

## Panorama® Non-Invasive Prenatal Test (NIPT)

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**SIGNED COPY SHOULD BE KEPT IN THE PATIENT MEDICAL RECORD (do not return with the blood kit)**

**Purpose of the Test(s):** Panorama™ Non-Invasive Prenatal Test (NIPT) screens a fetus for chromosome abnormalities that include whole extra or missing chromosomes. Panorama can also screen for fetal sex, microdeletions (small missing sections of a specified chromosome) and zygosity in twin pregnancies (determining if twins are identical or fraternal). Additional information requested regarding chorionicity in twin pregnancies (one placenta or two) is used for the purpose of test development and is not required for the interpretation of the test. Panorama™ is performed on a maternal blood sample that contains DNA (genetic material) from both the mother and fetus. The fetal DNA tested comes from the placenta; this DNA is identical to the DNA found in the cells of the fetus in ~98% of all pregnancies. Panorama™ is available to women who are at least 9 weeks pregnant. Your healthcare provider can provide you with more details about the conditions screened with this test, which are listed below.

**Whole Chromosome Abnormalities and Microdeletions Evaluated With the Panorama™ Test Options**

BASIC PANEL		
	Trisomy 21	This is caused by an extra copy of chromosome 21 and is also called Down syndrome. This is the most common genetic cause of intellectual disability and occurs in approximately 1 in every 830 live births. Individuals with Down syndrome have an average IQ of 50 and all have some degree of intellectual disability. Some children with Down syndrome have defects of the heart or other organs that may require surgery or medical treatment. Some have other medical conditions, including hearing or vision loss.
	Trisomy 18	This is caused by an extra copy of chromosome 18 and is also called Edwards syndrome. Trisomy 18 occurs in about 1 in every 7,500 live births and causes severe intellectual disability. Most babies with trisomy 18 have multiple severe birth defects of the brain, heart, and other organs. Poor growth during pregnancy is common and many babies are miscarried or stillborn. Of those babies born alive, most die before one year of age. Babies who survive have profound intellectual disabilities, as well as growth and development problems.
	Trisomy 13	This is caused by an extra copy of chromosome 13 and is also called Patau syndrome. Trisomy 13 occurs in about 1 in every 22,700 live births and causes severe intellectual disability. Most babies with trisomy 13 have multiple severe birth defects of the brain and other organs. Many babies are miscarried or stillborn. Of those babies born alive, most die before one year of age.
	Monosomy X*	This is caused by a missing copy of the X chromosome and is also called Turner syndrome. This only affects girls and is found in every 1 in 5,000 live births. Girls with Monosomy X are shorter than average. Some girls have heart or kidney defects, hearing problems, and some have minor learning disabilities. Girls with Monosomy X may need growth hormone treatments in early childhood and usually need sex hormone treatments at the time of puberty. As adults, they often have infertility.
	Triploidy	Triploidy is a condition in which the fetus has 3 copies of each chromosome instead of two. It is found in about 1 in 1,000 first trimester pregnancies. Features include brain and craniofacial abnormalities, neural tube defects, heart defects, as well as genitourinary and gastrointestinal abnormalities. Most pregnancies with triploidy will miscarry in the early part of the first trimester, although later miscarriage or stillbirth can occur. The majority of those that survive die shortly after birth. Triploidy may also put the mother's health at risk from pregnancy complications like pre-eclampsia, postpartum hemorrhage, and molar pregnancy with the potential for malignancy.
EXTENDED PANEL (OPT-IN)		
	22q11.2 Deletion Syndrome	22q11.2 deletion syndrome, also known as DiGeorge syndrome, is caused by a small missing piece of chromosome 22. It is found in about 1 in 2,000 live born babies <sup>1</sup> . Most children with 22q11.2 deletion syndrome have mild-to-moderate intellectual disability and delayed speech and language. Many have heart defects, immune system problems, and other health problems. Some people with 22q11.2 deletion syndrome have autism spectrum disorder and some develop psychiatric illnesses, such as schizophrenia.
	1p36 Deletion Syndrome	This syndrome is caused by a small, missing piece of chromosome 1 and is also called Monosomy 1p36. About 1 in every 5,000 live born babies has this condition <sup>2</sup> . Children with Monosomy 1p36 have moderate-to-severe intellectual disability. Most children have heart defects that may require surgery or medical treatment. Some children may need special physical and occupational therapies to help with weak muscle tone. About half of children with Monosomy 1p36 have seizures and/or behavioral problems, and can also have hearing and/or vision loss.
	Cri du Chat Syndrome (5p-)	This is caused by a small missing piece of chromosome 5 and is also called 5p minus (5p-) syndrome. About 1 in 20,000 live born babies has this condition <sup>4</sup> . Babies are usually small at birth with a small brain and head size. They often have breathing and feeding problems and need extra medical care. Children with cri du chat have severe intellectual disability.
	Angelman Syndrome (15q11.2 deletion maternal)	Angelman syndrome (AS) is caused either by a small, missing piece of chromosome 15 or from inheriting two copies of chromosome 15 from one parent and none from the other. Other rare causes also exist. About 1 in 12,000 live born babies has this condition <sup>3</sup> . They often have feeding difficulties and weak muscle tone. Children have severe intellectual disability and motor problems. Most have a small brain and head size, and some have seizures. Most children do not develop speech.
	Prader-Willi Syndrome (15q11.2 deletion paternal)	Prader-Willi syndrome (PWS) is caused either by a small, missing piece of chromosome number 15 or from inheriting two copies of chromosome 15 from one parent and none from the other; Other rare causes also exist. About 1 in 10,000 live born babies has this condition <sup>3</sup> . Babies have weak muscle tone and feeding problems. Children with PWS typically have intellectual disability, behavior problems, and delayed motor and language development. They also have excessive appetites and may become obese and develop diabetes.

