Racial and socioeconomic disparities in genetic evaluation and testing in the adult patient population

Authors

Jessica I. Gold, Yehuda Elkaim, Nina B. Gold, ..., Katherine L. Nathanson, Staci Kallish, Theodore G. Drivas

Correspondence

theodore.drivas@pennmedicine.upenn.edu

This study of over 14,000 adults in two major health systems reveals striking racial and socioeconomic disparities in access to genetics evaluation and testing. Black individuals and those from disadvantaged neighborhoods were less likely to be evaluated but more likely to undergo testing and receive positive results when seen.

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ARTICLE

Racial and socioeconomic disparities in genetic evaluation and testing in the adult patient population

Jessica I. Gold,[1,2] Yehuda Elkaim,[3] Nina B. Gold,[4] Stephanie Asher,[5] Anna Raper,[5] Courtney Condit,[5,6] Zoe Bogus,[5] Isaac Elysee,[5] Laura Hennessy,[5] Emma Kennedy,[5] Lauren C. Briere,[7] David A. Sweetser,[4,7] Colleen Kripke,[5] Anurag Verma,[5] Hojjat Salmasian,[8] Latrice Landry,[9] Katherine L. Nathanson,[5,10] Staci Kallish,[5] and Theodore G. Drivas[5,9,*]

Summary

Genetic information directs clinical management and leads to improved health outcomes. However, there are scant data regarding the role of race or social determinants of health (SDOH) on access to genetics evaluation or outcomes of genetic testing in the general adult population. Here, we present the results of a retrospective study of 14,669 individuals seen over a 5-year period within the University of Pennsylvania and the Mass General Brigham Health Systems' Adult Genetics Clinics. We assessed the effects of electronic health record-reported race and neighborhood-level measures of SDOH on likelihood of evaluation in an Adult Genetics Clinic, likelihood of having genetic testing sent, and outcomes of genetic testing. Black individuals (odds ratio [OR] < 0.65; $p < 2e{-}16$) and those from disadvantaged neighborhoods (OR < 0.99; $p < 0.0001$) were significantly less likely to be evaluated in an Adult Genetics Clinic, with Black individuals significantly less likely to be evaluated for clinical indications with more subjective presentations (OR < 0.33; $p < 0.003$). When evaluated, Black individuals were more likely to undergo genetic testing (OR = 1.35; $p = 0.005$), and individuals from disadvantaged neighborhoods, independent of race, were more likely to have pathogenic variants identified on genetic testing (OR > 1.01; $p < 0.002$). These findings highlight significant disparities in genetic healthcare delivery among adult individuals based on race and social determinants of health—disparities that are poised to widen unless proactively addressed as genetic testing becomes increasingly central to the practice of clinical medicine. We provide resources and information for providers and affected individuals to reduce barriers to medical genetics care and promote equity in precision medicine initiatives.

Introduction

Precision medicine, which is often contingent upon genetic diagnosis, is rapidly being implemented through targeted therapeutics and surveillance.[1] Medical geneticists and genetic counselors play a primary role in the evaluation and diagnosis of genetic disease.[2] However, access to these specialists is frequently impeded by clinicians' knowledge gaps, geographic limitations, and lengthy waitlists, all of which have the potential to exacerbate healthcare disparities and impact health outcomes.[3–5]

Recent reports by the American College of Medical Genetics and Genomics and the American Society of Human Genetics illustrate how race-, ancestry-, and socioeconomic-based biases may lead to inequitable care.[6,7] Individuals of lower socioeconomic status (SES) are less likely to undergo genetic testing,[8,9] and genetic testing more frequently returns inconclusive results in minoritized individuals[10,11]; these inequities lead to a lower likelihood of receiving a genetic diagnosis.[12–14] Additionally, minoritized individuals are less likely to be represented in dysmorphology atlases,[15] and other genomic medicine approaches, including polygenic risk scores and pharmacogenomic analyses, rely on population-level variant frequencies from databases with predominantly White participants and thus are often not applicable to individuals of other ancestries and ethnicities.[16–18] Furthermore, the use of race, a social construct, to suggest the existence of genetically distinct human populations, perpetuates the inappropriate and inaccurate categorization of individuals into groups that lack biological basis or clinical relevance.[19,20] However, race is often a factor in clinical algorithms and decision making, gatekeeping access to care, and increasing healthcare disparities.[21]

Race- and socioeconomic-status-based systemic barriers in obtaining a genetic diagnosis have been described in pediatric genetics,[8] pediatric neurology,[9] cancer genetics,[22] cardiomyopathy genetics,[12] and hearing loss.[13] However, the effects of race and other social determinants of health (SDOH) on genetic evaluation and testing in the broad

[1]Division of Clinical Genetics, Department of Pediatrics, Cohen Children's Medical Center, Northwell Health, Great Neck, NY 11021, USA; [2]Institute of Health System Science, Feinstein Institutes for Medical Research, Northwell Health, Manhasset, NY 11030, USA; [3]Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA 19104, USA; [4]Division of Medical Genetics & Metabolism, Massachusetts General Hospital for Children, Boston, MA 02114, USA; [5]Division of Translational Medicine and Human Genetics, Department of Medicine, Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA 19104, USA; [6]Kaiser Permanente, Tysons Corners Medical Center, McLean, VA 22102, USA; [7]Center for Genomic Medicine, Massachusetts General Hospital, Boston, MA 02114, USA; [8]Division of General Internal Medicine, Brigham and Women's Hospital, Boston, MA 02115, USA; [9]Department of Genetics, Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA 19104, USA; [10]Abramson Cancer Center, Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA 19104, USA
*Correspondence: theodore.drivas@pennmedicine.upenn.edu
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general adult patient population have not been examined. Here, we examine the impact of these factors on ~14,500 individuals evaluated at three general Adult Genetics Clinics within the University of Pennsylvania Health System (UPHS) and Mass General Brigham Healthcare System (MGB), each located in urban areas with high minoritized and underserved populations. We identify many race- and SDOH-based disparities in evaluation rate, likelihood of undergoing genetic testing, and outcomes of testing. As precision medicine continues to gain influence on clinical care, these inequities in access to a genetic diagnosis in the general adult patient population must be addressed.

