Quest Diagnostics to Present New Insights from Genetic Testing at the 2019 Annual American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting

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Eight presentations highlight Quest's Advanced Diagnostics testing and research insights into hereditary genetics

SECAUCUS, N.J., March 26, 2019 /PRNewswire/ -- Quest Diagnostics (NYSE: DGX), the world's leading provider of diagnostic information services, announced today that it will present results of eight studies at the 2019 American College of Medical Genetics and Genomics (ACMG) Annual Clinical Genetics Meeting, to be held April 2 – 6, 2019 in Seattle, Wash. (Quest Diagnostics booth 929). These studies by Quest Diagnostics researchers evaluate a broad range of laboratory technologies and best practices for aiding in the evaluation of risk for cancer and other diseases.

Among the research is a study that uses a "virtual panel" to estimate genetic disease prevalence in ethnic populations; and a study of associations between certain colorectal cancer genes and risk for breast cancer.

"With so much attention on new therapeutics for cancer and other disorders, people sometimes forget the crucial role of diagnostic insights in guiding treatments to improve outcomes. Genetics and genomics have a greater impact on the treatment and management of cancer and other disorders than ever before," said Felicitas Lacbawan, MD, FCAP, FACMG, Vice President and Executive Medical Director, Advanced Diagnostics Quest Diagnostics. "Quest is delivering diagnostic innovations that provide these insights through its advanced diagnostic services so more people can benefit from new, innovative treatments personalized for them."

A study titled "Anticipated Positive Rates for Genetic Testing in the General Population: a 'Virtual Panel' Approach" (Poster 736) evaluates a method for estimating the rate of positivity for multi-gene testing panels in the general population and ethnic sub-populations. The study uses hereditary cancer genes as an example. The level of disease prevalence in a population influences test design and results interpretation. The Quest team developed a method to better predict disease prevalence for inherited cancer genes in ethnic groups, many of which have been traditionally under-represented in genetic testing research.

Another study, "Lynch Syndrome Families with Breast Cancer: Majority of Observed Mutations Are in *MSH6* and *PMS2*," (Poster 220) examined the clinical and family histories of individuals positive for a Lynch syndrome mutation. Lynch syndrome, historically known as hereditary non-polyposis colorectal cancer (HNPCC), is an inherited cancer syndrome associated with a predisposition to colorectal and other cancers. The researchers analyzed the frequency of breast cancer in this population to determine the relationship between Lynch syndrome genes and breast cancer and observed an association with the *MSH6* and *PMS2* genes. These data, which are in line with previous findings, may help refine breast cancer risk assessment in families with Lynch syndrome, guiding clinical surveillance and management.

Quest is a leader in advanced diagnostics, including the areas of genetic and genomics. The company offers more than 1000 genetic tests, including whole exome sequencing, germline and somatic gene sequencing, noninvasive prenatal screening, pharmacogenomics as well as cytogenetics and biochemical genetic testing. The company's 650 MDs and PhDs and genetic counselors aid physicians in test selection and interpretation and publish hundreds of studies each year. In Advanced Diagnostics – Genetics, Genomics and R&D alone, there are at least 75 PhDs, MDs and MD/PhDs with 60 board-certified scientific and medical directors.

Abstracts can be accessed on the ACMG website at: https://acmg.expoplanner.com/index.cfm?do=expomap.sessResults&search_type=abstracts&event_id=13

Among the Quest Diagnostics scientific and clinical work being presented at the meeting are:

- Double Heterozygotes and Carriers of Biallelic Pathogenic Variants Identified During Genetic Testing for Hereditary Cancer Risk at a Diagnostic Laboratory (Poster: 211)
- Lynch Syndrome Families with Breast Cancer: Majority of Observed Mutations Are in MSH6 and PMS2 (Poster 220)
- Preliminary Insight from CADASIL Testing: Using Aggregate Patient Data to Correlate Position of Variants in NOTCH3 to Severity of Disease (Poster: 228)
- Standards for the Classification and Reporting of Constitutional Copy Number Variants: A ClinGen/ACMG Joint Consensus Recommendation (Poster: 682)
- Looming False Negatives: Reconsidering the Clinical Significance of Synonymous Variants (Poster: 705)

- Anticipated Positive Rates for Genetic Testing in the General Population: A "Virtual Panel" Approach (Poster 736)
- Maternal Chimerism as a Cause of Confounded Gender Reporting and Gender Discordance During cfDNA Screening (Poster: 897)
- Confirmatory methods used for germline NGS Testing Teach Us About the Strengths and Weaknesses of Molecular Technologies (Poster: 8781)

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 46,000 employees understand that, in the right hands and with the right context, our diagnostic insights can inspire actions that transform lives. www.QuestDiagnostics.com.

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