Non-Invasive Prenatal Testing (NIPT)

How does age affect fertility?

Age is one of the most important predictors of infertility and one of the most important factors in determining how successful fertility treatments are likely to be. The number of eggs in the ovaries (called the ovarian reserve) decreases naturally and progressively from the time someone is born until they reach menopause. The decline is gradual but accelerates quicker after the mid-30s. As age increases, the chance of having other conditions that can impact fertility, such as fibroids, also increases. And people who become pregnant at an older age have a higher risk of complications during pregnancy, such as gestational diabetes and preeclampsia. In addition to declining quantity, the percentage of eggs with chromosomal abnormalities increases with the age of the egg. Because of this, the chance of early pregnancy loss (miscarriage) and pregnancies with chromosomal abnormalities increases significantly with age.

Risk of Down Syndrome and Other Chromosome Abnormalities in Live Births by Maternal Age

Risk			Risk			Risk		
Maternal Age (at term)	Down Syndrome	Total Chromosome Abnormalities	Maternal Age (at term)	Down Syndrome	Total Chromosome Abnormalities	Maternal Age (at term)	Down Syndrome	Total Chromosome Abnormalities
25	1 in 1250	1 in 476	32	1 in 637	1 in 323	39	1 in 125	1 in 81
26	1 in 1190	1 in 476	33	1 in 535	1 in 286	40	1 in 94	1 in 63
27	1 in 1111	1 in 455	34	1 in 441	1 in 224	41	1 in 70	1 in 49
28	1 in 1031	1 in 435	35	1 in 356	1 in 179	42	1 in 52	1 in 39
29	1 in 935	1 in 417	36	1 in 281	1 in 149	43	1 in 40	1 in 31
30	1 in 840	1 in 385	37	1 in 217	1 in 123	44	1 in 30	1 in 21
31	1 in 740	1 in 385	38	1 in 166	1 in 105	>45	> 1 in 24	> 1 in 19

Source: Hecht CA, Hook EB. 1996
Reproduced from BC Prenatal Genetic Screening Program, Provincial Health Services Authority

What is NIPT?

Non-Invasive Prenatal Testing (NIPT), sometimes called Non-Invasive Prenatal Screening (NIPS), is a method of determining the risk that a specific fetus will be born with certain genetic abnormalities. This test does not screen for all possible genetic conditions. This testing is done in early pregnancy.

How is NIPT done?

NIPT is done through a blood sample from a pregnant person. DNA, or deoxyribonucleic acid, is the hereditary material in humans. NIPT analyzes small fragments of DNA that are circulating in a pregnant person's blood. Unlike most DNA, which is found inside a cell's nucleus, these fragments are free-floating and not within cells, and so they are called cell-free DNA (cfDNA). These small fragments arise through the normal process of the cells dying off, breaking down, and their contents (including DNA) releasing into the bloodstream.

During pregnancy, the pregnant person's bloodstream contains a mix of cfDNA that comes from their cells and cells from the placenta. The placenta is tissue in the uterus that links the fetus' and the pregnant person's bloodstreams. The DNA in placental cells is usually identical to the DNA of the fetus. Analyzing cfDNA from the placenta provides an opportunity for early detection of certain genetic abnormalities in the fetus.

What can NIPT screen for?

NIPT is most often used to look for chromosomal disorders that are caused by the presence of an extra or missing copy of a chromosome (Aneuploidy). NIPT primarily screens for the most common chromosomal abnormalities namely Trisomy 21 (also known as Down syndrome and caused by an extra copy of chromosome 21), Trisomy 18 (caused by an extra copy of chromosome 18), Trisomy 13 (caused by an extra copy of chromosome 13), and extra or missing copies of the sex chromosomes (the X and Y chromosomes).

Some versions of NIPT may include screening for additional chromosomal disorders that are caused by missing (deleted) or extra (duplicated) sections of a chromosome. NIPT is beginning to be used to test for genetic disorders that are caused by changes (variants) in single genes.

What are the risks and limitations of NIPT?

NIPT is considered non-invasive because it only requires a blood draw from the pregnant person during pregnancy. There are no risks to the pregnant person or developing baby outside of a normal blood draw.

NIPT is a screening test, which means that it will not give a definitive answer about whether or not a fetus has a genetic condition. The test can only estimate whether the risk of having certain genetic conditions is increased or decreased compared to the average risk for your age. In some cases, NIPT results indicate an increased risk for a genetic abnormality when the fetus is actually unaffected (False Positive), or the results indicate a decreased risk for a genetic abnormality when the fetus is actually affected (False Negative). Because NIPT analyzes both fetal and maternal cfDNA, the test may actually be detecting a genetic condition in the pregnant patient. The accuracy of the screening test varies for different genetic conditions.

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What types of results can I get?

A negative (normal) screening result suggests a reduced risk for the pregnancy to be affected by the genetic conditions screened. However, it does not fully eliminate the risk and there is a small risk for a false negative result.

A positive (abnormal) screening result suggests an increased risk for the pregnancy to be affected by one of the genetic conditions screened. A positive screen will require additional follow-up to determine if the result is a true positive or a false positive. Irreversible decisions about the pregnancy (termination of a pregnancy) should not be made based on an NIPT result alone without further confirmation.

A consultation with a Maternal-Fetal Medicine specialist will occur to discuss any positive results with you and the possible options for the next steps.

When can NIPT be done?

Cell-free DNA (cfDNA) screening can be performed as early as 10 weeks and 3 days into the pregnancy.

How long does it take to get NIPT results?

Results from NIPT screening are typically available 7 to 10 days after the blood sample is collected.

Where can I find more information?

You can find more information on NIPT here.
You can also watch these videos to learn more about NIPT, positive results, and negative results.

How can I schedule my NIPT?

If you are ready to proceed with scheduling your NIPT at ARC, please contact our Front Desk Team at 780-442-0461. This test is scheduled at ARC:

- Monday-Friday 9:30am-2:30pm
- Weekends from 9am-12pm.

How do I prepare for this test?

Please remember to bring your Alberta health care card and a government issued photo identification (eg. Driver's License). No fasting is required for this test but drinking lots of water prior to the blood test is recommended.