Subjects and methods

UPHS study population

Our study was approved by the University of Pennsylvania's Institutional Review Board (protocol #850058) and complies with the principles set out in the Declaration of Helsinki. Within the UPHS data, we collected records on 2,228,622 patient visits for all individuals (≥18 years) seen between October 1, 2016 and October 1, 2021. We generated data separately for the entirety of UPHS (consisting of six hospitals and ten multispecialty centers that serve south-central Pennsylvania, south-central New Jersey, and northern Delaware) and for the UPHS Perelman Center for Advanced Medicine (PCAM) endocrinology, cardiology, and genetics outpatient clinics. These data included 5,525 unique outpatient Adult Genetics Clinic visits (evaluations) at the UPHS PCAM Adult Genetics Clinic. We define a genetics "evaluation" as a clinical session with a genetics provider, and "testing" or "genetic testing" as the decision to undergo genetic testing within that evaluation; not all evaluations lead to testing, but all testing occurs in the context of evaluation. For the 5,525 genetics visits, we obtained individual-level demographic information, including age, gender, and electronic health record (EHR)-reported race and ethnicity, from the UPHS Epic Clarity database (Table S1) and joined this information with a clinical records database maintained by the UPHS Adult Genetics Clinic documenting the visit indication, whether genetic testing was sent, and the results of testing. The 1,147 outpatient visits with any missing observations were resolved by manual chart review. Clinical indications for outpatient visits were collapsed into 21 broad-indication groups (Table S3) by a panel of genetics providers blinded to both demographic information of individuals and outcomes of genetic testing. Patient neighborhoods were defined as home-address-based US census tracts, assigned to each individual using the tool Geocodio, and were joined with ARHQ's SDOH database from 2020 (available from https://www.ahrq.gov/sdoh/data-analytics/sdoh-data.html). SDOH variables were chosen based on previous work in pediatric[8,23] and cancer populations.[22] Neither study subjects nor the public were involved in the design, conduct, reporting, or dissemination plans of our research.

MGB study population

Our study was approved by the MGB Institutional Review Board (protocol #2023P003307) and complies with the principles set out in the Declaration of Helsinki. Within the MGB data, we collected records on 5,540,136 patient visits at Brigham and Women's Hospital (BWH) and 6,908,293 patient visits at Massachusetts General Hospital (MGH) for all individuals (≥18 years) seen between October 1, 2016 and October 1, 2021. We generated data separately for all outpatient visits at MGB in any department and for the MGB endocrinology, cardiology, and genetics outpatient clinics. These data included 9,144 unique outpatient Adult Genetics Clinic visits seen by a Medical Genetics-trained physician or genetic counselor. BWH has a specific Adult Genetics Clinic, while MGH has a single general genetics clinic serving both pediatric and adult individuals; for this study, we limited our analysis to only adults seen at the MGH general genetics clinic. We obtained individual-level demographic information, including age, gender, and EHR-reported race and ethnicity for all 9,144 visits from the MGB's data warehouse (Table S2). Patient neighborhoods were defined as home-address-based US census tracts, assigned to each individual using DeGAUSS, and were joined with ARHQ's SDOH database from 2020 (available from https://www.ahrq.gov/sdoh/data-analytics/sdoh-data.html). Neither study subjects nor the public were involved in the design, conduct, reporting, or dissemination plans of our research.

Additional data sources

Data regarding race and ethnicity for individuals living in Philadelphia and Boston was downloaded from the 2020 US Census (available from https://www.census.gov/programs-surveys/decennial-census/decade/2020/2020-census-results.html).

Genetic variant classification

Within the UPHS dataset, where we had access to genetic testing results, we considered genetic test results positive if the variant(s) identified was classified as likely pathogenic/pathogenic by the testing lab (for autosomal-recessive conditions, two variants in the same gene were required with at least one classified as likely pathogenic/pathogenic) and was consistent with the subject's presentation as determined by the clinical testing team. Results were classified as variants of uncertain significance (VUS) if the lab reported only VUS(s). Genetic test reports that included only likely benign/benign variants or identified no causative variants were considered negative. It is important to note that a negative or VUS result does not necessarily indicate the absence of a genetic diagnosis, only that no obvious pathogenic variant could be identified by the testing lab. Additionally, it is important to note that there is much literature suggesting that, on account of the overwhelming bias toward samples from individuals of European ancestry in our reference datasets, minoritized individuals may be less likely to receive positive results on genetic testing.[10,11]

Definitions of race and ethnicity

Race and ethnicity of the study populations (rather than genetic ancestry/similarity) were used as predictor variables for regression analyses, as this metric is best suited to capture the complex societal and political factors that directly produce healthcare disparities in minoritized and underserved patient populations.[20] Race and ethnicity of the UPHS and MGB study populations were extracted as reported in the EHR and collapsed into one of seven categories: Hispanic/Latino (defined as individuals with an EHR-defined ethnicity of "Latino" or "Hispanic," regardless of EHR-identified race), White (defined as all individuals identified in the EHR as of "White" race and not of Hispanic or Latino ethnicity), Black or African American (defined as all individuals identified in the EHR as "Black," "African American," or "Black or African American" race and not of Hispanic or Latino ethnicity), Asian (defined as all individuals identified in the EHR as "Asian" or "East Indian" race and not of Hispanic or Latino ethnicity), American Indian or Alaska

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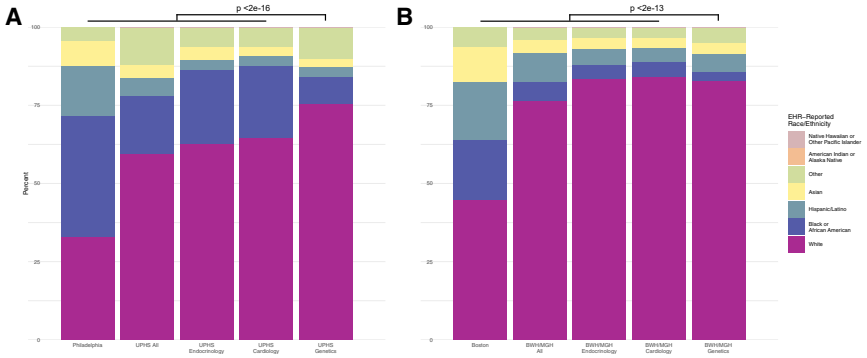

Figure 1. Racial disparities in adult individuals evaluated in the UPHS and BWH/MGH Adult Genetics Clinics

(A) The EHR-reported race and ethnicity of the population of the city of Philadelphia is shown (based on 2020 US Census data) compared to EHR-derived race and ethnicity for the entire UPHS patient population, and specifically the UPHS PCAM outpatient endocrinology, cardiology, and medical genetics clinics (for individuals seen from October 1, 2016 to October 1, 2021). Statistically significant differences were observed by logistic regression when comparing the proportion of Black individuals seen in genetics clinics to each other clinic group ($p < 2e{-}16$ by logistic regression).

