****

**What is prenatal screening?**

Prenatal screening provides a woman with her risk of having a child with:

[**Down syndrome**](http://www.prenatalscreeningontario.ca/Default.aspx?cid=15&pre=view) **(Trisomy 21)**

**T**[**risomy 18**](http://www.prenatalscreeningontario.ca/Default.aspx?cid=17&pre=view)

**O**[**pen Spina Bifida**](http://www.prenatalscreeningontario.ca/Default.aspx?cid=16&pre=view)

Each woman starts with a risk of having a child with a chromosome abnormality based on her age at the time of delivery.  Screening alters this risk, either lowering or raising it based on blood and ultrasound measurements.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| |  |  | | --- | --- | | Maternal Age | Chance of Down Syndrome | | 20 | **1/1650** | | 25 | **1/1250** | | 30 | **1/900** | | 35 | **1/385** | | 36 | **1/300** | | 37 | **1/225** | | 38 | **1/175** | | 39 | **1/135** | | 40 | **1/100** | | 41 | **1/80** | | 42 | **1/60** | | 43 | **1/50** | | 44 | **1/40** | | 45 | **1/30** | |

Screening doesn’t test for all chromosome abnormalities and it doesn’t screen for every birth defect. Screening doesn’t tell you for sure whether your baby has one of these conditions, only the CHANCE of this. Screening tests can be done starting at 11 weeks of pregnancy and are available for ALL pregnant women.

**Down syndrome**

Down syndrome, or trisomy 21, is a condition that is the result of an extra chromosome 21.  This can lead to health and learning difficulties.  Since the extra chromosome is found in every cell in the body, it cannot be fixed.

People with Down syndrome usually have mild to moderate intellectual delay.  People with Down syndrome are more likely to have health problems than the average person like heart, stomach, bowel, thyroid, vision and hearing difficulties. Treatment is available for many of these health conditions. People with Down syndrome generally live into their fifties. Each person with Down syndrome is different. There is no way to test how serious the disabilities will be.

**In general, about one in 1000 births has Down syndrome. The chance of having a child with Down syndrome increases with the mother’s age.**

**Trisomy 18**

Trisomy 18 is a genetic condition that is the result of an extra chromosome 18.  Babies with trisomy 18 have very serious mental and physical disabilities. Many pregnancies with trisomy 18 will miscarry. Most babies born with trisomy 18 do not survive past the first few months of life.

**In general about one in 6,000 births has trisomy 18. The chance of having a child with trisomy 18 increases with the mother’s age.**

**Neural tube defects**

Neural tube defects occur when the brain or spinal cord does not form properly.  Spina bifida causes physical disabilities such as difficulty walking, and controlling the bladder and/or bowel. People with spina bifida may also have mental disabilities. Treatment can help with many of the physical disabilities.

**In Canada, the chance of having a baby with an open neural tube defect is about 1 out of every 2000 births. The chance of a child being born with this condition does NOT increase with the mother’s age.**

**Is prenatal screening right for me?**

Many women have a difficult time deciding whether or not to have a prenatal screening test.

Remember, it is your choice whether or not to have prenatal screening.

Here are some things to think about that might help you make your decision and maybe discuss with your partner, health care provider, family or friends.

**Do you want to know the chance of your baby having Down syndrome, trisomy 18 or open neural tube defect?**

• there is no cure for Down syndrome, trisomy 18 or open neural tube defect.

• if your baby has one of these conditions, your health care provider may suggest additional tests and/or change in the way your baby is delivered.

• some people have screening because they would like the information before birth, or would like to prepare for a child with special needs.

• some people have screening because they would terminate their pregnancy if their baby were found to have one of these conditions.

**How will this information affect your feelings throughout your pregnancy?**

**If your screening test result is positive do you want further testing?**

**If your diagnostic testing shows your baby has one of these conditions, what would you do with the information?**

**Screening test choices if your first visit with your health care provider is before 14 weeks**

|  |  |  |  |
| --- | --- | --- | --- |
| **Test** | **First Trimester Screening (FTS)** | **Integrated Prenatal Screening (IPS)** | **Serum Integrated Prenatal Screening (SIPS)** |
| **Tests for** | **Down syndrome, trisomy 18** | **Down Syndrome, trisomy 18 spina bifida** | **Down Syndrome, trisomy 18, trisomy 13, X and Y chromosome abnormalities** |
| **1st blood sample** | 11-14 weeks | 11-14 weeks | 11-14 weeks |
| **Nuchal translucency ultrasound** | 11-14 weeks | 11-14 weeks | NONE |
| **2nd blood sample** | NONE | 15-20 weeks | 15-20 weeks |
| **Results available** | 2-5 days | 2-5 days after step 2 | 10 business days |
| **Detection rate** | Down Syndrome: 83%  Trisomy 18 91%  This means…of every 100 pregnancies with Down syndrome, about 80-85 will be detected | Down Syndrome: 88%  Trisomy 18: 90%  Open Spina Bifida: 85%  This means…of every 100 pregnancies with Down syndrome, about 85-90 will be detected | Down Syndrome: 99%  Trisomy 18: 98%  Trisomy 13: 80%  X and Y: 99%  This means…of every 100 pregnancies with Down syndrome, about 99% will be detected |
| **False positive rate** | 3 to 9 out of 100 pregnancies  (3-9%) | 2 to 4 out of 100 pregnancies (2-4%) | Less than 0.1% |
| **Pros and Cons** | + early result  - no spina bifida testing, higher false positive rate | + lower false positive rate, spina bifida testing  - slightly later result | + high detection rate, early result, low false positive rate  - cost if you don’t qualify for coverage |
| **Diagnostic test if prenatal screening test is positive** | CVS 11-13 weeks  If CVS is not available, you could have amniocentesis | Amniocentesis  15-22 weeks | Amniocentesis  15-22 weeks |

