Progenity launches first commercially available, custom-designed, noninvasive prenatal test for monogenic diseases

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Progenity also announces improvements to its Innatal® noninvasive prenatal test (NIPT), demonstrating market-leading sensitivity and specificity across all common chromosomal aneuploidies.

SAN DIEGO, April 2, 2019 – Progenity, Inc., a privately held biotechnology company, announced today the launch of the Resura® Prenatal Test for Monogenic Disease, the first of its kind, commercially available, customizable, noninvasive prenatal test (NIPT) for single-gene disorders.

This novel test broadens expectant parents' prenatal testing options to include a safe, noninvasive test tailored to their family's risk for a specific genetic disease. In addition to this launch, Progenity announced improvements to its Innatal® Prenatal Screen, a market-leading NIPT for chromosomal aneuploidies that now provides greater specificity and sensitivity in evaluating aneuploidy across all chromosomes tested in a recent validation study. Both products have been developed to provide increased confidence in genetic testing results, so informed and individualized care decisions can be made by patients planning for their future families.

"Since the advent of cell-free DNA testing for chromosomal abnormalities, healthcare providers and patients have been asking for a similar noninvasive testing pathway for monogenic disease," said Harry Stylli, PhD, CEO, Chairman of the Board and a founder of Progenity. "With the development of the Resura test, we accepted and overcame the challenges inherent in differentiating the fetal genotype from the maternal background, and we are proud to introduce a new, noninvasive testing option to families impacted by rare disease."

The Resura test is available to families with known risk for monogenic disease, which is typically caused by a mutation within a single gene. Common examples of monogenic disease include cystic fibrosis, sickle cell anemia, and Tay-Sachs disease. For many of these diseases, knowing the diagnosis before birth informs critical treatment decisions upon the infant's arrival. The Resura test can be performed on disease-causing variants of all inheritance types, including recessive, dominant, and X-linked genetic mutations. Currently, testing for these genetic variants in a fetus involves undergoing invasive prenatal testing, such as amniocentesis, or waiting for postnatal diagnosis. The Resura test uses fetal cell-free DNA (cfDNA) extracted from a sample of the mother's blood to test for genetic variants. Validation data for the Resura test was presented at the National Society of Genetic Counselors' 37th Annual Conference in November 2018 and can be found here.

Progenity also upgraded its Innatal Prenatal Screen, a noninvasive prenatal test for chromosomal aneuploidy, with the latest sequencing technology, improved chemistry, and bioinformatic analysis. These enhancements solidify the test's superiority compared to competitive tests to detect aneuploidies in chromosomes 21, 18, 13, X, and Y in our validation study of over 1,400 patients. The high sensitivity and specificity for this screening test ensure false positives or false negatives are very rare, which increases the positive predictive value (PPV) and negative predictive value (NPV) of the test. Together, this gives more assurance to providers and patients that the results accurately reflect the genetic makeup of the fetus. The Innatal test also has one of the lowest failure rates available, which is important because current ACOG guidelines recommend treating a failed test as a high-risk result and offering invasive diagnostic testing to the patient.

"Progenity is driving development of the next generation of noninvasive prenatal testing, fueled by our commitment to continually advance current technologies and develop innovative products for unmet medical needs, like the Resura test," said Matthew Cooper, PhD, Chief Scientific Officer at Progenity. "And with over one million next-generation sequencing tests performed in house at our CLIA-certified and CAP-accredited laboratory, we have proven experience helping physicians and patients make individualized care decisions."

To further support patients and providers in understanding prenatal test results, Progenity employs a team of board-certified genetic counselors with more than 250 years combined experience as a resource for its customers.

More information about Progenity's prenatal testing is available here.

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