MediaRoom

New Comprehensive Carrier Screening Research Assay Features Richest Pan-Ethnic Panel for Detection of Multiple Genetic Variations in Inherited Disease

CarrierScan™ Assay from Thermo Fisher Scientific consolidates conventional multi-assay approach for “all-in-one” research

Dateline:

CARLSBAD, Calif. & PHOENIX

Tuesday, March 21, 2017 8:00 am EDT

CARLSBAD, Calif. & PHOENIX--A new high-throughput, microarray-based assay designed to detect more than 6,000 genomic variations associated with inherited disease now provides molecular genetics laboratories with a consolidated and automated, single pan-ethnic solution for expanded carrier status research. Thermo Fisher Scientific announced the CarrierScan™ Assay today at the 2017 ACMG Annual Clinical Genetics Meeting and will make it available in April.

According to the American College of Obstetricians and Gynecologists (ACOG), carrier screening was traditionally targeted toward specific populations known to have an increased risk of particular disorders. However, the growing ethnic diversity among global populations, coupled with advances in genetic research technologies, has led ACOG to update its screening recommendations. Published in the March 2017 issue of *Obstetrics & Gynecology*, the new guidance calls for a standard approach that could be consistently offered and discussed during family planning regardless of ethnicity.

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“The recently updated recommendation from ACOG to offer a consistent approach for carrier screening speaks to the growing need for genetic information that could be used for family planning in the future, particularly for those with elevated risk of recessive mutations carrier status,” said Doron M. Behar, M.D., Ph.D., CSO of Gene by Gene. “Launch of CarrierScan will be an important advance for our lab’s clinical research. It’s optimized chemistry and coverage, plus consolidated format that is truly comprehensive for pan-ethnic screening research, can’t be matched.”

Traditionally, many laboratories are forced to use multiple assays and platforms, perhaps even outsource work, to capture complete carrier research data across ethnicities. This is taxing to resources, which is driving a need today to streamline this research through consolidation of assays onto a single solution.

CarrierScan Assay meets this need as the market’s first comprehensive, pan-ethnic research solution that enables laboratories to assess genomic variation associated with more than 600 genes involved in inherited diseases, such as cystic fibrosis and thalassemia. In all, it is designed to detect more than 6,000 mutations, including bi-allelic and multi-allelic single nucleotide variants (SNVs), in-dels and copy number variations (CNVs) in challenging regions such as highly orthologous genes and pseudogenes.

The ability to integrate expanded carrier research in a laboratory setting with a single, automated microarray platform, instead of outsourcing it, can help molecular research laboratories reduce time to results and remain competitive. The 600-gene content included in CarrierScan Assay is based upon empirical selection of probes and biological verification of the most common variants. This design and verification process enables reproducibility and confidence in results of broad screening research.

The assay is run on the Applied Biosystems GeneTitan Multi-Channel (MC) instrument for fully automated, high-throughput array processing. Laboratories have the choice of manual or automated sample preparation for complete flexibility to meet specific productivity needs. To optimize population-specific applications, the assay is customizable for the particular genetic diversity that is being studied. The CarrierScan Reporter software for rapid and simple analysis and annotation is also available at no additional fee.

“CarrierScan Assay represents our commitment to enable our clinical research and molecular laboratory customers stay competitive and on the cutting edge of expanded carrier research,” said Laurent Bellon, Ph.D. vice president and general manager of microarrays at Thermo Fisher Scientific. “This assay offers researchers access to a consolidated expanded carrier research analysis tool that will enable them to meet the demands of the rapidly growing reproductive health market.”

For more information on the CarrierScan Assay, visit Thermo Fisher's booth (#1009) at ACMG Annual Clinical Genetics Meeting March 22-24, or visit www.thermofisher.com/carrierscan.

CarrierScan Assay is for research use only; not for use in diagnostic procedures.

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