disorders, the results of the SIPS can also be used to identify:

- A multiple pregnancy
- Pregnancies that are more or less advanced than thought
- A higher risk of other pregnancy related complications

### What does and abnormal test mean

It is important to remember that SIPS is a screening test and not a diagnostic test. This test only notes that a mother is at a possible *risk* of carrying a baby with a genetic disorder. SIPS is known to have a high percentage of false positive results.

Abnormal test results warrant additional testing for making a diagnosis. A more conservative approach involves performing a second triple screen followed by a high definition ultrasound. If the testing still maintains abnormal results, a more invasive procedure like amniocentesis may be performed.

Invasive testing procedures should be discussed thoroughly with your healthcare provider and between you and your partner. Additional counseling and discussions with a counselor, social worker or minister may prove helpful.

### What are the reasons for further testing?

The SIPS is a routine screen that is not an invasive procedure and poses no risks to the mother or baby. An abnormal result often warrants additional testing or more careful pregnancy surveillance. The reasons to pursue further testing or not vary from person to

person and couple to couple. Performing further testing allows you to confirm a diagnosis and then provides you with certain opportunities:

- Pursue potential interventions that may exist (i.e. fetal surgery for spina bifida)
- Begin planning for a child with special needs
- Start addressing anticipated lifestyle changes
- Identify support groups and resources
- Make a decision about carrying the child to term

Some individuals or couples may elect not to pursue testing or additional testing for various reasons:

- They are comfortable with the results no matter what the outcome is
- Because of personal, moral, or religious reasons, making a decision about carrying the child to term is not an option
- Some parents choose not to allow any testing that poses any risk of harming the developing baby

It is important to discuss the risks and benefits of testing thoroughly with your healthcare provider. Your healthcare provider will help you evaluate if the benefits from the results could outweigh any risks from the procedure.

#### For patients who have had IVF and ICSI

It is important to note that the above-mentioned prenatal blood screening MAY NOT screen for some of the specific chromosomal abnormalities which may be associated with ICSI. Some of the genetic risks related to ICSI are unique – and may also be related to the degree of sperm abnormalities that the male has. For instance – if the male has a severely low sperm count – the genetic risks related to ICSI are higher than when the sperm count is only mildly to moderately abnormal. One of the specific problems related to ICSI is an increased risk of sex chromosome disorders. Overall the specific genetic risks related to ICSI are approximately 1 % (approximately half of these will be sex chromosome disorders and the other half somatic chromosome problems – such as Down's syndrome)

The only conclusive way to screen for all of the genetic problems which may be associated with ICSI would be to have an amniocentesis.

#### **Detailed Level 11 Ultrasound screening**

It is recommended to all pregnant women that they have a detailed ultrasound performed at between 18 and 20 weeks gestation. This allows a detailed examination of the baby, the amniotic fluid volume and the placenta. Approximately 70 % of major birth defects can be identified by this ultrasound.

Although ultrasound can carefully examine the baby's anatomy – it cannot evaluate function. So – an ultrasound may not be able to identify such problems as Autism, Mental function, or chromosomal abnormalities – unless they are associated with structural defects.

The above tests are all screening tests. If a problem is identified, or if you choose to have a more definitive test because you have been identified as being at higher risk for having a baby with a chromosomal problem – then you can proceed to having a definitive ( specific test) such as an amniocentesis

### **Amniocentesis**

Amniotic fluid is the thin watery substance that surrounds the developing fetus in the uterus/womb. Amniocentesis involves removing a small amount (about 4 teaspoons) of this fluid with a needle, for testing in the lab. This test can be done after the 15th week of pregnancy.

### **Who qualifies to have an amniocentesis ?**

Women who are at increased risk of having a baby with a chromosomal abnormality qualify for this test to be done. This includes the following...

1. All women over the age of 40 years
2. All women who have ICSI as part of In Vitro Fertilization
3. All women who have had an abnormal triple SIPS ( this is the blood test done at 15 – 18 weeks of pregnancy to check for various hormones and proteins produced by the developing baby and placenta – and can give a clue as the presence of an abnormality)
4. Women who have previously had a baby affected by a chromosome abnormality
5. If an ultrasound has identified certain risk factors.

Your doctor or midwife will book your test.

Amniocentesis is available at the following hospitals in BC.

- Victoria General Hospital
- Surrey Memorial
- Royal Columbian Hospital
- Kelowna
- Nanaimo
- BC Womens' hospital in Vancouver

You should have a light meal before coming to the hospital. No other special preparation is necessary.

