

ACMG NEWS

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The Expanded Carrier Screening Joint Statement will be published ahead of print in Current Commentary in [Obstetrics & Gynecology](#). Email Communications@acog.org for the full commentary.

ACMG NEWS

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Expanded Carrier Screening in Reproductive Medicine: New Joint Statement is Released in ACOG's *Obstetrics & Gynecology*

Bethesda, MD – Feb. 5, 2015 -- Carrier screening for inherited genetic disorders is an important part of preconception and prenatal care for the nearly 4 million women who give birth in the US annually. Carrier screening is meant to identify couples at risk for passing on such genetic conditions to their children. While there have been limitations to this approach in the past, new technology in genotyping and genetic sequencing allows for more efficient carrier screening of a greater number of conditions simultaneously.

In an important new statement, several of the nation's leading medical societies have collaborated to provide guidance on such advances and their use in reproductive medicine. The American College of Medical Genetics and Genomics (ACMG) along with the American College of Obstetricians and Gynecologists, the National Society of Genetic Counselors, the Society for Maternal-Fetal Medicine and Perinatal Quality Foundation have just released a new Joint Statement on "Expanded Carrier Screening in Reproductive Medicine - Points to Consider" published online ahead of print in *Obstetrics & Gynecology* ("the Green Journal") in Current Commentary at <http://journals.lww.com/greenjournal/toc/publishahead>.

Anthony R. Gregg, MD, FACOG, FACMG, vice-president, Clinical Genetics of the American College of Medical Genetics and Genomics and a co-author of the Joint Statement said, "This document is a

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sort of a blueprint of expanded carrier screening in clinical practice. It serves obstetric care providers by helping them navigate pretest information to share with patients and concepts applicable to posttest follow-up. Importantly, pitfalls surrounding expanded carrier screening are described. Readers will recognize that this document does not advocate for or against the universal implementation of expanded carrier screening. There is a paucity of scientifically sound information to guide professional organizations in taking a firm stance. For now currently available practice guidelines (summarized in the joint document) authored by ACMG and ACOG prevail and these represent a minimum screening standard. Professional organizations may, at a later time, determine whether and to what extent patients should be informed of expanded screening technology."

The five groups collaborated on the Joint Statement on Expanded Carrier Screening in order to provide education for clinicians and laboratories regarding the use of expanded genetic carrier screening in reproductive medicine. It states, "The current statement demonstrates an approach for health care providers and laboratories who wish to or who are currently offering expanded carrier screening to their patients."

While the new Joint Statement is not intended to replace existing practice guidelines and policy statements, it states that they "offer an opportunity for health care providers to better understand expanded carrier screening. Many more conditions, genes and variants are analyzed when expanded carrier screening is used compared with current screening approaches.... However, this approach introduces complexities that require special considerations."

ACMG President-Elect Gerald Feldman, MD, PhD, FACMG stated, "There are always advantages and disadvantages when a new technology is implemented, as is the case for expanded genetic testing. This document was written to provide a summary of the important points a physician should consider when discussing expanded carrier screening with his or her patient, because these tests offer testing for many more conditions than currently recommended by professional organizations. It is important that the patient fully understand and consent to such testing if they so choose. A referral to a genetics health care professional, such as a Board-certified clinical geneticist, should always be recommended when appropriate."

"Variation among people as to what they think justifies consideration when making reproductive decisions is varied and complicates generating a specific list of genes and variants that should be part of a test. Our goal for this document was to highlight the important aspects of genes and diseases that should be considered when developing expanded carrier screening panels," said co-author Michael S. Watson, MS, PhD, FACMG Executive Director of the American College of Medical Genetics and Genomics.

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About the ACMG and ACMG Foundation

Founded in 1991, the American College of Medical Genetics and Genomics (www.acmg.net) advances the practice of medical genetics and genomics by providing education, resources and a voice for more than 1750 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. ACMG is the only nationally recognized medical organization dedicated to improving health through the practice of medical genetics and genomics. The College's mission includes the following goals: 1) to define and promote excellence in the practice of medical genetics and genomics and to facilitate the integration of new research discoveries into medical practice; 2) to provide medical genetics and genomics education to fellow professionals, other healthcare providers, and the public; 3) to improve access to medical genetics and genomics services and to promote their integration into all of medicine; and 4) to serve as advocates for providers of medical genetics and genomics services and their patients. *Genetics in Medicine*, published monthly, is the official ACMG peer-reviewed journal. ACMG's website (www.acmg.net) offers a variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Find a Geneticist tool. The educational and public health programs of the American College of Medical Genetics are dependent upon charitable gifts from corporations, foundations, and individuals through the ACMG Foundation for Genetic and Genomic Medicine (www.acmgfoundation.org.)

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