(<sup>1</sup> Nussbaum et al 2007 Thompson and Thompson Genetics in Medicine (7th Ed) Oxford Saunders, Phila, PA; (<sup>2</sup> Arthur Robinson & Mary G Linden, 1993, Clinical Genetics Handbook, (2nd Ed). Cambridge, Mass, Blackwell Scientific Publications); (<sup>3</sup> GeneReviews: <http://genereviews.org/>; (<sup>4</sup> Genetics Home Reference: <http://ghr.nlm.nih.gov>)

\* Sex chromosome trisomies (XXY, XXX, and XYY) will also be reported, if identified.

**Methods:** Panorama™ was developed by Natera Inc.®, a laboratory certified under the Clinical Laboratory Improvement Act (CLIA). All testing is performed under prevailing regulations in accredited facilities. Two tubes of blood are required from the mother. The samples are only screened for those chromosome abnormalities listed below and options available may differ depending on the number of fetuses and zygosity. Incidental findings (e.g. findings unrelated to the specific conditions screened below) will **not** be reported.

		Singleton (1 baby)	Monozygotic (identical) twins	Dizygotic (fraternal) twins	Egg donor or Surrogate (singleton gestations only)
BASIC	Trisomies 21, 18, and 13	✓	✓	✓	✓
	Triploidy	✓	✗	✗	✗
	Sex chromosome abnormalities (including Monosomy X)	✓	✓	✗	✗
	Fetal Sex	✓	✓	✓	✓
OPT-IN	22q11.2 deletion syndrome	✓	✓	✗	✗
	Additional microdeletions syndromes: Cri-du-chat, 1p36 deletion, Angelman, and Prader-Willi	✓	✗	✗	✗

## Panorama® Non-Invasive Prenatal Test (NIPT)

For singleton (one baby) pregnancies, the Panorama™ Prenatal Test is performed by LifeLabs Genetics (LifeLabs) in Toronto, ON. For twin (two baby) pregnancies, pregnancies achieved using an egg donor or carried by a surrogate, the Panorama™ Prenatal Test is performed at Natera Inc.®, in San Carlos, California.

**Test Results:** Your test results will be sent to the healthcare provider who ordered the test.

- A **low risk** result indicates a reduced chance that your baby has the listed chromosome abnormalities but cannot guarantee normal chromosomes or a healthy baby.
- A **high risk** result indicates that there is an increased chance your baby has one of the chromosome abnormalities listed but does not confirm that the fetus has that abnormality. The recommended follow-up is a prenatal diagnostic test, such as chorionic villus sampling (CVS) or amniocentesis. Your healthcare provider will explain the test results and recommended follow-up steps to you, which may include a referral to a genetic counsellor in addition to the prenatal diagnostic testing. In the event of a high risk result, **LifeLabs may contact your healthcare provider to obtain follow-up diagnostic information to ensure quality and accuracy in reporting.**
- Panorama® is not a diagnostic test – **DECISIONS ABOUT YOUR PREGNANCY SHOULD NEVER BE MADE BASED ON THESE SCREENING RESULTS ALONE, AS THEY NEITHER CONFIRM NOR RULE OUT THE PRESENCE OF A CHROMOSOME ABNORMALITY IN THE FETUS.** Follow-up diagnostic testing should always be performed during pregnancy or at birth to confirm or rule out a chromosome abnormality or microdeletion.
- There is a chance that the sample(s) submitted will not return results. In this case, your healthcare provider will be informed by LifeLabs and you may be asked to provide a second blood sample to repeat the test. There is no charge for a repeat. In rare cases, a result cannot be provided on a subsequent sample, and, if you have self-paid for NIPT, you will receive a full refund, in this case.

**Test Limitations:** Although this screening test will detect the majority of pregnancies in which the fetus has one of the above listed chromosome abnormalities, it cannot detect 100% of pregnancies with these conditions. The result of this test does not eliminate the possibility of other abnormalities of the tested chromosomes, and it does not detect abnormalities of untested chromosomes, other microdeletions, genetic disorders, birth defects, or other complications in your fetus or pregnancy. The Panorama® prenatal test has not been cleared or approved by the U.S. Food and Drug Administration (FDA) or Health Canada.

