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CRS Report for Congress

Genetic Ancestry Testing

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Summary

Interest in genetic testing to determine ancestry has increased rapidly since its introduction in 2000. Many individuals are eager to learn more about their ancestors, and believe genetic testing will be able to provide information where other traditional genealogical methods have failed. While genetic ancestry testing may in some cases be able to provide very general information, it also currently has many limitations. The very complex relationship between race, genetics, and ancestry further complicates this testing and the interpretation of results. Genetic ancestry testing may, therefore, raise several policy issues. This report describes genetic ancestry testing, outlines the basic scientific limitations of the testing currently, and provides an overview of the policy issues this testing may raise.

Overview of Genetic Ancestry Testing

There is significant interest in the United States, a country predominantly composed of fairly recent immigrants, in utilizing genealogical research to learn more about individual ancestral roots. Until recently, genealogists had only the traditional tools of their trade available to them to conduct this research, such as historical documents and other paper records. More recently, with the commercialization of genetic ancestry testing, genealogists and others have gained access to a new tool. Genetic ancestry testing may be used to identify biological relatives, validate genealogical records, and to fill in gaps in family histories.[1] In addition, this testing may be used to aid in medical risk calculation, forensic investigations, admixture mapping,[2] and the assessment of ancestry for socio-political purposes such as affirmative action qualification or Native American tribal

[1] Bolnick D.A. et al., "The Science and Business of Genetic Ancestry Testing," *Science*, 318: 399-400, October 19, 2007.

[2] Admixture mapping can be defined as estimating the proportions of an individual's genome that have ancestry from different subpopulations.

affiliation.[3] This report considers only non-health and non-forensic applications of genetic ancestry testing.

Commercial genetic ancestry testing was first offered in 2000 by the company Family Tree DNA.[4] Since that time, this testing has grown in popularity, especially among African-Americans attempting to learn more about their history and culture.[5] Currently, almost two dozen companies offer genetic ancestry testing, and it is estimated that roughly 460,000 people have undergone testing at a cost ranging anywhere from $100 to $900 per test.[6] These tests are termed "recreational" in deference to and in contrast with genetic tests performed for health or forensic purposes.

Broadly, there are three types of genetic ancestry tests available: Y chromosome testing; mitochondrial DNA (mtDNA) testing; and autosomal marker testing. Since the Y chromosome is passed down essentially unchanged from father to son, Y chromosome testing of a male can help to determine the origin of paternal ancestors. Mitochondrial DNA, on the other hand, is passed down from mother to child unchanged; therefore, mtDNA testing of an individual may be used to help determine the origin of maternal ancestors. Importantly, mtDNA or the Y chromosome represents only a small portion of an individual's total DNA. The third type of testing, autosomal marker testing, utilizes markers located on autosomes (non-sex chromosomes). It is currently offered by a company called DNAPrint Genomics. Autosomal marker testing is the most challenging type of testing since it requires a substantial amount of data to ensure the accurate interpretation of the results.

Scientific Considerations

Despite the recent proliferation of commercially available genetic ancestry tests, the literature has reported several scientific limitations to current tests.[7] These limitations are important because they raise certain policy issues, as discussed in the next section.

Genetic ancestry testing cannot always definitively determine the ethnicity or geographical location of an individual's ancestors. Scientists point out that we have very limited genetic data available from the ethnic groups in Africa (less than 1% have been analyzed), and that the migration of Africans makes it difficult to match African-Americans to exact ethnic groups.[8] They also note that "some less common lineages can be traced to particular ethnic groups or locales," but that "tracing the more common

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[3] Shriver M.D. and R.A. Kittles, "Genetic ancestry and the search for personalized genetic histories," *Nature Reviews*, 5:611-617, 2004.

[4] [http://www.familytreedna.com/].

[5] Ibid.

[6] Bolnick D.A. et al., "The Science and Business of Genetic Ancestry Testing," *Science* 318:399, October 19, 2007.

[7] Ibid.

[8] Coleman, T., "Regaining a Lost Heritage," *Diverse Issues in Higher Education*, 23(26): 29-31, 2007.

haplotypes to a particular location is problematic."[9] In a recent study comparing African-American mtDNA with a database of sub-Saharan mtDNA, it was found that about 50% of the African-Americans were a match to multiple African ethnic groups, 40% did not match any sequence in the database, and only 10% were a perfect match for a single African ethnic group.[10]

Difficulty determining an individual's exact ethnic match has generated mixed results for consumers. Henry Louis Gates Jr., who has done much to draw public attention to genetic ancestry testing with his PBS special "African American Lives," reportedly had testing performed twice. He was first told that his maternal ancestors were most likely Egyptian (Nubian). The second test concluded that his maternal ancestors were most likely European.[11] Ron Nixon, a *New York Times* journalist, reports that he had his DNA tested by multiple companies and was told that he was a match with the Mende and Kru people from Liberia; the Songhai in Mali; various ethnic groups from Mozambique and Angola; and the Bambara from Mali. While some of these ancestral links were confirmed by multiple tests, each additional test also indicated several unique links of its own.[12] As these reports demonstrate, pinpointing one's ancestral ethnicity may be difficult, and often not possible, using current genetic testing techniques.

