Yukon

Congenital Anomalies Support Yukon

An Aid to Decision-Making for Prenatal Screening

What is prenatal screening?

Prenatal screening is a type of non-invasive testing that is available to all pregnant individuals in Yukon. It uses blood tests to tell you the chance (e.g. one in 1000, one in 20) that your unborn baby may have certain genetic conditions (birth defects). These tests cannot tell you for sure whether or not your baby has one of these conditions, only the chance of it happening. A diagnostic test can do that.

Prenatal screening tests are optional – it is your choice whether to have a prenatal screening test.

What conditions does the screening test for?

Prenatal screening tests for the following conditions:

- **Trisomy 21 (T21)**, also known as *Down syndrome*.
- **Trisomy 18 (T18)**, also known as *Edwards syndrome*.
- **Trisomy 13 (T13)**, also known as *Patau syndrome*.
- **Neural tube defects (NTD)**, such as spina bifida and anencephaly.

You will find more information about these conditions below.

Who might think about prenatal screening?

All pregnant individuals can choose to have prenatal screening. The chance that a baby will have a genetic condition increases with the mother's age.

AGE-RELATED CHANCE OF BIRTH DEFECTS				
Mother's age	Chance of T21 (Down syndrome)	Chance of T18	Chance of T13	Chance of neural tube defect
25	1 in 2500	1 in 25000		1 in 1000 for all ages
30	1 in 840	1 in 8400		
35	1 in 356	1 in 3560	1 in 5000	
38	1 in 166	1 in 1660	1 in 2000	
40	1 in 94	1 in 940	1 in 909	

A decision to make

Doing and not doing prenatal screening are both good choices. Making the decision might be easier if you:

- Base your decision on the best scientific information available.
- Share your thoughts with your doctor/midwife and your family.
- Base your decision on your values and preferences. It can be helpful to ask yourself:
  - *Do I want to know the chance of my baby being affected by one of these conditions?*
  - *How would I use this information?*
  - *How might doing the screening test affect my feelings during the pregnancy?*

Information to help you make the decision

What is Trisomy 21 (T21) or Down syndrome?

- Trisomy 21 occurs when a baby has an extra copy of chromosome 21, which affects how the baby develops and grows.
- People with T21 have almond-shaped eyes, a round face, poor muscle tone, greater risk of vision and hearing problems, heart, stomach and bowel defects, and intellectual disabilities that can be mild or moderate.
- Sixty per cent of children with T21 need specialized home care.
- Some adults with T21 have jobs and are almost completely independent.
- People with T21 can have meaningful emotional relationships and lead lives that are fulfilling for themselves and their family and friends.

What is Trisomy 18 (T18) or Edwards syndrome?

- Trisomy 18 occurs when a baby has an extra copy of chromosome 18.
- Many pregnancies with T18 will miscarry.
- Babies that are born with T18 rarely live more than a few days or months because of serious heart and brain defects and poor growth before and after birth.
- Fifty per cent of babies born with trisomy 18 survive beyond their first six to nine days and about 12 per cent of babies survive the first year of life.

What is Trisomy 13 (T13) or Patau syndrome?

- Trisomy 13 occurs when a baby has an extra copy of chromosome 13
- Many pregnancies with T13 will miscarry.
- Due to multiple serious medical defects, only five to ten per cent of babies born with T13 will live to one year of age.

What is a neural tube defect?

- An open neural tube defect (NTD) occurs when the brain or spinal cord does not form properly.
- Spina bifida is an NTD in which the spine does not completely close. People with spina bifida may have both physical and mental disabilities.
- Anencephaly is an open NTD involving the brain. A baby with anencephaly will be stillborn or die shortly after birth.

Types of prenatal screening

There are two types of non-invasive prenatal screening tests available in Yukon: **Serum Integrated Prenatal Screening** (SIPS) and **Non-Invasive Prenatal Test** (NIPT).

What is Serum Integrated Prenatal Screening (SIPS)?

SIPS tells you if you have a higher chance of carrying a baby with T21, T18, or an NTD.

SIPS requires two blood samples taken:

- One between week 9 and the end of the week 13.
- The second between week 15 and the end of the week 20 (preferably between 15 and 16 weeks).

