

Genetic Screening: Your options

Every pregnant woman in British Columbia may choose to have their pregnancy tested for certain genetic diseases. This is a blood test for the mom. Not all women choose to do this testing. This page is designed give you an outline of your testing options. **We would like you to schedule an appointment to review this information with your physician in more detail, so please book a time to see your doctor with your questions.**

First

This testing is *optional*. It is important that we talk to all expectant parents about this testing, but it is *more important* for you to decide if this testing is right for you.

Second

This testing doesn't check for *all* genetic problems. It really only tests for 3 things:

1. **Trisomy 21 (“Down’s Syndrome”)**: this is where the baby has three copies of chromosome 21, when you should only have 2. People with this all have learning disabilities (ranging from at least moderate, to very profound), and many also have other health problems that may involve their heart or other organs.
2. **Trisomy 18 (“Edward’s Syndrome”)**: this is where the baby has three copies of chromosome 18, when you should only have 2. People with this problem have more profound difficulties than Down’s syndrome. It is also less common.
3. **Neural Tube Defects (“Spina Bifida”)**: this is where the baby had a problem growing its spinal cord and, rarely, it’s brain. People with this problem often have difficulties walking or using their legs, and may have difficulties with controlling their bladder or bowel. Often, people with this problem have normal thinking powers and don’t have learning disabilities.

All of these problems are life-long and have no known cure. Parents are encouraged to consider how they might feel about raising a child with a disability, as this often guides parents about whether they want to pursue testing or not. Options for ending the pregnancy are available, if parents choose.

Third:

Timing is important. This type of testing is started early in your pregnancy, often between 10 to 12 weeks. We need to meet with you SOON to review this.

Fourth:

This testing doesn't give you a YES or NO answer. The blood testing, once completed, will come back to your doctor as a result, either:

1. Your pregnancy is at low risk for these problems.
2. Your pregnancy is at higher risk for these problems.

In the event that your pregnancy is found to be at increased risk, we will call you to come into the office to review the results. The majority of these babies are normal, however, you may choose to have further testing to “know for sure”.

Please turn over...

Genetic Screening: Your options, cont...

There are 2 testing options that are available to you:

Serum Screening:

This is covered by MSP, and is free. There are 2 blood tests, one done between 10-14 weeks, and a second one done after 15 weeks. If you are 35 years old, or older, when the baby is born, there is an additional ultrasound done between 11 -14 weeks. The testing results are generally available about 7 to 10 days after your last blood test. This means that women's results are usually available around 16 to 17 weeks. If further testing is required (such as amniocentesis), this is organized immediately, but there is also a 2-3 week wait to obtain these results afterwards.

This is a useful weblink for more information:

<http://www.bcprenatalscreening.ca/>

First Trimester Screening:

Currently, this is offered by 2 fertility clinics in Vancouver. You must pay for this privately (approximately \$500). You contact the clinic yourself and book an appointment for this service. They collect a blood test and do an ultrasound, between 11 – 14 weeks. A second blood test is not required. You will be given your screening results very quickly, usually within 1-5 days. This means that most women's results are available early, around 12-14 weeks. If further testing is required (such as amniocentesis), this is covered by MSP, but there may be a delay involved in booking this testing. For example, amniocentesis is usually only performed after 16 weeks of pregnancy. Remember again, testing results from amniocentesis take 2-3 weeks.

Useful weblinks for the clinics are listed here. Both clinics offer excellent service.

Genesis Fertility Centre:

<http://www.genesis-fertility.com/fertility-services/first-trimester-screening>

Pacific Centre for Reproductive Medicine:

http://www.pacificfertility.ca/?page_id=17

This information is complex! As expectant parents, we encourage you to read this over, take a look at the websites and consider if any of this testing might be meaningful to you. Please book an appointment to talk to your physician, whether you think you might do the testing or not.

Family Practice Maternity Service, Feb 2011