

Targeted panel testing superior for neuromuscular diseases

February 13 2015



(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 <u>exome</u>



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 fillin 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 <u>molecular diagnosis</u> 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)

Copyright © 2015 HealthDay. All rights reserved.

Citation: Targeted panel testing superior for neuromuscular diseases (2015, February 13) retrieved 5 May 2024 from https://medicalxpress.com/news/2015-02-panel-superior-neuromuscular-diseases.html

This document is subject to copyright. Apart from any fair dealing for the purpose of private study or research, no part may be reproduced without the written permission. The content is provided for information purposes only.