Targeted panel testing superior for neuromuscular diseases

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(HealthDay)—Targeted panel testing has the highest clinical yield for molecular diagnosis of neuromuscular diseases (NMDs), according to a study published in the February issue of the *Annals of Neurology*.

Arunkanth Ankala, Ph.D., from the Emory University School of Medicine in Atlanta, and colleagues designed and validated several next generation sequencing (NGS)-based comprehensive gene panel tests. The validation established that the targeted gene panel tests have 100 percent sensitivity and specificity for detection of single nucleotide variants. The clinical diagnostic yields of single gene tests were compared with NMD NGS panels using data from all clinical tests performed at the Emory Genetic Laboratory. The clinical utility of the targeted NGS panel test was also compared with that of exome

sequencing.

The researchers found that the diagnostic yield was three-fold higher for the NMD comprehensive panel testing than for single gene testing (46 versus 15 to 19 percent). Panel testing was complemented by Sanger fill-in of low coverage exons, copy number variation analysis, and thorough in-house validation of the assay, allowing detection of all types of causative pathogenic variants, of which about 18 percent may be missed by exome sequencing.

"Our results strongly indicate that for molecular diagnosis of heterogeneous disorders such as NMDs, targeted panel testing has the highest clinical yield and should therefore be the preferred first-tier approach," the authors write.

More information: Abstract
Full Text (subscription or payment may be required)