harmony™
PRENATAL TEST

Clear ANSWERS to Questions that Matter

Dynacare®

What is a Trisomy?

Humans have 23 pairs of chromosomes, which are strands of DNA that carry genetic information. A trisomy is a chromosomal condition that occurs when there are three copies of a particular chromosome instead of the expected two.

Trisomy 21

Trisomy 21 is due to an extra chromosome 21 and is the most common trisomy at the time of birth. Trisomy 21, also called Down syndrome, is associated with mild to moderate intellectual disabilities and may also lead to digestive disease and congenital heart defects. It is estimated that trisomy 21 is present in 1 out of every 800 births in Canada.[1]

Trisomy 18

Trisomy 18 is due to an extra chromosome 18. Trisomy 18, also called Edwards syndrome, is associated with a high rate of miscarriage. Infants born with trisomy 18 often have congenital heart defects as well as various other medical conditions, shortening their lifespan. It is estimated that trisomy 18 is present in approximately 1 out of every 6,000 births.[2]

Trisomy 13

Trisomy 13 is due to an extra chromosome 13. Trisomy 13, also called Patau syndrome, is associated with a high rate of miscarriage. Infants born with trisomy 13 usually have severe congenital heart defects and other medical conditions. Survival beyond the first year is rare. It is estimated that trisomy 13 is present in approximately 1 out of every 16,000 newborns.[3]

[1]Canadian Down Syndrome Society
[2]Ontario Ministry of Health and Long-Term Care
[3]U.S. National Library of Medicine

Harmony™ Prenatal Test

When you're pregnant, your blood contains fragments of your fetus' DNA. The Harmony Prenatal Test is a new type of test that analyzes this DNA in a sample of your blood to determine the risk of trisomy 21, 18 and 13.

For Women of Any Age or Risk Category

Traditional screening tests can miss as many as 20% of trisomy 21 cases in pregnant women.[1] The Harmony Test was developed to be a more accurate prenatal screening test for women of any age or risk category. It is a new DNA-based blood test that has been extensively tested in pregnant women ages 18 to 50.[1-2]

A More Accurate Test

The Harmony test has been shown in clinical testing to identify 99% of trisomy 21 cases and to have a false-positive rate of less than 0.1%.

Clarity Early

Harmony Prenatal Test requires a single blood draw and can be done as early as 10 weeks or later in pregnancy. Results are available within 10 business days.

Minimizes Need for Follow-Up Tests

The greater accuracy and low false-positive rate of the Harmony test compared to traditional tests minimizes the chance further testing would be recommended due to a positive result. Follow-up testing might include an invasive procedure, such as amniocentesis.

[1]Norton M, *et al. N Engl J Med.* 2015; 372:1589-1597.
[2]Nicolaides KH, *et al. Am J Obstet Gynecol.* 2012 Nov;207(5):374.e1-6.

Ask Your Provider

Fetal Sex Chromosomes

The Harmony Prenatal Test can also be used as early as 10 weeks to evaluate X and Y sex chromosomes. You have the option to test for conditions caused by having an extra or missing copy of the X or Y chromosomes, including Turner and Klinefelter syndromes.

Ask your provider

The information in this brochure is provided to inform you about the Harmony Prenatal Test. Talk to your healthcare provider to decide if the Harmony Prenatal Test is appropriate for you.

Genetic Counselling

Dynacare offers genetic counselling services to support both patients and physicians by identifying genetic risks, explaining appropriate genetic testing options, discussing the implications of the test results, and helping patients make better healthcare decisions. Genetic counselling is included in the price of Harmony Prenatal Test.

Customer Care
T 888.988.1888
F 450.663.4428
DynacareNext@dynacare.ca

dynacare.ca

Understanding Your Results

The Harmony Prenatal Test detects trisomies of chromosomes 21, 18 and 13 in the fetus, but does not rule out all fetal abnormalities.

Low Risk result

If the Harmony Prenatal Test results show a Low Risk, the chance of having a fetus with trisomy 21, trisomy 18, or trisomy 13 is low. As with any test, a low risk result reduces, but does not eliminate, the chance of having an affected pregnancy.

High Risk result

If the Harmony Prenatal Test results show a High Risk, there is an increased chance of having a fetus with trisomy 21, trisomy 18, or trisomy 13. If your result is High Risk, your healthcare provider may offer genetic counseling and/or diagnostic testing to determine if your fetus is affected with one of these conditions.

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Diagnostic Tests

If I am High Risk, what additional testing is available?

If the Harmony Prenatal Test shows you are High Risk, it does not necessarily mean that the pregnancy has one of these birth defects. Your healthcare provider may offer you one of the following procedures*:

- **Chorionic villi sampling (CVS)**: Procedure that takes a small amount of tissue from the developing placenta. The tissue is then sent to a laboratory to test the chromosomes. CVS is typically performed between 10 and 12 weeks of pregnancy. CVS is associated with a small risk of miscarriage.

- **Amniocentesis**: Procedure that withdraws a small amount of fluid that surrounds the fetus. The fluid is then sent to the laboratory to test the chromosomes. An amniocentesis is usually performed around or after the 16th week of pregnancy. Amniocentesis is associated with a small risk of miscarriage.

*Dynacare does not offer these diagnostic tests.

