nature genetics

The unexamined population

We agree with our colleagues at Howard University that it is scientifically and socially imperative to examine the state of the science of human variation and it is time to engage everyone in this discussion. As we explained in an editorial called "The unexamined 'Caucasian'" (*Nat. Genet.* **36**, 541; 2004), the use of race as a proxy is inhibiting scientists from doing their job of separating and identifying the real environmental and genetic causes of disease. Similarly, we think it is not useful for society to spend time in an inflammatory debate on whether data from human genome variation can confirm or refute pre-genetic concepts of 'races'. Finally, unless we discuss human genomic variation, we will all miss the emerging story of who we are and where we come from.

We selected participants from the Howard University meeting to provide a broad and deep overview of the state of the science of human genetic variation. Our aims are restricted in scope but ambitious. We are not going to be able to solve the social problem of racism scientifically, because its source lies with racists and not in the intrinsic biology of their victims. Early in the history of the science, geneticists made the mistake of fixing group labels to biological markers and thereby contributed to racist atrocities. Geneticists are painfully aware of this ugly past, and some have presented reassuring but incomplete assertions that do not match common-sense observations of our diversity. But now, by discussing all we know, by presenting the marvels of the ways the human genome varies and the reasons why it does so to the human population, we hope to gain your interest, advice and priorities to promote the use of what information geneticists have found for the benefit of all members of the human race.

Everyone is genetically unique. We are all one species. Between these obvious extremes, the question you ask determines how closely and for how long you are prepared to look at the ways in which the human genome varies. How hard you are prepared to look depends on your funds and on your level of open curiosity. By reporting the discoveries of the last few years to a wider public, we hope to stimulate interest in who we are and in the ways we are similar to and different from one another. A detailed and accurate view of the diversity of the human genome can overwhelm the legacy of inequity with enthusiastic inquiry into our common ancestors and into the variant genomes we share, compare and exchange among ourselves.

In our first foggy view of the species we can compare the groups that ancestrally populated each of the continents with conspicuous mountaintops above the clouds. As our view clears, the valleys connecting the mountaintops can be seen to be far more fertile. The beauty of the whole continuous landscape can be appreciated, and we begin to note similarities and contrasts in various features. Our first approximate view of human genetic variation must therefore be one that enables the next look, and the next, in an ever more accurate refinement of our view of who we are. Future research will certainly spring some surprises; for example, we now know that the copy number and order of genes on chromosomes can vary greatly between individuals in ways that may have functional implications (*Nat. Genet.* **36**, 931–932; 2004). But we shall have established a new and accommodating framework within which to understand and to apply these discoveries.

In "The unexamined 'Caucasian'" we argued that researchers should specify as much ancestral and environmental information as they have so that their data can be grouped flexibly to serve the needs of medical geneticists, epidemiologists and biological anthropologists. What is helpful to the researchers may also be helpful to people seeking to understand human genome variation. Thinking of our genetic identities flexibly, gene by gene, level by level, region by region, with criteria for grouping examined and made explicit, fits well with the contingent way in which people describe their own ethnicity, ancestry, nationality, phenotype, family ties and genetic legacy.

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