Genetics, race, ethnicity, and health

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Genetics plays only a small part in ethnic differences in health, and other factors are often more amenable to change

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In the past few decades spectacular advances have occurred in molecular biology techniques. Researchers have been eager to use these new techniques to study ethnic or racial differences in health that are commonly assumed to have genetic causes. We argue that this assumption is based on confusion between three very different concepts: genetics, race, and ethnicity. This is invalid both scientifically and in terms of public health policy. We concentrate on research in Maori and Pacific (Polynesian) people, but the issues considered are of more general relevance.

Genotypes and phenotypes

It is a common misconception that the genotype determines the phenotype. Genetic factors do have a large influence on health, but they are just one piece of a much larger picture. We are all continuously developing throughout our lives with a constant interaction between our genes and the environment.[1] Any discussion of genetic tendencies thus makes assumptions about who is normal and what is a normal environment.[2] For example, some researchers have argued that Polynesians have a thrifty genotype with a greater tendency towards obesity and non-communicable diseases such as diabetes when they adopt a European diet.[3] It now seems, however, that almost everyone except Europeans may have the thrifty genotype.[4]

Genetics, heritability, and the environment

The constant interaction between genes and the environment means that few diseases are purely hereditary (even if they are genetic). Purely hereditary diseases are very rare (1/2300 births for cystic fibrosis, 1/3000 for Duchenne's muscular dystrophy, and 1/10 000 for Huntington's disease) and account for a small proportion of overall disease.[2]

It is often assumed that diseases are genetic because they run in families, but this often reflects a common environment and lifestyle rather than a genetic influence. For example, in 1900 the Pellagra Commission investigated the problem of pellagra (a vitamin deficiency disease) in the southern United States and concluded that it was genetic because it ran in families; "apparently, no one on the Commission realized that poverty and malnutrition run in families too."[5] In fact, the characteristic that is most strongly inherited is religion.[2]

Even for classic genetic diseases, environmental factors usually also have an important role and are easier to modify.[6] This may change with the continuing development of techniques such as gene therapy, but to date it has promised much and delivered little.[7] In fact, genetic studies can show the importance of environmental factors. Mendelian randomisation means that rare genetic polymorphisms that mimic common environmental factors (such as low dietary folate intake) can be studied epidemiologically to establish the causal associations of these environmental factors with disease (such as neural tube defects).[8]

Race

So what does this have to do with race? The term race has been commonly defined in terms of biological differences between groups that are assumed to be genetic. For example, Risch et al argued that five major racial groups can be identified (Africans, Caucasians, Pacific Islanders, Asians, and Native Americans).[9] However, human races are not and never were pure,[10] and such broad continental groupings explain little in terms of the overall genetic variation of humanity. For example, Lewontin[2] writes that:

> Skin colour, hair form, and nose shape are certainly influenced by genes, but we do not know how many such genes there are, or how they work. On the other hand…

Environmental factors have more influence on health than genetic differences

about 85 percent of all identified human genetic variation is between any two individuals from the same ethnic group. Another 8 percent of all the variation is between ethnic groups within a race—say, between Spaniards, Irish, Italians, and Britons—and only 7 percent of all human genetic variation lies on the average between major human races such as those of Africa, Asia, Europe, and Oceania.

This is not to say that there are no genetic differences between races, but very few differences have been found which directly relate to health.[7] Those that have been shown are usually not absolute; rather, they concern differences in the percentage of people that have particular gene types. Furthermore, most known genetic variants that are health related are random mutations in subpopulations or result from regional selection and are not related to continental race.[7]

Race and health

A New Zealand study of alcoholism provides one example of a health related genetic difference.[11] The authors found that the ADH2-2 gene, which is believed to protect against alcoholism, was relatively common in Maori people but was not found in New Zealand Europeans. In fact, alcohol related health problems are more common in Maori, suggesting that the hypothesised protective genetic factors are being outweighed by social, economic, cultural, and political factors.

Further evidence that genetic factors are unlikely to explain racial differences in health is provided by trends in death rates. Maori death rates fell steeply in the last century, although the rate of progress has slowed in the last two decades.[12] Major problems remain, but the rapid reduction in many of these diseases strongly suggests that they are not primarily genetic.

Ethnicity and health

The fact that racial categorisation based on genetic criteria is inaccurate and misleading[13] does not change the historical reality of the effects of colonisation on indigenous peoples, nor the realities of indigenous health today. It does, however, change our interpretation of the causes of high mortality and morbidity in indigenous peoples, which primarily relate to issues of ethnicity, rather than of race or genetics.

