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## Supplemental Data

# Clinical Genetics Lacks Standard Definitions and Protocols for the Collection and Use of Diversity Measures

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## Supplemental Note

Survey Questions (All respondents)

1. Do you collect or use any information about population identity (E.g., population allele frequencies, self-reported race or ethnicity, and/or ancestral origin(s) of a patient or family members) in any aspect of your research or clinical practice? (Yes/No)
2. In your opinion, and based on your own understanding, how well does the term "biological group" describe each of the following? (Required) – [Race/Ethnicity/Ancestry]
3. In your opinion, and based on your own understanding, how well does the term "cultural group" describe each of the following? (Required) – [Race/Ethnicity/Ancestry]
4. In your opinion, and based on your own understanding, how well does the term "genetic lineage group" describe each of the following? (Required) – [Race/Ethnicity/Ancestry]
5. In your opinion, and based on your own understanding, how well does the term "lifestyle/behavioral group" describe each of the following? (Required) – [Race/Ethnicity/Ancestry]
6. In your opinion, and based on your own understanding, how well does the term "population group" describe each of the following? (Required) – [Race/Ethnicity/Ancestry]
7. In your opinion, and based on your own understanding, how well does the term "religious group" describe each of the following? (Required) – [Race/Ethnicity/Ancestry]
8. In your opinion, and based on your own understanding, how well does the term "social identity group" describe each of the following? (Required) – [Race/Ethnicity/Ancestry]
9. In your opinion, and based on your own understanding, how well does the term "species group" describe each of the following? (Required) – [Race/Ethnicity/Ancestry]
10. How often do you interpret or curate genetic variants in either a clinical or research setting? (Multiple choice)

The following questions were asked only to those who reportedly conduct variant interpretation:

11. How often do you order genetic tests or return results to patients in a clinical care setting? (Multiple choice)
12. For what purpose(s) do you typically interpret genetic variants? (Select all that apply)
13. When interpreting genetic variants, which of the following kind(s) of data are most likely to inform your interpretation? (Select all that apply / Free text option)
14. If information about race, ethnicity, and/or ancestry is used in clinical variant interpretation, from which of the following source(s) are these data most likely obtained? (Select all that apply / Free text option)
15. Which of the following resource(s) do you use frequently in the curation of genetic variants? (Select all that apply / Free text option)

16. Which of the following resources are you likely to use when applying population-level data to variant curation or interpretation? (Select all that apply / Free text option)

17. To your knowledge, what information about population-level data is typically included in reports from clinical testing laboratories? (Select all that apply)

18. How important are the following in the clinical interpretation of genetic variants? – [Race/Ethnicity/Ancestry]

    a. Since you indicated that "it depends" how important race, ethnicity, and/or ancestry are for clinical variant interpretation, please feel free to elaborate here. (Free text response)

19. In your opinion, what is the relevance of race, ethnicity, and/or ancestry information in the context of clinical variant curation or interpretation? (Multiple choice)

20. How often do you calculate genetic ancestry from a patient's DNA for the purpose of clinical interpretation? (Multiple choice)

    a. When genetic ancestry is calculated by you and/or your colleagues for the purpose of clinical interpretation, what method(s) are most often used?

    b. Which reference population panel(s) do you and/or your colleagues use when estimating genetic ancestry for clinical interpretation?

<u>The following questions were asked only to those who reportedly work in genomic medicine and clinical care, including ordering and interpreting genetic tests, reporting results to patients, etc.</u>:

21. How often do you receive estimates of a patient's genetic ancestry (calculated from DNA) in addition to carrier or diagnostic test results on a clinical lab report? (Multiple choice)

22. If information about a patients' race, ethnicity, or ancestry is used in any aspect of your work as a clinician, from where are these data likely obtained? (Select all that apply / Free text response)

23. For the purposes of ordering a genetic test, how important do you consider each of the following type(s) of data? (Likert scale of importance)

    a. Race of the patient
    b. Ethnicity of the patient
    c. Ancestry of the patient
    d. Geographic Origin(s) of the patient
    e. Disease prevalence in a of which the patient is a member

        i. Since you indicated that "it depends" how important certain identity or other measures are for ordering a genetic test, please feel free to explain here. (Free text response)

24. In your opinion, what is the relevance of race, ethnicity, and/or ancestry information in decision-making about ordering a genetic test? (Free text response)

25. Race, ethnicity, and/or ancestry may be relevant for obtaining consent from patients for a clinical diagnostic genetic test. (True/False)

26. Race, ethnicity, and/or ancestry may be relevant for contextualizing genetic test results in discussions with a patient. (True/False)

27. Race, ethnicity, and/or ancestry may be relevant for tailoring treatment options to patients. (True/False)

28. A positive test result would likely motivate me to discuss race, ethnicity, or ancestry (REA) for relevant traits and conditions. (True/False)

29. A negative test result would likely motivate me to discuss REA for relevant traits and conditions. (True/False)

30. If the patient has a variant of uncertain significance (VUS) in a gene of interest, I am more likely to discuss REA with them. (True/False)

31. If the patient is a member of an ethnic or racial minority population, I am more likely to discuss REA with them. (True/False)

32. Are there other factors (beyond those listed in the previous questions) that might motivate you to discuss race, ethnicity, and/or ancestry with a patient? (Yes/No)

    a. Since you indicated that there are other factors that might motivate you to discuss race, ethnicity, and/or ancestry with a patient, please list or describe them here.

