Sema4 Launches Supplemental Newborn Screening Test for 190+ Childhood-Onset Diseases

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Simple, at-home DNA test gives parents early insights into treatable health conditions

Stamford, CT — February 7, 2018 — Sema4, a health information company providing advanced genomic testing, today announced the launch of Sema4 Natalis, a supplemental newborn screening test designed to detect 193 childhood-onset diseases or disorders so parents can gain early insight into their baby's health. This simple genetic test can be performed at home with a gentle cheek swab.

In the United States, newborns are typically screened at the hospital for 34 health conditions on the Recommended Uniform Screening Panel (RUSP), but the selected conditions vary by state and represent only a fraction of the genetic diseases that can manifest in a child's first decade of life. Advances in DNA analysis have made it possible to detect the presence of far more childhood-onset health conditions long before symptoms appear. As a supplemental test, Sema4 Natalis screens for more than five times the number of genetic diseases than a state's standard hospital test. For all conditions covered by the test – including atypical epilepsy, spinal muscular atrophy, and childhood cancers, among many others – there are validated medical interventions that may positively influence a baby's future wellbeing when introduced early enough. Parents can order Sema4 Natalis online, and all orders will be reviewed and approved by a physician to ensure they are medically appropriate for the child.

"Until now, families have been likely to be caught off-guard by these early-onset diseases, and prognosis is often poor by the time symptoms have manifested. Thanks to breakthroughs in science and medicine, we can now identify babies at risk for these broader set of diseases and deliver interventions – sometimes as simple as vitamin supplements – in time to make a real difference," said Eric Schadt, PhD, Founder and Chief Executive Officer of Sema4. "We believe Sema4 Natalis will give parents the advantage of early insight in support of the care of their children."

Every Sema4 Natalis order also includes a pharmacogenetic analysis of how a baby is likely to respond to 38 medications commonly prescribed at an early age. This information can help pediatricians guide prescription choice to avoid adverse effects or incorrect doses of medications, including antibiotics.

Sema4 Natalis uses advanced DNA sequencing to analyze a baby's genes with the accuracy of next generation technology. For children who screen positive for a health condition, genetic counseling is available to the parents and pediatricians to discuss results and help navigate next steps. For parents who missed the opportunity when their babies first came home, the test can be used for children up to 10 years of age.

As noted in its recent comments to the Centers for Medicare and Medicaid Services, the National Organization for Rare Disorders (NORD) finds that "on average, individuals with a rare disease wait seven to 10 years to obtain an accurate diagnosis, leaving many individuals with chronic conditions still waiting for a diagnosis. There are millions of patients in the U.S. who are still undiagnosed, and [next-generation sequencing] may be their only hope."

Sema4 Natalis was developed to help address this issue of undiagnosed pediatric illness by using next-generation DNA sequencing and analysis to supplement traditional newborn screening.

For more information, or to order Sema4 Natalis online, visit www.sema4.com/Natalis.

About Sema4

Sema4 is a health information 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, Conn., 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 and connect with Sema4 on Facebook, Twitter and YouTube.

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