

# Improving the Use of Race and Ethnicity in Genetic Research: A Survey of Instructions to Authors in Genetics Journals

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## Abstract

To explore the guidance provided by genetics journals on the use of race and ethnicity, this survey examined the instructions for authors and recent content of 120 journals indexed under “Genetics and Heredity” within the Web of Science. Only two of the journals had instructions for authors that directly referred to race or ethnicity, and both implicitly validated the genetic utility of ethnicity by encouraging its use to identify or classify genetic variation. The instructions for authors of five other journals referred to the American Medical Association’s *Manual of Style* or the “Uniform Requirements for Manuscripts Submitted to Biomedical Journals” developed by the International Committee of Medical Journal Editors (ICMJE); one more journal was listed on the ICMJE Web site. Both those more generic style guides had dedicated sections advising prospective authors to “justify” and “define” or “explain” their use of race and ethnicity. However, it is unclear whether any of the six genetics journals linked to either of the style guides actually intended authors to comply with their particular sections on race and ethnicity, particularly inasmuch as the instructions of three of the journals referred only to the sections of

the style guides related to the formatting of references. Despite the paucity of guidance on race and ethnicity, from 1994 to 2004 55.8% of the journals had published empirical articles using “racial”, “ethnic”, or related census categories. In the absence of dedicated guidelines, the responsibility rests with journal editors to draw the attention of authors and reviewers to longstanding concerns over the reliability, validity, and sensitivity of using race and ethnicity in genetic research and to encourage critical reflection and debate and better standards of measurement and reporting.

## Introduction

Debates about whether racial and ethnic categories should be used in genetic research have peppered the genetic, biomedical, and anthropologic literature for over a half-century. Recently, the debate has hinged on two sets of seemingly contradictory studies. The first have demonstrated that there is greater genetic variation within racial or ethnic groups than between them<sup>1,2</sup> and that traditional “racial” categories are associated with less than 10% of human genetic variation (perhaps as little as 5%). The second have shown that genetic researchers are often able to predict the racial or ethnic affiliation of people by using an array of genetic markers even though racial and ethnic categories do not reflect genetically distinct populations.<sup>3,4</sup> Thus, although it has been popular to claim that “there’s no such thing as race”<sup>5</sup> and to emphasize that “in genetic terms, all human beings, regardless of race, are more than 99.9 percent the same”,<sup>6</sup> most geneticists accept that modest differences exist in the frequency of some genetic traits among racial and ethnic groups, however these groups are categorized. The debate about the use of

race and ethnicity in genetic research has therefore moved on<sup>7</sup> to explore the extent to which racial and ethnic categories might provide reliable, valid, and sensitive markers for these modest “collective genetic affinities”<sup>8</sup> and how useful such markers might be.<sup>9</sup>

Some critics of this more pragmatic approach have pointed out that racial and ethnic categories are fluid dimensions of social identity that defy definition or measurement and that vary over time and place.<sup>10</sup> Others have argued that using race and ethnicity as proxies for genetic variation legitimizes the discredited view that racial and ethnic categories are genetically *determined* and reflect substantive, rather than minor, genetic differences.<sup>11</sup> Indeed, both race and ethnicity are socially constructed,<sup>12</sup> and their incidental association with modest differences in the frequency of some genetic traits seems unlikely to explain much of the variance in biology or behavior observed among racial or ethnic groups. There is therefore increasing concern that using race or ethnicity in any scientific research is politically inflammatory because it might suggest that race and ethnicity are essential (if not “natural”) determinants of biologic or social phenomena rather than components of the sociocultural and structural frameworks (which include “gender” and “class”) through which biologic and social differences are recognized and produced. For those reasons, the reliability, validity, and sensitivity of using race and ethnicity as proxies for genetic variation remain highly contested; for geneticists, much of the ensuing debate has focused on whether, when, and how race and ethnicity should be used as research variables.<sup>13,14</sup>

