Study: Major Gaps Block Genetics Evaluation and Testing for Black and Low Income Patients

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Perelman School of Medicine at the University of Pennsylvania December 19, 2025

Newswise — PHILADELPHIA— Black patients and patients from socioeconomically disadvantaged neighborhoods are dramatically under-represented in genetics clinics—often at rates half or less than those of White patients or individuals from wealthier areas—according to researchers at the Perelman School of Medicine at the University of Pennsylvania and Massachusetts General Hospital. But once patients were evaluated for genetic testing, the disparities reversed: Black patients were more likely to have genetic testing ordered, and patients from lower-income neighborhoods were more likely to get a definitive, actionable result that can guide treatment and improve outcomes. The study was published today in the *American Journal of Human Genetics.*

“These findings indicate that minorities or lower-income patients are not hesitant about genetic testing, they simply haven’t been informed about the option to receive it,” said the study’s corresponding author, Theodore G. Drivas, MD, PhD, an assistant professor of Translational Medicine and Human Genetics at Penn.

The study focused on germline genetic testing, which looks for signs of inherited variants in a patient's DNA that could raise a red flag for hereditary cancers, mitochondrial diseases, or cystic kidney disease, for example. Researchers analyzed records from more than 14,000 patients seen in adult genetics clinics at the University of Pennsylvania Health System and Mass General Brigham over five years. “This work shows that when patients from underrepresented communities access genetics services, they not only accept testing at higher rates but often uncover critical answers.” said Latrice Landry, PhD, an instructor of Genetics at Penn and a co-author of the study.

Guidelines could help the trickle-down effects from fewer genetic testing referrals

“The real bottleneck is likely at the referral stage; systemic barriers in primary care, such as knowledge gaps, clinic hours, and referral patterns, are preventing patients who stand to benefit the most from ever reaching our door,” said Drivas.

The lack of national guidelines on the importance of genetic testing plays a key role in these gaps. Without clear-cut indications to suggest the importance of genetics referral or testing, providers are less likely to refer their patients to genetics, and insurances are less likely to cover such testing. The study found that large concerted efforts to develop such guidelines, for example in

cancer genetics, where genetic testing is now common, helped eliminate racial disparities. The researchers say similar steps could help other adult genetic conditions, where coverage isn't always guaranteed and can make doctors or patients hesitate.

Closing the gap

Promising interventions identified in the study include electronic health record flags to prompt referrals, embedding genetic counselors in primary care settings, and offering virtual genetic counseling options, which have been successfully implemented in cancer genetics programs. The researchers also call for expanded workforce training and diversification, noting the severe shortage of genetics specialists and the lack of diversity in the field. "The findings have urgent implications as genetic testing becomes central to routine care," said Landry. "We owe it to all patients to build equitable systems — through better education, virtual care, and policy changes— so that precision medicine truly benefits everyone."

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