### **Who will do my procedure?**

A doctor who has completed special training in this procedure will perform your amniocentesis. A nurse and a sonographer (ultrasound technician) will be assisting.

Who should come with me?

It is advisable that you not drive yourself, or take public transportation (bus) after the test.

For this reason, we recommend that your partner, or another person of your choosing, accompany you for your test. If they do not wish to view the actual procedure, they may wait in the lobby, or sit just outside the procedure room. If you have to come alone, we recommend taking a taxi home, if possible.

Another adult must accompany children in case they need to be taken out of the procedure room.

### **What will happen?**

Before having your test, the doctor will review the procedure with you, make sure you are aware of the available options, and answer your questions. We will obtain your written consent. You will receive information to take home about what to expect after your test.

It is not necessary for you to change your clothes. Wear something loose and comfortable that can be slipped down below your hips easily. Towels are used to help maintain your privacy and to protect your clothing.

Ultrasound is used to locate the baby and placenta. The baby's heart rate is recorded before (and after) the procedure. Some measurements are taken to check the baby's growth and to confirm your weeks of pregnancy. With the ultrasound picture on the screen, the doctor finds the safest and easiest place to insert the needle. An antiseptic solution is used to clean the skin. (The pink stain on your skin will wear off in a few days.) The needle is carefully guided to the selected spot and the small sample of fluid is withdrawn through the needle.

During the test, you may experience some cramping and a slight sensation of pressure. Most women find it no worse than having a blood test taken from their arm.

## **What can Amniocentesis detect?**

Amniocentesis is a specific test that is able to tell you if your baby has a normal number of chromosomes (46). It can find Down syndrome and other major chromosome abnormalities. A second test is done to measure the amount of alpha-fetoprotein (AFP) in the amniotic fluid. A higher than normal amount may suggest the possibility of a neural tube defect, such as spina bifida. Normal results can take up to 3 weeks. Concerns are usually identified and given to your doctor or midwife within 10-14 days.

If there is a family history of a known problem, other special tests may be done on the amniocentesis sample. You need to discuss this with your doctor or midwife who will refer you to Medical Genetics for additional tests in advance of your procedure.

Five in one hundred pregnant women ( 5 % ) have a chance of having a baby with a birth defect that can be found during the first year of life. There are many forms of birth defects that may occur however some include the following : cleft lip and palate, various forms of hernias, clubfeet, and heart defects . The results from Amniocentesis, CVS or fetal blood sampling will indicate whether or not there are chromosome abnormalities or neural tube defects, but will not be able to rule out other abnormalities. A detailed ultrasound at 18 weeks gestation will identify 75% of major congenital birth defects

What are the risks involved in Amniocentesis?

- **Miscarriage:** It is important to note that the risk of miscarriage unrelated to any procedure is about 6 in 100 at nine to twelve weeks of pregnancy, decreasing to 1 in 100 by the fifteenth week of pregnancy. Following amniocentesis, there is an additional risk that one to two in two hundred women will have a miscarriage (1-2 in 100 for twin pregnancy). Two in one hundred women will have problems, such as amniotic fluid leakage or bleeding.
- **Damage to the Fetus:** It is rare for the needle to touch the fetus because ultrasound is used as a guide, and the needle can be moved in another direction. Sometimes amniocentesis must be repeated because the cells do not grow in the laboratory, or there are not enough cells in the fluid sample.
- **Bleeding, Cramping or Infection:** Cramping during or following the procedure is not uncommon and usually gets better with rest. Small amounts of bleeding or amniotic fluid leakage may occur following the procedure. Infection following amniocentesis rarely occurs, and would likely begin to show with a fever and flu-like symptoms.

## **What happens after my procedure?**

You do not need to stay in the hospital following your test.

We recommend that you rest for the next 24 hours. You do not have to go to bed; resting on the couch or in a reclining chair is fine. Avoid heavy lifting and use the time to relax with a favourite book, video, etc. If you need a note to be off work, the doctor who does your procedure will sign it for you.

**When are the results available ?**

The cells from the amniotic fluid are cultured in a laboratory until there are enough to evaluate the chromosomes. This usually takes about 10 – 14 days. Your doctor will normally receive the results within 2 – 3 weeks of your having the test done. No news is good news – in that if the result is abnormal the Geneticist will normally notify your doctor within 12 – 14 days. It is important that you check with your GP/Midwife/Obstetrician if you have not heard of the result within 3 weeks of the test being done.

Please make sure that you read and understand what this is all about – and contact me if there is anything further you wish to discuss.

Dr.Stephen Hudson