Inaccurate test results or a failure to obtain test results may occur due to one or more of the following rare occurrences: courier/shipping delay; sample mix-up; laboratory failure or error; biological factors; other circumstances beyond our control; or unforeseen problems that may arise. Biological factors can include, but are not limited to: sample contamination or degradation; too little DNA from the fetus in the maternal blood sample (low fetal fraction); other genetic variants in the mother or fetus; an unrecognized twin pregnancy; or mosaicism (a mixture of cells with normal and abnormal chromosomes) in the fetus, placenta, or mother. About 1 to 2% of all pregnancies have confined placental mosaicism, a situation in which the placenta has cells with a chromosome abnormality while the fetus has normal chromosomes or vice versa. This means that there is a chance that the chromosomes in the fetus may not match the chromosomes in the DNA screened.

This test cannot be performed on patients who are carrying more than two babies (triplets or more), on patients who are carrying multiple babies (twin, triplets, etc.) where there is also an egg donor or surrogate, on pregnancies with a vanishing twin, or pregnancies in which the mother had a prior bone marrow/solid organ transplant.

**For patient who opted into microdeletion testing:** It is possible during analysis that you may be identified to be at increased risk to be a carrier of a 22q11.2 deletion. If this occurs, the Panorama report will state that there is a 1 in 2 or 50% chance to have an affected pregnancy (as fetal status cannot be determined in this case). Finding out you carry a microdeletion may cause feelings of anxiety or concern about your own health, as well as concerns about your pregnancy. Women who do not wish to risk finding out whether they carry this microdeletion should consider opting out of the microdeletion portion of the screening test. If the mother of the pregnancy is found to be a carrier of one of the other microdeletions on this panel, this screen will not be able to return results on the fetus. If you know you carry one of the microdeletions on this screen, it is recommended that you use another form of testing to detect the presence or absence of that microdeletion in your fetus. If the percentage of fetal (placental) DNA in the sample is below 7%, screening for Angelman syndrome will not be performed and the results will be reported as “risk unchanged”. A redraw will not be recommended and, if so chosen by the ordering healthcare provider, the cost will be borne by the patient.

**Alternatives:** There are multiple other prenatal screening options available, which you can discuss with your healthcare provider. You also have the option to decline all chromosome screening tests during your pregnancy. If you want or need more conclusive information about the fetal chromosomes, invasive diagnostic tests, such as CVS or amniocentesis, are available.

**Confidential Reporting Practices:** Natera® and LifeLabs comply with applicable American and Canadian privacy laws. Test results will be reported to the ordering healthcare provider(s) or genetic counsellor(s) involved. You must contact your provider to obtain the results of the test. Additionally, your personal information could be released to others, as permitted or required by law (e.g. – law enforcement).

**Genetic Counselling:** If you have remaining questions about non-invasive prenatal testing after talking with your healthcare provider, we recommend you speak with a genetic counsellor who can give you more information about your testing options. You can find a genetic counsellor in your area through the Canadian Association of Genetic Counsellors (<https://cagc-accg.ca/>).

**Cancellation, Disposition, or Retention of Samples:** If a test is cancelled prior to test set-up, LifeLabs will send a cancellation report free of charge. Once testing is initiated, the full price of the analysis will be charged. LifeLabs and/or Natera® may also keep your leftover de-identified samples for ongoing research and development. You and your heirs will not receive any payments, benefits, or rights to any resulting products or discoveries. If you do not want your de-identified sample and/or data used for the purposes listed above, you may send a request in writing to LifeLabs at 175 Galaxy Boulevard, Toronto ON, M9W 0C9 within 60 days after test results have been issued and your sample will be destroyed. You may also make this request by email to Ask.Genetics@LifeLabs.com and indicate “Sample Retention” in the subject line. LifeLabs will forward your request to Natera®, if your sample has been tested at their laboratory.

### PATIENT CONSENT STATEMENT

I have read or have had read to me the above informed consent information about the Panorama™ Non-Invasive Prenatal Test (NIPT). I have had the opportunity to ask questions of my healthcare provider regarding this test, including the reliability of test results, the risks, and the alternatives prior to giving my informed consent. I understand that my personal health information and my blood samples will be sent to LifeLabs Genetics in Toronto, ON, and/or to Natera’s® testing facility in San Carlos, California, USA. I request and authorize LifeLabs to test my sample(s) for the chromosome conditions listed above as indicated on my test requisition. I acknowledge that LifeLabs will send the results to my ordering healthcare provider. In the event of a high risk or no result, I acknowledge that LifeLabs may contact my healthcare provider to obtain follow-up diagnostic information to ensure quality and accuracy in reporting. I acknowledge that I must sign the consent statement located on the test requisition form that will be sent with my sample(s) to LifeLabs. I understand that I must also sign this consent form, which will remain in my clinic chart.

Signature of Patient

Printed Name

Date