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Next Generation Genomic Diagnostics

What if there was a predictive diagnostic test that would help families prepare for the challenges of early childhood diseases and disorders? Early, precise diagnosis leads to better long-term outcomes and researchers at Cold Spring Harbor Laboratory (CSHL) are developing the next generation of genomic diagnostics.

Michael Ronemus, PhD

Drs. Michael Wigler, Michael Ronemus and colleagues at CSHL have been at the forefront of researching a segment of the human genome, copy number variants (CNV), and identifying their roles in human disease and disorders such as autism spectrum disorders and congenital heart disease. There are also studies showing the importance of CNV in cancer, schizophrenia, and other disorders where SMASH could potentially prove to be a useful diagnostic tool.

The researchers have developed technology for "Short Multiply Aggregated Sequence Homologies" ("SMASH"), which has the potential to provide a drastic improvement in detection of copy number variants (CNV) with much greater resolution than current detection methods at a much lower cost than currently available technologies. Dr. Ronemus' research into the application of the SMASH technology for CHD received funding from the Long Island Bioscience Hub (LIBH), one of three National Institutes of Health Research Evaluation and Commercialization Hubs (REACH), in 2015.

Derek Brand, MBA

An improved method of detection could provide geneticists with earlier information on the presence of causal mutations in newborn infants and allow for earlier interventional work in autism and earlier interventions in CHD. These disorders are often diagnosed much later in development, and this technology will enable appropriate therapeutic or behavioral action to be taken earlier in a child's life—when it can be of even greater efficacy in improving outcomes.

SMASH has also caught the eye of LIBH BioEntrepreneur-in Residence Derek Brand, a senior executive and entrepreneur with experience in multiple bio-medical startup companies. Together, Drs. Wigler, Ronemus and Mr. Brand co-founded Marvel Genomics, launched from Cold Spring Harbor Laboratory as a vehicle to commercialize the SMASH technology.

"This technology has the potential to provide earlier detection and greatly improved outcomes for autism spectrum disorders and congenital heart defects; it's an ideal fit with emerging use of next-gen sequencing technologies for early detection and will provide a true opportunity for improved outcomes in these areas, especially at a time where we are learning how to better work with children with autism spectrum disorders" said Mr. Brand. "We have been really fortunate to have terrific technology development support from the Long Island Bioscience Hub (NIH-REACH). Drs. Wigler and Ronemus from LIBH Partner Institution Cold Spring Harbor Laboratory (CSHL) are world recognized leaders in genomics and autism spectrum disorders, and CSHL Executive in Residence Andrew Whiteley serves as executive chairman, and brings a wealth of commercial and development experience to the table."

There is a provisional patent on the SMASH technique filed, and the company expects to partner with a genomics facility with CLIA/CLEP certification and the capacity to offer the tests at clinical scale. The goal is to have a commercially ready test for clinical uses in the second half of 2017. In addition to CHD and ASD, the SMASH technology has the potential to be applied in other research markets including cancer, schizophrenia and other neuro-psychiatric disorders.