The result of SIPS is available about 10 days after the second blood test.

This information can help you decide whether to prepare for a child with special needs, offer the child for adoption, or consider ending the pregnancy.

If the chance is high for either T21 or T18, your doctor/midwife will offer you NIPT or an amniocentesis.

What is Non-Invasive Prenatal Test (NIPT)?

NIPT is more accurate than SIPS. It identifies whether you are at high risk for T21, T18, or T13. It can also tell you the risk of certain sex chromosome anomalies (see supplemental information).

NIPT requires a single blood draw taken any time after 10 weeks of pregnancy.

The results for NIPT are available within about 10 days.

NIPT is covered by Yukon Health **ONLY** for individuals at higher risk for T21/T18/T13. Those who are not considered high risk but choose NIPT instead of SIPS must cover the cost of $495.

Individuals who are considered high risk are those who:

- Are 35 years or older when their baby is born.
- Are found to be at higher risk based on their SIPS results.
- Are pregnant with twins.
- Other factors may be considered in rare situations.

NIPT does not screen for neural tube defects (NTDs). If you chose NIPT, screening for NTDs will be done by your detailed ultrasound at 19-20 weeks' gestation.

If the chance is high for either T21, T18 or T13, your doctor/midwife will offer you an amniocentesis to confirm the result.

Although SIPS can detect a pregnancy with an increased risk of T18 and NTDs, and NIPT can detect a pregnancy with an increased risk of T18 and T13, most cases will also be detected by ultrasound. For this reason, the benefits and harms of doing or not doing SIPS or NIPT will focus on screening for T21.

Glossary of terms

Amniocentesis: Sometimes called "amnio", this is a prenatal test that is used when the baby is at a higher risk of having an abnormality. While watching an ultrasound, a needle is inserted through the birth parent's abdomen to take a small sample of the fluid around the baby. There is a risk (about one in 200) that the test could cause a miscarriage.

Birth defect: Birth defects are physical differences in a person's development that are present at birth. Some birth defects have a genetic cause, others an environmental one.

Chromosome: Chromosomes carry the genetic information that we inherit from our mother and father. They are found inside almost all of the cells in our bodies. Humans have 46 chromosomes – 23 from our mother, and 23 from our father. If a person doesn't have the usual number of chromosomes, this usually leads to serious health issues and can even cause death.

Non-invasive: A non-invasive prenatal test (like an ultrasound) poses no known risk to the fetus. An invasive test (like an amnio) could cause a miscarriage.

Prenatal diagnostic testing: This test will give you a definitive answer on whether or not your baby has a genetic condition. Amniocentesis is an example of a diagnostic test.

Prenatal screening: These tests will give you an indication of whether you are at higher risk of carrying a baby with a specific condition. SIPS and NIPT are examples of screening tests.

Trisomy: A trisomy is when a person has three copies of a specific chromosome. Normally humans have only two copies of any given chromosome. The most common is Trisomy 21 syndrome, better known as Down Syndrome, where a person has three copies of chromosome 21.

The benefits and harms of prenatal screening

Again, doing and not doing prenatal screening are both good choices. The information provided here can help you make the choice that's best for you.

Serum Integrated Prenatal Screening (SIPS)

Benefits

- **Know your chances of carrying a baby with T21**

Out of every 5000 people screened, 500 have a result that says they are at higher risk for carrying a baby with T21. If these 500 have NIPT or an amniocentesis to know for sure, only 13 would actually be carrying a baby with T21.

- **Prepare to end the pregnancy**

Some who know they are carrying a baby with T21 will choose to end the pregnancy.

- **Prepare for a child with T21**

Some who know they are carrying a baby with T21 will choose to continue the pregnancy and can prepare for a child with T21 or may consider an adoption plan.

- **Reassurance**

Out of 5000 individuals who take the test, 4500 have a result that means they are at low risk for carrying a baby with T21. These people are reassured.

- **No risk**

There is no risk that this test will cause harm to the baby, making it an extremely safe test.

Potential Harms

- **False alarm**

Out of the 500 people whose screening results show they are at increased risk of carrying a baby with T21, 487 are actually NOT carrying a baby with T21.

