McMaster University Medical Centre

Welcome to the Prenatal Diagnosis Clinic (PND)

At your consult visit (by phone or in person), you will first speak with a Genetic Counsellor, who will discuss the reason you were referred. She will review your family and medical history with you. She will provide information about the findings in your pregnancy, and any genetic implications.

You will also speak with the doctor who is a Maternal Fetal Medicine (MFM) specialist (specialized high risk obstetrician). The MFM will review any ultrasound you may have had as part of your consult, or before your consult), and the issues concerning your pregnancy. Together, the team will make a plan for your care. This may include asking other health professionals or specialists to see you.

To assist us, please complete the personal and family history questionnaire.

Please note, this is a teaching hospital. We may have residents, medical students, midwifery students, or genetic counselling interns participating in patient care. If you have any concerns about the role of learners in your care, please speak with a staff member.

The information in this booklet
is intended to accompany a consultation in the PND clinic.
Some of the information in this booklet may not apply to your pregnancy.

Contact Information

Business Clerk (905) 521-2100 ext 73135
General inquiries, directions, confirm or change an appointment

Fax number (905) 521-4955

Table of Contents

Information about the Prenatal Diagnosis Clinic

This clinic will help you learn about findings in your pregnancy or your medical / family history that may affect the health of your baby.

During your initial consult with the Prenatal Diagnosis Clinic, you will:

- Complete a review of your family history and medical history.
- Speak with a genetic counsellor to discuss your pregnancy.
- Speak with a doctor to review the findings and the plan of care.

There are different types of tests that could be done to learn about your baby's health.

- Tests that tell you the **chance** that your baby has a genetic condition or other health condition are called **prenatal screening tests**. Routine prenatal screening is available for all pregnant women of any age regardless of any findings or previous history.

- Tests that tell you **for certain** if your baby has a genetic condition are called **prenatal diagnostic tests.** Prenatal diagnostic testing is available for all pregnant women who have an increased chance of certain genetic conditions or other health conditions (from blood tests, ultrasound findings, or previous history).

It is <u>your</u> choice whether to have any screening or diagnostic tests in your pregnancy.

Your doctor or midwife may have already discussed the tests that are available to you. You may already have results of prenatal screening or testing, which we can review with you. Please feel free to ask questions at any time. Take the time you need to understand what the tests can and cannot tell you.

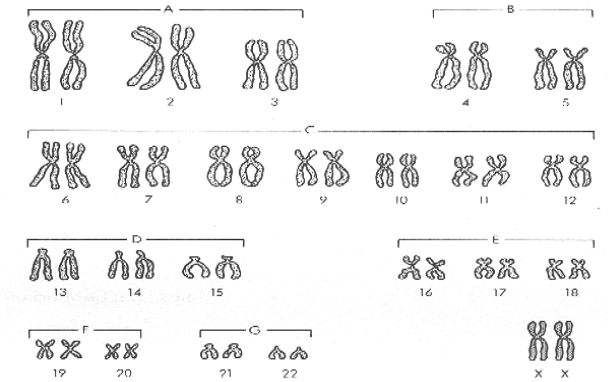
If you choose to have any screening or diagnostic tests, we will make the arrangements. We will explain and discuss the results with you when they are ready, and help you decide on the next steps in your care.

We know that decisions about testing your pregnancy can be stressful for you and your family. The PND (prenatal diagnosis) health care team is available to support you. Other specialists are also available to you, such as a Social Worker.

Learning about chromosomes

- Chromosomes carry genetic information known as "genes" (the instructions for growth and development).
- A complete set of 46 chromosomes is needed for normal growth and development.
- Most females have two X chromosomes, while most males have one X and one Y chromosome.
- The lab test that checks a person's chromosomes is called a "karyotype".

This is a picture of the regular number of chromosomes (the karyotype). There are two copies of each chromosome, with one copy inherited from the mother and one copy inherited from the father.



