Six Innovative Laboratories Win GenomeQuest Grants to Bring Next Generation Sequencing Diagnostics to the Clinic

Program Provides GenomeQuest Software, Services, and Expertise to Convert and Consolidate Sanger Gene Test Panels

WESTBOROUGH, Mass., Jan. 10, 2012 — GenomeQuest Inc., a global provider of large-scale genomic software applications, announced today that it has awarded six grants totaling more than $120,000 in software and services to help six laboratories facilitate the transition from multiple Sanger-based gene tests to consolidated next generation sequencing (NGS) based tests.

"As a clinician, I am intrigued by the potential benefits of NGS for the long-term and the practical benefits in the near-term," said Dr. Roberta Pagon, Professor, Department of Pediatrics, University of Washington, Principle Investigator of GeneTests and GeneReviews. "At this point it appears that the entry path to NGS for a number of labs listed in the GeneTests Laboratory Directory will be through consolidation of existing single gene testing into multi-gene panels — such panels will benefit many patients whose physicians are seeking to identify the genetic basis of an inherited disorder. The GenomeQuest Lab Grant program offers a complete NGS bioinformatics solution that promises to help labs advance to NGS and next generation testing services."

GenomeQuest will provide the software, annotation data, and infrastructure required to process and store NGS data, produce diagnostic reports, and perform follow-up research on individual and aggregate results. Recipients will use the resources to more quickly and cost-effectively perform a variety of diagnostics for rare genetic disorders, autism, cystic fibrosis, congenital heart disease, muscular dystrophy, and harmful bacterial pathogens.

"We look forward to collaborating with the six grant recipients who were selected from a competitive field of applicants based on their clear, actionable vision for utilizing NGS-based diagnostics in the clinic," said GenomeQuest CEO Richard Resnick. "Lab directors are recognizing the enormous opportunity to improve patient care by screening larger and larger regions of the genome while simultaneously lowering costs and turn-around times with next generation sequencing. This program supports GenomeQuest's commitment to empowering laboratories with the knowledgebase and interpretation tools they need to bring new NGS-based diagnostics to the clinic."

The grant recipients and areas of intended research include the following:

- **Cincinnati Children's Hospital Medical Center (Cincinnati, Ohio)** will use the grant to apply NGS diagnostics to diagnosing congenital heart disease, adult and pediatric cardiomyopathy, and cardiovascular disorders with genetic syndromes.

- **The Institute of Genomic Medicine at the University of Medicine & Dentistry of New Jersey (Newark, New Jersey)** initially plans to use the grant for cystic fibrosis gene testing. Future tests may focus on NGS testing for metabolic diseases.

- **Nationwide Children's Hospital (Columbus, Ohio)** is conducting tests for congenital muscular dystrophies and is interested in validating NGS testing for Noonan syndrome and related disorders, holoprosencephaly, a birth defect of the brain, and infertility.

- **The Molecular Diagnostics Laboratory at the University of Massachusetts Medical School/UMass Memorial Medical Center (Worcester, Mass.)** will use NGS for genetic testing of cystic fibrosis, Ashkenazi Jewish diseases, heart disease, lysosomal storage disorders, and dwarfism, as well as for the identification of bacterial pathogens.

- **The Department of Laboratory Medicine and Pathology of the University of Minnesota Medical School (Minneapolis, Minn.)** will apply NGS to eight areas of interest: pediatric bone marrow transplantation, congenital eye disorders, familial cancer syndromes, cardiology, congenital hearing loss, neurogenetics/neuromuscular disorders, genetic and metabolic pediatric disorders, and intersex disorders.

- **The University of Nebraska Medical Center (Omaha, Neb.)** plans to focus its testing on developmental disability/autism, connective tissue and cardiomyopathy, bone diseases, neurology, and amyotrophic lateral sclerosis (ALS).

"The application of NGS to interrogate many genes simultaneously provides a major advance in Clinical Diagnostic Testing that will benefit patients from a wide range of clinical services," said Dr. Edward I. Ginns, MD, PhD, Director of the Molecular Diagnostics Laboratory at UMass Memorial Medical Center. "Working with GenomeQuest on software for analysis of the massive amounts of data generated by NGS and on building an interface with our clinical laboratory information systems will further enhance the accuracy and timeliness of reporting results to providers," added Marzena Galdzicka, PhD, Associate Director of the Laboratory.

NGS-based diagnostics are informing treatment decisions today. As an example in the diagnosis of inherited deafness, the University of Iowa has consolidated over 20 Sanger-based gene-panels tests into a single NGS test. They report this advance will reduce the total cost of a molecular diagnosis from $75,000 to under $2,000 and the total time from one year to five weeks. More information about the program is available at: www.genomequest.com/2011/06/gq-dx-iowa/.

About GenomeQuest
GenomeQuest is a global provider and consistent pioneer in large-scale genomic software applications. Based on a whole/multi-genome engine and hosting platform, the company's products now include GQ-IP for global research and management of intellectual property on genomic sequences, GQ-Dx for decision-support and research in molecular diagnostics, and GQ-Ag for agricultural research. GenomeQuest serves major pharmaceutical companies, global agriculture firms, biotech firms, IP legal teams, genome centers, academic research centers, diagnostic labs, and universities around the world. Learn more at www.genomequest.com.

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