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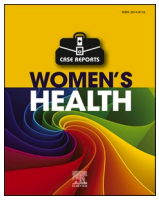

Invited Editorial

Terminological use of 'African ancestry' Vis-à-Vis 'black race' in relation to genetically linked healthcare conditions

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The terminological use of population descriptors related to ancestry, ethnicity and race in research concerning genetically linked health conditions, including case reports, has a long, convoluted history and is prone to controversy. This may be due to the lack of standardised definitions for these terms, resulting in misuse and confusion in their application [1]. Genetic studies attempt to provide a scientific basis for the trans-ancestry genetic variation among populations that influence the aetiological description, incidence and prevalence rates of disease, as well as the efficacy and safety of treatments.

Social scientists assert that race is both a social construct, categorising groups of people according to a small subset of phenotypes, and a political construct, where categories can be based on self-designation, skin colour, citizenship, geographic origin or home language [2]. The use of the term 'race' may have harmful consequences, as it reinforces the false belief that race categories are an appropriate proxy for biological differences, including genetic differences, and perpetuates 'race science' [1]. Owing to the challenges associated with using 'race' in health research, some researchers favour the term 'ethnicity', which refers to commonality in social practices, traditions, language and geopolitical factors [3]. Since ethnicity is not a static biologic variable, its categorisation differs across locations. For instance, in the United Kingdom (UK) the ethnic group 'Asian' comprises individuals from the Indian subcontinent [4], whilst in the United States of America the term is used to describe people from East Asia [4]. Therefore, delineating ethnic groups to understand health disparities is a complex undertaking. To address this complexity, the National Health Service (NHS) in the UK utilises standardised, harmonised ethnic monitoring questions and specific response codes (A – Z and unknown coded as 99) as outlined in the NHS data dictionary, which aligns with the 10-yearly census conducted by the Office for National Statistics (ONS) [5].

Although race and/or ethnicity could serve as appropriate markers for certain socio-economic determinants of health, including housing, education and/or income, they are poor predictors of genetic ancestry. Incorporating race and ethnicity data into case reports without acknowledging its socially constructed nature and without discussing its relevance to treatment and disease prevalence is likely to perpetuate stereotypes and reinforce existing implicit biases. Thus, 'ancestry' is the preferred objective population descriptor in the genetic context. In practice, however, 'ancestry' is ambiguous and lacks a standardised definition [6], which means that the use of the term alone should be avoided and always qualified, such as 'genealogical ancestry', 'genetic ancestry' or 'geographic ancestry', to enhance an understanding of the processes and dynamics underlying health outcomes [1,6]. Genetic ancestry identifies the ancestors from whom an individual is biologically descended [1,6], while genealogical ancestry refers to a family pedigree [1]. As offspring in each generation inherit half of their deoxyribonucleic acid (DNA) from each parent, an individual's genetic ancestry comprises a small fraction of all their genealogical ancestors. Finally, geographic ancestry refers to genetically related individuals who originate from a similar geographic region [1]. Therefore, accurately defining sub-populations with shared genetic ancestry will enable a better understanding of disease susceptibilities, therapeutic outcomes and health disparities, as in pre-eclampsia [7].

Scientists agree that the *Homo sapiens* lineage of anatomically modern humans originated in Africa ~300,000 years ago [8], with people of African ancestry manifesting diverse phenotypes and a unique genetic structure. As African genomes become increasingly available, their analyses have revealed that ancient divergence events occurred within African populations and that they began to display population structure, i.e., population subdivision, migration, and subsequent admixture, ~200,000 years ago. Furthermore, African ancestry is largely subdivided by geography and language, with a notable discontinuity in genetic terms between North Africa and sub-Saharan Africa warranting an independent review [8]. In addition, evolutionary forces, such as founder effect [9], genetic drift and natural selection [10], have ensured that while the frequency of genetic variants and haplotypes vary

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worldwide, researchers can infer approximate geographic origins based on similarities in certain subsets of genome [11]. However, human population studies have also indicated that there is greater genetic variation within, rather than between racial groups [2], and that ancestral admixture is a confounding factor in genetic ancestry that must be considered when interpreting genetic results [11]. Moreover, African populations have evolved genetic adaptations in response to the diverse climates, infectious agents and diets that are unique to the African continent [8]. Thus, the term 'black race' offers an insufficient description when considering the biological and genetic complexity ensconced in the African continent.

The value of using the term African ancestry as opposed to black race, given its direct link to health disparities in genomic medicine, is well described, with individuals of African ancestry both underutilising genetic counselling and testing services and being underrepresented in genetic studies [12]. Furthermore, this underrepresentation may result in an inadequate understanding of the role of genetic variants in disease outcomes as health disparities are not only influenced by environmental and sociocultural factors but also by biological and genetic factors. In tandem, the extent to which the findings from genetic studies conducted in one population can be applied to another is restricted by differences in linkage disequilibrium and allele frequencies [8]. Thus, developing ancestry-specific hereditary disease panels using the appropriate population subgroups will improve the accuracy of the data and the reproducibility of results. Furthering this agenda are the indispensable efforts of the Human Heredity and Health in Africa (H3Africa) population study and the Southern African Human Genome Programme pilot study [8]. Undertakings to capture the genetic diversity across the continent, including whole-genome sequence data from previously under-studied African populations, is necessary to enrich this repository of human variants [8].

In conclusion, amending the misuse of terminology is critical, as race, ethnicity and ancestry have a complex and interlinked relationship that is nuanced. Genetics does not supersede other factors, such as socioeconomic status and environment, but rather is ancillary to them. Thus, healthcare research, including case reports, require appropriate methods of investigation that consider race/ethnicity information, while accounting for or recognising the differences in ancestral genetic structure, gene-gene and gene-environment interactions.

Contributors

Habiba Ishmail contributed to conception of the editorial, drafting the manuscript, undertaking the literature review and revising the article critically for important intellectual content.

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