33. In your opinion, are there particular traits or conditions for which you think it most important to consider race, ethnicity, and/or ancestry? (Yes/No)

34. How confident are you in describing the similarities and differences among race, ethnicity, and ancestry? (Multiple choice)

35. How confident are you in discussing how race, ethnicity, and ancestry are related to genomics and clinical care? (Multiple choice)

<u>The following questions were asked to all respondents:</u>

36. Do you believe that new guidelines would be helpful to guide the collection, use, and/or communication of race, ethnicity, and ancestry information in clinical genomics? (Multiple choice)

    a. Please feel free to elaborate on why you indicated "other" in response to the previous question about whether new guidelines would be helpful to guide the collection, use, and/or communication of race, ethnicity, and ancestry information in clinical genomics. (Free text response)

    b. For which of the following purposes do you agree that it would it be useful to develop standards or guidelines in clinical genomics? (Select all / Free text)

    c. Please feel free to elaborate on how standards or guidelines about the use of race, ethnicity, and/or ancestry would be helpful for researchers and professionals working in clinical genomics. We welcome any suggestions or comments.

    d. Why do you believe that no additional guidelines are needed for the use of race, ethnicity, and or ancestry information? (Select all that apply / Free text response)

    e. Since you indicated "other" reasons that there is no need for additional guidelines for the use of race, ethnicity, and/or ancestry information, please feel free to elaborate here.

37. Which of the following organizations or consortia are you a member of, or affiliated with? (Select all that apply)

38. Which of the following entities best represents your employment affiliation(s)? (Select all that apply / Free text response)

39. Please indicate the country in which your primary employer is located. (Multiple choice)

    a. Please indicate in which U.S. state your primary employer is located.

40. Please specify your role(s) with regard to clinical genomics. (Select all that apply / Free text response)

41. For how many years have you been working in this field?

    a. Clinical professionals only: Do you work primarily in Prenatal, Pediatric or Adult Medicine? (Select all that apply)

42. How do you personally identify on the basis of each of the following? (Fill in the blank with your own words to describe your identity.)
    a. Sex
    b. Gender
    c. Race
    d. Ethnicity
    e. Ancestry

43. The following choices are designated by the U.S. Office of Management and Budget (OMB) as the official racial categories used for reporting demographics in the federal government. Please check the box(es) that best correspond(s) to your personal identity.

44. Which of the two previous questions about your own personal identity would you prefer to be asked in the future?

    a. Why did you prefer to answer the open-ended identity question?

    b. Why did you prefer to answer the multiple-choice identity question?

45. Have you seen any version of these survey questions before (E.g., as part of the development or validation of the survey)? (Yes/No/I’m not sure/Free text response)

## Supplemental Data

**Table S1. Survey Dissemination and Response Rates.** The number of individuals solicited to conduct the survey is shown for each professional organization, consortium, and a manually curated list of clinical geneticists. The number of surveys opened through these solicitations is also shown, in addition to the number and percentage of those who completed the survey among those who opened it, and the overall response rates. Response rates are adjusted to account for overlap in organizational membership among survey respondents (starred).

| Dissemination Approaches | Individuals Emailed (N) (Adjusted*) | Opened Survey (N) | Completed Survey N (%) | Response Rates (%) |
|---|---|---|---|---|
| Total numbers | 11930 | 527 | 448 | 4.2% |
| American Board of Genetic Counselors (ABGC) | 4661 (4633*) | 195 | 122 (27.2%) | 2.6% |
| American College of Medical Genetics and Genomics (ACMG) | 2218 (2118*) | 3 | 1 (0.2%) | 0.05% |
| American Society of Human Genetics (ASHG) | 2659 | 98 | 75 (16.7%) | 2.8% |
| The Clinical Genome Resource (ClinGen) | 788 (743*) | 138 | 98 (21.9%) | 12.4% |
| Clinical Sequencing Evidence-generating Research (CSER) | 184 (161*) | 56 | 41 (9.2%) | 22.3% |
| Clinical Geneticists Manually Identified Online by State | 638 | 66 | 38 (8.5%) | 5.9% |
| Snowball Approach (Social Media, Personal Correspondence) | N/A | 137 | 73 (16.3%) | N/A |