The debate has spilled over onto the

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editorial pages of genetics and biomedical journals, and editors are increasingly concerned with what advice or guidelines they should provide to authors and reviewers.<sup>15-21</sup> For example, before its recent (2004) special issue on the genetics of race and ethnicity, "Genetics for the Human Race", *Nature Genetics* had published three editorials that tried to alert researchers to the potential pitfalls of using race and ethnicity. The first, "Census, Race and Science",<sup>22</sup> used changes in how racial and ethnic categories were classified in the 2000 US Census to show how the classifications are sociopolitically derived and subject to change. The second, "Genes, Drugs, and Race",<sup>23</sup> reflected on a pharmacogenetic study published in the same issue of the journal that concluded that "commonly used ethnic labels are both insufficient and inaccurate representations of the inferred genetic clusters".<sup>24</sup> Finally, "The unexamined 'Caucasian'"<sup>25</sup> reviewed the use of racial and ethnic categories in *Nature Genetics* and reported that only 36.7% (22 of 60) of papers that "included descriptions of geographic or ethnic groups of people" had conformed to the recommendations made in the first *Nature Genetics* editorial that advised authors to "explain why they make use of particular ethnic groups or populations, and how the classification was achieved".<sup>22</sup> Although there clearly is room for improvement, the editorials appear to have encouraged authors to adopt and report more specific methods to facilitate scientific reproducibility<sup>26</sup> and to develop a more consistent style in keeping with a scientific journal, particularly with respect to terminology and nomenclature.<sup>22,27</sup>

The aims of our survey were, first, to establish what guidelines are offered to authors, reviewers, and editors regarding the use of race and ethnicity in genetics journal articles and, second, to analyze the content of any available guidelines. The survey sought to assess what effect concerns about the use of race and ethnicity have had on the instructions for authors or style guides provided, or referred to, by genetics journals.

## Methods

The genetics journals examined in this survey were those included in the "Genetics and Heredity" "subject category" of the Web of Science's Journal Citation Reports. Each journal's Web site was located and searched to identify its instructions for authors. We recorded any sections that referred to racial or ethnic categorization and references to any generic style guides containing dedicated sections on race or ethnicity, such as the "Uniform Requirements for Biomedical Journals" published by the International Committee of Medical Journal Editors (ICMJE)<sup>28</sup> and the *Manual of Style* published by the American Medical Association (AMA).<sup>29</sup> To assess whether the availability of guidelines on race or ethnicity was confined to journals that published research using racial or ethnic categories, we used the Web of Science to search the abstracts of articles describing empirical research that had been published by "Genetics and Heredity" journals from 1994 to 2004, using the terms *race*, *racial*, *ethnic*, and *ethnicity* and the racial and ethnic categories used in the last two US and UK Censuses (see Ellison, Table II<sup>9</sup>).

## Results

A total of 120 journals were listed under the category "Genetics and Heredity", for which instructions for authors were located online for 108. Of those, only two had instructions for authors that referred directly to race or ethnicity (*Clinical Dysmorphology* and *Human Mutation*); both appeared to encourage authors to use ethnic categories when exploring or reporting unusual and noteworthy genetic variations between human populations:

Priority will be given to distinct, previously undescribed conditions and cases confirming these entities and to rare findings and ethnic differences in existing syndromes. [*Clinical Dysmorphology*]

Mutations in Brief (MIB) . . . [for this type of article] Report robust

population studies of known and novel mutations in ethnic groups in conjunction with genetic databases. [*Human Mutation*]

That advice was very different from the guidance provided by the ICMJE's "Uniform Requirements" or the AMA's *Manual of Style*, both of which encouraged authors to "justify" and "define" or "explain" their use of race and ethnicity:

When authors use variables such as race and ethnicity, they should define how they measured the variables and justify their relevance. [ICMJE<sup>28</sup>]

Racial categories should not be used automatically. Authors should explain and justify racial designators used, perhaps in the methods section of the manuscript. Any such terms should be used accurately. [AMA<sup>29</sup>]

However, it is not clear how many genetics journals expected prospective authors to comply with either the ICMJE's or the AMA's specific guidance on race and ethnicity. A search of the 601 journals listed on the ICMJE Web site found one genetics journal whose instructions for authors contained no reference to race or ethnicity or the ICMJE (*Journal of Assisted Reproduction and Genetics*); that suggests that the ICMJE's "Uniform Requirements" were meant to be applied by the editorial staff rather than by authors (or that the journal no longer subscribed to the ICMJE). And although the instructions of four other journals (*Human Heredity*, *Journal of Medical Genetics*, *Genetics in Medicine*, and *Psychiatric Genetics*) referred to the ICMJE's "Uniform Requirements", the first and second of these did so only for specific reasons unrelated to race and ethnicity (for formatting references). Likewise, both journals that referred to the AMA's *Manual of Style* (*Genetics in Medicine* and *Cancer Gene Therapy*) did so specifically for formatting references. Therefore, only two journals' instructions for authors referred to either of the generic

style guides in a way that did not exclude their sections on race and ethnicity:

