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ACMG Provides Roadmap for Screening Couples Before or During Pregnancy: New ACMG Clinical Practice Resource for Autosomal Recessive and X-linked Conditions

Bethesda, MD –July 20, 2021| The American College of Medical Genetics and Genomics (ACMG) has released an important new Clinical Practice Resource (CPR) that reviews the current status of carrier screening, provides answers to emerging questions, and recommends a new tiered, consistent and equitable approach for offering carrier screening to all individuals during pregnancy or preconception. The paper, "Screening for autosomal recessive and X-linked conditions during pregnancy and preconception: a practice resource of the American College of Medical Genetics and Genomics (ACMG)," was published in ACMG's official journal, *Genetics in Medicine*.

Carrier screening is used to identify individuals or couples who are at-risk to have a child with an autosomal recessive or X-linked genetic disorder. Once identified, carriers of these disorders can become educated about their risks and consider a range of reproductive options.

In 2015, the ACMG, along with other professional organizations, published a "Points to Consider" joint statement focused on expanded carrier screening. The principles emphasized in that document remain important today. This current document considers more recently published information and closes gaps in the previously published "Points to Consider," while acknowledging technological advances in sequencing and the need for equity and distributive justice of genomic technologies.

"This CPR redefines carrier screening and promotes the values of equity and access to reproductive choices for carriers," said co-author and ACMG President-elect Susan Klugman, MD, FACMG, FACOG. "We have the technology to make a meaningful improvement in health outcomes. Patients should be able to utilize this testing, of course with appropriate pre- and post-test counseling."

"This document is a major step forward in delineating a new approach to carrier screening," said Jeffrey S. Dungan, MD, FACMG, FACOG, co-author and chair of the ACMG Professional Practice and Guidelines Committee. "The need for having a professional organization recommend a well-defined and explicit approach to carrier screening is long overdue. The concept of a tiered structure in defining sets of genes for analysis will help the clinician and the patient to better communicate with each other. More notably, ACMG has now expressly created a set of 113 genes, both autosomal recessive and X-linked (Tier 3), that should be considered the standard offering to preconception and prenatal patients," he concluded.

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"To come up with a list of candidate genetic conditions for screening, this document utilized the large genomic datasets that have become available in the past few years in various populations," said co-author Mahmoud Aarabi, MD, PhD, FACMG. "This is only the first step. I expect the list of genes will be updated regularly, based on the new genomic and clinical findings, to deliver the greatest benefit to the individuals of various ancestral backgrounds."

The authors developed a series of questions that are important for clinicians and reproductive-age patients to consider as part of the carrier screening process. Among the notable considerations:

What screening approach should be offered to patients considering carrier screening?

Professional organizations have an obligation to define the conditions appropriate for carrier screening. Until now, molecular testing laboratories have assumed this responsibility with the consequence that conditions screened for are not uniform across laboratories. This consensus group recommends establishing a tier-based system of carrier screening, which is detailed in this CPR, and which will enhance communication and precision while advancing equity in carrier screening. ACMG recommends:

- All pregnant patients and those planning a pregnancy should be offered Tier 3 carrier screening. Genetic conditions qualified for inclusion in the Tier 3 carrier screening are listed in the Clinical Practice Resource document.

What should be emphasized during pre-test and post-test counseling when performing carrier screening?

- Education and counseling are critical in carrier screening.
- Informed decision-making with carrier screening is complex and ideally should be a part of preconception care to allow any of the reproductive decision-making options.
- Knowledgeable and appropriately trained healthcare professionals should inform patients of the risks, benefits and consequences of carrier screening, pre- and post-test. After appropriate counseling that considers the patient's needs and values, patients should be supported to make informed and autonomous decisions including the decision to not undergo carrier screening. This process is time- and labor-intensive and new models need to be developed to train non-genetics providers and to counsel patients.

Over time, we believe clinicians will become comfortable with the concepts, specific genes, and their associated conditions which are proposed in this document. Importantly, molecular testing laboratories are called on to adapt and innovate to keep carrier screening costs low and throughput high.

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"For all patients to benefit from carrier screening, the carrier frequency needed to expand, and X-linked conditions needed to be included," said lead author Anthony R. Gregg, MD, MBA, FACOG, FACMG. "The benefits of carrier screening are clear. The greatest benefits can be achieved by accepting the challenge that all women be offered carrier screening not during pregnancy, but as they move from being pediatric patients to patients requiring well-women care. Professional organizations must respond to this call," he added.

About the American College of Medical Genetics and Genomics (ACMG) and ACMG Foundation

Founded in 1991, the American College of Medical Genetics and Genomics (ACMG) is the only nationally recognized medical professional organization solely dedicated to improving health through the practice of medical genetics and genomics, and the only medical specialty society in the U.S. that represents the full spectrum of medical genetics disciplines in a single organization. The ACMG is the largest membership organization specifically for medical geneticists, providing education, resources and a voice for more than 2,300 clinical and laboratory geneticists, genetic counselors and other healthcare professionals, nearly 80% of whom are board certified in the medical genetics specialties. ACMG's mission is to improve health through the clinical and laboratory practice of medical genetics as well as through advocacy, education and clinical research, and to guide the safe and effective integration of genetics and genomics into all of medicine and healthcare, resulting in improved personal and public health. Four overarching strategies guide ACMG's work: 1) to reinforce and expand ACMG's position as the leader and prominent authority in the field of medical genetics and genomics, including clinical research, while educating the medical community on the significant role that genetics and genomics will continue to play in understanding, preventing, treating and curing disease; 2) to secure and expand the professional workforce for medical genetics and genomics; 3) to advocate for the specialty; and 4) to provide best-in-class education to members and nonmembers. *Genetics in Medicine*, published monthly, is the official ACMG journal. ACMG's website, www.acmg.net offers resources including policy statements, practice guidelines, educational programs and a 'Find a Genetic Service' tool. The educational and public health programs of the ACMG are dependent upon charitable gifts from corporations, foundations and individuals through the ACMG Foundation for Genetic and Genomic Medicine.

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