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FOR IMMEDIATE RELEASE

NOVEL NEXT GENERATION SEQUENCING ASSAY FOR CARRIER SCREENING IN PLAIN POPULATIONS IDENTIFIED

STRASBURG, PA- A new study has identified a novel next generation sequencing assay to carrier test for autosomal recessive disorders found in the Old Order Amish and Old Order Mennonite (Plain) populations. Due to the small number of founders and a phenomenon known as genetic drift, the Plain communities show relatively high carrier rates for a small subset of genetic diseases.

The study included 48 samples based on prior whole exome sequencing (WES) results. In order to assess the accuracy of the panel, 168 unique genes were targeted, with a focus on 202 genetic variants associated with 162 different syndromes found in the Plain communities. An additional 15 samples were used to validate *SMN1* and *SMN2* copy number analyses. Mutations or deletions in *SMN1* causes Spinal Muscular Atrophy (SMA) and the number of *SMN2* copies correlates to disease severity. Collectively, the panel detected 273 pathogenic (single nucleotide or small insertion/deletion) variants, 35 copy number variants (CNVs), and one chromosomal abnormality (Klinefelter syndrome). The panel matched 100% with the previous WES analyses. The researchers utilized Anchored Multiplex PCR (AMP™) technology, with its use of a unique molecular index created by ArcherDX, to target numerous classes of variants with a wide range of allele frequencies.

The study, by clinicians and researchers at the Clinic for Special Children in Strasburg, PA, Nemours/A.I. duPont Hospital for Children in Wilmington DE, and ArcherDX, Inc. in Boulder, CO, appears in the April 2019 issue of *Journal of Molecular Diagnostics.* This expanded carrier screening method has allowed the successful creation of Plain population-wide carrier testing and has the potential to drastically reduce overall medical costs and improve patient outcomes by identifying at-risk couples and informing

them of their carrier status before affected children are born. This also allows the opportunity for presymptomatic treatment, a tantalizing prospect in the age of gene therapy. This panel will be available for clinical samples at the Clinic for Special Children by Summer of 2019, and is aptly named the Plain Insight Panel.

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The research was conducted by a team including the study's first author Erin L. Crowgey from Nemours Biomedical Research, Nemours Alfred I. duPont Hospital for Children, Wilmington, DE; senior author Erik G. Puffenberger from the Clinic for Special Children, Strasburg, PA; Michael C. Washburn from ArcherDX, Inc., Boulder, CO and Anders Kolb from Nemours Biomedical Research, Nemours Alfred I. DuPont Hospital for Children, Wilmington, DE.

About the Clinic for Special Children
The Clinic for Special Children (CSC) is a non-profit organization located in Strasburg, PA, which provides primary pediatric care and advanced laboratory services to those who suffer from genetic or other complex medical disorders. Founded in 1989, the organization provides services to over 1,050 active patients and is recognized as a world-leader in translational and precision medicine. The organization is primarily supported through community fundraising events and donations. For more information, please visit www.ClinicforSpecialChildren.org