Chromosome differences

- A chromosome difference occurs when a person has a change in the number, size or structure of his/her chromosomes.
- A chromosome difference may result in problems in growth, development and/or functioning of the body systems.
- Most chromosome differences occur when the egg or sperm cells are forming, during conception or just after. Some chromosome differences can be inherited from a parent.
- One common chromosome difference is Down syndrome, which has an extra copy of chromosome 21 (called trisomy 21).
- People with Down syndrome can have a range of physical and intellectual differences. There is no way to predict the severity before birth.
- Other well-known conditions are trisomy 13 and trisomy 18, which usually have very serious birth defects. These infants rarely survive for very long after birth.

- Sometimes there can be a change in only part of a chromosome, or even in a single gene. Standard chromosome testing will not find these kinds of changes, but there are other DNA based tests that can be used if needed.

Introduction to prenatal screening and testing

Nuchal translucency (NT) ultrasound (page 4)

NT ultrasound measures a small fluid collection at the back of the baby's neck between 11 and 14 weeks of pregnancy.

The NT measurement can be combined with mother's age and blood testing for routine prenatal screening.

Routine prenatal screening (page 4)

Routine prenatal screening uses blood testing and sometimes an NT ultrasound to tell you **the chance** for the baby to have Down syndrome or trisomy 18.

The routine screening tests that are commonly used are **First Trimester Screening** (FTS) or **Second Trimester Screening** (STS).

Non-Invasive Prenatal Screening (NIPS) (page 5)

NIPS looks at pieces of cell free DNA (cfDNA) in a pregnant woman's blood to screen for the most common chromosome differences (trisomy 21, trisomy 18, trisomy 13, and differences with the sex chromosomes).

The NIPS tests that are commonly used are **Harmony** (offered through Gamma-Dynacare labs) and **Panorama** (offered through LifeLabs).

Prenatal diagnostic testing (amniocentesis / CVS pages 6 and 7)

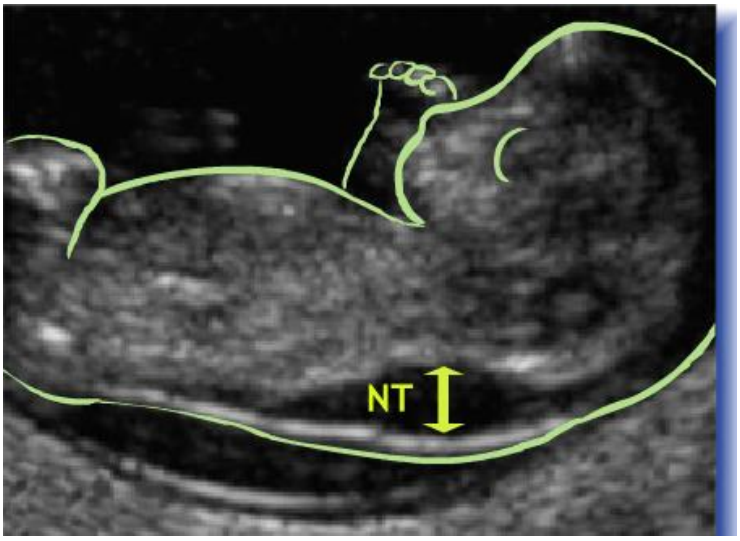
Prenatal diagnostic testing looks at the baby's chromosomes/DNA. These tests will tell you for sure if the baby has one of the common chromosome differences, such as Down syndrome. Some other genetic conditions can also be detected with these tests.

These tests are only used if there is a reason to suspect a chromosome difference or genetic condition, such as:

- There are abnormal ultrasound findings
- Your prenatal screening reported an increased chance of a chromosome difference such as Down syndrome
- You have a family history or are a carrier of a genetic syndrome.

The procedures that are commonly used are **Amniocentesis** or **Chorionic Villus Sampling (CVS**) (see pages 6 and 7).

Increased nuchal translucency (NT)



- Increased NT means the measurement is bigger than expected.
- When the NT is increased there is a higher than usual chance for the baby to have a chromosome difference or birth defect.
- In some cases, the extra fluid disappears later in the pregnancy, and the baby can develop normally.
- It might be helpful to complete prenatal screening or prenatal diagnostic testing.
- A detailed ultrasound should be done at approximately 19 weeks of pregnancy to check the baby's anatomy carefully.

