



## **Diagnostic & Treatment Guidelines Development Fund**

*Providing the resources to build the guidelines for better health through genetics and genomics.*

### **Purposes**

The purposes of “The ACMG Foundation’s Diagnostic & Treatment Guidelines Development Fund” (the “Guidelines Fund”) are to advance education, research and standards of practice in medical genetics and genomics and to develop and expand clinical and laboratory expertise worldwide by supporting the development of a compilation of evidence-based diagnostic and treatment guidelines for genetic and genomic diseases.

### **Description of the Guidelines Fund**

The completion of the Human Genome Project has made tools widely available for understanding the role of genetics and genomics in human health and disease, and for the development of new tests and treatments for heritable conditions. The pace of scientific discovery is now precedent setting. Never have individuals and families at risk for or suffering from genetic conditions realized such promise in the way these often-rare conditions are being diagnosed, managed and treated. However, translating discovery and innovation into uniform clinical care and improved outcomes is a complex process.

A key step in this process is the evidence-based assessment of new tests and treatments and the formulation of clinical practice guidelines that evolve into standards of care across the U.S. and around the world.

The American College of Medical Genetics and Genomics (ACMG) has remained a highly trusted and respected medical organization since 1993. ACMG’s timely development and review of practice guidelines have helped safely and effectively diagnose, manage, and treat genetic conditions. Gene conditions extend over the human lifecycle. Genetic disease detected in utero, by newborn screening, or involving common disorders of adulthood (heart disease, diabetes, and gene-based responses to medications) have all been the focus of past guidelines.

ACMG’s leading role as the recognized authority and trusted expert in the diagnosis and management of genetic disorders is due, in no small measure, to the network of physicians and specialists throughout the country and worldwide who comprise its membership. The recognition of Medical Genetics as the 24<sup>th</sup> primary specialty board of medicine with corresponding recognition by the American Board of Medical Specialties (ABMS) and the American Medical Association (AMA) have placed ACMG in a strong position within organized medicine to work with other specialties as medical genetics and genomics continue to integrate into health care delivery.

Both the acceleration of scientific discoveries and the emergence of new therapeutics for genetic diseases puts increasing demands on ACMG to intensify its work in updating or developing new diagnostics, practice and treatment guidelines.

The Guidelines Fund supports ACMG’s development and dissemination of trustworthy evidence-based guidelines that incorporate new scientific discoveries in the diagnosis, management and treatment of genetic and genomic conditions.

## Objective

To improve health and to assist healthcare providers worldwide by supporting the development, dissemination and implementation of practice guidelines that address the diagnosis, management and treatment of genetic and genomic disorders and, thereby, improve the health and quality of life of individuals and families at risk for or suffering from genetic conditions.

## Overview

Developing practice guidelines is a time and labor-intensive process. It involves:

- Identifying and convening a group of subject matter experts representing a breadth of perspectives;
- Working with this group and key staff to determine the questions that must be resolved;
- Reviewing the scientific literature, existing practice guidelines, and the evidence base;
- Identifying knowledge gaps; and,
- Making recommendations only after rigorous deliberation and a weighing of risks and benefits.

In the past, ACMG was able to promulgate these detailed documents (as well as the popular Newborn Screening Act(ion) Sheets and accompanying algorithms) using its volunteer committee structure. Because staff time was limited, substantial grant support was not needed – but a volunteer effort is not always timely or efficiently managed during larger complex projects.

Today's science demands an intensified approach. The pace of scientific discovery and translational research has quickened. There is an expanded and justified attention to conflict of interest issues between industry and professional organizations. And we face an increasing number of genetic conditions for which treatments are on the horizon. All these factors demand a more formal process and structure. The Evidence Based Guidelines Program must be timely and get the best-practices to the point of care quickly, while systematically addressing both real and perceived conflicts of interest.

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With the launch of our Evidence Based Guidelines Program in the summer of 2019, ACMG Foundation seeks to raise funds to support the evaluation, writing, and updating of evidence-based guidelines for the diagnosis, management and treatment of specific genetic disorders. The ACMG Foundation, a 501(c)(3) nonprofit organization, is a community of supporters and contributors who understand the importance of medical genetics, genomics, and genetic counseling in healthcare. The Foundation supports ACMG's mission to “translate genes into health.”

