Newborn screening conference examines privacy concerns

Newborn screening programs exist in all 50 states. Conducted when an infant is only a day or two old, newborn screening checks a baby for rare, serious and mostly treatable health conditions such as sickle cell disease, cystic fibrosis, phenylketonuria, hypothyroidism and more. It is done through a blood test by pricking a baby's heel to get a few drops of blood. A hearing test and heart screening are also conducted.

Whole genome sequencing — which maps out an individual's entire DNA sequence — can be used to diagnose a disease in a sick child, but is also used in healthy newborns to look for potential issues. It is not routinely done as part of newborn screening. But will that change in the future? And what are the potential privacy consequences?

"We are at a time where newborn screening is sitting between changing technologies, changing panels, the use of whole genome and exome sequencing in both families experiencing rare disease and in healthy newborns. … At the same time, on the other side, we have an increased concern among families and communities around genetic privacy," said session co-moderator Aaron Goldenberg, PhD, MPH, professor and vice chair in the bioethics department at Case Western Reserve University and the co-director for Case Western's Center for Genetic Research Ethics and Law.

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