# Poll Finds Most Americans Want to Know About Their Baby's Genetic Risk for Treatable Childhood Disorders

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*88% would want to find out their baby's genetic risk factors for treatable early onset disorders, and 92% think it's important for new parents to be able to learn about their child's risk of experiencing these conditions. 87% say they would likely request a non-invasive DNA test to supplement their state's newborn screening panel if their state test did not include testing for many treatable conditions that could affect a child in their first years of life.*

**STAMFORD, CT – February 28, 2018** – Sema4, a health information company providing advanced genomic testing, today announced the results of a public opinion poll of among more than 2,000 Americans about newborn genetic screening. According to the survey, commissioned by Sema4 and conducted online by Harris Poll, 88% of Americans say that if they were new parents and could find out just after their baby is born about their child's genetic risk of getting a treatable early onset disease, they would want to know. Similarly, 92% of Americans think it is important for new parents to be able to learn about their baby's genetic risk of treatable early-onset conditions that typically manifest in a child's first 10 years.

"This poll to assess the attitudes of consumers toward genetic screening for newborns is just one component of our overall program to gain new insights into genomic health," said Eric Schadt, PhD, Founder and CEO of Sema4. "We seek to understand, in partnership with our patients and the scientific and medical communities, the benefits of obtaining supplemental genetic information at birth. To this end, we are in the process of launching a clinical study to explore the utility of supplemental newborn screening to both patients and physicians, including how individuals respond to this new type of actionable information, what actions they take as a result, and how this ultimately impacts health outcomes and healthcare utilization."

The results of the poll were announced on Rare Disease Day, an international day of awareness that shines a light on rare diseases and their impact on patients' lives. According to Global Genes and the Shire Rare Disease Index Report, more than 7,000 rare diseases have been identified, 80% of which are caused by genetic variations. These diseases collectively affect 30 million Americans, half of whom are children. Today, it takes most rare disease patients an average of 8 years to obtain an accurate diagnosis.

While most rare diseases still have no FDA-approved treatment, some genetic diseases have known effective treatments, especially when administered early. These can be as simple as a change in diet or vitamin therapy, as is the case in pyridoxine-dependent epilepsy, which is treatable with a form of vitamin B6. Such a lifesaving and simple intervention for this rare form of epilepsy was dramatically illustrated in a recent case at Rady Children's Hospital in San Diego.

In the United States, newborns are typically screened at the hospital for 34 health conditions on the Recommended Uniform Screening Panel (RUSP), but the conditions screened vary by state and represent only a fraction of the genetic diseases that can manifest in a child's first decade of life. Pyridoxine-dependent epilepsy, for example, is not currently on any state panel.

**Parents Would Like More Information Than Most State Newborn Panels Provide**

According to the survey, 87% of Americans say that if the newborn screening test in their state did not cover many treatable conditions that could affect a child in his or her first years of life, and a supplemental non-invasive DNA test was available to screen for such conditions, they would be likely to request this supplemental test.

"In my practice, I find that parents generally want as much information as possible related to the health of their babies, and access to genomic data now opens up a whole new era," said Dr. Joanne Stone, Director of Maternal Fetal Medicine for the Mount Sinai Health System, and Professor of Obstetrics, Gynecology and Reproductive Science at the Icahn School of Medicine at Mount Sinai. "When accompanied by appropriate genetic counseling, high-quality DNA testing can provide valuable, actionable insights and help guide decision making as part of the parental journey, from pre-conception carrier screening to expanded screening for newborns. In general, we are finding that interest is growing among parents in taking more control of the health of their family."

Sema4, which commissioned this poll, was founded on the idea that more information, deeper analysis, and increased engagement will improve the diagnosis, treatment, and prevention of disease. As part of its clinical laboratory services, the company offers optional DNA tests for parents and prospective parents who are interested in gaining more insight into their own genetic carrier status as well as their newborn's genetic risk factors. For example, Sema4 Natalis screens for more than five times the number of genetic diseases on a standard state-mandated test, as well as the potential for adverse reactions to medications commonly prescribed during childhood. Testing takes places in a rigorously controlled, CLIA-certified laboratory and is overseen by qualified physicians.

Rare disease advocate Kim Tuminello, Co-Founder, Association of Creatine Disorders, commented: "The idea of every family having screening information about Guanidinoacetate Methyltransferase Deficiency following birth, as opposed to the average

8-year odyssey that most families currently endure, is a concept that is now possible through the Sema4 Natalis newborn screening test.  We're excited to share news with all of our member families about the availability of Sema4 Natalis."

Reproductive health experts Dr. Joanne Stone, Director of Maternal Fetal Medicine at The Mount Sinai Hospital and Dr. Alan Copperman, Chief Medical Officer of Sema4 and Medical Director of Reproductive Medicine Associates, will host a Facebook LIVE discussion about newborn screening and the new Natalis test on March 7th from 1-1:45pm EST at www.facebook.com/sema4.

**Survey Methodology**
This survey was conducted online within the United States by Harris Poll on behalf of Sema4 from February 22-26, 2018, among 2,037 U.S. adults ages 18 and older. This online survey is not based on a probability sample and therefore no estimate of theoretical sampling error can be calculated. For complete survey methodology, including weighting variables, please contact glenn.farrell@sema4.com.

**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 LinkedIn.