**If your first visit to your health care provider is AFTER 14 weeks, there is still a screening option for you. It is called Second Trimester Screening or MSS. It tests for the same conditions as the IPS. The detection rate is about 75-85% and the false positive rate is 5-10%.**

**At the present time, FTS, IPS and MSS are covered by the Ministry of Health. SIPS is NOT covered in many cases but is available to patients who wish to pay for it. It currently costs approximately $800.**

**Results of screening tests**

Prenatal screening results are reported as either SCREEN NEGATIVE or SCREEN POSITIVE

Screening results are not definitive - they provide you with a CHANCE (a numerical probability) for your baby to have Down syndrome, trisomy 18 and/or an open neural tube defect.

Screen Negative Results

The chance that your baby has Down syndrome, trisomy 18 or an open neural tube defect is **LOWER** than the specific cut-off set by your center.

* There is a less than 1 in 1 000 (less than 0.1%) chance of your baby having Down syndrome that was not identified by the screening test.
* Having a normal screen doesn’t guarantee the birth of a healthy baby. Some health problems are not detected by prenatal screening.

Screen Positive Results

The chance that your baby has Down syndrome, trisomy 18 or an open neural tube defect is **HIGHER** than the specific cut-off set by your center.

* A screen positive result does NOT mean that your baby has one of these conditions. In fact, most of the time when the result is POSITIVE, the baby doesn’t have any health problem. This is called a “false positive”.
* A screen positive result means you have the option to have genetic counseling and / or further testing to learn if your baby has one of the conditions.
* About 1 out of 1 000 women will have a TRUE POSITIVE result, which means that after diagnostic testing or birth, the baby does have one of these conditions.

**Resources for parents**

**Canadian Down Syndrome Society** [www.cdss.ca](http://www.cdss.ca/) 1 800 883 5608

More information about Down syndrome with links to local parent support groups.

**S.O.F.T. Support Organization for Trisomy 18, 13 and Related Disorders**

[www.trisomy.org](http://www.trisomy.org/) 1 800 716 7638 or 585 594 4621 More information about trisomy 18

**Spina Bifida and Hydrocephalus Association of Canada**

[www.sbhac.ca](http://www.sbhac.ca/) 1 800 565 9488 More information about spina bifida

**The Genetics Home Reference - Your Guide to Understanding Genetic Conditions**

[www.ghr.nlm.nih.gov](http://www.ghr.nlm.nih.gov/)

An excellent site to learn more about genetics including Down syndrome, trisomy 18 and more.

**Motherisk** [www.motherisk.org](http://www.motherisk.org/) 416 813 6780 or 1 877 327 4636

Information service for women who have been exposed to environmental agents, drugs and alcohol during pregnancy.

**The Genetics Education Project** [www.mtsinai.on.ca/FamMedGen](http://www.mtsinai.on.ca/FamMedGen)

**March of Dimes**

[www.marchofdimes.com](http://www.marchofdimes.com/) excellent source of information about pregnancy including common prenatal tests, complications during pregnancy, things to avoid during pregnancy, birth defects.



**What is prenatal screening?**

Prenatal screening tests tell you the chance that your baby has [**Down syndrome,**](http://www.prenatalscreeningontario.ca/Default.aspx?cid=15&pre=view) **T**[**risomy 18**](http://www.prenatalscreeningontario.ca/Default.aspx?cid=17&pre=view) **or O**[**pen Spina Bifida.**](http://www.prenatalscreeningontario.ca/Default.aspx?cid=16&pre=view) These are conditions that can cause health difficulties or even serious disabilities in a child.

**Is prenatal screening right for me?**

Lots of women have a hard time deciding whether or not they want to have testing. Here are some things to think about that might help you make your decision and maybe discuss with your partner, health care provider, family or friends.

**Do you want to know the chance of your baby having one of these conditions?**

**How will this information affect your feelings throughout your pregnancy?**

**If your test result is positive would you want further testing?**

**If further testing showed your baby has one of these conditions, what would you do with the information?**

**The tests**

If you see your doctor before 14 weeks, you can have:

**FTS:**  ultrasound and bloodwork between 11-14 weeks, tests for Down Syndrome and Trisomy 18

**IPS:** ultrasound and bloodwork between 11-14 weeks and more bloodwork after 15 weeks, tests for Down Syndrome, Trisomy 18 and Open Spina Bifida.

**SIPS:** bloodwork after 11 weeks, tests for Down Syndrome and Trisomy 18, many patients need to pay for this test and the cost right now is about $800

If you see your doctor after 14 weeks, you can have:

**MSS:** bloodwork after 15 weeks, tests for Down Syndrome and Trisomy 18 and Open Spina Bifida.

There are pros and cons for each of these tests—some give you results EARLY, some are more ACCURATE. Most give you results quickly. The biggest decision is whether or not you want testing in the first place.

**The Results**

Remember that these screening tests don’t test for all birth defects or illnesses and only tells you the CHANCE that your baby has one of these conditions. If your test is NEGATIVE (not abnormal), we recommend no more testing. If your test is POSITIVE (abnormal), we recommend that you speak to genetic counselors in Hamilton who can talk to you about options for more testing. Unfortunately, because of how screening tests work, most POSITIVE tests are false alarms and rare NEGATIVE tests miss babies who have the conditions above.

Ask us questions and we are happy to help you decide!