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-to-moderate 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.

panorama™ prenatal test

MICRODELETION SYNDROME	FREQUENCY PER LIVE BIRTHS
22q11.2 Deletion Syndrome/ DiGeorge Syndrome	1 in 2,000[1]
1p36 Deletion Syndrome	1 in 5,000[2]
Angelman Syndrome	1 in 12,000[2]
Cri-du-chat Syndrome	1 in 20,000[3]
Prader-Willi Syndrome	1 in 10,000[2]
TOTAL FREQUENCY OF THESE SYNDROMES	**Approximately 1 in 1,000**

1. Nussbaum *et al* 2007. *Thompson and Thompson Genetics in Medicine* (7th edn). Oxford Saunders: Philadelphia.
2. http://www.genetests.org.
3. http://www.ncbi.nlm.nih.gov/entrez/disponim.cgi?id=123450.

The tests were developed by Natera, Inc., a laboratory certified under the Clinical Laboratory Improvement Amendments (CLIA). These tests have not been cleared or approved by the U.S. Food and Drug Administration (FDA).

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Information for you about Panorama's Microdeletion Screening

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Conceive. Deliver.

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. Panorama was first made available in early 2013 and looked specifically at chromosomes 21, 18, 13, X, and Y.

A revolutionary new capability – Panorama now screens for microdeletion syndromes.

Until now, there has never been a way to screen for microdeletions. Ultrasound is unreliable, especially in the first trimester of pregnancy. The only way to know for sure is with an invasive procedure like amniocentesis, which carries a small risk of miscarriage. Panorama is a non-invasive, risk-free, and highly accurate way to screen for microdeletions.

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.

Does the chance of having a baby with a microdeletion increase with the mother's age or family history?

No. Unlike Down syndrome and some other conditions whose risk increases with the age of the mother, a 20-year-old has the same risk as a 45-year-old. In most microdeletion cases, there is no previous family history.

Should I get screened for microdeletions?

That is a decision you should make with your doctor or healthcare provider. If you are looking for greater reassurance about the health of your baby, this test is available as early as 9 weeks into your pregnancy.

How does the microdeletion screening work?

The Panorama test is able to look at the baby's DNA through a simple blood test from the mother. The test differentiates between the mother's DNA and the baby's DNA, and then it screens to see if the baby is at high risk for having a specific microdeletion. The test screens for only microdeletions that cause syndromes with major health impacts. These syndromes and their frequencies are listed on the back panel of this brochure.

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.

Where can I get more information about these conditions?

You can ask your healthcare provider or seek a genetic counselor near you at www.nsgc.org. You may also visit the Support Foundation page of the Panorama website: http://www.panoramatest.com/resources where you will find links to organizations that offer information about specific syndromes and related support networks.