

Clinical, technical and environmental biases influencing equitable access to clinical genetics and genomics testing

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With the goal of fostering awareness and identifying strategies to reduce bias within the medical genetics field and to improve health equity,

members of the ACMG's Social, Ethical and Legal Issues (SELI) and Diversity, Equity and Inclusion (DEI) Committees collaborated to address factors in which bias can occur in clinical genetic testing in a just-published statement, "Clinical, technical, and environmental biases influencing equitable access to clinical genetics/genomics testing: A points to consider statement of the American College of Medical Genetics and Genomics (ACMG)".

Published in *Genetics in Medicine*, this is the first joint statement of the ACMG's SELI and DEI Committees.

Co-lead author and workgroup co-chair Dena R. Matalon, MD, FACMG, said, "By teaming up with our ACMG DEI colleagues, we were able to address more topics thoughtfully than if we pursued this complex issue in each committee alone. Our goal in working together was to raise awareness about how [bias](#) affects all of us, ways in which it does that we may not have realized, how bias affects underrepresented minorities, and what we can do to begin to mitigate it."

"We ultimately want to provide more equitable clinical [genetic](#) care for all, and hope this document helps us take a step in that direction."

"I am proud of the [collaborative efforts](#) of these two ACMG committees in developing this paper in that it reflects an honest and timely response to a set of issues of great importance to society, to the American healthcare enterprise and to members of the clinical team delivering genetic and genomic care to the patients and communities we serve," said ACMG Interim CEO Robert G. Best, Ph.D., MS, FACMG.

"This paper is a response to the growing recognition that disparities in medicine are common and that they propagate along the lines of race, geography and socioeconomic status. The authors of this Points to Consider document articulate an interplay between nested systems within [health](#)

[care](#) related to biases within environmental, clinical and technical domains and have begun to delineate the need for action that takes stock of the complex systems in which patient care plays out."

"ACMG is committed to learning, understanding and taking responsibility for the part that our discipline has played in sustaining a system that accepts inequity as being somehow beyond the level of our agency to meaningfully address, and this paper represents an early step along a better path."

As genetic testing becomes more accessible within the US and the world, multiple inherent scientific and clinical biases must be overcome to achieve [health equity](#) for individuals for which care is provided, including those belonging to historically marginalized populations. The ACMG SELI and DEI workgroup members reviewed and categorized bias into three main areas: environmental, clinical and technical.

Environmental bias factors in which individuals may face discrimination include employment, [medical insurance](#), housing, and interactions with colleagues, peers and medical institutions. Clinical factors that have contributed to inequities in genetic testing interpretation can encompass limited clinician diversity of representation, access to genetic services, education of both healthcare professionals and patients, and complexities associated with unknown family history and/or genetic ancestry.

For example, with few genetics professionals being bilingual, non-English speaking patients are at a disadvantage in receiving culturally appropriate care. Thus, diversifying the genetics workforce must continue to be a high priority.

Lastly, difficulties have arisen in variant interpretation for historically marginalized populations undergoing clinical genetic testing, as most individuals that take part in genomic research studies have been of

European descent. Multiple examples are provided in the new ACMG statement regarding these "technical" biases, and the statement highlights the need for an ongoing effort to recruit individuals from ancestral and biogeographically diverse populations to improve database genomic representation.

This document illustrates a framework that supports a positive and constructive dialogue among all stakeholders and lawmakers to continually address biases and keep advancing [genetic testing](#) in the clinic. The goal is to recognize and reduce bias, ensuring equitable care and avoiding unfair discrimination for all.

Cinthya J. Zepeda Mendoza, Ph.D., FACMG, co-lead author and workgroup co-chair, said, "Since its inception almost three years ago, the ACMG's Diversity, Equity, and Inclusion Committee has been working hard to prioritize DEI principles within the College and the medical genetics specialties."

"This document is the culmination of a rewarding collaboration with our ACMG SELI colleagues, in which we addressed biases in clinical testing and their significant impact on individuals and communities, particularly those from underrepresented and marginalized backgrounds. Our goal with this publication is to disrupt our bias and to remain accountable in ensuring that individuals from all backgrounds receive the best possible care."

More information: Dena R. Matalon et al, Clinical, technical, and environmental biases influencing equitable access to clinical genetics/genomics testing: A points to consider statement of the American College of Medical Genetics and Genomics (ACMG), *Genetics in Medicine* (2023). [DOI: 10.1016/j.gim.2023.100812](https://doi.org/10.1016/j.gim.2023.100812)

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