## What does Panorama tell me?

Panorama gives you a personalized risk score and tells you if your baby is at high risk or low risk for certain genetic conditions.

### **Genetic Conditions Tested**

- Down syndrome (T21)
- Edwards syndrome (T18)
- Patau syndrome (T13)
- Certain sex chromosome abnormalities:
- Turner syndrome (monosomy X)
- Klinefelter syndrome (XXY)
- Jacob syndrome (XYY)
- Triple X (XXX) or vanishing twin
- Triploidy
- Sex of the child (if requested)

## **Additional Tests**

#### Microdeletions

- 22q11.2 Deletion Syndrome/DiGeorge Syndrome
- 1p36 Deletion Syndrome
- Angelman Syndrome
- Cri-du-chat Syndrome
- Prader-Willi Syndrome



Innovative Diagnostics is a medically managed service practice led by a group of doctors and supported by a dedicated team of senior and experienced industry professionals. We are forging a new path and aspire to set industry benchmarks in doctor and patient experiences.

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## Non-Invasive Prenatal Test

Learn about the health of your baby



# innevative DIAGNOSTICS

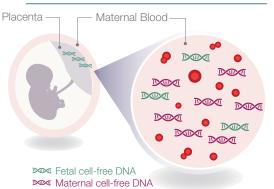
## Congratulations on being Pregnant!

What an exciting time. There are many things you are thinking about right now, one of which may be: "How healthy is my baby?" This is where Panorama™ can help you.

## What is Panorama?

Panorama™ is a Non-Invasive Prenatal Test (NIPT) that screens for Down syndrome and other genetic abnormalities caused by extra or missing chromosomes in the baby's DNA.

#### Baby's DNA in Mother's Blood



## How does this test work?

During pregnancy, some of the DNA from the baby crosses into the mother's bloodstream. Panorama looks at this DNA to see if there is evidence of certain conditions that could affect the baby's health. This is done by a simple blood test.

## When can I get a Panorama test?

You can have this test as early as 9 weeks gestation. And your doctor gets your results back in 7-10 working days.

## What other tests are available?

There are various other tests available. Traditional screening tests are not as accurate as Panorama, and diagnostic tests such as amniocentesis or chorionic villus sampling (CVS) have a slight risk of pregnancy complications including miscarriage.

## What syndromes does Panorama screen for?

#### • Down Syndrome (T21)

Babies with Down Syndrome have intellectual disabilities and specific physical features. They may have heart defects, weak muscle tone and can be smaller than average. Other health and learning problems can also occur in children with Down syndrome but these problems will vary from child to child.

#### • Edwards Syndrome (T18)

Babies born with Down Syndrome usually have lifethreatening birth defects and severe intellectual disabilities. They often have heart and brain defects along with birth defects of other organs. Babies with Trisomy 18 may be born small and have slow growth.

#### • Patau Syndrome (T13)

Babies born with Patau Syndrome have severe intellectual disabilities and life threatening birth defects, such as heart defects, brain or spinal cord problems, extra fingers and/or toes, cleft lip, cleft palate and weak muscle tone.

#### Triploidy

Most pregnancies with triploidy miscarry. The few babies that are born with triploidy have problems affecting the brain, heart, kidneys and other internal organs.

#### Jacob Syndrome (XYY syndrome)

Most babies with XYY syndrome do not have any birth defects. In childhood, boys with XYY may be tall for their age and some have an increased risk for learning, speech and behavioural problems.

#### • Turner Syndrome (Monosomy X)

Children with Monosomy X can vary from mild to more severe problems. Girls with Monosomy X may have heart defects and kidney problems. Most girls will be shorter than average height, some may have learning problems in school and hearing loss as they get older.

#### • Klinefelter Syndrome (XXY syndrome)

Most babies with XXY syndrome do not have any birth defects. In childhood, boys with XXY syndrome may be tall for their age and some have an increased risk for learning problems. Males with this condition often have lower levels of male hormones that can affect puberty and fertility.

#### Triple X Syndrome (Trisomy X)

Most babies with trisomy X do not have any birth defects. In some cases, babies with trisomy X may have heart defects, kidney problems and low muscle tone. In childhood, girls with trisomy X may be tall for their age and some have an increased risk for learning problems.



## What is a microdeletion?

A microdeletion is when a small piece of a chromosome is missing. Some microdeletions are known to cause specific genetic syndromes with major health impacts to the baby—including intellectual disabilities, heart and breathing issues, immune system problems, trouble feeding and other problems that may need immediate care upon birth.

## What microdeletion syndromes does Panorama screen for?

22q11.2 Deletion syndrome/DiGeorge syndrome
Babies born with 22q11.2 deletion syndrome often have
heart defects, immune system problems, and mild-tomoderate intellectual disability. They may also have
kidney problems, feeding problems, and/or seizures.

#### • 1p36 deletion syndrome

Babies born with 1p36 deletion syndrome have weak muscle tone, heart and other birth defects, intellectual disabilities, and behavior problems. About half will have seizures.

#### Angelman syndrome

Babies born with Angelman syndrome often have delayed milestones (like sitting, crawling and walking), seizures, and problems with balance and walking. They also have severe intellectual disability and most do not develop speech.

Cri-du-chat syndrome, also known as 5p minus
 Babies born with Cri-du-chat syndrome typically have low birth weight, small head size, and decreased muscle tone.

 Feeding and breathing difficulties are also common. They have moderate-to-severe intellectual disability.

#### • Prader-Willi syndrome

Babies born with Prader-Willi syndrome have low muscle tone and problems with feeding and gaining weight. They also have intellectual disability. As children and adults, they have rapid weight gain and often develop obesity-related medical problems.

## How do I get a Panorama test?

Your doctor orders the test, which is a simple blood draw from you. The baby's father can provide a cheek swab too, but it is not required, and will not affect the accuracy of the test. However, unlike other NIPTs that cannot use the father's cheek swab, this may increase the chance Panorama will be able to give you a result.

## Should I get Panorama?

If you do have concerns about the health of your baby and want peace of mind, consider Panorama. The first step is to talk with your doctor or medical specialist.

Some women have a higher chance of their baby being affected with certain conditions: women over the age of 35, women with certain family histories or abnormal ultrasound findings or blood test results.

However, the risk of having a baby with a microdeletion like 22q11.2 deletion syndrome is the same no matter what the mother's age. The American College of Obstetricians and Gynecologists recommends these women be offered an NIPT like the Panorama test (Committee Opinion No.545, Dec 2012).

## What if I receive a high risk result?

This is a screening test, not a diagnostic test. It is important to know that not all women who have a high-risk Panorama result will have a baby with a microdeletion, and not all women carrying a baby with a microdeletion will have a high-risk Panorama result.

Women who receive a high-risk result should follow-up with confirmatory diagnostic testing such as amniocentesis or chorionic villus sampling with microarray analysis. It is important to receive genetic counseling and to consult your doctor or healthcare provider about next steps.