(B) The EHR-reported race and ethnicity of the population of the city of Boston is shown (based on 2020 US Census data) compared to EHR-derived race and ethnicity for the entire MGB patient population, and specifically the MGB outpatient endocrinology, cardiology, and medical genetics clinics (for individuals seen from October 1, 2016 to October 1, 2021). Statistically significant differences were observed by logistic regression when comparing the proportion of Black individuals seen in genetics clinics to each other clinic group ($p < 2e{-}13$ by logistic regression).

Native (defined as all individuals identified in the EHR as "American Indian," "Alaska Native," or "American Indian or Alaska Native" race and not of Hispanic or Latino ethnicity), Native Hawaiian or Other Pacific Islander (defined as all individuals identified in the EHR as "Native Hawaiian," "Pacific Islander," or "Native Hawaiian or Other Pacific Islander" race and not of Hispanic or Latino ethnicity), and other/unknown (defined as all individuals identified in the EHR as "Other," "Some other race," "Declined," or "Unavailable" race, or any individual listing multiple races and not of Hispanic or Latino ethnicity). These data likely represent a combination of self-reported and clinician/staff-assigned identities. Prior studies suggest that, although inaccuracies exist, EHR-derived race and ethnicity is highly concordant with self-reported data.[24] Using this collapsing scheme, the UPHS Adult Genetics Clinic population included 4,107 individuals of White race/ethnicity, 399 of Black or African American race/ethnicity, 138 of Asian race/ethnicity, 101 of Hispanic/Latino race/ethnicity, 6 of American Indian or Alaska Native race/ethnicity, 5 of Native Hawaiian or Other Pacific Islander race/ethnicity, and 769 of other race/ethnicity (Table S1). The MGB Adult Genetics Clinic population included 7,562 individuals of White race/ethnicity, 273 of Black or African American race/ethnicity, 336 of Asian race/ethnicity, 525 of Hispanic/Latino race/ethnicity, 10 of American Indian or Alaska Native race/ethnicity, 4 of Native Hawaiian or Other Pacific Islander race/ethnicity, and 416 of other race/ethnicity (Table S2).

Statistical analysis

Statistical analyses were carried out by constructing linear/logistic regression models, adjusting for covariates as indicated in the figure and table legends. All analyses were performed with R software, version 4.2.2. For analyses specifically comparing the effects of race on genetics evaluation and testing outcomes, we first compared the relative representation of individuals of each race/ethnicity group seen in the Adult Genetics Clinic to all other clinical populations at both UPHS and MGB (Figure S1). The only cross-site, statistically consistent difference observed was in an over-representation of White and under-representation of Black or African American individuals in Adult Genetics Clinics at both sites, and we thus focused the remainder of our analyses on comparisons between these two groups, excluding all other individuals from analysis. Odds ratios (ORs), with corresponding confidence intervals, are reported for many analyses to quantify the relative odds of an outcome occurring in one group compared to another. An OR greater than 1 indicates increased odds in the group of interest, whereas an OR less than 1 indicates decreased odds. For example, an OR of 2.0 would indicate twice the odds of the outcome (e.g., a positive test result), while an OR of 0.5 would indicate half the odds. For continuous predictors (e.g., percentage of the population on Medicaid), regression coefficients were exponentiated to obtain ORs, which therefore represent the change in odds associated with a one-unit increase in the predictor (e.g., the change in odds of obtaining a positive genetic testing result associated with a 1% increase in the percentage of the population on Medicaid).

Results

Under-representation of Black individuals in Adult Genetics Clinic patient populations

We compared the relative representation of individuals of each race/ethnicity group seen in the Adult Genetics Clinic to all other clinical populations at both UPHS and MGB (Figure S1). The only cross-site, statistically consistent difference observed was in an over-representation of White and under-representation of Black or African American individuals (hereafter identified as "Black" for succinctness) in Adult Genetics Clinics at both sites; no other race/ethnicity showed a similarly significant and consistent effect, and we focused the remainder of our analyses on individuals of White and Black race/ethnicity. Only 8.9% of individuals seen in the UPHS Adult Genetics Clinic during the study time frame were identified in the EHR as Black, compared to 18.8% of the overall UPHS patient population (OR = 0.42 [0.40–0.45], $p < 1e{-}16$), 23.4% of the outpatient cardiology clinic population (OR = 0.32 [0.30–0.34], $p < 1e{-}16$), 23.7% of the outpatient endocrinology clinic population (OR = 0.31 [0.29–0.33], $p < 1e{-}16$), and 38.6% of the population of the City of Philadelphia (OR = 0.15 [0.15–0.16], $p < 1e{-}16$, Figure 1). These ORs suggest that Black individuals were about 58% less likely to be represented in the UPHS Adult Genetics Clinic patient population compared with the

overall UPHS population, about 68% less likely compared with cardiology and endocrinology outpatients and about 85% less likely compared with the City of Philadelphia population. These findings were replicated in MGB, where only 3.0% of adult individuals seen in the MGB Adult Genetics Clinic during the study time frame were identified in the EHR as Black, compared to 6.4% of the overall MGB patient population (OR = 0.45 [0.40–0.51], $p < 1e{-}16$), 5.1% of the outpatient cardiology clinic population (OR = 0.58 [0.51–0.65], $p < 1e{-}16$), 4.7% of the outpatient endocrinology clinic population (OR = 0.63 [0.56–0.71], $p = 1.8e{-}13$), and 19.1% of the population of the City of Boston (OR = 0.13 [0.12–0.15], $p < 1e{-}16$). As with the UPHS data, these ORs suggest that Black individuals were about 55% less likely to be represented in the MGB Adult Genetics Clinic compared with the overall MGB population, about 42%–37% less likely compared with cardiology and endocrinology outpatients, and about 87% less likely compared with the City of Boston population.

Socioeconomic differences between White and Black individuals in the UPHS and MGB Adult Genetics Clinics

Comparing demographic differences specifically between the White and Black individuals evaluated at either the UPHS or MGB Adult Genetics Clinic, no differences were observed in age or gender (Tables S1 and S2). However, significant differences were observed for health insurance payor and for five of the six census tract/neighborhood-level SDOH metrics that we examined (Tables S1 and S2). In both UPHS and MGB, Black individuals who presented to an Adult Genetics Clinic were from neighborhoods that were significantly more disadvantaged ($p = 3.6e{-}06$ to $5.6e{-}244$) based on measures including median household income and educational attainment.

