

POPULATION HEALTH FORUMS

New Developments in Genetic/Genomic Testing: Implications for Population Health

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Vivian Coates, MBA, is Vice President of Information Services and Health Technology Assessment at [ECRI Institute](#) where she develops and leads the evidence-based medicine and health technology assessment program, including the Evidence-based Practice Center (EPC) and the Health Technology Assessment Information Service (HTAIS) for health plans, hospitals/health systems and health policymakers. Ms. Coates' most recent initiative is the development of ECRI's personalized medicine resource on genetic/genomic testing, [ECRIgene](#). This interactive database includes critical information on hundreds of genetic, genomic and proteomic tests meeting ECRI's criteria for inclusion.

ECRI Institute is a nonprofit health services research organization with a history of over 47 years of laboratory-based medical device evaluations, and 25 years of conducting health technology assessment, forecasting and comparative-effectiveness research (CER). ECRI has a special relationship with the Agency for Healthcare Research and Quality (AHRQ), where they have maintained their role as an evidence-based practice center since 1997. ECRI has also created the National Guideline Clearing House and National Quality Measures clearing house for AHRQ.

Ms. Coates' presentation focused on the human genome. A genetic test analyzes a single gene, while a genomic test analyzes an entire or large portion of the genome. These tests involve analysis of human

chromosomes, DNA, RNA, genes and/or gene products predominately used to detect heritable or somatic mutations and genotypes related to disease and health. Ms. Coates explained that genetic/genomic tests are more widely available due to less expensive, quicker, and improved technologies, and advent of the [Human Genome Project](#).

Personalized medicine, explained Coates, is not a new concept. It's a way of customizing treatment for individual patients. It's an area that has evolved during the past few decades due to the advance in genetic science and technologies. Genetic testing can provide crucial information to accurately predict risk of developing disease, disease progression, and response to treatment. The effectiveness of personalized medicine really depends on how well clinicians understand each person's unique characteristics.

Coates described ways in which genetic tests can pose challenges related to costs, practice, and policy. There are concerns regarding: the increasing complexity of multigene test panels and underlying platforms in the face of huge gaps in evidence; aggressive direct-to-consumer and provider marketing by the labs; ordering, interpretation, patient counseling, and a shortage of genetic counselors; and intensive time and resource requirements. Many tests serve different purposes, from carrier screening to monitoring to risk assessment.

Adding to the challenges are issues of regulation and reimbursement, with a plethora of federal and state certifications and pathways and varying levels of rigorous evaluation. Coates emphasized that the FDA is very concerned about genetic tests and may increase their oversight of Laboratory

Developed Tests (LDTs). It has been found that some LDTs have high false negative and/or false positive results, inflated claims of accuracy, and weak clinical validity.

Coates went on to describe the challenges for payers. Lack of evidence showing clinical utility of a test creates a major barrier for insurance reimbursement. Sources for informing coverage decisions have limitations and are not available for all genetic tests. Although payers such as Medicare may be influential, their decisions may not translate well to genetic tests for those under the age of 65.

The most important domains that ECRI examines to assess genetic tests include: analytic validity; clinical validity; and clinical utility. Coates explained that poor analytic validity will often compromise clinical validity and clinical utility, therefore efforts are focused on evidence for clinical validity and utility. She referred to a "chain of evidence," which includes some of the following issues: whether or not the test detects the genetic variant accurately and reliably; whether the test detect the disease accurately; and whether the treatment will lead to improved health outcomes.

Coates ended her presentation by discussing the impact of genetic testing on population health. She pointed out that, in many instances, precision medicine has caused a paradigm shift in treatment and helped to achieve superior health outcomes. There are, however, challenges in development of a comprehensive genetic/genomic testing approach for population-based care. Gaps in evidence do exist and the underlying science and data analytics must continue to improve.