

PROFESSION

AMA meeting: Standards sought as clinical use of gene sequencing speeds up

Diagnostic mysteries are being solved using genomic-based technologies. Overcoming barriers to clinical implementation is the next step, delegates say.

By **KEVIN B. O'REILLY**, amednews staff. *Posted Nov. 26, 2012.*

Honolulu Appropriate professional standards and regulation will help ensure that recent advances made in the use of genomic-based technologies benefit more patients, according to policies adopted at the American Medical Association Interim Meeting.

In its action, the AMA House of Delegates recognized the utility of these genomic technologies and encouraged the development of standards to guide clinical use as well as best practices for the laboratories performing such tests. The AMA also will support regulatory and payment policies to enable doctors to use these diagnostic tools when clinically appropriate, while protecting patient rights such as confidentiality and freedom from genetic discrimination. It took more than 10 years and cost \$2.7 billion for the Human Genome Project to sequence the entire human genome. Next-generation sequencing of the human genome, known as NGS, has sped the process dramatically, said the AMA Council on Science and Public Health report that the house adopted. An entire individual genome can be sequenced within two to three days for less than \$5,000, with the cost expected to fall to less than \$1,000 in the near future. The amount of data in that sequencing would occupy more than 400,000 pages when printed, with as many as 50,000 clinically important gene variants requiring examination using specialized clinical software. Incidental findings could require hours of genetic counseling for patients and families, the AMA report said.

Improved health outcomes seen

"NGS-based technologies have the potential to drive significant improvements in patient care," said Sandra A. Fryhofer, MD, chair of the science council. "Already, these technologies have shown remarkable ability to end the diagnostic odyssey for patients with disorders that are resistant to standard diagnostic procedures and targeted genetic testing.

"Cancer patients also stand to gain from improved molecular analysis that enables accurate tumor classification, and improved diagnosis and management options," she said. "But challenges such as managing extremely large data sets, return of results, and regulation and reimbursement must be addressed in order to fully realize the incredible potential of NGS-based technologies to improve health outcomes."

Genomic-based technologies such as whole-genome sequencing and whole-exome sequencing can identify disease-causing gene mutations and will be used increasingly in diagnosis and treatment, the science council's report said.

For example, doctors using whole-exome sequencing were able to arrive at a definitive diagnosis for a boy with a life-threatening form of inflammatory bowel disease. After the mutation involved was identified, a cell transplant was performed that relieved the boy's symptoms and enabled him to eat and drink again. Studies have provided evidence that these genomic-based technologies should be considered in cases when a genetic condition is suspected but targeted genetic testing has proven negative.

"Advances in DNA sequencing technology offer patients and physicians a new diagnostic tool to help improve health outcomes, but clinical challenges must be addressed to realize the full potential benefits of this technology," said Robert M. Wah, MD, a McLean, Va., reproductive endocrinologist and immediate past chair of the AMA Board of Trustees. "While cost and time barriers to genetic sequencing have been reduced, important privacy, practice, payment and regulatory issues — including how to compensate physicians and other health care professionals for the considerable work-related demands required — must still be addressed."

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RESOURCES

- Our coverage of the meeting
- Central meeting archive [🔒](#)
- AMA official proceedings

Sandra A. Fryhofer, MD, chair of the AMA science council, sees much potential in NGS-based technology.
[Photo by Ted Grudzinski / AMA]



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