MediaRoom

Thermo Fisher Scientific Customers to Showcase Innovations in Precision Genomics Research for Inherited Disease and Reproductive Health at ASHG

AmpliSeq On Demand NGS Panels nearly tripled to 3,000 genes for investigating inherited disease-causing variants

CARLSBAD, Calif. and ORLANDO, Fla., Oct. 17, 2017 /PRNewswire/ -- Inherited diseases are genetically passed down from parent to child through abnormalities in the genome or DNA sequence. These variants can be caused by a single-gene mutation or a host of complex aberrations that combine with environmental factors. At the American Society of Human Genetics (ASHG) Annual Meeting October 17-21, 2017, Thermo Fisher Scientific will host a lunch seminar that highlights the work of several investigators that is being enabled by new technologies the company is introducing at ASHG to advance precision genomics for research in inherited disease and reproductive health.

Dr. Pan Zhang, Director of the Sequencing and Microarray Center at the Coriell Institute for Medical Research, and her research team focus on identifying biomarkers associated with adult-onset hearing loss, a condition that affects about 15 percent of Americans (37.5 million), according to the National Institute on Deafness and Other Communication Disorders (NIDCD)[1]. During her presentation, Dr. Zhang will demonstrate how Thermo Fisher's newly expanded Ion AmpliSeq On-Demand Panels for targeted sequencing, in combination with Ion Torrent next-generation sequencing (NGS) technology, is playing a key role in elucidating novel genetic underpinnings of hearing loss. The Coriell Personalized Medicine Collaborative (CPMC) research study, which has been ongoing for the last 10 years, is part of the United States Air Force Personalized Medicine Program.

"Genome-wide associated studies have not proven fruitful in hearing loss research. For this poorly understood genetic condition, you need a discovery tool that is both rapid and cost-effective for analyzing multiple genes in depth in many samples," says Dr. Zhang. "By combining the medical history, family history and lifestyle data we've collected over the last decade with the genetic information we're getting with the Ion AmpliSeq On-Demand NGS panels, we are now able to more efficiently identify novel genetic factors that are linked to adult onset hearing loss."

The lunch seminar (12:30-1:45 pm, Oct. 19 - Hilton Orlando, Lake Mizell Room) will also feature presentations that highlight clinical research in reproductive health using complementary platforms to NGS, including data from one of the first studies using the Applied Biosystems SeqStudioGenetic Analyzer, the new capillary electrophoresis (CE) system that offers a low-throughput, cartridge-based system for Sanger sequencing and fragment analysis, and Thermo Fisher's CytoScan XON Suite for exonic deletions and duplications analysis.

An early access customer, Dr. Alka Chaubey, Director of the Cytogenetics Laboratory at the Greenwood Genetic Center, has provided valuable early feedback after running more than 50 samples on the CytoScan XON Suite and says the assay will serve as a critical tool to efficiently move her clinical research forward.

"The initial data that we have seen is just phenomenal. There is such a beautiful tiered-level approach for the analysis of genes, and the way the aberration calls show up is extremely user friendly for us," Dr. Chaubey says. "The biggest hurdle today is that there is nothing on the market that will give us comprehensive coverage of the deletions and duplications [at the exome level] we are analyzing. But with CytoScan XON Suite, we can be confident about the genes we are interrogating and have that data available to us in less than five days from sample to analyzed data."

More than 30 Mendelian disorders, including both neurodevelopmental and neurodegenerative diseases, are known to be associated with expansions of repetitive DNA sequences. Recent genome-wide studies have also linked thousands of previously uncharacterized DNA repeat elements to changes in gene expression. As a result, there is a rapidly growing interest and appreciation of repetitive "dark" DNA, which is often difficult to analyze.

To this end, Dr. Gary J. Latham, Sr. Vice President, Research and Development at Asuragen, will present his work investigating DNA repeat expansions in inherited disease and reproductive health using the Applied Biosystems SeqStudio Genetic Analyzer. In combining with the SeqStudio Genetic Analyzer and Asuragen's AmplideX PCR technology, he will describe preliminary data for the sensitive, accurate and streamlined analyses of GC- and AT-rich repeat polymorphisms associated with Fragile X Syndrome, ALS/FTD, Myotonic Dystrophy, Huntington's Disease and Alzheimer Disease.

For a full list of customer presentations or to register for the seminar, visit the Thermo Fisher ASHG 2017 website. For more information on the wide range of genetic analysis platforms in the Thermo Fisher portfolio, stop by the company's booth (#631) during the conference.

1. Blackwell DL, Lucas JW, Clarke TC. Summary health statistics for U.S. adults: National Health Interview Survey, 2012 (PDF). National Center for Health Statistics. Vital Health Stat 10(260). 2014.

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