Information about routine prenatal screening

First Trimester Screening (FTS)

- NT ultrasound and blood testing done at 11 to 14 weeks of pregnancy
- No longer used for twin pregnancies (see NIPT information on page 5)
- Detects 85-90% of babies with Down syndrome and 90% of babies with trisomy 18

Second Trimester Screening (STS)

- Blood testing done between 15 and 21 weeks of pregnancy
- Can be done for singleton pregnancies only
- Detects 80% of babies with Down syndrome and 60-70% of babies with trisomy 18

Results of routine prenatal screening

- Results typically take a few days
- "Screen negative" means the chance for the baby to have one of these conditions is low and you probably do not need any more testing. This does not guarantee the birth of a normal baby.
- "Screen positive" means there is an increased chance for the baby to have one of these conditions. You could have more testing to find out for sure.

Non-Invasive Prenatal Screening (NIPS)

How is it done?	Blood draw
When is it done?	Anytime after 9 weeks, preferably after 11 weeks
What is tested?	DNA from chromosomes 21, 18, 13, X and Y
Length of time for results	Up to 2 full calendar weeks
Detection rate	99% Down syndrome (Trisomy 21) 98% Trisomy 18 80% Trisomy 13 99% X and Y *Detection rates may be lower for twin pregnancies.
False positive rate	Less than 0.1%
Chance of inconclusive result	5% no result 1% inconclusive result
Cost	Up to $550 if private pay Covered by OHIP if: twin pregnancy, abnormal ultrasound findings, "screen positive" routine prenatal screening, previous trisomy or maternal age over 40 and with certain ultrasound findings.

- cfDNA comes from both the mother and the pregnancy (placenta).
- The amount of cfDNA that comes from the pregnancy is called the "fetal fraction".

Results of NIPS

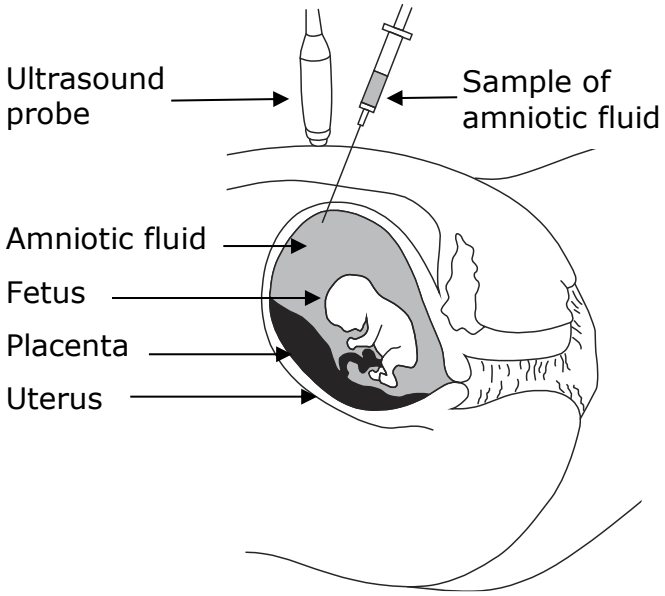
- "High risk" (abnormal) results can be confirmed using CVS or amniocentesis (see page 6), or a blood sample from the infant after delivery.
- "Low risk" (normal) results do not require further testing in most cases. This does not guarantee the birth of a normal baby.
- If there is "no result", "atypical/inconclusive" or "partial result", a repeat test may give a result. Sometimes, it is recommended to have a diagnostic test instead. Your Genetic Counsellor will talk to you about this.

Amniocentesis

This procedure removes a small amount of fluid from the sac of fluid around the baby (amniotic sac). The amniotic fluid contains cells from the baby. The baby's cells and the fluid can be tested.

How is amniocentesis done?

- A specially trained obstetrician does the procedure, usually between 15 and 22 weeks of pregnancy.
- An ultrasound confirms the position of the baby and placenta. The doctor uses the ultrasound to find a safe spot to insert a thin needle through the mother's abdomen, into the amniotic sac.
- A small amount of amniotic fluid is removed through the needle, then the needle is taken out.
- The fluid sample is sent to the laboratory for testing.

