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American College of Medical Genetics and Genomics
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Q&A with Dr. Laurie Demmer, Chair of the Topic Selection Committee for ACMG's New Evidence-Based Guidelines Program

In a recent interview with *The ACMG Medical Geneticist*, Laurie Demmer, MD, FACMG, ACMG board member and chair of the Topic Selection Committee, described ACMG's exciting new Evidence-Based Guidelines Program and how it aligns with the College's new strategic plan.

ACMG Medical Geneticist: What is the new ACMG Evidence-Based Guidelines Program and why is it important?

Laurie Demmer (LD)*:* This new program is a comprehensive and sustainable way for ACMG to develop, publish, and regularly update evidence-based guidelines that address validity and utility of clinical genetic services.

The College has already been developing guidelines for several decades. Government and private health insurers use our guidelines to help determine coverage for clinical genetic tests, and the guidelines help inform our clinical and lab practices.

We're hoping the new Evidence-Based Guidelines Program, supported by external funding (see sidebar article), will expand upon and expedite this process so we can disseminate guidelines more often and more quickly than we have in the past.

ACMG*:* Why is this program happening now?

LD*:* We need evidence-based guidelines to help clinicians and insurers make the best decisions regarding testing and new treatments. Guidelines that are backed by a rigorous evidence review can have a greater impact on care. We also need to keep pace with the rapid technological advances in genetic and genomic medicine. Technology is changing so quickly that it's hard for clinicians to know what the best test is and what is the best treatment. Having up-to-date guidelines will help them in their practice.

I also want to emphasize that this program is a direct result of the member-driven new strategic plan, and it's one of the top priorities identified by the member survey that preceded the strategic plan. Members want us to develop standards and guidelines to enhance the practice of clinical genetics and genomics. Now, members themselves will be nominating the topics for those guidelines.

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***ACMG**:* Who will be involved with the program, and how will it work?

***LD**:* Once a key question is developed, we will be using an evidentiary framework—systematic evidence reviews (SER)—to review the literature. Information derived from the SER, along with other informational sources and input from an expert panel, will contribute to the evidence-based guideline.

The first step is for members to consider where the evidence base surrounding a specific topic is diffuse, contradictory, or misrepresented. These topics should have an important clinical impact so that after a systematic review of the evidence, conclusions from the SER can be used to develop an evidence-based practice guideline. Members submit their suggested topics to the Topic Selection Committee, which will review and prioritize the nominations and make recommendations to the ACMG Board of Directors, who will approve the topics chosen for SER. Our goal is to choose topics that are relevant and timely so the program will benefit all the stakeholders in genetics and genomic medicine. In today's world, that would include both geneticist and nongeneticist clinicians, laboratory geneticists, providers, payors, and healthcare systems.

Once a topic is selected, volunteers from the ACMG membership, the Systematic Evidence Review Work Group (SER WG), will perform the SER. The Evidence-Based Guideline Work Group (EBG WG) then will develop the practice guideline based on the conclusions of the SER and other sources. The EBG WG members will be experts in the field and will be required to disclose and keep current any potential conflicts of interest.

SERs will also be performed for topics derived from previously published ACMG policy statements, practice guidelines and points to consider documents. Revised and reissued documents will be supported by a greater evidence base. ACMG committee chairs are reviewing documents for consideration.

ACMG: Must members who are interested in participating be familiar with SERs or guideline panel work to volunteer for an SER WG?

LD: It's not necessary to already have worked on a guideline or a SER, though that experience is helpful. Our methodologists will provide appropriate training to each work group at every step along the way.

ACMG currently has a part-time methodologist and is in the process of hiring a full-time methodologist. They will facilitate development of key questions related to SER topics. The next step is to conduct the evidence reviews and grade the evidence, leading to a SER manuscript. Finally, the EBG WG will develop a manuscript for publication.

New guidelines that result from the SERs will be published in *Genetics in Medicine*, as well as in the Practice Resources section of the ACMG website.

***ACMG**:* What should members watch for as this project develops?

***LD**:* Recently, all members received an invitation to submit topics of their choice. The deadline for those submissions was September 23 and we received seven nominations of topics to review. The first topic selected for an SER is "Noninvasive Prenatal Screening for Fetal Aneuploidy in Average Risk Populations."

We don't have a specific timeline yet for when the first round will be completed, because the pace of guideline development will be determined largely by funds raised for this program by the ACMG Foundation. But ACMG's communications staff will share information as it becomes available, including when the new guidelines have been published and when the next round of topics will be considered.

We will be formally soliciting nominations for future topics periodically. Members should watch for a blast email that contains a link to the SER nomination form for our next round of review or members can submit SER nominations anytime at https://forms.gle/EfCpPvN1VxZ2e7B66.

Sidebar: The ACMG Foundation is working to secure $1,000,000 annually to support the new Evidence-Based Guidelines Program. The goal is to gather funding from a wide range of individual, corporate, and nonprofit donors.

Thanks to an early commitment from Illumina, our Guidelines Program has the seed funding to start work immediately. But to sustain this effort long term, ACMG will need many other committed donors to step forward.

Rapid advances in medical science demand new guidelines in the genetic and genomic space. An investment is needed for this new guideline development and to help update some of ACMG's past guidelines. Keeping medical genetics and genomics in line with our ever-expanding understanding of the human genome will aid payors, providers, and patients alike.

To support this effort, please contact Karl Moeller at kmoeller@acmgfoundation.org. You may also visit our website at www.acmgfoundation.org and follow the Donate button to the contribution page. Be sure to indicate "Diagnosis and Treatment Guidelines" if you wish to support the guideline initiative specifically.