Ethnicity is a complex construct that includes biology, history, cultural orientation and practice,[14] language, religion, and lifestyle, all of which can affect health. The lack of major systematic genetic differences between ethnic groups, together with the extensive differences in lifestyle (diet, alcohol, housing, smoking, etc), means that ethnic differences in mortality and morbidity to some extent provide evidence against the importance of genetic factors and for the importance of environmental factors.

For example, the European colonisation of the Pacific and the Americas after 1492 saw indigenous populations decimated by imported communicable diseases.[15] In the Pacific, indigenous people experienced high mortality from imported infectious diseases mainly when their land was taken, disrupting their economic base, food supply, and social networks. When land was not taken in large amounts by European settlers, the death rate was relatively low.[15] Similar effects of social disruption resulting in increased mortality have been seen more recently in Eastern Europe and again in the Pacific.[15 16]

Summary points

- The concepts of genetics, race, and ethnicity are commonly confused
- Genetic factors are important for health but are a small part of a large and complex picture
- Few systematic genetic differences exist between races, at least with regard to genes that affect health
- Ethnic differences in health are due to historical, cultural, and socioeconomic factors, which in turn influence lifestyle and access to health care.
- Overemphasis on genetic explanations may divert attention and resources from more important influences on health

Ethnic differences in access to health care

Access to health care is also important for ethnic differences in health. For example, the high death rates from asthma in Maori people in the 1980s led some researchers to assume that Maori have a genetic tendency to asthma. However, the high death rate was primarily due to the drug fenoterol, which affected Maori people particularly severely. This is probably because problems accessing health care meant that Maori people were more likely to overuse the drug.[17] When fenoterol was restricted in 1989, the asthma death rate fell sharply and is now low in both Maori and non-Maori populations.[17] Furthermore, until recently the prevalence of asthma was about the same in Maori and non-Maori children, but non-Maori children tended to lose the disease as they grew older whereas Maori children did not, producing a greater prevalence in adult Maori.[18] This is probably due to poor access to health care and asthma education.[18]

Similar considerations may apply to other diseases requiring long term medical management, such as diabetes and cardiovascular disease. Ibrahim and colleagues recently argued that issues of access to care include a complex mix of cultural factors and individual preferences of patients, characteristics, and practices of healthcare professionals (such as racism, stereotyping, bias, discrimination, and lack of cultural safety), and the system of delivery of health care (such as composition of the workforce, location of facilities, costs of access, and involvement of different ethnic groups in shaping health policy and allocation of resources).[19] They argued that "racial and ethnic disparities in health and health care are rooted in historic socioeconomic inequalities that persist…today." Thus race may be important in terms of the perceptions of healthcare workers, rather than in terms of the underlying disease aetiology.

Gene hunting: the new research colonialism

Genetic research will undoubtedly lead to important discoveries and new forms of treatment. However, such

benefits are a long way off and require large investments with potential benefits for a few high risk individuals (and researchers). This leaves little to promote the health of the majority. The emphasis on genetic explanations for population differences in health has also led to controversial instances of "gene hunting" by multinational institutions.[20] The affected countries may have inadequate or non-existing legislation on ethical and social protection for human subjects of health research in general, or genetic research in particular. The wave of health research that has swept the Pacific since the late 1960s has turned the "Pacific into a laboratory for exotic researchers."[20] In response, it has been argued that there must be a strong community base for health research that is of community relevance, with a sustained community involvement in research planning and implementation.[15]

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Contributors and sources: NP is an epidemiologist, with particular experience of research in non-communicable disease. AS and CC are Maori health researchers with extensive experience working with European New Zealand colleagues on studies of Maori health, as well as conducting their own Maori health research. SF is a Pacific health researcher specialising in epidemiology, with a joint appointment between the Massey University Centre for Public Health Research and the Ministry of Health of Tonga.

