Study concludes insurers should provide better coverage for cutting-edge genetic test

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UCLA researchers have found that a state-of-the-art molecular genetic test greatly improves the speed and accuracy with which they can diagnose neurogenetic disorders in children and adults. The discovery could lead directly to better care for people with rare diseases like spinocerebellar ataxia, leukodsystrophy, spastic paraplegia and many

other conditions.

The test, called exome sequencing, involves determining the order of all of the genes in a person's genome. When used in concert with a complete patient evaluation and family medical history, the approach can help doctors identify disorders that may have gone undiagnosed for years, said Dr. Brent Fogel, first author of a review that appears in the April issue of *Neurology Clinical Practice*.

Exome sequencing is more efficient and less costly than the type of genetic testing that has been more commonly used, Fogel said—and a proper diagnosis can end what for many patients is an agonizing journey just to find a name for their conditions.

The growing body of evidence supporting the use of the test, and the demonstrated benefits to patients, should lead to greater insurance coverage of the test, said Fogel, who is director of the UCLA Neurogenetics Clinic and an associate professor of neurology and human genetics.

"Despite extensive literature supporting the use of this technology, many insurance companies still consider it to be investigational and may refuse coverage," he said. "Our article outlines the appropriate use, benefits and limitations of exome sequencing that these companies need to consider when making coverage decisions."

UCLA has been a leader in using the test as a diagnostic tool since 2012. Fogel and his team were among the first to adopt the technology for routine neurological practice, and he has been a strong advocate for wider use.

Fogel and colleagues wrote a 2014 study about exome sequencing that was published in the *Journal of the American Medical Association*

Neurology. That research found that 20 percent of a group of people with spinocerebellar ataxia could be diagnosed immediately using the technique, and useful genetic information could be identified in more than 60 percent of the subjects, regardless of their age when the disease began or their family history.

More information: B. L. Fogel et al. Clinical exome sequencing in neurologic disease, *Neurology: Clinical Practice* (2016). DOI: 10.1212/CPJ.0000000000000239

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