While the ACMG Foundation will drive fundraising efforts, staff and Board members for the College, a 501(c)(6) membership organization, will write, update, prioritize work to be completed through independent committees of ACMG members and experts. These committees will be appointed by the Board of Directors of the College. Unless specifically needed and approved by the ACMG Board, experts from industry should generally not serve on the independent committees.

The guidelines will be written and updated on the same schedule as other ACMG practice guidelines as based on the latest scientific evidence.

All needed funds will be raised and administered by the ACMG Foundation and contributions in support of the program will be earmarked for the Guidelines Fund.

The Guidelines fund will act as a clearing house for all contributions to the EBG Program. Individuals, foundations, and corporations should have no expectation that their gift to the Foundation will be used to write or update a specific guideline by the College. It is the goal of ACMG and the ACMG Foundation to write and update guidelines for all common genetic and Newborn Screening panel (NBS) disorders in the order of priority established by the independent Topic Selection Committee. While the guidelines will not be “sponsored” by a single individual, foundation, or corporation, in the spirit of full disclosure, an acknowledgement of all contributors to the Guidelines Fund will be part of each guideline.

ACMG will be responsible for the effective dissemination of the final guidelines.

### **Selection of Guidelines**

The Guidelines developed through the Guidelines Fund will be selected by a Topic Selection Committee nominated by the ACMG Medical Director (or the ACMG Board if not staffed), with consent by the ACMG Board of Directors. This Topic Selection Committee may include members of the ACMG Professional Practice and Guidelines Committee, the Laboratory Quality Assurance Committee, or the Therapeutics Committee.

Each guideline proposal will be evaluated based on common criteria (see “Review Criteria” below) that weigh potential contributions to the improvement of medical genetics practice. The Selection Committee will determine other appropriate selection criteria, all of which will be consistent with the purposes of EBG Program and the other qualifications and criteria.

Corporate Donors will not have a representative on the Topic Selection Committee or have any role in selecting specific guidelines.

### **Submitting a Request**

ACMG committees, ACMG members, patient advocacy groups, interested individuals and corporations may submit a request that a specific guideline be developed and may also suggest past guidelines to be reviewed and updated. A request form template is available to the public at:

[www.ACMG.net/EBGRequestForm](http://www.ACMG.net/EBGRequestForm)

The Topic Selection Committee is under no obligation to accept or develop a guideline based on a submitted request.

### **Review Criteria**

The goal of the Guidelines Fund is to develop guidelines that improve the practice of medical genetics and genomics and to promote better health through genetics. The following review criteria may be applied by the Selection Committee, based on the nature of the request:

- The importance and timeliness of the proposed guideline,
- The adequacy of the scope and content of the proposed guideline,
- The appropriateness of the guideline for achieving the stated goals of the Fund,
- Is the proposed guideline clearly described,
- Will the proposed guideline produce significant or immediate changes in practice,
- Is the proposed product or outcome needed,
- Can the guideline be executed in an appropriate time period,

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- Are the resources adequate,
- Can the request be improved
- Are the proposed guidelines likely to be effective,
- Can the proposed guidelines be easily used,
- Can the guidelines be easily distributed,
- Can the proposed guidelines be integrated into another guideline, and
- Does the request revise or update an existing guideline?

### **Guidelines Writing Committees**

Once a guideline is approved for development by the Topic Selection Committee, the ACMG Board of Directors will establish a Guideline Writing Committee of no less than three experts, and may include members from the ACMG Professional Practice and Guidelines Committee, Laboratory Quality Assurance Committee, Therapeutics Committee, or other appropriate committees and experts.

ACMG intends to maintain communication between work groups over time, so any required updates or modifications are more easily executed.

### **Receipt and Review Schedule**

Requests for proposed guidelines are accepted throughout the year, but will be considered in no less than two rounds annually, based on the following schedule:

- Request Submission Deadline: December 1 and May 1,
- Reviews and Decisions on Guidelines: January 1 and June 1,
- Guidelines Writing Committee begins work: February 1 and July 1,
- Guideline Announcement: at the earliest ACMG Annual Meeting.

