NEWS RELEASE

Quest Diagnostics and Blueprint Genetics to Present New Insights from Genetic Testing at the 2021 Annual American College of Medical Genetics and Genomics Annual Clinical Genetics Meeting

4/13/2021

10 studies highlight Quest Diagnostics' commitment to specialty genetics testing and research in areas such as mitochondrial mtDNA analysis and next generation sequencing

Multiple studies focus on genetic disorders impacting children, who are more likely to achieve favorable outcomes when diagnosed and treated early

SECAUCUS, N.J. and HELSINKI, Finland, April 13, 2021 /PRNewswire/ -- Quest Diagnostics (NYSE: DGX), the world's leading provider of diagnostic information services, and Blueprint Genetics announced today that they will present results of 10 studies at the virtual 2021 American College of Medical Genetics and Genomics (ACMG) Annual Meeting, to be held April 13–16, 2021. These studies demonstrate the value of a broad range of genomic sequencing and other technologies to help diagnose several inherited disorders across various medical specialties.

In January 2020, Quest Diagnostics acquired Blueprint Genetics, a leading specialty genetic testing company with deep expertise in gene variant detection using next generation sequencing (NGS), proprietary bioinformatics, and clinical interpretation. Since that time, Quest and Blueprint Genetics have broadened access to actionable insights in genetic disorders and inherited diseases for patient care and anticipatory management as well as pharmaceutical drug research and development and clinical trials, particularly in the United States.

"Genomic testing is an essential component of patient care as results can impact treatment and management on many levels. Too often, patients experience a diagnostic odyssey, spending months, years or even a lifetime searching for a diagnosis because they lack access to genomic testing insights," said Carrie Eglinton Manner, Senior Vice President, Advanced Diagnostics, Quest Diagnostics. "Quest and Blueprint Genetics are working

together to bring innovative advanced diagnostics – from test ordering to gene variant interpretation and clinical reporting – to patient populations with unmet medical needs."

Featured studies focus on mitochondrial disease, hearing loss and skeletal dysplasias

Among the research is the study "Retrospective review of mitochondrial genome analysis in over 6600 cases using clinical grade mtDNA sequencing" (Poster: eP345), which demonstrates that including high-quality mitochondrial mtDNA analysis by next generation sequencing (NGS) in panels in multiple medical specialties increases the ability to make diagnoses for patients with mitochondrial disease. Mitochondrial disorders can be difficult to diagnose, as many of the symptoms, such as vision or hearing loss, seizures or poor muscle tone, can be mistaken for other diseases. While mitochondrial disorders have no cure, patients often do better when the underlying cause of their symptoms is diagnosed and addressed early.

"It's exciting to witness first-hand how mtDNA analysis increases diagnostic yields: Greater than a 1 percent increase in diagnostic yield, on average, across all panels, and a greater than 5 percent increase in multiple panels. The NGS-based technology we developed and extensively validated is specifically optimized for the detection of large mtDNA deletions and low levels of heteroplasmy. Mitochondrial disorders need to be considered in the diagnostic workflow for patients with suspected inherited disorders to provide more molecular diagnoses for all patients, not just those with complex presentations," said Jennifer Schleit, Blueprint Genetics Laboratory Director, North America.

Molecular genetic testing is now considered a standard part of the evaluation of hearing loss in infants. However, comprehensive genetic testing in hearing loss using standard NGS methods is complicated. A comprehensive testing strategy that includes difficult-to-sequence regions is needed for the most accurate diagnosis. A study titled "Next-generation sequencing panels for hereditary hearing loss testing with approaches for difficult-to-sequence regions" (Poster: eP345) demonstrates that the inclusion of difficult-to-sequence genes, such as STRC and OTOA, contributed to more than 10 percent of the diagnostic yield.

Another study, "Diagnostic utility of next-generation sequencing panel tests in the diagnosis of skeletal dysplasias" (Poster: eP346), found that NGS panels enabled diagnosis in 42 percent of patients. Skeletal dysplasias involve more than 450 heritable conditions that cause abnormalities of cartilage and bone, but diagnosis is challenging given significant overlap in symptoms. The analysis also demonstrated a diagnostic yield of 62 percent in prenatal cases, suggesting that testing in prenatal situations has significant clinical utility.

Abstracts can be accessed on the ACMG **website**.

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

- Diagnostic yield and clinical utility of genetic testing in children with seizure onset after 2 years of age: An update (Poster: OP086)
- Retrospective review of mitochondrial genome analysis in over 6600 cases using clinical grade mtDNA sequencing (Poster: eP345)
- Next-generation sequencing panels for hereditary hearing loss testing With approaches for difficult-to-sequence regions (Poster: eP345)
- Biallelic NRAP variants are a significant cause of dilated cardiomyopathy (Poster: eP343)
- Diagnostic utility of next-generation sequencing panel tests in the diagnosis of skeletal dysplasias (Poster: eP346)
- Novel BCL11A variant Arg3Cys identified in male with intellectual disability and persistence of fetal Hb (Poster: eP138)
- A web-based educational program to support the updated ACMG/ClinGen technical standards for constitutional copy number variant classification (Poster: eP366)
- Expanding the mutational spectrum for branchiooculofacial syndrome: a novel nonsense variant (Poster: eP415)
- Arginine to ornithine ratio as a diagnostic marker in patients with positive newborn screening for hyperargininemia (Poster: eP012)
- Retrospective review of genetic testing for inherited bone marrow failure syndromes (eP363)

Quest Diagnostics and Blueprint Genetics are improving patient outcomes through high-quality genomic testing. Quest Diagnostics is the leader in advanced diagnostics, including in genetics and genomics. The company offers more than 1,000 genetic tests, including whole exome sequencing, germline and somatic gene sequencing, noninvasive prenatal screening, pharmacogenomics as well as cytogenetics and biochemical genetic testing. With a global customer base in over 70 countries, Blueprint Genetics brings specialty genetics knowledge in sequencing and bioinformatics and variant interpretation and reporting to Quest, which complements and extends its existing genetics leadership. Quest Diagnostics' 600 MDs and PhDs and genetic counselors aid physicians in test selection and interpretation and publish hundreds of studies each year.

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 Diagnostics annually serves one in three adult Americans and half the physicians and hospitals in the United States, and our nearly 50,000 employees understand that, in the right hands and with the right context, our diagnostic insights can inspire actions that transform lives. **www.QuestDiagnostics.com**.

About Blueprint Genetics

Blueprint Genetics, a Quest Diagnostics company, is a leading specialty genetics and bioinformatics company focused on providing genetic testing for inherited diseases. The company is based in Helsinki and Seattle, with a customer base spanning over 70 countries. **www.blueprintgenetics.com**

View original content to download multimedia:**http://www.prnewswire.com/news-releases/quest-diagnostics-and-blueprint-genetics-to-present-new-insights-from-genetic-testing-at-the-2021-annual-american-college-of-medical-genetics-and-genomics-annual-clinical-genetics-meeting-301266902.html**

SOURCE Quest Diagnostics