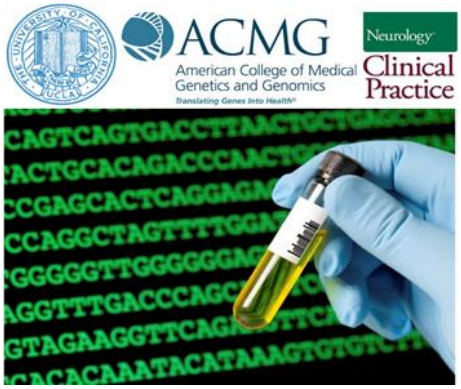
Genetic Science Spotlight

Whole-Exome Sequencing Improves Clinical Diagnosis



Whole-exome sequencing (WES) is increasingly being used in clinical diagnostics for a variety of indications to identify the underlying genetic cause of disease, and as costs continue to decline it may become a routine part of health care. Early applications of the technology include solving genetically heterogeneous disorders, such as complex neurologic diagnoses and multiple congenital anomalies, offering hope to millions of individuals with previously undiagnosed or untreated rare disorders. WES is a laboratory process that determines, all at once, the order of all of the expressing genes in a person's genome. Researchers from UCLA carried out a study to investigate how WES could serve as a viable diagnostic approach for identifying rare inherited neurogenetic disorders. Their study, published in Neurology Clinical Practice, found that 20% of a group of individuals with spinocerebellar ataxia could be diagnosed promptly using WES, and useful genetic information could be identified in over 60% of the affected individuals, regardless of their age when the disease began or their family history. WES has not only aided in the diagnosis of rare disorders and solved diagnostic dilemmas, it has also been used as a method of gene discovery in large series of patients with autism, epilepsy, brain malformations, congenital heart disease, and neurodevelopmental disabilities, and it has effectively identified many novel disease genes and pathways. The growing body of evidence supporting the use of WES, and the demonstrated benefits to patients on a diagnostic odyssey, indicates that WES will soon be widely adopted in all routine clinical practice globally.

http://www.nature.com/gim/journal/vaop/ncurrent/full/gim2015148a.html

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