The accuracy of genetic ancestry testing is dependent on the size and sampling of the database used to match test samples to populations or geographical regions. Scientists note that "many databases that are derived from published research are too small and lack samples in certain geographical regions."[13] At this point in time, because of limited data, the interpretation of test results often relies on a significant amount of inference. The information provided by these tests, therefore, is only probabilistic in nature, and is not definitive.

In addition, scientifically suspect inferences may sometimes be made, such as assuming that specific genetic variation is diagnostic of a particular population, when it may also be present in many other populations, just at lower rates.[14] In other words, an individual may carry a particular variant or pattern of variation and be told that he or she definitely has ancestors belonging to a sub-population where that particular variant or pattern of variation is common. However, because the particular variant or pattern of variation may in fact be present in other populations, albeit at lower rates, it is possible,

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[9] Shriver M.D. and R.A. Kittles, "Genetic ancestry and the search for personalized genetic histories," *Nature Reviews*, 5:611-617, 2004.

[10] Ely B. et al., "African-American mitochondrial DNAs often match mtDNAs found in multiple African ethnic groups," *BMC Biology* 2006, 4:34.

[11] Keith J. Winstein, "Harvard's Gates Refines Genetic Ancestry Searches for Blacks; Scholar Founds a Firm After DNA Tracer Put Forebear in Wrong Place," *Wall Street Journal*, page D5, November 15, 2007.

[12] Ron Nixon, "DNA ancestry testing leaves some in doubt: Former proponents question accuracy of the science," *New York Times*, December 2, 2007.

[13] Shriver M.D. and R.A. Kittles, "Genetic ancestry and the search for personalized genetic histories," *Nature Reviews*, 5:611-617, 2004.

[14] Bolnick D.A. et al., "The Science and Business of Genetic Ancestry Testing," *Science*, 318: 399-400, October 19, 2007.

although less so, that he or she has ancestors belonging to this group as well (or exclusively). The possibility for subjectivity in the interpretation and presentation of results concerns some scientists, while others maintain that appropriate methods used in a rigorous manner may achieve valid results, although with clear limitations that should always be communicated to the consumer.[15]

Lineage-based testing, that is, testing of the mtDNA or Y chromosome, examines only a small percentage of an individual's total DNA, and therefore provides limited information about an individual's complete ancestry. "One major disadvantage of ... lineage-based analyses is that they focus on a single maternal or paternal lineage and therefore neglect the contribution of the vast majority of an individual's ancestors to their genome."[16] This testing may be informative in certain cases, but scientists stress that some customers may lack the understanding that "their maternal and paternal lineages do not necessarily represent their entire genetic make-up."[17]

Policy Considerations

Genetic ancestry testing raises several policy considerations. These include concerns about direct-to-consumer marketing of the tests; reinforcement of scientifically questionable ideas about the relationship between race and genetics; and privacy of the test results.

Direct-to-Consumer Testing. Although genetic ancestry tests are not health-related genetic tests, they are nonetheless very complex tests that provide qualified information about issues of significant social and personal importance, such as racial or ethnic identity or geographical origin. The results of such tests are highly qualified, and the public may not fully appreciate these limitations in the absence of thorough pre-test education on the part of the company offering the testing. Companies often argue that direct-to-consumer testing empowers individuals, and this premise may be true, but is predicated on providing the consumer with complete information.

In addition, a concern with any direct-to-consumer marketed genetic test is that it might exploit particularly vulnerable groups of people such as the elderly, the infirm, or those with a personal or family history of disease. In the case of genetic ancestry testing, some African-Americans have been seeking out testing in an attempt to learn more about a history that was obscured by the slave trade. African-Americans pursuing the identification of their ancestral communities is in fact cited as "the most prominent example" of those seeking genetic ancestry testing.[18] Understandably, the emotional investment in seeking and finding such information may result in a less critical appraisal

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[15] Coleman, T., "Regaining a Lost Heritage," *Diverse Issues in Higher Education*, 23(26): 29-31, 2007.

[16] Shriver M.D. and R.A. Kittles, "Genetic ancestry and the search for personalized genetic histories," *Nature Reviews*, 5:611-617, 2004.

[17] Ibid.

[18] Ibid.

of the test or acknowledgment of its limitations. Appropriate pre-test education may help address this issue.

Proprietary Databases. Currently, many companies performing genetic ancestry testing maintain proprietary databases of sequences that are used to make matches with a test taker's DNA sample. From a commercial perspective, maintaining proprietary databases is understandable. However, in terms of producing a more reliable analysis of ancestry, pooling data might be a more productive approach. In addition, the propriety nature of the databases makes an open assessment of scientific claims about a test difficult.

