

Perspectives on U.S. Policies that Address the Involvement of Groups in Genomic Data Sharing

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Challenges

- Social groups are not constituted based on biological characteristics
- Social groups nonetheless tend to have non-random frequency distributions of biological characteristics
- Those frequency distributions of biological characteristics often are mistaken for biological definitions of group membership

Challenges

- Social groups have differing kinds of “groupiness”
- All social groups are to some extent heterogeneous in viewpoint (including views on genetics and genetic data sharing)
- Nonetheless, group labels, when used, can have uniform implications for all members

Background of ongoing debate about race, ethnicity, and genetics

- Many scientists naively use social labels for biological data
- Media and public continue directly to link race and ethnicity with unique biological characteristics
- Individual risks for disease are more important than group risks for disease in understanding personal health
- Non-random frequency distributions of biological characteristics among social groups are meaningful
- Population stratification is a valid analytic approach for aggregate data
- Health disparities at the group level are a product of both non-random social and non-random biological frequency distributions of contributing factors

If there were a straightforward solution to the challenges of using group labels in biological and biomedical research, we likely would have adopted it by now.

So far, concerns about group risk have out-weighed concerns about group benefit

- Non-Maleficence: How do we protect groups from collective harms as a result of inappropriate equation of social labels and biological data?
- Justice: How do we insure that non-majority groups have equal access to genetic-based diagnostics and therapeutics?

In the U.S., concerns about group risk have been complicated by respect for individual autonomy

- Except where a legally-constituted group controls access to a facility or territory, group consent has not been taken to trump individual consent
- Guidelines for community consultation and other stakeholder approaches have not resolved challenges of representativeness and heterogeneity
- Self-reported identity almost never is further interrogated

In the U.S., group benefit has been complicated by legal definitions of inclusiveness

- OMB guidelines for racial and ethnic inclusion often do not result in scientifically meaningful inclusion of non-majority participants
- Identities that are legally and politically relevant may not be biologically or biomedically relevant, particularly in the context of specific research questions

Is the race and genetics debate a fundamentally non-Darwinian creation that is incapable of adapting and evolving?

Oddly, several scientific developments may suggest a way forward

Population stratification using genotypes

- Genotyping to profile population stratification is more accurate than using self-reported social identity
- Decreases in cost of genotyping make for a less expensive filter
- Potential risk: genotypes (such as AIMs) could become surrogates for social and ancestral identities

Rare variants and structural rearrangements may be more important than common variants as contributors to common diseases

- Emphasizes the benefit to members of non-majority groups in taking part in genetic research and sharing data
- Refocuses the scale of relevant groups on smaller populations with more recently shared ancestral histories
- Potential risk: “rare” could mistakenly be interpreted to mean “unique” or “group-specific”

Will personal genomics save us all?

- Greater emphasis on individual data than on aggregate group data
- Greater emphasis on individual risk
- Much more individual data
- However, DTC providers tend to rely on convenient, reductionary categories when interpreting personal data and also use very “groupy” ancestry analyses -- not encouraging for overcoming social labels

Policy suggestions

- Continue to help geneticists and other scientists in using social labels in association with biological data appropriately and only when scientifically meaningful
- Continue to educate media and the public about the significance of the non-random frequency distribution of biological characteristics by social group
- Continue to develop social science approaches to community consultation and stakeholder analysis that are more robust with respect to representativeness and heterogeneity of viewpoint
- Disentangle legal and regulatory requirements for and definitions of inclusiveness from scientific design and evaluation
- Emphasize smaller scale groups and non-random frequency distribution of biological characteristics among those groups
- Don't mandate policy prescriptions -- that just results in another set of OMB-like categories and check-offs