Correlation between social determinants of health and likelihood of evaluation in Adult Genetics Clinics

To determine whether SES affects the likelihood of evaluation in an Adult Genetics Clinic, we determined the number of individuals evaluated in the UPHS and MGB Adult Genetics Clinics from each US Census tract per 1,000 of the total census tract population. Utilizing census-tract-level SDOH metrics, we performed linear regressions to find out whether any SDOH metrics were predictive of the likelihood of being evaluated (Figure 2). We performed our analyses separately for UPHS and MGB and subsequently performed a meta-analysis across both sites. Nominally significant and consistent positive associations were seen between evaluation in an Adult Genetics Clinic and median household income (Figure 2A; UPHS OR = 1.02 [1.02–1.03], $p < 1e{-}04$; MGB OR = 1.05 [1.04–1.06], $p < 1e{-}04$; meta-analysis OR = 1.03 [1.03–1.04], $p < 1e{-}04$) and the percentage of the population with a masters, professional, or doctoral degree (Figure 2B; UPHS OR = 1.01 [1.01–1.02], $p < 1e{-}04$; MGB OR = 1.03 [1.03–1.04], $p < 1e{-}04$; meta-analysis OR = 1.02 [1.01–1.02], $p < 1e{-}04$). These findings indicate that each $10,000 increase in neighborhood median household income was associated with a 2%–5% greater likelihood of evaluation, and that each 1% increase in the proportion of neighborhood residents with a graduate or professional degree was associated with a 1%–3% greater likelihood of evaluation in an Adult Genetics Clinic.

Nominally significant and consistent negative associations were seen between evaluation in Adult Genetics Clinics and the percentage of the population with less than a high school education (Figure 2C; UPHS OR = 0.99 [0.98–0.99], $p < 1e{-}04$; MGB OR = 0.98 [0.96–0.99], $p = 4.8e{-}03$; meta-analysis OR = 0.99 [0.98–0.99], $p < 1e{-}04$) and the percentage of the population with Medicaid (Figure 2F; UPHS OR = 0.99 [0.99–1.00], $p < 1e{-}04$; MGB OR = 0.99 [0.98–1.00], $p = 1.2e{-}02$; meta-analysis OR = 0.99 [0.99–1.00], $p < 1e{-}04$). These findings indicate that each 1% increase in either the proportion of neighborhood residents with less than a high school education or the proportion of neighborhood residents with Medicaid coverage was associated with a 1% lower likelihood of evaluation in an Adult Genetics Clinic.

Conflicting associations were seen between evaluation in an Adult Genetics Clinic and the percentage of limited-English-speaking households; these were negatively correlated in the Penn data and positively correlated in the MGB data (Figure 2D; UPHS OR = 0.99 [0.99–1.00], $p = 1.1e{-}03$; MGB OR = 1.09 [1.07–1.11], $p < 1e{-}04$; meta-analysis OR = 1.0 [0.99–1.00], $p = 0.69$). No significant association was seen between evaluation in an Adult Genetics Clinic and the percentage of the population with Medicare (Figure 2E).

A subgroup analysis considering only White individuals (Figure S2) found the same associations, suggesting that SDOH, independent of race, have significant associations with the likelihood of undergoing genetics evaluation in the general adult population.

Association of race and SDOH with outcomes of genetics evaluation and genetic testing

In the UPHS dataset, we had access to individual-level data on outcomes of Adult Genetics Clinic evaluation, including whether genetic testing had been sent, and results of genetic testing. For individuals evaluated in the UPHS Adult Genetics Clinic, Black individuals were significantly more likely than White individuals to have genetic testing sent following evaluation (OR = 1.35 [1.10–1.66], $p = 0.005$). This corresponds to Black individuals being about 35% more likely than White individuals to undergo genetic testing after evaluation. However, race was not significantly associated with any change in the likelihood of obtaining positive or VUS results on genetic testing (Figure 3A).

We next performed a secondary analysis of the association of neighborhood-level SDOH metrics with outcomes of genetics evaluation, including whether genetic testing

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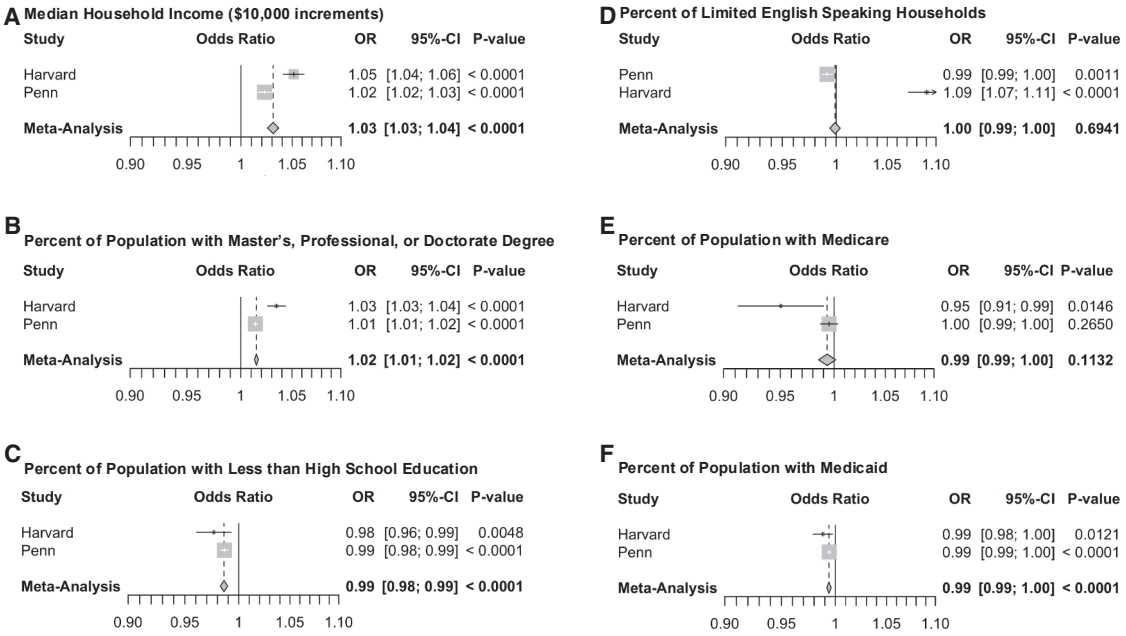

Figure 2. Socioeconomic disparities in adult individuals evaluated in the UPHS and MGB Adult Genetics Clinics