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1 Potter JD. Reconciling the epidemiology, physiology, and molecular biology of colon cancer. *JAMA* 1992;268:1573-7.
2 Lewontin RC. *The doctrine of DNA*. London: Penguin, 1991.
3 Neel JV. Diabetes mellitus: A "thrifty" genotype rendered detrimental by "progress"? *Am J Hum Genet* 1962;14:353-62.
4 McMichael A. *Human frontiers, environments and disease: past patterns, uncertain futures*. Cambridge: Cambridge University Press, 2001.
5 Lewontin RC. *Human diversity*. San Francisco: Scientific American, 1982.
6 Cruickshank JK, Mbanya JC, Wilks R, Balkau B, McFarlane-Anderson N, Forrester T. Sick genes, sick individuals or sick populations with chronic disease? The emergence of diabetes and high blood pressure in African-origin populations. *Int J Epidemiol* 2001;30:111-7.
7 Cooper RS. Race, genes and health—new wine in old bottles? *Int J Epidemiol* 2003;32:23-5.
8 Davey Smith G, Ebrahim S. Mendelian randomization: can genetic epidemiology contribute to understanding environmental determinants of disease? *Int J Epidemiol* 2003;32:1-22.
9 Risch N, Burchard E, Ziv E, Tang H. Categorization of humans in biomedical research: genes, race and disease. *Genome Biol* 2002;3: comment2007.
10 Templeton AR. Human races: a genetic and evolutionary perspective. *Am Anthropologist* 1999;100:632-50.
11 Marshall SJ, O'Brien D, Wheeler K, Hermans IF, Chambers GK, Robinson GM, et al. Genes and alcoholism: a preliminary report. *NZ Med J* 1994;107:106-7.
12 Ajwani S, Blakely T, Robson B, Tobias M, Bonne M. *Decades of disparity*. Wellington: Public Health Intelligence, Ministry of Health, 2003.
13 Schwartz RS. Racial profiling in medical research. *N Engl J Med* 2001;344:1392-3.
14 Durie MH. Te Hoe Nuku Roa Framework: A Mâori identity measure. *J Polynesian Soc* 1995;104:461-70.
15 Foliaki S, Pearce N. Changing pattern of ill-health for indigenous people. *BMJ* 2003;327:406-7.
16 Foliaki S, Pearce N. Prevention and control of diabetes in Pacific people. *BMJ* 2003;327:437-9.
17 Ellison-Loschmann L, Cheng S, Pearce N. Time trends and seasonal patterns of asthma deaths and hospitalisations among Mäori and non-Mäori. *NZ Med J* 2002;115:6-9.
18 Ellison-Loschmann L, Pearce N. He Mate Huango: an update on Mäori asthma. *Pacific Health Dialogue* 2000;7:82-93.
19 Ibrahim SA, Thomas SB, Fine MJ. Achieving health equity: an incremental journey. *Am J Pub Health* 2003;93:1619-21.
20 Fitzgerald M. Our DNA, ourselves. *Time* 2001;33(Aug 20-27):46.

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Q&A

Effects of ginkgo extract

Question
What are the benefits and side effects of *Ginkgo biloba*? Does it cause high blood pressure?

Mahmood M Althahabi, *pilot, Bahrain*

Answer
The dried leaf of the ginkgo tree has been used in medicine for thousands of years. More than 400 studies over the past 30 years have investigated its ability to improve blood flow in a variety of conditions including memory impairment, dementia, peripheral vascular diseases, and tinnitus. The results have shown modest improvements, not to some remarkable levels. The results much depend on the patient's psychological framework.

In general, it is well tolerated. Allergic skin reactions, gastrointestinal upsets, and headaches occur in fewer than 2% of patients. There are theoretical concerns about a risk of increased bleeding because of inhibition of platelet activating factor. Although no bleeding complications have been reported in any clinical trials, caution should be exercised when taking ginkgo with aspirin, warfarin, or other anticoagulant. No effects on blood pressure have been documented.

Ali K Yousuf, *senior house officer, Karachi, Pakistan*

Answer
Ginkgo biloba is said to have neutral effects on blood pressure. However, there are reports suggesting that it may have antihypertensive actions.

In a study on healthy, young volunteers, ginkgo in a dose of 120 mg given twice daily for seven days was not found to have any immediate or short term effects on systolic or diastolic blood pressure.[1] In a randomised, double blind trial a significant decrease in diastolic blood pressure was reported in elderly patients with age related cognitive dysfunction who were given an extract of ginkgo (GB-8) for three months.[2] In one study on healthy volunteers, a single treatment with an extract of ginkgo (EGb 761, 120 mg) was found to reduce stress induced rise in blood pressure without affecting the heart rate.[3]

Vikas Soni, *medical advisor, Ahmadabad, India*

1 Kalus JS, Piotrowski AA, Fortier CR, Liu X, Kluger J, White CM. Hemodynamic and electrocardiographic effects of short-term Ginkgo biloba. *Ann Pharmacother* 2003;37:345-9.
2 Winther K, Randløv C, Rein E, Mehlsen J. Effects of Ginkgo biloba extract on cognitive function and blood pressure in elderly subjects. *Curr Ther Res* 1998;59:881-8.
3 Jezova D, Duncko R, Lassanova M, Kriska M, Moncek F. Reduction of rise in blood pressure and cortisol release during stress by Ginkgo biloba extract (EGb 761) in healthy volunteers. *J Physiol Pharmacol* 2002;53:337-48.

Answer
I started taking gingko before exams as it had been suggested that it helped memory, and I have noticed that, since starting it, I have not had a migraine. This is, of course, a purely anecdotal benefit, but it worked for me.

Beth Bain, *specialist trainee in public health, West Midlands Specialised Services Agency*

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