Applying genomic medicine into clinical practice for chronic diseases still in the early stages

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A large gap exists between what knowledge is available about genomic medicine and incorporating it into clinical practice for assessing the risk of and treating common chronic diseases, such as cardiovascular disease, diabetes mellitus, and cancer, according to a systematic review in the March 19 issue of JAMA, a theme issue on *Genetics and Genomics*.

Maren T. Scheuner, M.D., M.P.H., of the RAND Corporation, Santa Monica, Calif., presented the findings of the study at a JAMA media briefing at the National Press Club in Washington, D.C.

“The greatest public health benefit of advances in understanding the human genome will likely occur as genomic medicine expands from its focus from rare genetic disorders to inclusion of more common chronic diseases, such as coronary heart disease, stroke, diabetes mellitus, and cancer,” the authors provide as background information in the article. “With genomics discoveries relating to common chronic diseases, numerous genetic tests may emerge that hold promise for significant changes in the delivery of health care, particularly in preventive medicine and in tailoring drug treatment.”

Dr. Scheuner and colleagues analyzed the medical literature for research articles and systematic reviews published between Jan. 2000 and Feb. 2008 dealing with common chronic adult-onset conditions. The authors included 68 articles in the analysis and assessed four key areas: outcomes

of genomic medicine, consumer information needs, delivery of genomic medicine, and challenges and barriers to integration of genomic medicine.

“Generally there were modest positive effects on psychological outcomes such as worry and anxiety, behavioral outcomes have shown mixed results, and clinical outcomes were less well studied,” the authors report. “The most important and consistent finding from our literature review is that the primary care workforce, which will be required to be on the front lines of the integration of genomics into the regular practice of medicine, feels woefully underprepared to do so.”

The authors note that consumers are unsure about the value of genetic testing and have concerns about privacy issues and discrimination in health insurance and employment. However, the consumers were interested in the technology to help better identify diseases for which they and their family members are at increased risk.

The analysis identified the need to better understand the outcomes of genomic medicine interventions. “More research describing clinical outcomes is needed: do patients who receive counseling and testing have better clinical outcomes in terms of mortality, decreases in incidence of disease, and better clinical responses to pharmaceuticals? And at what cost?”

Other barriers to the clinical integration of genomic medicine for common chronic diseases were identified by these authors in addition to the perceived inadequacy of the primary care workforce. “The most prominent of these include health professionals’ lack of basic knowledge about genetics and their lack of confidence in interpreting familial patterns of disease, which limits their ability to appropriately counsel their patients, order and accurately interpret genetic tests, and refer their patients for genetics consultation.”