For each clinic site—UPHS (Penn) and MGB (Harvard)—the number of individuals evaluated in an Adult Genetics Clinic from each census tract, per 1,000 of the total census tract population, was taken as the outcome variable for linear regression with each of six different census-tract-level social determinant of health (SDOH) metrics as predictor variables: (A) median household income ($10,000 increments), (B) percentage of the population with master's professional or doctorate degree, (C) percentage of population with less than high school education, (D) percentage of limited-English-speaking households, (E) percentage of population with Medicare, and (F) percentage of population with Medicaid. For each SDOH metric, a forest plot is shown depicting the odds ratios (OR, indicated as a vertical tick), 95% confidence intervals (95% CI, indicated as a horizontal line), and unadjusted p values, derived from two-sided logistic regression. The size of the boxes represents the weight of each study in the meta-analysis, with larger boxes corresponding to studies with higher precision (i.e., smaller standard errors). The OR and CI results of meta-analysis are shown at the bottom of the plot as diamonds, summarizing the meta-analysis results under a fixed-effect model. A Bonferroni-corrected significance threshold of $p = 0.0083$ was used for the results of meta-analysis, accounting for six independent tests. Significant and consistent positive associations were seen between evaluation in an Adult Genetics Clinic and median household income (A) and the percentage of the population with master's professional or doctorate degree (B). Significant and consistent negative associations were seen between evaluation in an Adult Genetics Clinic and the percentage of the population with less than a high school education (C) and the percentage of the population with Medicaid (F). Conflicting associations were seen between evaluation in an Adult Genetics Clinic and the percentage of limited-English-speaking households (D); these were significantly negatively correlated in the Penn data and significantly positively correlated in the Harvard data. No significant association was seen between evaluation in an Adult Genetics Clinic and the percentage of the population with Medicare.

had been sent and the results of genetic testing. In the UPHS data, individuals were significantly more likely to obtain positive results of genetic testing if presenting from neighborhoods with a higher percentage of the population on Medicaid (OR = 1.01 [1.00–1.02], $p = 0.002$) or with less than a high school education (OR = 1.02 [1.01–1.04], $p = 0.001$). These findings indicate that each 1% increase in neighborhood Medicaid coverage or in the proportion of residents with less than a high school education was associated with a 1%–2% greater likelihood of a positive result. Patients were significantly less likely to receive a positive result if presenting from neighborhoods with higher median household incomes (OR = 0.96 [0.94–0.98], $p = 0.001$). This corresponds to about a 4% lower likelihood of a positive result per $10,000 increase in median household income. After adjusting for multiple hypothesis testing, no SDOH metrics were associated with the likelihood of genetic testing being sent or the likelihood of receiving a VUS result. A subgroup analysis considering only White individuals replicated these findings (Figure S3). These data suggest that race and SDOH have significant independent associations with outcomes of genetics evaluation, with an overall trend that, if evaluated, minoritized individuals are more likely to have genetic testing sent, and those presenting from socioeconomically disadvantaged neighborhoods are more likely to receive positive genetic testing results, potentially altering their care.

Association of race and SDOH with the likelihood of genetics evaluation for different clinical indications

We compared the association of race with the likelihood of evaluation in the UPHS Adult Genetics Clinic for 20 different specific clinical indication groups (Figure 4A and Table S3). After adjusting for multiple hypothesis testing, Black individuals were significantly less likely than White counterparts to be evaluated for lysosomal storage disorders (OR = 0.29 [0.13–0.64], $p = 2.2e{-}03$) and hypermobility spectrum disorders (OR = 0.32 [0.21–0.48], $p = 6.2e{-}08$), indicating that Black individuals

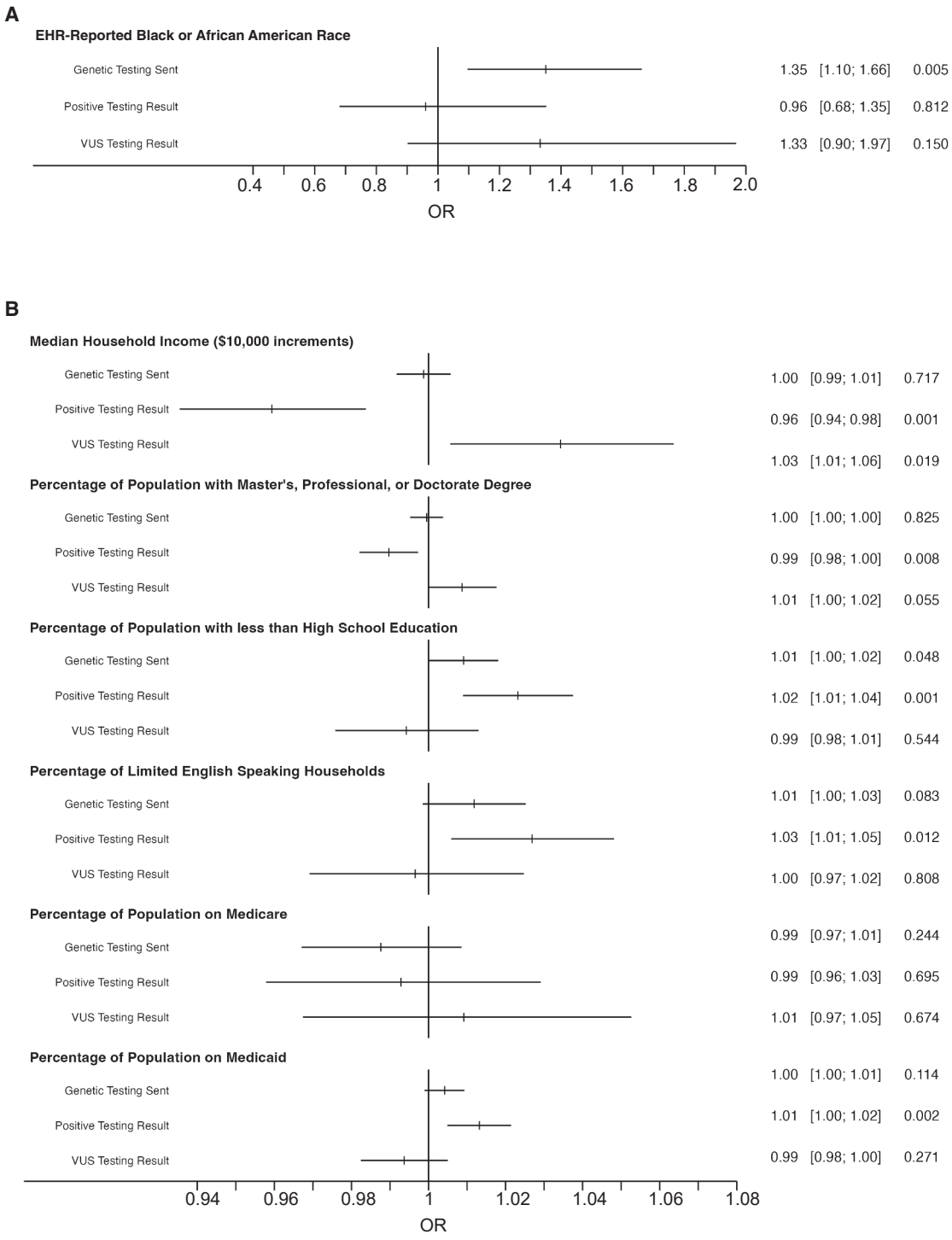

Figure 3. Forest plots illustrating the effects of race and SDOH on genetic testing outcomes for all individuals seen in the UPHS Adult Genetics Clinic

