

# Should we screen all newborns for neurodevelopmental disorders?

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Expanding newborn screening (NBS) to include identifying genes associated with an increased risk for neurodevelopmental disorders (NDD) would cause more harm than good, according to an article published in *Pediatrics*. While some experts believe early identification of NDDs in the newborn period would provide an equitable way to flag and treat disabilities early, the authors of the new paper contend that

broader genomic sequencing would worsen existing health disparities.

"The benefit of early genomic screening hinges upon the ability to identify [children](#) with NDD early and then provide timely access to therapeutic supports," said lead author, Sarah Sobotka, MD, assistant professor of Developmental and Behavioral Pediatrics at the University of Chicago. "The reality is that we're practicing in a context where there are few supports and disparate access to care for children already identified as having an NDD."

Given the dire shortage of genetics experts and diagnosticians in the U.S., Sobotka and co-author Lainie Ross, MD, Ph.D., chair of the Department of Health Humanities and Bioethics and director of the Paul M. Schyve, MD Center for Bioethics at the University of Rochester, recommend strategic use of NDD screening on populations of children who actually show signs of developmental delays.

## **Ambiguity and the problem of access**

Researchers have yet to prove that genotype ([genetic makeup](#)) and phenotype (signs or behavior that are manifested outwardly) are scientifically matched. This can raise alarms, and in some cases, lead to overtreating disorders that may never manifest in a child.

Families may receive a diagnosis and label children based on the premise that a genetic variant means a child has an NDD or will develop one in the future. This can lead to stigma and self-fulfilling prophecy, causing harm to those who never display signs or symptoms.

Further exacerbation stems from the fact that there is an inadequate supply of trained therapists to provide the services needed for children with NDDs. Increasing early identification of those at risk who may never present with developmental delays would inevitably favor those

with resources—children whose parents are more poised to advocate—widening the gap for minoritized groups who already struggle to access care.

## **Data that lack diversity**

The authors argue that our current body of genetic population data is also skewed because the majority of participants in genetic research have been members of the white middle class. This could naturally lead to more diagnoses for children in those groups—hence, quicker access to [early interventions](#) that would leave others who need urgent help behind, especially if they are in a minoritized group that has not yet been adequately studied.

Not only do we not have enough genetic diagnosticians to meet demand for broad testing, we don't have adequate genetic information about variants in [diverse communities](#), Ross explains.

"If we wait to test the children until they manifest signs of delays, we can actually start looking for other genetic variants within those populations so when early genomic testing is proposed again in 10 or 20 years, we will have a better understanding of the extent to which these genotypes will correlate with NDD phenotypes," said Ross, who is both a bioethicist and a pediatrician. "We'll also have collected a much more diverse set of genotypes, so we will actually be looking for these differences in all populations."

## **A new approach to early screening**

While Sobotka and Ross oppose broad genomic sequencing of newborns, they are not against early screening for NDD. They argue the best way to do that is not through universal NBS programs but focusing resources on

early identification by general pediatricians during routine well-child visits by using standardized screening assessment tools as well as referrals from head-start programs or other early childhood education programs.

"We need to identify neurodevelopmental delays in children early," said Sobotka, "and we need to provide enrichment opportunities, when possible, to prevent delays from developing." As Sobotka points out, such supports include not only educational interventions but also supportive family leave policies that encourage early parent-child attachment and parent-child interactive activities.

"There are population-based interventions proven to improve neurodevelopmental outcomes that we are still not doing. These should be our first public-health initiatives; not expanding NBS," she concluded.

**More information:** Newborn screening for neurodevelopmental disorders may exacerbate health disparities, *Pediatrics* (2023).

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