Interaction with Existing Policy. Some national policies have as their goal fair compensation for individuals who were disadvantaged due to their racial or ethnic affiliation. Especially since racial and ethnic affiliation are often culturally determined as well as self-reported, ancestry testing might undermine the social goals of these policies. For example, the *New York Times* reports that some white-skinned applicants have applied as minority candidates for both employment opportunities and college admission based on results of genetic ancestry tests.[19] This is illustrative of the problems that genetic ancestry testing may create for social policy. It also clearly demonstrates the problems inherent in equating race with genetics (discussed below).

There is some concern that genetic ancestry testing may become a requirement for receiving benefits based on racial or ethnic background. This is particularly a concern with respect to Native American ancestry. In 2000, a bill was introduced in the Vermont Legislature recommending that the Commissioner of Health "establish standards and procedures for DNA-HLA testing to determine the identity of an individual as a native American, at the request and the expense of the individual. The results of such testing shall be conclusive proof of the Native American ancestry of the individual."[20] The bill did not pass, and triggered criticism by some who maintained that it would potentially allow a genetic identity to supercede a culturally determined one. Proponents of the bill stated that it was meant to allow genetic testing to be used as an additional method for determining Native American heritage (not to replace culturally and socially determined heritage).

Race, Genetics and Ancestry. The relationship between race, genetics, and geographical origin is extremely complex and the subject of significant academic debate. Experts in anthropology and sociology maintain that race is a social and cultural concept. Race, they argue, has been defined by observable (i.e., phenotypic) traits, such as skin color or eye shape, which have been arbitrarily vested with a degree of social importance or relevance. Thus, race is viewed by these groups as a fluid concept whose definition has been influenced more by social and historical forces than by true genetic differences. To some degree, race correlates with patterns of genetic variation, but this correlation is seen primarily when race acts as a proxy for geographical origin. Geographical origin does reliably correlate with specific patterns of genetic diversity, reflecting the evolution of specific genetic variants or patterns of variation in early human populations.

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[19] Amy Harmon, "Seeking Ancestry in DNA Ties Uncovered by Tests," *New York Times*, April 12, 2007.

[20] DNA Testing and Native Americans, H. 809, 1999-2000 Sess., Vermont Legislature.

Besides the scientific concerns about "geneticizing" race, social concerns are raised as well. Attempts have been made in the past to inaccurately use the science of genetics to legitimize various prejudiced policies and positions.[21] For example, claims have been made that genetically based differences in IQ exist between different races.[22] In this social context, genetic ancestry testing may create a false impression in the public that race is primarily genetic in nature. Genetic ancestry testing for race and ethnicity implies that race and ethnicity may be uncovered utilizing a genetic test, a premise that many argue is false.[23] This could serve to reinforce common misconceptions that there is a biological basis for differences between racial groups. This misconception might be employed to support policies that claim that differences between racial groups are inherent and therefore unable to be addressed or remedied through social policy. This is particularly relevant with respect to health disparities policy, because such policy attempts to redress differences in health outcomes by race.

Privacy. Finally, the information generated by genetic ancestry testing is not viewed as personally identifiable health information and so is not protected under federal privacy protections pursuant to the Health Information Portability and Accountability Act.[24] The results of genetic ancestry tests do not fall under the definition of "protected health information" under the Privacy Rule, and testing companies do not fall under the definition of "covered entities" under the Privacy Rule. Protecting the privacy of the results of these tests is at the discretion of the company offering the test. For example, at the company Family Tree DNA, DNA samples are bar-coded in order to protect an individual's identity. Customers are asked if they will provide their consent to the banking of their DNA for future testing, and of more than 70,000 individuals undergoing testing, only two requested that their samples be destroyed.[25] This raises several important issues. First, even if samples are bar-coded to protect individual's identities, they may be linked to personally identifiable information held by the company in order to facilitate future testing. Second, given the extremely low number of individuals requesting that their DNA sample be destroyed, questions naturally arise about the degree of education that companies provide to customers during the consent process. One senior research scientist noted that "How much responsibility a company has in educating a potential client prior to sample submission and credit card number is a value judgment matter."[26] Third and finally, as these databases grow in size, they will grow in potential value as well. Family Tree DNA maintains that its database currently is not for sale; however, as its size and potential value increase, this may be reassessed. On the other hand, as the databases grow in size, their value to individual companies may be greater if they are kept proprietary.

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[21] Andrews L.B. and D. Nelkin, "The Bell Curve: A Statement," *Science* 271:13-14, January 5, 1996.

[22] See, for example, Richard J. Herrnstein and Charles Murray, *The Bell Curve: Intelligence and Class Structure in American Life*, Free Press Paperbacks, New York, N.Y., 1994.

[23] Bolnick D.A. et al., "The Science and Business of Genetic Ancestry Testing," *Science*, 318: 399-400, October 19, 2007 and Coleman, T., "Regaining a Lost Heritage," *Diverse Issues in Higher Education*, 23(26): 29-31, 2007.

[24] [http://www.hhs.gov/ocr/privacysummary.pdf].

[25] Wolinsky, H. "Genetic genealogy goes global," *EMBO reports*, 7(11): 1072-1074, 2006.

[26] Ibid.