(A) Forest plots are shown illustrating the odds ratios (OR, indicated as a vertical tick), 95% confidence intervals (95% CI, indicated as a horizontal line), and with *p* values displayed, for the effects of EHR-reported Black race (derived from individual-level data) on three different genetics clinic evaluation outcomes (whether genetic testing was sent, whether genetic testing yielded a positive result, and whether genetic testing yielded a VUS). Regression models were adjusted for individuals' sex and age. A Bonferroni-adjusted significance threshold of 0.016 was used to account for three independent tests. A significant positive association was seen between EHR-reported Black race and the likelihood of genetic testing being sent.

(B) Forest plots are shown for the effects of six different SDOH metrics (derived from census-tract-level data) on three different genetics clinic evaluation outcomes as in (A). A Bonferroni-adjusted significance threshold of 0.0028 was used to account for 18 independent tests. Nominally significant positive associations were seen between median household income and the likelihood of receiving a VUS result, percentage of the population with less than a high school education and the likelihood of receiving a positive testing result, percentage of limited-English-speaking households and the likelihood of receiving a positive testing result, and percentage of the

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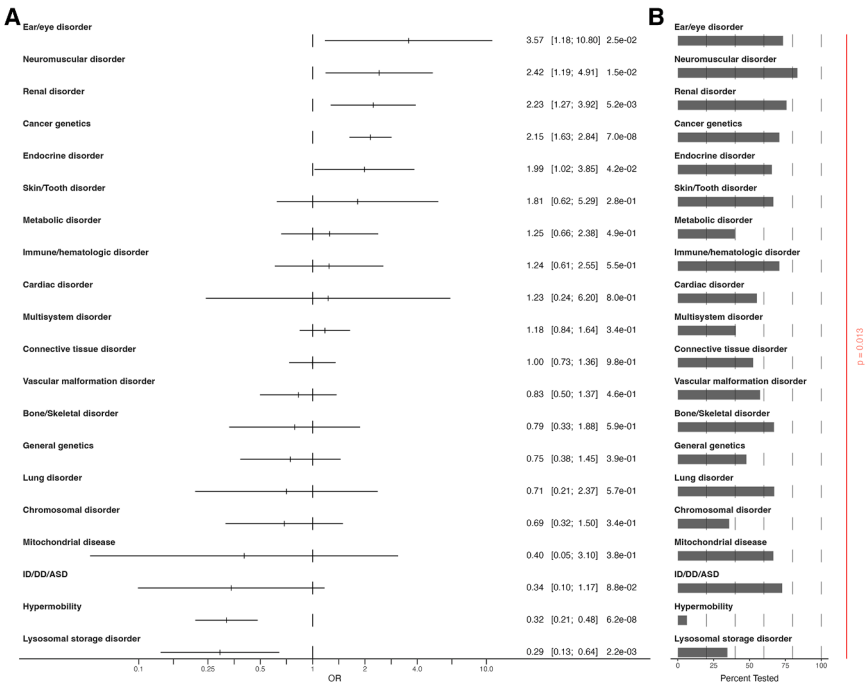

Figure 4. The effect of race on the likelihood of evaluation in the UPHS Adult Genetics Clinic for specific clinical indications

(A) Forest plots are shown illustrating the odds ratios (OR, indicated as a vertical tick), 95% confidence intervals (95% CI, indicated as a horizontal line), and with *p* values displayed, for the effects of EHR-reported Black race on the likelihood of evaluation in the UPHS Adult Genetics Clinic for each of 20 different clinical indications. Regression models are adjusted for sex, age, and all six SDOH metrics shown in Figure 2. A Bonferroni-adjusted significance threshold of 0.0025 was used to account for 20 independent tests. Black individuals are significantly more likely to be evaluated for the indications of cancer genetics, while they are significantly less likely to be evaluated for hypermobility and lysosomal storage disorders.

(B) The overall rate at which genetic testing was sent for each clinical indication across all individuals evaluated in the UPHS Adult Genetics Clinic from October 1, 2016 to October 1, 2021. There is a significant association ($p = 0.013$ by linear regression) between the overall rate at which genetic testing is sent for each specific clinical indication and the OR of evaluation for Black individuals for that same indication.

were about 70% less likely to be evaluated for lysosomal storage disorders and about 68% less likely to be evaluated for hypermobility spectrum disorders compared to White individuals. For intellectual disability/developmental delay/autism spectrum disorders and mitochondrial disease, both rare indications for evaluation, no significant differences in evaluation rate were identified. Nonetheless, the ORs were substantial in magnitude (OR = 0.34 [0.10–1.17] and OR = 0.40 [0.05–3.10], respectively), suggesting that Black individuals may be less likely to be evaluated for these important conditions, although the wide confidence intervals underscore the need for confirmation in larger datasets.

On the other hand, Black individuals were significantly more likely to be evaluated for cancer genetics (OR = 2.15 [1.63–2.84], $p = 7.0e{-}08$). This OR indicates that Black individuals were more than twice as likely to be evaluated for cancer genetics than White individuals. Additionally, there was a significant positive association between the OR of evaluation of Black individuals for a given indication and the overall likelihood of genetic testing being sent for that indication (Figure 4B, $p = 0.01$), suggesting that Black individuals were, in general, more likely to be evaluated for clinical indications that have clinical practice guidelines recommending genetic testing, such as cancer genetics.

A secondary analysis comparing the association of the same six SDOH metrics with the likelihood of evaluation for the same 20 clinical indications revealed several significant associations (Figure S4), with five associations surpassing the Bonferroni-adjusted significance threshold. Three of these five associations were for hypermobility spectrum disorders, where individuals with markers of higher SES were more likely to be evaluated in our clinic. Additionally, individuals coming from neighborhoods with a higher percentage of the population on Medicaid were more likely to be evaluated for multisystem disorders, while individuals coming from neighborhoods with a higher percentage of the population with an advanced degree were more likely to be evaluated for renal disorders.