### **Administration**

The Guidelines Fund will be administered under the direction of the ACMG Foundation President, the Foundation Board of Directors, and the Executive Director of the ACMG Foundation or, barring none, by the CEO of the College. The development of Guidelines will be administered under the direction of the ACMG Medical Director, ACMG President and Chief Executive Officer.

### **Dissemination**

The Guidelines Fund is responsible for the dissemination of all final guidelines and will use all available outlets and resources. Including but not limited to the following:

- Genetics in Medicine journal,
- Other medical and/or professional journals,
- ACMG and ACMG Foundation websites,
- Other medical society or professional association websites,
- Interested corporations and patient advocacy organizations' websites and newsletters,
- Press releases,
- Email blasts and social media postings (LinkedIn, Facebook, and others),
- Other ACMG and ACMG Foundation publications

### Where to Direct Inquiries

Questions about the Guidelines process:

Maximilian Muenke, MD, FACMG  
Chief Executive Officer,  
American College of Medical  
Genetics and Genomics  
7101 Wisconsin Avenue, Suite 1101  
Bethesda, MD 20814  
301-718-9603  
[mmuenke@acme.net](mailto:mmuenke@acme.net)  
Website: [www.acmg.net](http://www.acmg.net)

Questions about donating to the Guideline Fund:

Karl Moeller  
Executive Consultant  
American College of Medical  
Genetics and Genomics Foundation  
7101 Wisconsin Ave, Suite 1101  
Bethesda, MD 20814  
[kmoeller@acmgfoundation.org](mailto:kmoeller@acmgfoundation.org)  
240-204-9641 (phone)  
301-718-9604 (fax)

### Proposed Budget

The Board of the College has set an aggressive goal of publishing two guidelines in the first year of the program, then expanding that number as the program and staff's capacity grows. It is estimated that between \$70,000 and \$100,000 are needed to publish each guideline. Clearly, slightly less is needed to revise an already published ACMG guideline. These costs depend on the complexity of the disease, the extent of the studies available for review, and the number of questions the Topic Selection Committee identifies for the project.

To maintain the program, a significant amount will need to be raised in order to sustain staffing levels and ensure job stability for those hired. The ACMG Foundation is seeking \$1 million in funding by July 1 of 2020 to stand up the program and hopes to identify a core group of interested donors late in 2019 and early in 2020 who are willing to serve as inaugural donors.

### Giving Levels

- Corporate Friend – Under \$10,000
- Silver Partner - \$10,000 to \$49,999
- Gold Partner - \$50,000 to 99,999
- Platinum Partner - \$100,000 to \$250,000
- Diamond Partner – More than \$250,000 per yr.

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*The ACMG Foundation ([www.acmgfoundation.org](http://www.acmgfoundation.org)), a 501(c)(3) nonprofit organization, is a community of supporters and contributors who understand the importance of medical genetics and genomics and genetic counseling in healthcare. Established in 1992, the ACMG Foundation supports the American College of Medical Genetics and Genomics' mission to "translate genes into health" by raising funds to promote the profession of medical genetics and genomics to medical students, to fund the training of future medical geneticists, to support best-practices and tools for practicing physicians and laboratory directors, to promote awareness and understanding of our work in the general public, and much more.*

*Founded in 1991, the American College of Medical Genetics and Genomics ([www.acmg.net](http://www.acmg.net)) advances the practice of medical genetics by providing education, resources and a voice for more than 1400 biochemical, clinical, cytogenetic, medical and molecular geneticists, genetic counselors and other healthcare professionals committed to the practice of medical genetics. ACMG's activities include the development of laboratory and practice standards and guidelines, advocating for quality genetic services in healthcare and in public health, and promoting the development of methods to diagnose, treat and prevent genetic disease. Genetics in Medicine, published monthly, is the official ACMG peer-reviewed journal. ACMG's website ([www.acmg.net](http://www.acmg.net)) offers a variety of resources including Policy Statements, Practice Guidelines variety of resources including Policy Statements, Practice Guidelines, Educational Resources, and a Medical Geneticist Locator. The educational and public health programs of the*

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*American College of Medical Genetics and Genomics are dependent upon charitable gifts from corporations, foundations, and individuals.*