The right to be screened: Identifying and addressing inequities in genetic screening

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Genetic testing has increasingly been incorporated into clinical practice to identify patients in need of additional screening, stratify risk among patients with cancer, and allow doctors to intervene sooner in potentially high-risk tumors. While already a mainstay in the risk stratification for several cancer types, such as enhanced breast cancer screening for people with *BRCA1* or *BRCA2* pathogenic variants and colon cancer screening for people with Lynch Syndrome, genetic testing is currently being considered for new cancers, such as prostate cancer. Bancroft et al. recently demonstrated that patients who have been identified with germline mismatch repair pathogenic variants in *MLH1, MSH2*, or *MSH6* genes may benefit from targeted PSA screening to identify clinically significant prostate cancer.[1] Despite the numerous advantages in genetic testing, we contextualize this technology in a disparities framework, highlighting the persisting equity challenges that remain, and potential steps forward.

Prior studies have acknowledged significant inequalities in access to genetic counseling and testing. Muller et al. noted that, despite similar rates of mismatch repair deficiencies across racial and ethnic groups, members of minority populations were less likely to receive a referral for genetic testing.[2] Furthermore, Sivakumar et al. found that, although there may be similar rates of targetable pathogenic variants amongst Black and White men with prostate cancer, Black men were less likely to receive comprehensive genomic profiling earlier in their treatment course.[3]

Unequal referrals and low levels of genetic testing have a damaging cyclic effect; when certain subpopulations receive lower levels of care, reference databases provide an incomplete picture of the epidemiology and penetrance of pathogenic variants. Consequently, the lack of evidence reinforces the incorrect assumption that genetic testing may be unnecessary in these communities.

Disparities in access to testing likely reflect both systemic influences and patient preferences. Providers are less likely to encourage genetic testing in non-White populations despite similar or increased risk, highlighting biases in provider behavior.[2,3] Financial barriers to testing also persist. We must also acknowledge atrocities committed against minority populations by the healthcare system, which continue to influence patient behavior: in a survey-based study, African Americans were found more likely to believe that genetic testing will lead to racial discrimination, while also acknowledging the value of screening for disease.[4]

Therefore, we suggest key steps to improve equity in genetic screening and cancer diagnosis:

1. **Build trust -** Direct, sustained engagement with community partners is a necessary investment to improve relationships between minority populations and the medical community. Trust also involves better cultural competency and appropriate discussion of risks and benefits of preventative care. By framing conversations in a way that considers the patient's background as well as what matters most to them, providers are better able to both understand their patient's needs and increase equitable care.
2. **Equity-focused implementation** - There is a need for equity-focused care delivery.[5] Changes to screening practices should prospectively consider influences on equity rather than encourage intervention once disparities have already been exacerbated. Efforts should focus on financial barriers to care, screening access among ethnic, racial, linguistic, geographic, and sex/gender minorities, and increased education about genetic testing. Implementation strategies should actively include populations who are at highest risk of experiencing disparities in access to genetic risk assessment. These strategies may be informed by adult learning theory and cultural humility.[6,7]

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3. **Address variations in provider behavior** - Further research is needed to explain the observed variations. Findings from these studies will allow for more directed and tailored interventions that challenge current unequal practices in referral and access to genetic testing. Individually, healthcare professionals should take time to explore their own unconscious biases they bring into patient encounters, and how these may influence conversations. Systemic change is undoubtedly necessary, but individual work can help to mitigate harms.

Access to genetic counseling and testing can guide the ways in which patients are screened for various cancers. Recent advances suggest screening patterns for other cancers, such as prostate cancer, may also be guided by genetic testing. Therefore, it is critical to identify and address disparities in genetic screening as we work to promote equity in cancer care for all.

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