Medical ethics experts outline strategy for overcoming reimbursement barriers for clinical genome sequencing tests

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Genomic tests using next generation sequencing technologies are increasingly being offered in a range of clinical settings, but these tests may only transform clinical practice if patients and clinicians have access to them, said medical ethics experts from Baylor College of Medicine, Johns Hopkins University and the University of North Carolina at Chapel Hill in a commentary published today in the *Journal of the American Medical Association*.

In the commentary, authored by Dr. Amy McGuire, director of the Center for Medical Ethics & Health Policy at Baylor, Dr. Patricia Deverka, adjunct research associate professor at UNC Chapel Hill and Dr. David Kaufman, director of research at The Genetics & Public Policy Center at Johns Hopkins, the experts outline a strategy to help public and private payers evaluate and understand this emerging technology when they are deciding whether to pay for such testing.

"We are starting to see large studies about the promise of these technologies to diagnose rare genetic disorders," said McGuire, who specializes in ethical issues in genomics and genomic tests.

Yet, the current reimbursement environment is not structured to necessarily recognize the value of next generation sequencing tests, the authors wrote.

Payers base coverage decisions for genomic tests on an evaluation of the clinical validity (accuracy with which a test can predict the presence or absence of a disorder or disease), as well as evidence of clinical utility (whether the test affects treatment decisions and patient health outcomes), they said.

Some technologies, such as genetic testing of fetal blood cells found in the circulation of pregnant women, have rapidly generated this evidence and are widely covered by private insurers. However, other technologies such as whole exome sequencing (which looks at the protein coding region of the genome) for the diagnosis of suspected rare genetic disorders, has yet to generate evidence that is as clear-cut and thus, payment is less assured, they noted.

For example, the experts said that at one clinical laboratory, approximately 49 percent of whole exome tests ordered at one academic clinical laboratory could not be performed because of denial of coverage. A majority of these claims were denied because whole exome sequencing is considered experimental or investigational.

The biggest issue, the authors noted, can be attributed to the clinical utility evidence "gap," a previously described problem for molecular diagnostic tests that look for single gene mutations. This issue becomes even more complicated for tests that look at millions of variations at the same time.

To help close this gap and ensure access to tests that are likely to benefit patients, while avoiding the clinical and economic harms of potentially ineffective tests, the authors suggested a four-pronged approach:

- Test developers should invest in robust validation studies to determine analytic and clinical validity. These reports should be made readily available to payers and technology assessment

groups.

- Establish a system for prioritizing research to assess clinical utility and the quality of existing evidence and give priority to tests that have demonstrated analytical and clinical validity, some existing evidence of clinical utility and for which it is feasible to conduct additional utility studies. The authors used an example of a next generation sequencing test, which recently received U.S. Food and Drug Administration approval for targeted sequencing for cystic fibrosis and is being evaluated for utility as a companion diagnostic in cancer.
- Use existing evidentiary frameworks, such as those recommended by technology assessment groups and large payers, for assessing clinical utility as a starting point for designing clinical utility studies.
- Evolve the evidence review process to account for the full range of benefits that are theoretically possible with next generation based sequencing, which the authors characterize as "compound utility." Compound utility includes the concepts of personal utility (value of information to the patient and/or family beyond its intended clinical use), as well as the unique potential value of this type of testing that is likely to occur over the long term.

Adopting this approach could help increase the quantity and quality of information payers need to make evidence-based coverage decisions. The goal is to help ensure that payment decisions for next generation sequencing tests are based on the evidence that best supports their clinical use and value to the health care system, the authors concluded.

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