Discussion

Despite initiatives over the last decade to improve equity in medical genetics evaluation and genetic testing, race- and SDOH-based barriers persist. Our results demonstrate that minoritized individuals (Figure 1) and individuals presenting from low-SES neighborhoods (Figure 2) are under-represented in our adult medical genetics patient populations. However, our data also show that, when evaluated, minoritized individuals are more likely to undergo genetic testing (Figure 3A) and that individuals from low-SES neighborhoods are more likely to receive a positive result (Figure 3B).

population on Medicaid and the likelihood of receiving a positive testing result. Nominally significant negative correlations were seen between median household income and the likelihood of receiving a positive test result, and percentage of the population with an advanced degree and the likelihood of receiving a positive test result.
Note that (A) and (B) utilize different horizontal-axis scales, as the magnitude of ORs observed in (A) are much larger than those in (B).

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The significant difference we observe in evaluation rates for minoritized individuals suggests the presence of systemic barriers that are specific to medical genetics. If general access factors, such as business hours, insurance, cost, or other opportunity debts, were the primary deterrents to adult genetics evaluation, we would expect similar underrepresentation in other adult subspecialty clinics within our health systems. However, the disparities we observe appear to be most pronounced in the Adult Genetics Clinics, suggesting unique barriers to evaluation in this field.

Several factors may contribute to this discrepancy. First, the influence of inherited disease on adult health remains under-recognized, both by affected individuals and by providers. Many adult-medicine-trained clinicians have significant knowledge gaps in medical genetics, particularly regarding non-traditional presentations of genetic conditions in minoritized populations.[25] Additionally, primary care providers often underestimate the importance of genetics evaluation in routine patient care, with those serving underserved populations being even less likely to refer individuals for genetics evaluation.[26] The absence of widely adopted clinical practice guidelines addressing genetic testing in adults further limits clinician exposure and utilization of genetic services. On the outpatient side, awareness of genetics is a strong predictor of seeking genetics care,[27] and minoritized and underserved individuals are known to be disproportionately unaware of the relevance of genetic testing to their health.[28] These differences in provider knowledge and patient awareness likely contribute to the disparities observed in our data. To empower clinicians and affected individuals to use existing educational resources that address genetic testing indications, genetic counseling availability, and the potential for a health-altering diagnosis, and to highlight initiatives to diversify and expand the genetics workforce,[29] we present a list of resources in Table 1 as a starting point and quick reference for affected individuals and providers to begin the process of dismantling these profound disparities.

Interestingly, despite a lower evaluation rate, we find that minoritized individuals are more likely to complete genetic testing once evaluated. This challenges race-based stereotypes about disinterest in genomics among Black and minoritized patient groups and aligns with recent studies showing that Black or Latino individuals highly value genetic testing.[23,30,31] Moreover, we find that Black individuals and those presenting from low-SES neighborhoods are not more likely to receive negative or uncertain genetic testing results, contrasting with studies in pediatric and select adult populations suggesting that genetic testing is more frequently non-diagnostic in minoritized individuals.[12,13,32]

In fact, we find that individuals from lower-SES neighborhoods are more likely to obtain positive genetic testing results, despite no evidence of higher rates of genetic conditions in these populations to explain this trend. Our data also reveal that minoritized individuals are more frequently evaluated for clinical indications involving a single organ system with well-recognized signs of genetic disease (e.g., retinal degeneration and cystic kidney disease) but are less likely to be evaluated for multisystemic conditions requiring specialized knowledge for diagnosis (e.g., intellectual disability and lysosomal storage disease). This suggests that minoritized individuals may be primarily referred based on objective signs consistent with a known genetic condition, making them more likely to undergo testing and receive a positive result. However, less obvious genetic diagnoses in this population may be overlooked by referring providers, highlighting the need for future studies to investigate the root causes of these disparities.

The increased likelihood of genetics evaluation for minoritized adults affected by cancer is a bright spot among our data. Despite a concerted effort over the last two decades to expand oncologic precision medicine for minoritized individuals with breast, ovarian, and colorectal cancer,[33–35] genetic testing for cancer predisposition syndromes is significantly less likely to be offered to individuals of minoritized ancestry.[36–38] Here, the data from our center contradict national data, likely due to multilevel educational interventions at UPHS specifically in the area of cancer genetics. These interventions have included embedding dedicated genetics physicians within oncology specialties across the health system, weekly multidisciplinary clinical conferences, and active genetics participation in tumor boards. Cancer genetics providers have also delivered grand rounds and educational sessions across a wide range of clinical departments and have developed reflex testing pipelines when somatic testing reveals possible germline variants. In parallel, the cancer genetics group has conducted quality improvement initiatives, pragmatic clinical trials, and mainstreaming of genetic testing at the point of care.[39–42] Together, these efforts have reinforced the importance of germline testing regardless of patient demographics and have likely contributed to the higher rates of evaluation for minoritized individuals seen in our data. Ongoing efforts at UPHS to replicate these outcomes outside of oncology include embedding genetic counselors in other subspecialty clinics, integrating genomic data and best practice advisories into our EHRs, and diversifying research recruitment.[40,43] Such approaches require buy-in from multiple stakeholders, but seem likely to make the largest difference in combating inequalities in genetics and precision medicine and could be replicated in other health systems.

Our datasets and analyses have several limitations. First, we are unable to distinguish between disparities in physician referral rate and disparities in patient follow-through with scheduling such that the source of the disparities in Adult Genetics Clinic evaluations cannot be pinpointed. Additionally, it is possible that our findings from two major academic health systems are not generalizable to other clinical practices, although our results likely represent a best-case scenario: the UPHS and MGB Adult Genetics

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Table 1. Selected educational resources for non-genetics clinicians, affected individuals, and genetics workforce diversity

