FOR IMMEDIATE RELEASE

GOOD START GENETICS PUBLISHES VARIANT DATABASE VALIDATION STUDY

CAMBRIDGE, Mass., June 8, 2015 - Good Start Genetics®, Inc., a commercial-stage molecular genetics information company, today announced the publication of a study demonstrating its robust variant database validation process. In the paper, the authors confirm that Good Start Genetics’ next generation sequencing (NGS) represents a comprehensive, clinically proven method to screen entire genes, and is thereby able to detect a much larger set of sequence variants across ethnic groups. However, interpreting the large number of variants detectable using NGS requires a rigorous variant selection process for carrier screening that is not only highly sensitive, but also highly specific for pathogenic variants. The paper was published in Molecular Genetics & Genomic Medicine and can be accessed here.

“The clinical utility of NGS for genetic screening is its ability to detect a wider range of disease-causing genetic mutations with high sensitivity, but also with high specificity,” said Don Hardison, president and chief executive officer of Good Start Genetics. “This paper validates our rigorous methodology underlying the selection of genetic variants, and reinforces the power and accuracy of our NGS-based carrier screening approach.”

The variant selection approach illustrated in the publication used NGS-derived sequencing data from 22,864 pan-ethnic individuals being screened for Bloom syndrome, a disorder with an increased incidence among the Ashkenazi Jewish (AJ) population. Eleven pathogenic variants, and 16 novel variants previously not linked to Bloom syndrome but likely to be pathogenic were detected in the pan-ethnic population, in addition to the AJ-specific, ‘common’ variant.

“Our objective was to compile a comprehensive collection of variants for relevant genes in a database and then apply stringent classification criteria in order to retain only those variants with clear evidence for pathogenicity,” said Valerie Greger, Ph.D., Good Start Genetics’ director of genetic research.

More than 1,000 publications were reviewed by company geneticists to compile a comprehensive set of genetic variants linked to 15 commonly screened inherited diseases. Computer simulations were performed to assess NGS detectability. The variant selection approach validated in this study was designed by Good Start Genetics, and is routinely implemented to ensure that its variant panel for carrier screening is current, comprehensive and clinically relevant. Without rigorous validation, a laboratory could miss important pathogenic mutations and/or provide results of uncertain or unknown clinical utility.

“We’re very proud of the rigor that our scientific, development and clinical operations teams apply to processes such as this,” continued Mr. Hardison. “Good Start Genetics now has published in major, peer-reviewed medical journals studies concerning its analytical validation, its clinical validation and its variant database validation. It is this scientific rigor that has allowed
Good Start Genetics to set the standard in carrier screening, and become the first to offer NGS-based carrier screening in the clinical setting.”

About Good Start Genetics, Inc.
Good Start Genetics is a molecular genetics information company transforming the standard of care in reproductive medicine by providing clinicians and patients with clinically relevant and actionable information concerning inherited and other genetic disorders. Good Start Genetics’ suite of reproductive genetics products are designed to promote successful pregnancies through advanced technologies. The Company’s flagship genetic carrier screening service, GoodStart Select™, provides a comprehensive menu of tests for known and novel mutations that cause inherited genetic disorders. Good Start complements its proprietary next-generation DNA sequencing (NGS) capabilities at the core of GoodStart Select with other proven genetic screening technologies, as well as world-class customer care and genetic counseling. For more information, please visit www.goodstartgenetics.com.

###

CONTACT:
Katie Engleman
Pure Communications
910-509-3977
katie@purecommunicationsinc.com