Race/Ethnicity in biomedical research and clinical practice

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ABSTRACT

There is ongoing debate as to whether persons of different racial/ethnic groups are biologically significantly different, and, if such differences exist, whether they are relevant in relation to disease susceptibility and to treatment outcomes. There is also debate about the benefits of using race/ethnicity as a factor in clinical decision making, and as a variable in biomedical or public health research, because of the emotional sensitivities attached to race/ethnic categorisation. Such categorisation may also divert attention from underlying issues such as socioeconomic status and lack of access to modern health care. In this short article we will discuss these controversies, and will emphasize the importance of responsible and sensitive use of race/ethnicity as a variable in biomedical research and in clinical practice.

Key words: race, ethnicity, biomedical research, BRCA, Tay-Sachs.

INTRODUCTION

The term race refers to a group of genetically related persons who share certain physical characteristics such as skin colour and facial features, and who have for a long time been isolated geographically, or have in common cultural or religious practices.[1,2] On the other hand, the term ethnicity, which has been found to be more socially acceptable, applies to a group of people of the same nationality, language or culture, and who may or may not have genetic markers in common.[1,2]

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There are two ways of considering race. The first is in terms of anthropological classifications of negroid, caucasoid etc; and the second is based either on overt physical characteristics or, as in the United States, on self racial-identification.[3] Almost universally throughout the world, this second system of classification is applied.

As there is no clear distinction between race and ethnicity, many researchers no longer distinguish between these, but rather use the single term race/ethnicity.[1,2] Analysis of genetic markers show that many people possess a high degree of genetic admixture, with the majority of self-reported blacks (African Americans) in the United States being in fact of mixed racial origin.[4] Thus, from a genetic point of view, categorizing persons by physical features into racial/ethnic groups is imprecise, arbitrary and subjective, and only genotyping tests can assign persons to genetic subgroups.[2,5]

The biological concept of race/ethnicity is emotionally charged because throughout history it has been used by political and social forces to discriminate between humans on the basis of skin colour, customs or religion. Therefore it is claimed that using race/ethnicity in biomedical research and in clinical practice is unwarranted because it may lead to stereotyping and to preconceived judgements, and to inequalities in medical care.[2,4,6,7]

However, others consider race/ethnicity to be informative in relation to the genetic make-up since individuals assigned to different racial/ethnic groups largely differ in allele frequencies at a variety of loci. This can provide valuable information about racial/ethnic risk factors for disease susceptibility and for adverse treatment outcomes.[3] This information may aid in developing preventive treatment strategies.[8]

Racial/ethnic groups not infrequently differ from one another in relation to socioeconomic status, education and access to good quality health care, which are well known factors influencing the incidence and outcomes of treatment of disease. Nevertheless, racial/ethnic differences in incidence of disease and response to treatment may sometimes remain after socioeconomic status and access to health care have been removed from the equation. This strongly suggests that race/ethnic-specific genetic factors, environmental factors, or a combination of both, do indeed play a role in susceptibility to disease and in response to medication.

Some points illustrating the limitations of using self-reported race/ethnic categorisation in biomedical research and clinical practice.

- Self-reported self-identification of race/ethnicity is problematic because of the high degree of genetic admixture of members of the same race/ethnic group. Therefore race/ethnicity should be viewed as a social construct and not as a scientific, genetically-derived objective category.[5]
- There are as many genetic variations within members of the same racial/ethnic group as there are between members of different racial/ethnic groups and there is a 99% commonality of DNA sequences among all humans.[4,9,10] Evidently therefore, race/ethnicity is not a rational basis for categorisation of humans.[4,9]
- The use of racial/ethnic categories in biomedical research seeks to validate the controversial genetically-driven biological concept of race/ethnicity, thus diverting the attention from more important issues such as socio-economic conditions, education and access to health care which are critical factors in health status of different racial/ethnic groups.[4]
- In clinical practice, overemphasis on the role of race/ethnicity in determining disease susceptibility and in selecting drug therapies puts patients at risk of receiving health care based on their presumed racial/ethnic biological characteristics. Clearly this can be detrimental because such race/ethnicity biased clinical judgements may result in more important clinical factors being ignored.[4, 11]
- According to Schwartz, both the development of disease and the outcome of drug therapy are complex multifactorial processes which include interactions between disease-susceptibility genes, genetic variants of drug metabolizing enzymes, the age of the patient, environmental factors and other undetermined genetic factors, and therefore can seldom if ever be attributed solely to genetic racial/ethnic differences.[5] Furthermore, general health, life-style habits, socioeconomic status and education can also indirectly influence susceptibility to disease and treatment outcome.[4,12]

Table 1: Some diseases/conditions which are more prevalent in particular racial/ethnic groups.

Disease/condition	Racial/ethnic groups
Tay-Sachs disease [5,15]	Ashkenazi Jews in association with Hex A mutation
Type 2 Diabetes [8,16]	Pima Indians in the United States and Mexico • susceptibility genes • environmental factors • life-style factors
Systemic lupus erythematosus[4]	Young black women • susceptibility genes
Breast Cancer[5]	Ashkenazi Jewish women in association with BRCA 1 germline mutation
Sickle cell diseases [17]	Persons of West African ancestry in association with globin S (β^s) mutation
Haemochromatosis[18]	Whites of Northern European descent in association with C282Y mutation
Pulmonary cystic fibrosis[19,20]	White children in association with CFTR mutation

Some points illustrating the importance of using race/ethnicity as a variable in biomedical research and as a factor in clinical practice

- The frequency of certain genetic variants (genetic polymorphism) is higher among populations of the same geographic location or origin, or of the same culture (Table 1). For example, the frequency among Ashkenazi Jewish women of germline BRCA 1 mutation is higher than in some other racial/ethnic groups, rendering them susceptible to breast cancer; and the frequency of the variant gene that causes Tay-Sachs disease is also higher among Ashkenazi Jews than in the general population.[5,13] This is because the Jewish population descended from a small number of founders and remains largely endogamous.[3,8] Tay-Sachs disease is a fatal, autosomal recessive metabolic disease caused by impaired functional activity of the lysosomal enzyme Hex A.[13] As it is known which populations are at risk, parental carrier screening for Tay-Sachs disease in the Ashkenazi Jews has become a standard of care.[14]
- Although racial/ethnic categorisation does not correlate with a high degree of reliability to genetically-driven biological traits, race/ethnicity remains a convenient and useful clinical parameter providing insight into the probability that certain disease susceptibility genes will be expressed, thus enhancing clinical judgement and improving diagnostic focus and consequent treatment outcomes.[3,8]
- The appropriate use of racial/ethnic categorisation can be used to identify and describe differences in the health risks and needs of populations, that warrant further investigation and intervention.[7]

CONCLUSION

- Screening programs to identify genetically susceptible persons within high-risk racial/ethnic groups are essential and justifiable.
- In order to minimize the risk of validating the spuriously derived genetic biological concepts of race, one should avoid the use of race-based analyses in clinical and research studies when there is no plausible role for race in the hypothesis. If race/ethnicity is used as a variable, one should clearly state the context in which it is being used, describe the method that was used to assess and categorize it, and discuss all significant findings.[14]
- In order to reduce health disparities among and between members of different racial/ethnic groups, and to provide appropriate individualised healthcare strategies, it is essential to research how race/ethnicity impacts upon health.

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