

# New genetic test detects hundreds of harmful mutations among high-risk groups

April 25 2019

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Researchers at Nemours Children's Health System have developed a new low-cost genetic test that accurately identified more than 200 known disease-causing gene variations in two high-risk populations, the Old Order Amish and Old Order Mennonites of Lancaster County, Pennsylvania. The findings, published today in the *Journal of Molecular Diagnostics*, could help reduce morbidity and mortality from these rare genetic disorders, and dramatically reduce costs of care through early diagnosis of newborns.

"These are genetically isolated communities with a higher incidence of certain disorders," said Erin Crowgey, Ph.D., lead author of the study and associate director for bioinformatics at Nemours. "Robust, expanded screening and diagnostic testing in these populations for people who may carry these gene variants could potentially improve patient outcomes and reduce [medical costs](#), through a single low-cost procedure."

The research team developed a next-generation sequencing test capable of identifying 202 known gene variants, or alleles, associated with 162 conditions found in these Amish and Mennonite communities, also known as Plain communities, to identify carriers of these variants in a simple blood test. To do so, the Clinic for Special Children (CSC), a medical home for uninsured Plain community children with genetic disorders, selected 63 patients to participate by providing [blood samples](#) for the DNA analysis.

The research team created a targeted gene panel using a custom

configuration of the ArcherDX Anchored Multiplexed PCR (polymerase chain reaction) technology. In total, 309 different gene variants were detected. To assess the test's accuracy, they used an alternate method to validate all genetic variants. For instance, they compared 48 samples with prior whole exome sequencing results, and found 100 percent agreement between the two methods.

"Due to their small number of community founders, the Plain populations over time have come to exhibit relatively high carrier rates for a small set of genetic diseases," said Erik Puffenberger, Ph.D., a study author and laboratory director at CSC. "We needed a methodology for a single procedure to test individuals for all known genetic variations related to those conditions."

The researchers note that population-wide carrier screening may help decrease morbidity and mortality, as well as treatment costs associated with genetic conditions in these high-risk groups by helping to identify asymptomatic newborns when many of these conditions are most treatable.

"For many disorders that disproportionately occur in the Plain communities, detection of asymptomatic infants is critical to prevent the devastating effects of these diseases," said Crowgey. "Without rapid diagnosis and treatment of newborns, children can have shorter lifespans or lasting disabilities, even after intensive, lengthy, and costly medical care."

The study authors estimate that use of this low-cost screening tool (approx. \$165 per person) by Plain communities could result in hundreds of thousands of dollars health care costs savings annually.

Other members of the study team include Michael Washburn, Ph.D., of ArcherDX, Inc., and E. Anders Kolb, MD, of Nemours Children's

Health System.

Matthew Demczko, MD, a pediatrician who cares for Amish children at Nemours Kinder Clinic in Dover, Delaware, and was not involved in this research, said: "In our ongoing study of the Delaware Amish, we've found that the community is aware of their increased risk for inherited disorders and knowledgeable about carrier screening. They generally perceive screening to be useful to avoid surprises and guide care decisions for their children. We hope that data from this study will help promote larger screening efforts throughout Plain communities in North America."

Provided by Nemours Children's Health System

Citation: New genetic test detects hundreds of harmful mutations among high-risk groups (2019, April 25) retrieved 6 May 2024 from <https://medicalxpress.com/news/2019-04-genetic-hundreds-mutations-high-risk-groups.html>

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