



## Quest Diagnostics to Present New Insights from Genetic Testing at the 2018 American Society of Human Genetics Annual Meeting

October 16, 2018

### Four Presentations Reveal Genetic Insights from Quest's Advanced Diagnostics

SECAUCUS, N.J., Oct. 16, 2018 /PRNewswire/ -- Quest Diagnostics (NYSE: DGX), the world's leading provider of diagnostic information services, announced today that it will present results of four studies at the 2018 American Society of Human Genetics Meeting (ASHG), October 16 – 20, 2018 in San Diego (Quest Diagnostics booth 1514).



These studies by Quest Diagnostics researchers evaluate a broad range of laboratory technologies and best practices for aiding in genomic sequencing of data for the diagnosis of patients with a broad range of genetic diseases and disorders. These include next generation sequencing (NGS) to detect alpha-thalassemia common deletions, which can affect the body's production of hemoglobin, causing anemia and other blood disorders. Other research describes best practices for identifying clinically relevant gene variants from sequenced genetic data.

Additionally, Quest utilized ClinVar data to demonstrate a scalable approach to classifying genes and genomic regions related to neurodegenerative disorders. ClinVar is a public gene database, and Quest is a participating laboratory.

"The science behind the treatment and management of genetic diseases is making leaps and bounds, but in order to deliver that care, you first need diagnostic insights that come from unlocking the genetics of the patient," Steven Keiles, MS, LCGC, Senior Director, Genomic Services, Quest Diagnostics. "Quest is at the forefront of these diagnostic innovations, and through our expertise, collaborations and scale, Quest is broadening access to advanced diagnostic services so more people everywhere can benefit."

Quest is a leader in advanced diagnostics, including in the areas of genetic and genomics. The company offers more than 700 genetic tests, such as BRCA and tumor sequencing to noninvasive prenatal screening. The company's 650 MDs and PhDs aid physician test selection and interpretation and publish hundreds of studies each year. Additionally, our genetic counselors are a resource for clinicians.

Abstracts can be accessed on the ASHG 2018 Annual Meeting website at: <http://www.ashg.org/2018meeting/pages/online-planner.shtml>

Among the Quest Diagnostics research being presented at this year's annual meeting are:

#### Thursday, October 18: 3 p.m. – 4 p.m.

- "Detection of alpha-thalassemia common large deletions by next generation sequencing" (Poster: 2860)
- "Scaling the resolution of sequence variant classification discrepancies in ClinVar" (Poster: 3046)

#### Friday, October 19: 3 p.m. – 4 p.m.

- "Evaluating dosage sensitivity of genes associated with neurodevelopmental disorders" (Poster: 2936)
- "A straightforward method to improve variant calling sensitivity for a MSH2 splicing mutation (c.942+3A>T) using GATK tools" (Poster: 1568)

#### About Quest Diagnostics

Quest Diagnostics empowers people to take action to improve health outcomes. Derived from the world's largest database of clinical lab results, our diagnostic insights reveal new avenues to identify and treat disease, inspire healthy behaviors and improve health care management. Quest annually serves one in three adult Americans and half the physicians and hospitals in the United States, and our 45,000 employees understand that, in the right hands and with the right context, our diagnostic insights can inspire actions that transform lives. [www.QuestDiagnostics.com](http://www.QuestDiagnostics.com).

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