SMART Study Finds 22q11.2 Microdeletion Prevalence Much Higher than Expected

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Natera introduces Panorama AI, improving upon its industry-leading detection for 22q11.2 microdeletions

SAN CARLOS, Calif., Feb. 1, 2021 /PRNewswire/ -- Natera, Inc. (NASDAQ: NTRA), a pioneer and global leader in cfDNA testing, presented key results from its SMART study at the SMFM 41st Annual Pregnancy Meeting.[1] The SMART study sets a new standard as the largest prospective NIPT study to date (N = 20,927 enrolled from 21 medical centers), and the only large-scale study to collect genetic outcomes in most of the subjects. The study includes the validation of a new artificial intelligence-based algorithm for Panorama® called Panorama AI, which utilizes information from over 2 million cfDNA tests performed by Natera.

Key results related to the 22q11.2 microdeletion:

- Study showed a higher-than-expected prevalence of 1/1,524 in the general population that compares well with conditions broadly recommended for routine testing such as trisomy 21 in average risk pregnancies (~1/1,000), cystic fibrosis (~1/2,500), and spinal muscular atrophy (~1/10,000).[2-5]
- Study reaffirmed Panorama's highest commercially available sensitivity in detecting the most common 2.54Mb microdeletions.
- Study expanded Panorama's capabilities by enabling accurate identification of small microdeletions, which no massively parallel shotgun sequencing (MPSS)-based NIPT has validated.
- Small deletions accounted for at least 41% of the disease load, providing a significant competitive advantage for Panorama compared to MPSS-based NIPTs that exclude small microdeletion sizes in their performance claims.
- Combined performance, even when including small microdeletions, was better than any other published NIPT result to date (sensitivity 83%, PPV 53%).

"This is the first prospective NIPT study in which genetic outcomes were confirmed in the vast majority of the patients enrolled, and provides a wealth of data about the real-world performance of NIPT across a diverse group of global centers and patients," said Mary Norton, MD, Professor, UCSF, and one of the Principal Investigators of SMART. "The findings related to high prevalence of 22q11.2 deletion syndrome, the limited ability of ultrasound to detect all cases prenatally, and the performance of NIPT in detection of these cases with high accuracy provide exciting data to inform discussions around testing for a broader set of conditions beyond common aneuploidies."

"The diagnostic odyssey related to 22q11.2 deletion syndrome is well documented, with median time to diagnosis of almost 5 years.[6] And in the meantime, a window of opportunity might be lost to intervene and impact outcomes. Delivery of a child with 22q11.2 deletion syndrome should be at a tertiary facility well-equipped to deal with short-term complications that are associated with the disorder.[7] Depending on the issue at hand (e.g., cardiac, endocrine), appropriate interventions are warranted. For example, timely administration of neonatal calcium has been shown to correlate with preventing the intellectual decline commonly seen in affected children,"[8,9] said Pe'er Dar, MD, Albert Einstein College of Medicine, Bronx NY, and one of the Principal Investigators of SMART. "With the ability to detect more accurately in combination with a low false positive rate, I believe that the findings of the SMART study provide professional societies with sufficient evidence to consider including screening for 22q11.2 deletions in routine prenatal genetic screening."

In 2020, Natera performed over 400,000 tests for the 22q11.2 microdeletion. Natera has established a CPT code and favorable pricing for microdeletion testing. Based on high prevalence and excellent performance in the study, Natera looks forward to engaging professional societies for routine testing of pregnancies for the 22q11.2 microdeletion, and will then pursue broader insurance coverage.

About Panorama®

Panorama reveals a baby's risk for severe genetic disorders as early as nine weeks into pregnancy. The test uses a unique single-nucleotide polymorphism (SNP)-based technology to analyze fetal/placental DNA obtained through a blood draw from the mother. It is the only commercially available test that differentiates between maternal and fetal DNA to assess the risk of aneuploidies. The test also screens twin pregnancies for zygosity and fetal sex of each baby, and identifies risk for more genetic conditions in twin pregnancies than any other NIPT. Panorama is one of several genetic screening tests from Natera designed to help families on the path to parenthood. Natera has published 23 papers, studying over 1.3 million patients, since the launch of Panorama – the largest body of evidence in the space today. Panorama has been developed and its performance characteristics determined by Natera, the CLIA-certified laboratory performing the test. The test has not been cleared or approved by the US Food and Drug Administration (FDA). CAP accredited, ISO 13485 certified, and CLIA certified.

About Natera

Natera is a pioneer and global leader in cell-free DNA testing from a simple blood draw. The mission of the company is to change the management of disease worldwide with a focus on women's health, oncology, and organ health. Natera operates ISO 13485-certified and CAP-accredited laboratories certified under the Clinical Laboratory Improvement Amendments (CLIA) in San Carlos, California and Austin, Texas. It offers proprietary genetic testing services to inform obstetricians, transplant physicians, oncologists, and cancer researchers, including biopharmaceutical companies, and genetic laboratories through its cloud-based software platform. For more information, visit natera.com. Follow Natera on LinkedIn.

Forward-Looking Statements

All statements other than statements of historical facts contained in this press release are forward-looking statements and are not a representation that Natera's plans, estimates, or expectations will be achieved. These forward-looking statements represent Natera's expectations as of the date of this press release, and Natera disclaims any obligation to update the forward-looking statements. These forward-looking statements are subject to known and unknown risks and uncertainties that may cause actual results to differ materially, including with respect to our efforts to develop and commercialize new product offerings, our ability to successfully increase demand for and grow revenues for our product offerings, whether the results of clinical or other studies will support the use of our product offerings, our expectations of the reliability, accuracy and performance of our tests, or of the benefits of our tests and product offerings to patients, providers and payers. Additional risks and uncertainties are discussed in greater detail in "Risk Factors" in Natera's recent filings on Forms 10-K and 10-Q and in other filings Natera makes with the SEC from time to time. These documents are available at www.natera.com/investors and www.sec.gov.

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