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GUARDIAN Study to Explore Newborn Screening by Genome Sequencing in 100K Babies in NYC

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Premium

BOSTON – A new research study in New York City called GUARDIAN (Genomic Uniform-screening Against Rare Diseases in All Newborns) is exploring the utility of whole-genome sequencing for screening newborns for rare and, for the most part, treatable genetic conditions.

The study, which hopes to enroll 100,000 babies in New York City within the next four years, is a partnership between Columbia University Irving Medical Center, NewYork-Presbyterian, the New York State Department of Health, Sema4, and Illumina. It is open to all babies born at one of NewYork-Presbyterian's hospitals.

Wendy Chung, a professor of pediatrics and medicine at Columbia University and the study's principal investigator, presented an outline of the project at the inaugural International Conference on Newborn Sequencing in Boston on Thursday.

Also, Sema4 announced earlier this week that its GeneDx subsidiary will provide whole-genome sequencing and interpretation services as a partner in the program. In addition, Illumina is contributing reagents, and support "for everything else" is coming from philanthropic sources, Chung told *GenomeWeb*.

GUARDIAN is among a number of new projects around the world presented at the meeting that are testing the clinical use of genome sequencing for newborn screening.

All women delivering at a NewYork-Presbyterian hospital will be invited, mostly in person, to participate in GUARDIAN and are consented through an online portal. Parents can also sign up in advance of their baby's delivery and enroll once their child has been born. Information about the study in English, Spanish, Russian, and Chinese — with additional languages to be added — is available on a website and through flyers distributed at Ob/Gyn offices and elsewhere.

According to Chung, about 70 percent of families invited to the study have decided to participate so far, and 250 babies have been enrolled.

The study uses the same dried blood spots that are already collected in the hospital for the state newborn screening program, and results are available to parents within three to six weeks, a time frame that is likely to get shorter as the project gets going. They are also shared with the child's pediatrician and go into the medical record. In addition, test results are reported to the New York State Department of Health's newborn screening program, which already looks for about 50 disorders through metabolic testing.

GUARDIAN researchers will screen for several hundred rare genetic conditions, most of which are currently not part of standard newborn screening. Chung said the criteria for including a disease were onset before the age of 5 years, a 90 percent penetrance, and the availability of an "effective treatment" — for example, a special diet, medication, or a bone marrow transplant — that lessens symptoms.

While all babies enrolled in the study will be screened for 158 conditions that have a treatment available, parents can also choose to learn about a second set of about 100 conditions, many of them neurodevelopmental, that are currently not treatable. This might allow their child to receive other types of medical support early, such as physical, occupational, or speech therapy, and to be considered for clinical trials of treatments currently in development.

Chung said that, so far, 90 percent of parents who enrolled have opted to learn about both groups of conditions.

With the current list of diseases, about one in 1,000 babies are expected to screen positive, meaning approximately 100 newborns in the entire study. However, Chung said that her team has already identified additional disorders to screen for, including more than 100 treatable ones, and the study will soon shift to version 2.0 of the disease list for new enrollments, thus doubling the number of expected positive cases.

If a baby tests positive, parents will talk to a genetic counselor or a geneticist to discuss further steps and receive help in finding follow-up care with a specialist, as well as information about research studies they might be eligible for. However, the study will not pay for any additional care or treatment.

The sequencing data generated for the study will be stored on a private, secure server at Columbia University, with a backup copy at GeneDx, Chung said.

According to the study website, all genomic data will be stored in an identifiable manner for the duration of the study. Afterwards, all identifiable information will be removed, but the data will be kept. Pooled, anonymous allele frequency data will also be made available to researchers at some point through an online browser.

No analyses beyond those for the GUARDIAN study will be conducted unless the family or their pediatrician asks for them, or the family consents to additional research. For example, if a child develops symptoms of a condition that the study didn't screen for, the parents can ask for the genome to be re-analyzed, though they will need to bear the costs, according to the website.

Editor's Note: The inaugural International Conference on Newborn Sequencing (ICoNS) conference was organized by Genomes2People and Ariadne Labs in partnership with GenomeWeb.

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