These instructions comply with those formulated by the International Committee of Medical Journal Editors. For further details, authors should consult the following article: International Committee of Medical Journal Editors. "Uniform Requirements for Manuscripts Submitted to Biomedical Journals." *N Engl J Med* 1997;336:309-315. The complete document appears at [www.icmje.org](http://www.icmje.org). [*Psychiatric Genetics*]

The submission requirements of *Genetics in Medicine* conform to those presented by the International Committee of Medical Journal Editors in "Uniform Requirements for Manuscripts Submitted to Biomedical Journals". [*Genetics in Medicine*]

Although it cannot be assumed that either of those journals intended for prospective authors to comply with the ICMJE's specific guidance on race and ethnicity, there was no indication that authors should ignore the section of the "Uniform Requirements". It therefore seems fair to assume that authors might have felt obliged to comply with them and that a total of four (of the 108) genetics journals directly or indirectly offered guidance—albeit very different guidance—on race and ethnicity.

Notwithstanding the paucity of guidance on race and ethnicity, more than half the journals (67 of 120, 55.8%) had published articles describing empirical research using racial or ethnic categories during the preceding 10 years. Indeed, assuming that guidelines on race and ethnicity would be relevant only for those 67 journals makes only a slight improvement in the proportion offering guidance on race and ethnicity (four of 67, 6.0%).

### Discussion

Despite concerns about the reliability, validity, and sensitivity of race and ethnicity as measures of "collective genetic affinity",<sup>8</sup> our survey found that most genetics

journals have yet to develop their own guidelines for improving how racial and ethnic categories are used or reported. In the two instances where ethnicity was specifically mentioned in a journal's instructions for authors, no guidance was given on how it should be measured or reported. Instead, the instructions implicitly validated the genetic utility of ethnicity by encouraging its use to identify or classify genetic variation. That is very different from the guidance provided by the ICMJE's (2004) "Uniform Requirements" and the AMA's *Manual of Style*, which advised authors to "justify" their use of racial and ethnic categories and to "define" or "explain" how they were applied. However, it remains unclear whether either of the genetics journals whose instructions referred more generally to the "Uniform Requirements" or the other genetics journal listed on the ICMJE Web site intended prospective authors to follow all aspects of the "Uniform Requirements" (including the section related to race and ethnicity). If they all did, it at least means that a small number of genetics journals (three of 108) referred prospective authors to advice intended to improve rather than simply promote the use of ethnicity. If not, it means that none of the journals did. Either way, it seems clear that the sorts of concerns voiced by editorials and commentaries in *Nature Genetics* and by contributors to its recent special issue have not resulted in the development of dedicated guidance for genetics journals.

That is extraordinary, given that over half (55.8%) the genetics journals surveyed had recently published empirical articles using "race", "ethnicity", and related census categories. However, our interviews with genetics journal editors suggest that many felt ill-equipped to tackle the issues involved and were reluctant to develop or enforce guidelines without the support of the genetics research community<sup>20,21</sup>—an issue that Virginia Barbour raised at a recent CSE meeting.<sup>30</sup> Some were unconvinced that guidelines could or should be developed and felt they might undermine or constrain the role of peer review and academic debate in refining how race and

ethnicity are used. Their skepticism seems entirely reasonable, given the minimal effect that such guidelines have had on the use of race and ethnicity in articles published in the *British Medical Journal*,<sup>31</sup> although there is evidence that journal guidelines on other topics (such as criteria for authorship<sup>32</sup> and the reporting of controlled trials<sup>30</sup>) can be effective. Nonetheless, concerns remain about the reliability, validity, and sensitivity of race and ethnicity in genetic and related biomedical research,<sup>8</sup> and journal editors have a responsibility to draw these concerns to the attention of authors and reviewers and to encourage critical reflection and debate and better standards of measurement and reporting.<sup>33</sup>

There can be little doubt that the disaggregation of genetic findings with ill-defined, imprecisely measured, inappropriately labeled, and inadequately reported racial and ethnic categories undermines our understanding of population genetics and the translation of such findings to clinical and social contexts. Moreover, because all research findings are open to misinterpretation by scientific, clinical, and lay audiences alike, the unsystematic use of racial and ethnic categories in genetic research increases the risk of reifying unfounded beliefs in both the meaning and the scale of genetic differences between populations—beliefs that undermine the fabric of society and threaten to discredit genetic research, if only by association.<sup>34</sup> Genetics journal editors seem well placed to address these issues, particularly as gatekeepers of how research findings are described and presented. Given the high profile afforded this issue in the scientific and popular press,<sup>35,36</sup> the time seems ripe for editors to discuss it and to map out a unified strategy to improve, support, and safeguard legitimate research into genetic variation within and between human populations.

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