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Thermo Fisher unveils new NGS based solution

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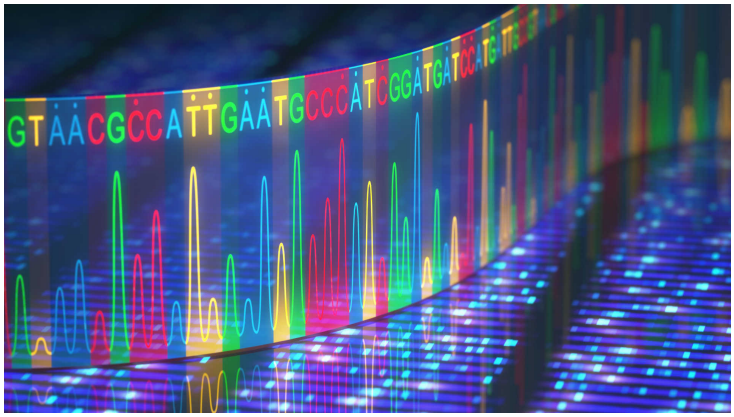
Ion Torrent CarrierSeq ECS Kit consolidates multi-testing approach into a single, end-to-end workflow to facilitate implementation globally



Thermo Fisher Scientific has introduced a new next-generation sequencing (NGS)-based solution that enables reproductive health researchers to more efficiently analyze a broad range of key genetic markers with an end-to-end workflow. The Ion Torrent CarrierSeq ECS Kit for the Ion GeneStudio S5 System consolidates a multi-platform approach to expanded carrier screening (ECS) into a single solution that extends the company's reproductive health research portfolio, which includes tools for prenatal, postnatal, invitro fertilization and newborn screening analysis.

The CarrierSeq ECS Kit consists of a 420-gene, AmpliSeq-based panel that combines many stand-alone tests into a single assay, including difficult-to-sequence genes, such as SMN1 and SMN2 for spinal muscular atrophy, GBA for Gaucher disease, CYP21A2 for 21-hydroxylase deficient congenital adrenal hyperplasia, and HBA1 and HBA2 for alpha thalassemia. The kit detects more than 28,000 non-benign ClinVar variants, including both single nucleotide variants (SNVs) and copy number variants (CNVs), and contains optimized reagents and a customizable Carrier Reporter Software for data analysis and reporting.

"In the past, we've typically used many different platforms to cover the plethora of tests, from NGS and qPCR to capillary electrophoresis. With CarrierSeq ECS kit, we can consolidate many of the tests which really improves our lab efficiency," said Luis A. Alcaraz, Ph.D., scientific and lab director at Bioarray, Spain, an early access customer. "In addition, because Carrier Reporter Software is intuitive to use, we can confidently perform various analyses and customize our reporting quickly."

The global carrier screening market is estimated to reach $2.7 billion by 2024 at a CAGR of 19.6 percent, according to an October 2019 report by *MarketsandMarkets*. Advancements in NGS is making ECS more efficient, economical, and an accessible method to screen for a large number of heritable genetic conditions during family planning. However, the high development cost to overcome lack of bioinformatics and assay development expertise has presented implementation challenges for many reproductive health researchers in the past.

"We designed the CarrierSeq ECS Kit by tapping into our deep R&D expertise in panel design, algorithm development, assay optimization, and software development," said Yan Zhang, vice president and general manager of reproductive health at Thermo Fisher. "The end result is a straightforward, ready-to-use solution that our customers can confidently implement in their lab. The addition of this kit underscores our mission to enable our customers to make the world healthier."