Resource	Description	Link
Clinician focused		
AMA/ACMG Genetics 101 Series for Healthcare Providers	CME-accredited modules for non-genetics providers. Includes general genetics topics and subspecialty-specific courses	https://www.acmgeducation.net/URL/Genetics101
Genome Ed	NHGRI-supported compendium of peer-reviewed genomics educational materials for clinicians	https://www.genome.gov/GenomeEd/resources
UTHealth Adult Cardiovascular Genomics Certificate Program	online, CME-accredited case-based modules on adult-onset inherited cardiac conditions for non-geneticist clinicians	https://uthealth.catalog.instructure.com/browse/ms/courses/acgcp
USPSTF recommendations	screening guideline for BRCA-related cancer risk published in 2019; guidelines in progress for Lynch syndrome-related cancer, hemochromatosis, and hereditary lipid disorders	https://www.uspreventiveservicestaskforce.org/uspstf/recommendation-topics
Society guidelines (selected examples)	ACOG Non-invasive Prenatal Testing	https://www.acog.org/advocacy/policy-priorities/non-invasive-prenatal-testing/current-acog-guidance
	NCCN Familial/High Risk: breast, ovarian, and pancreatic cancer	https://www.nccn.org/guidelines/guidelines-detail?category=2&id=1503
Podcasts	The Curbsiders, episode #139: Genetic testing in primary care	https://thecurbsiders.com/curbsiders-podcast/139-genetic-testing-in-primary-care
	The Beagle Has Landed: medical, ethical, and social implications in clinical genetics	https://beaglelanded.substack.com/
Genetic Counselor Directory	NSGC-supported directory to identify local or telehealth-based genetic counselor by areas of specialization	https://findageneticcounselor.nsgc.org/
Patient focused		
Hudson Alpha Information is Power	subsidized counseling and genetic testing for risk of breast, ovarian, prostate, and colorectal cancer via non-invasive buccal swab	https://www.hudsonalpha.org/information-is-power/
Genetics Journeys	individuals from URM communities describe their experience with genetic testing and the importance of diagnosis	https://nymacgenetics.org/dc/
Personal Genetics Education Program	inclusive workshop and lesson plans for community spaces, faith-based groups, and schools on the role of genetics in society	https://pged.org
Podcast	In Those Genes: presents genetics concepts through the lens of Black culture	https://inthosegenes.com
Workforce development		
Minority Genetics Professionals Network	mentoring and support group to increase diversity in the genetic workforce to improve health care equity	https://minoritygenetics.org
GOLDEN	initiative to increase awareness of genetic counseling career pathways among students at HBCUs and provide support through the application process	https://nymacgenetics.org/golden
ASHG Human Genetics Scholars Initiative	mentorship and financial support for career development to enhance diversity and inclusion in early-career human genetics researchers	https://www.ashg.org/membership/awards/hgsi
The Alliance for Genetic Counseling Fellowship	full 2-year scholarships and mentorship for 40 genetic counseling students across five institutions to increase diversity	https://agcfellowship.org/

AMA, American Medical Association; ACMG, American College of Medical Genetics; CME, Continuing Medical Education; NHGRI, National Human Genome Research Institute; USPSTF, United States Preventive Services Task Force; AHA, American Heart Association; ACOG, American College of Obstetricians and Gynecologists; NCCN, National Comprehensive Cancer Network; NSGC, National Society of Genetic Counselors; URM, under-represented minority; HBCU, historically Black colleges and universities; ASHG, American Society of Human Genetics.

Clinics are among the most established, longest-running programs among the eight adult-centered general genetics practices nationally. Geneticists are highly visible beyond the outpatient clinic, seeing inpatient consults and participating in trainee education. In contrast, most genetics divisions nationally are embedded in pediatrics departments and have limited adult outreach.[3,44] Disparities that exist in UPHS and MGB are likely to be magnified in other systems.

An additional limitation of our study is that our datasets were predominantly composed of individuals of White race, resulting in both statistical imbalance that may affect regression estimates and insufficient sample sizes to evaluate disparities among the least represented racial/ethnic groups. We chose not to downsample the White patient group, as this would have reduced statistical power and discarded valuable data; however, the resulting imbalance remains a limitation of our analysis. Furthermore, many individuals at both study sites were identified as "other" race or ethnicity; this group likely represents a highly heterogeneous population of individuals, highlighting some of the limitations of EHR-derived race and ethnicity data, which misses much of the nuance of these axes of personal identity. Lastly, some of our analyses relied heavily on census-tract-level SDOH data, which may be erroneously assigned by home-address-based geocoding (the methodology used here), which is an imperfect proxy for individual-level metrics of SES and which may be subject to confounding and bias. This may partially explain the modest OR we observed when comparing evaluation/testing outcomes by census-tract-level SDOH metrics. Additionally, while most analyses of SDOH yielded highly concordant results between UPHS and MGB, the census-tract-level percentage of limited-English-speaking households showed divergent associations between the two sites, highlighting another important limitation of using broad metrics of SDOH; the category "limited-English-speaking households" likely encompasses highly heterogeneous populations, and the differences observed may reflect underlying variation in the demographic and socioeconomic characteristics of non-English-speaking communities in each city. Future studies utilizing individual-level SES metrics are needed to more critically analyze the effects of SDOH on access to genetics evaluation and testing.

Genomic and precision medicine are transforming healthcare but, without deliberate intervention, these advances will only widen existing disparities, deepening race- and SES-based gaps in outcomes. The severe shortage of Medical Genetics-trained practitioners—particularly those serving adult individuals—is unlikely to improve in the next decade, making it imperative to explore alternative solutions. Expanding provider education, implementing targeted patient outreach, and integrating genetics into routine care through innovative approaches, such as EHR-driven automation to identify individuals who may benefit from genetic testing, as is being implemented at UPHS,[45] will be essential to ensure equitable access. Our findings underscore the urgent need to dismantle barriers to genetic evaluation and testing, particularly for minoritized individuals and those of lower socioeconomic standing. Achieving true equity in precision medicine requires not only recognizing these disparities but actively working to correct them.

Data and code availability

The summary data supporting the findings of this study are available upon request. Individual-level data, aside from what has been included in the paper and supplemental information, cannot be shared due to individuals' privacy concerns.

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Author contributions

J.I.G. and T.G.D. conceived and planned the experimental design and analysis; J.I.G., C.K., A.V., H.S., and T.G.D. carried out the analyses; J.I.G., Y.E., N.B.G., S.A., A.R., C.C., Z.B., I.E., L.H., E.K., L.C.B., D.A.S., K.L.N., S.K., and T.G.D. contributed to data preparation and analysis; J.I.G., N.B.G., L.L., K.L.N., S.K., and T.G.D. contributed to interpretation of the results; T.G.D. supervised the project; and J.I.G. and T.G.D. took the lead in writing the manuscript. All authors provided critical feedback and helped shape the research, analysis, and manuscript.

Declaration of interests

The authors declare no competing interests.

Supplemental information

Supplemental information can be found online at https://doi.org/10.1016/j.ajhg.2025.11.010.

Web resources

ARHQ SDOH database 2020, https://www.ahrq.gov/sdoh/data-analytics/sdoh-data.html

US Census 2020, https://www.census.gov/programs-surveys/decennial-census/decade/2020/2020-census-results.html

For more resources, see Table 1

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