Many of these people will experience anxiety.

- **Anxiety while waiting for results**

Those waiting for test results have anxiety levels 10 times higher than normal.

- **May have to face difficult decisions**

The 500 people whose screening results show they are at increased risk of carrying a baby with T21 will need to decide about having further testing. Those who have testing and are shown to actually have a baby with T21 will need to make a decision about whether to continue or end the pregnancy.

- **False reassurance**

Of the 4500 individuals whose screening results show they are at low risk for carrying a baby with T21, two will actually be carrying a baby with T21. These two are falsely reassured.

Discussion with Your Care Provider

What is your personal risk of having a baby born with T21, T18, T13 or neural tube defect based on your age? Check the table on page 1 to know your risks.

Be sure that you understand the following:

- What the tests are for.
- How and when you get results.
- Options for further testing if your screen result shows a high risk.
- Private pay options.
- Benefits and harms of the tests.

Non-Invasive Prenatal Test (NIPT)

Benefits

- **High detection rate**

There is a high detection rate for T21 (greater than 99 per cent). Most babies with T21 will be detected by NIPT.

- **Prepare to end the pregnancy**

Some people who know they are carrying a baby with T21 will choose to end the pregnancy.

- **Prepare for a child with T21**

Some who know they are carrying a baby with Down Syndrome will choose to continue the pregnancy and can prepare for a child with T21. These people may consider an adoption plan.

- **Reassurance**

Those who choose NIPT and have a low risk result are reassured.

- **No risk**

There is no risk that this test will cause harm to the baby, making it an extremely safe test.

Potential Harms

- **False alarm**

Although the rates for false positives are low at less than 0.1 per cent, a person's positive results could be false which will cause unnecessary stress.

- **May have to face difficult decisions**

People who are faced with a positive test result must make a decision about whether to continue or end the pregnancy.

- **False reassurance**

False negative results can happen; this means that a person who tests negative is actually carrying a baby with T21. This happens rarely, but these people are falsely reassured.

- **May need to pay for NIPT**

Since Yukon Health only covers NIPT if a pregnancy is considered high risk, those with a low risk pregnancy will need to pay for the service. Alternatively, they may choose SIPS, or to not undergo screening.

- **Inconclusive results**

Inconclusive results happen in about 3 per cent of those who undergo NIPT. This may cause high levels of stress and anxiety. Further testing can be pursued.

The benefits and harms of NOT doing prenatal screening

Benefits

- **Avoid anxiety and extra testing**

Some people wouldn't consider invasive testing and/or terminating a pregnancy regardless of their result.

- **Stay true to your personal convictions and values**

For some, not doing the test is the right choice for their personal or family's convictions.

- **Avoid difficult decisions**

Not doing the test can avoid the anxiety and stress of making a decision about continuing or ending the pregnancy if the baby has T21, T13 or T18.

Potential Harms

- **Not knowing your risk of carrying a baby with T21**

Out of 5,000 individuals who do not undergo any screening, 15 are carrying a baby with T21. These people do not have the knowledge to prepare for giving birth to a baby with T21.

- **Anxiety from not knowing**

Those who don't take the test may be anxious because they don't know if their child will have T21 or not.

- **Possible social pressure to do screening**

What are the benefits and harms that matter most to you?

Doing SIPS or NIPT	NOT doing SIPS or NIPT
Benefits	**Benefits**
_____	_____
_____	_____
_____	_____
Harms	**Harms**
_____	_____
_____	_____
_____	_____

What is your decision?

☐ **Do SIPS** ☐ **Do NIPT** ☐ **No Screening** ☐ **I don't know**

Are you comfortable with this decision?

