



## Sema4 Applies Data-Driven Approach to Develop and Launch New NIPT and Expanded Carrier Screen Tests

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*Company broadens offerings across the reproductive health journey*

**Stamford, CT — August 21, 2018**— Sema4, a patient-centered predictive health company, today announced the launch of two reproductive health tests: the new Sema4® Noninvasive Prenatal Select and an enhanced version of its Expanded Carrier Screen. These products, developed with an informatics-driven approach using Sema4's Health Intelligence Platform (SHIP), add to the company's industry-leading portfolio of tests designed to support providers and patients from preconception to pediatrics.

Using only a maternal blood sample, Sema4 Noninvasive Prenatal Select extensively screens the fetal DNA in that sample for common chromosome aneuploidies, sex chromosome aneuploidies, and microdeletion syndromes, while also offering the flexibility to order select components of the test. Built on cutting-edge technology with advanced statistical modeling, the test delivers highly accurate results (>99% for common aneuploidies) as early as nine weeks into pregnancy. This is the only non-invasive prenatal test (NIPT) offering on the market that routinely screens for trisomy 15 to aid in detection of Prader-Willi and Angelman syndromes. The American College of Obstetricians and Gynecologists (ACOG) and the Society for Maternal-Fetal Medicine recommend that all pregnant women are offered screening or diagnostic testing for fetal aneuploidy.

Sema4 has also updated its Expanded Carrier Screen of more than 280 inherited conditions. DNA-based carrier screening, which is recommended by ACOG to help guide family planning for all women who are pregnant or planning for pregnancy, is an important tool that allows people to determine their risk of passing certain diseases or disorders on to their children. An informatics-driven approach was used to select the most clinically relevant genetic variants for inclusion in the Expanded Carrier Screen, by querying data from publicly available databases in combination with Sema4's proprietary, curated knowledge base. The resulting Expanded Carrier Screen uses multiple technologies for robust and accurate detection of a broad range of genetic variants, including copy number variation, and is offered with flexible options to allow providers and patients to choose subsets of genes if the entire panel is not needed. With the new Expanded Carrier Screen, Sema4 has added full exon sequencing via long-range PCR to improve detection rates for genes traditionally difficult to sequence, including the *CYP21A2* (congenital adrenal hyperplasia due to 21-hydroxylase deficiency), *GBA* (Gaucher disease) and *HBA1/HBA2* (alpha-thalassemia) genes.

"Genetic testing is revolutionizing the reproductive health journey by giving parents and prospective parents unprecedented and actionable information ahead of, during, and after pregnancy," said Eric Schadt, PhD, Founder and Chief Executive Officer of Sema4. "Our newest tests were developed using advanced data science technologies, leveraging our proprietary knowledge base of genomic information, to provide patients and healthcare providers with the most meaningful and actionable insights."

For both tests, customers may use Sema4's patient portal to seamlessly track the status of the test and view results. Sema4 is committed to enabling patients to better manage all their health information, including electronic medical records, for those who electronically consent to do so. The patient portal also includes helpful educational resources for Noninvasive Prenatal Select, including videos for pre-test patients and videos for post-test patients with negative results. Phone-based genetic counseling is available to all patients.

Sema4's other reproductive health offerings include CarrierCheck, a carrier screening test of 67 conditions, and Natalis, a highly accurate test of 193 treatable, childhood-onset conditions that parents can use to gain early insight into a baby's health. The Expanded Carrier Screen test was first launched in 2015 at Mount Sinai Health System.

For more information about Sema4 Noninvasive Prenatal Select or the Expanded Carrier Screen, including lists of all genes analyzed, visit [sema4.com/products](http://sema4.com/products).

### About Sema4

Sema4 is a patient-centered predictive health company founded on the idea that more data, deeper analysis, and increased engagement will improve the diagnosis, treatment, and prevention of disease. A Mount Sinai Health System venture based in Stamford, Connecticut, Sema4 is enabling physicians and consumers to more seamlessly engage the digital universe of health data, from genome test results and clinical records to wearable sensor metrics and more. The company currently offers advanced genome-based diagnostics for reproductive health and oncology and is building predictive models of complex disease. Sema4 believes that patients should be treated as partners, and that data should be shared for the benefit of all.

For more information, please visit [sema4.com](http://sema4.com) and connect with Sema4 on [Facebook](#), [Twitter](#) and [YouTube](#).

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