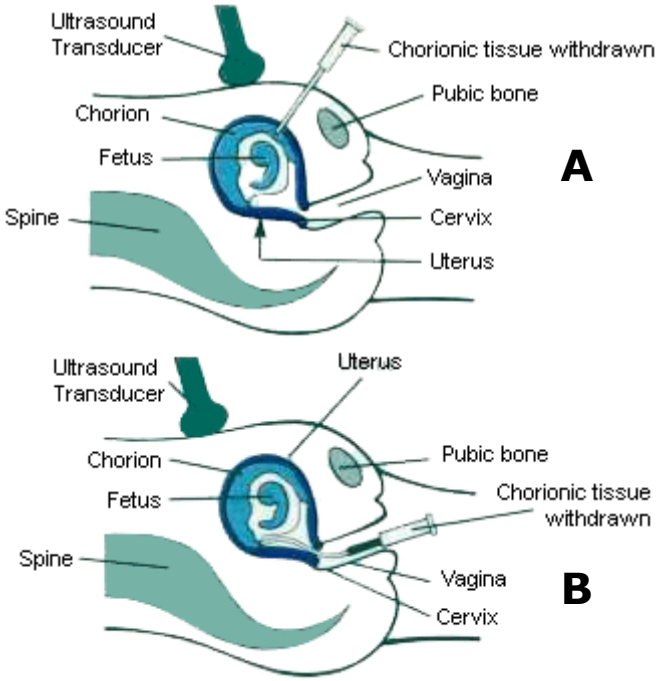


Chorionic Villus Sampling (CVS)

This procedure removes a small amount of tissue from the developing placenta (afterbirth). Cells from the placenta are genetically the same as the baby in most cases.

How is CVS done?

- A specially trained obstetrician does the procedure, usually between 11 and 13 weeks of pregnancy.
- An ultrasound scan confirms the position of the baby and placenta, and helps to find a safe way to do the procedure.
- There are two ways to perform CVS:
  A. Transabdominal CVS: A thin needle is put in through the mother's abdomen, into the developing placenta.
  B. Transcervical CVS: A thin instrument is inserted through the cervix (birth canal) and into the developing placenta.
- A small tissue sample is sent to the laboratory for testing.



What are the risks of amniocentesis or CVS?

- **Miscarriage:** For amniocentesis, there is a risk of between 1 in 800 to 1 in 1000 (about 0.1%) to have a miscarriage due to the procedure. For CVS, there is a risk of about 1 in 300 (0.3%) to have a miscarriage following the procedure.
- **Cramping, bleeding or fluid leakage:** After amniocentesis, some women have cramping or fluid leakage from their vagina. After CVS, some women have cramping or bleeding from their vagina. This may last a few days. It may be helpful to limit your activity and to consider bedrest for 24 hours if you are concerned about any of these symptoms.
- **Discomfort:** Most women say amniocentesis and CVS are uncomfortable, but not more painful than having a blood test or a Pap test. No freezing is used, since the needle for freezing is more uncomfortable than the one used for amniocentesis or CVS.

Genetic testing from amniocentesis or CVS

- **QF-PCR**: This is a quick test for extra or missing copies of chromosomes 21, 18, 13, X or Y. This test will diagnose Down syndrome, trisomy 18, trisomy 13, and sex chromosome differences. You may be able to find out if the baby is male or female. Results take 2 to 5 business days.
- **Chromosomal microarray (CMA):** The DNA from all 23 pairs of chromosomes is tested for extra or missing pieces. Results take 3 to 4 weeks.
- **Karyotype**: The whole chromosomes are studied under the microscope to look for any extra or missing large pieces, or any obvious differences in how the chromosomes are arranged. This test is mostly used after there has been a finding in either the QF-PCR or the CMA. Results take 2 to 4 weeks.
- **Other genetic testing:** Depending on your history or the findings in your pregnancy, specific genetic tests may be done for one or more less common genetic conditions. Your Genetic Counsellor will talk to you about whether these tests are needed. Timing for results depends on the test ordered.

Results of diagnostic tests

- One of the Genetic Counsellors will call you to tell you about your results.
- Sometimes it may be helpful to come back to the PND clinic for more discussion.