The Harmony Prenatal Test was developed by Ariosa Diagnostics (San Jose, CA, USA). The test is performed at Dynacare's facilities in Bowmanville, Ontario. Non-Invasive Prenatal Testing (NIPT) based on fetal cell-free DNA analysis is not a diagnostic test. No irrevocable obstetrical decision should be made on a positive result generated from a NIPT based on fetal cell-free DNA analysis, without confirmation by other invasive diagnostic testing. Data have not been submitted or evaluated by Health Canada or other federal regulatory agencies and the test is not for sale as an In Vitro Diagnostic test in Canada.

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The quality choice for 22q11.2 deletion screening

NEW TEST OPTION: 22q11.2 is now part of the Harmony Prenatal Test menu. Early prenatal screening for 22q11.2 deletion combined with diagnosis enables informed choices and appropriate obstetrical and neonatal management.[1] Order 22q11.2 by selecting the check box on the Harmony requisition form.

Recommend the Harmony Test

Benefits of the Harmony test:

- Flexible testing options and clinically relevant testing
- Reliable timely results regardless of test options ordered
- Minimize unnecessary invasive procedures due to false-positives[2]

Choose tests that are clinically relevant to your patients, not test panels for rare microdeletions. Each condition tested has an associated false-positive rate and adds to the total false-positive rate of the test.

Performance

The Harmony test has an overall low cumulative false-positive rate. For trisomies 21, 18, 13, and 22q11.2, the **cumulative false-positive rate is less than 0.6%.**

22q11.2 Deletion

	Detection Rate	False-positive Rate
within the 3 Mb region*	75%[3]	0.5%[3]

*including smaller nested deletions

Combined cumulative false-positive rate for trisomies 21, 18, 13, and 22q11.2 deletion	<0.6%

The false-positive rate for T21, T18, and T13 combined is less than 0.1%.[4]

Options for Ordering

- ☑ Harmony Prenatal Test: Assesses the probability of fetal trisomy 21, trisomy 18, trisomy 13. Validated for use in twin and IVF pregnancies, including self and non-self donor pregnancies.[4]

Additional Test Offerings:

- ☐ Fetal sex (singletons and twins)
- ☐ Monosomy X (singletons only)
- ☐ Sex chromosome aneuploidy panel (singletons only)
- ☐ 22q11.2 (singletons only)

For more information, please contact us at **888.988.1888** or **DynacareNext@dynacare.ca** or go to **dynacare.ca**

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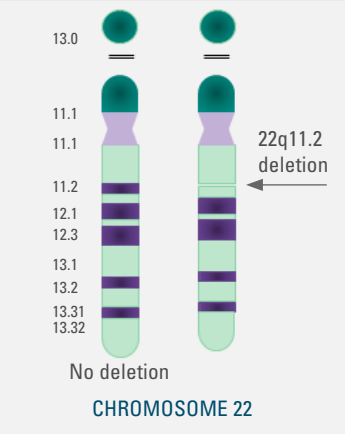
New Screening Option: 22q11.2 deletion

22q11.2 deletion is the **most common** chromosomal microdeletion[5]

This condition may occur in as many as **1 in 1000** pregnancies.[6]

It is the second most common cause of developmental delay after Down syndrome.[7]



Identify pregnancies which may be at increased risk early:

- Maternal age is not a risk factor for microdeletions[6]
- More than 90% of affected individuals have no family history of 22q11.2 deletion[8]
- 22q11.2 deletion is not reliably detected by routine screening or karyotype[7]

Clinically Relevant

22q11.2 deletion is the underlying cause of conditions described as DiGeorge syndrome and velocardiofacial syndrome (VCFS). Clinical presentation demonstrates a wide range of severity that cannot be predicted prenatally.

Features are diverse and may include the following:[9]

- congenital heart disease
- palatal anomalies
- immune deficiency
- hypocalcemia

Other features may include renal anomalies, learning difficulties, developmental delays, and psychiatric illness.[9]

Early screening and diagnosis of 22q11.2 deletion affects management of pregnancy. If a pregnancy is affected with 22q11.2 deletion, the following is recommended:[10]

- Level II ultrasound with fetal echocardiogram to evaluate for anomalies such as congenital heart defect, cleft palate, etc.
- Screening for and coordinated management of associated conditions
- Delivery at a tertiary care center

NIPT is a screening test. If a pregnancy is known to be at increased risk for 22q11.2 deletion based on family history or ultrasound findings, diagnostic testing should be considered.

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1. McDonald-McGinn et al. Nature Reviews Disease Primer. 2015 Nov 19.
2. Wax et al. J Clin Ultrasound. 2015 Jan;43(1):1-6
3. Data on file, pending publication
4. Stokowski et al. Prenat Diagn. 2015 Oct; DOI: 10.1002/pd.4686
5. McDonald-McGinn DM, Emanuel BS, Zackai EH. 22q11.2 Deletion Syndrome. 1999 Sep 23
6. Grati et al. Prenat Diagn. 2015 Aug;35(8):801-9.
7. Bassett et al. J Pediatr. 2011 Aug;159(2):332-9.
8. McDonald-McGinn et al. Genet Med. 2001 Jan-Feb;3(1):23-9.
9. McDonald-McGinn et al. Genet Couns. 1999;10(1):11-24.
10. McDonald-McGinn et al. GeneReviews (2013) http://www.ncbi.nlm.nih.gov/books/NBK1523/