Sure of myself	1) Do you feel sure about the best choice for you?	Yes	No
Understand information	2) Do you know the benefits and harms of doing or not doing the test?	Yes	No
Risks and Benefits	3) Are you clear about which benefits and harms matter most to you?	Yes	No
Encouragement	4) Do you have enough support and advice to make a choice?	Yes	No

References

Schieve et. al. Disabil Health J. 2011; (4): 68–77. ACOG Practice Bulletin No. 77. Obstet Gynecol. Jan 2007;109(1): 217-227. Morris et al. J Med Screen. 2002; 9(1): 2-6. Malone et al. N Engl J Med. 2005; 353(19): 2001-2011. Wald et al. Health Technol Assess. 2003;7(11): 1-77. Green et al. Health Technol Assess. 2004; 8(33): iii, ix-x, 1-109. Won et al. Prenatal diagnosis. 2005; 25(7): 608-611. Harper, Peter. Practical Genetic Counselling, Seventh Edition. Harold Arnold, 2010. Chitayat, David. "Trisomy 18 (Edwards Syndrome)". About Kids Health, http://www.aboutkidshealth.ca/En/HealthAZ/ConditionsandDiseases/Genetic Disorders/Pages/trisomy-18-edwards-syndrome.aspx. 2016.

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Supplementary information – sex chromosome anomalies (SCA)

Chromosomes are the structures inside our cells that contain our DNA. Everyone has 23 pairs of chromosomes. Twenty-two of these pairs are the same between men and women, while the 23rd pair, the sex chromosomes, are different:

- Boys are typically born with one X and one Y chromosome
- Girls are typically born with two X chromosomes

Sex chromosome anomalies occur when a person is missing an X or Y chromosome, or has an extra one. This type of disorder is fairly common, and can affect a person's development and health. These people can lead fulfilling lives and develop meaningful relationships. These changes can be detected by NIPT but are not screened for by SIPS. Some people choose not to screen for SCA.

<table>
<tr><th colspan="5">AGE RELATED RISK OF SEX CHROMOSOME ANOMALIES IN BABY AT BIRTH</th></tr>
<tr><th>Mother's age</th><th>Turner (TS)</th><th>Triple X</th><th>Klinefelter (XXY)</th><th>XYY</th></tr>
<tr><td>35</td><td rowspan="3">1 in 2500
female births</td><td>1 in 1000
female births</td><td>1 in 825
male births</td><td rowspan="3">1 in 1000
male births</td></tr>
<tr><td>38</td><td>1 in 550
female births</td><td>1 in 450
male births</td></tr>
<tr><td>40</td><td>1 in 385
female births</td><td>1 in 300
male births</td></tr>
</table>

What is Klinefelter syndrome (XXY)?

Klinefelter syndrome occurs when a boy is born with an extra X chromosome. Most babies born with Klinefelter syndrome appear normal and do not begin showing physical symptoms until puberty.

Not every person with Klinefelter syndrome is affected in the same way. Some may not even know they carry an extra X chromosome. Some individuals are taller, have more breast tissue and wide hips, and almost all of them have smaller testicles and are infertile. Some boys have language and learning difficulties or behavioral or mental health issues requiring extra support.

Individuals with Klinefelter syndrome have a normal life expectancy.

What is Turner syndrome (TS) ?

Turner syndrome occurs when a girl is missing part of, or all of, an X chromosome. Many pregnancies with TS will miscarry.

Most individuals with TS will have short stature and cardiac problems, some may have extra skin folds on the back and side of the neck. During puberty, most girls develop very little breast tissue, do not menstruate, and will have problems with fertility later in life. Most people with TS have normal intelligence but some have difficulty developing social skills.

Due to the risk of cardiac conditions the life expectancy of people with TS is shortened compared to the general population.

What is triple X syndrome (XXX)?

Triple X syndrome occurs when a girl is born with an extra X chromosome.

People with triple X tend to be taller than average, some have language and learning difficulties but are otherwise healthy and do not typically have fertility problems. Triple X is often discovered unintentionally; many women do not know they have this condition.

Individuals with Triple X have a normal life expectancy.

What is XYY syndrome?

XYY syndrome occurs when a boy is born with an extra Y chromosome.

Individuals with XYY may be taller than average but otherwise have typical male physical features and normal fertility. Some boys have language and learning difficulties and may have behavioral problems such as attention deficit disorder or autism.

Individuals with XYY syndrome have a normal life expectancy.

References

Gardner RJM, Sutherland GR, Shaffer LG. Chromosome Abnormalities and Genetic Counseling. Parental age counseling and screening for fetal trisomy 2012.

NOTES