Geneticists sound note of caution over DNA ancestry testing

By John Timmer | Published: November 13, 2008 - 12:15PM CT

If you do an Internet search for "DNA ancestry testing," the results that show up (below the ads, naturally) are a series of commercial services that promise to take your DNA and let you know what it says about where you came from. Stories have appeared in the press about how these tests have helped people discover unexpected connections with bits of a past they never knew existed. But just how accurate are these tests? The American Society of Human Genetics is releasing a Statement on Ancestry Testing today in conjunction with their annual meeting, and it sounds some significant cautions about these services.

The statement points out what's perhaps the biggest danger with these services: there's no objective way to evaluate the results they produce. The services may use any of a variety of methods—mitochondrial and/or Y-chromosome DNA, scans of the entire genome, etc.—any of which could potentially produce contradictory results, all of which are valid within the limits of the DNA being looked at. Since all of them are "right," as it were, the only thing that's left to discriminate among the services is price. Although the ASHG doesn't say so, it's effectively a race to the bottom.

That doesn't mean that the tests are meaningless, but it's important to recognize their limits. For much of its history, humanity has largely bred on a local level, allowing DNA differences to accumulate in populations. As we've started to do research on human genetic diversity, we're developing an ever-improving ability to identify some changes that are prevalent in specific populations.

But note the term "prevalent." As the ASHG statement points out, most of the frequencies of DNA markers have known error rates. Something that's prevalent in say, Pacific Islanders, will often show up in an unrelated population, like Scandinavians, at a measurable rate. There's also the question of what sort of pattern is really relevant. As the statement notes, "present-day West Africans are the most frequently used proxy for inferring African-American ancestry even though the African origins of African Americans are quite heterogeneous."

Then there's the issue of what ancestors a given test is looking at. "Every person has hundreds of ancestors going back even a few centuries and thousands of ancestors in just a millennium," the statement notes. Although, on average, we can figure out what each ancestor may have contributed to someone, just like the DNA markers, these numbers are only probabilities; there's no way of knowing if, by chance, a given ancestor is over- or underrepresented in their modern descendent.

Between these three aspects: choice of test, its probability of finding a spurious link to a geographic origin, and the probability that a given DNA marker isn't representative, then only one thing is inevitable: if you do enough tests, you're going to get a wrong answer now and again, even if the average test is more or less correct.

That wouldn't be such a problem if it weren't for two things. The first is that many people assign an enormous

psychological weight to their ancestry, as it's intertwined with their sense of identity. The second is that many people are aware that certain genetic diseases and predispositions are prevalent in some ethnic groups, and the tests may trigger unnecessary worries—especially unnecessary, in that some of the same tests may be precisely the ones used to assign these risks in a more fine-grained manner.

The statement ends with a series of recommendations, the first of which is that the scientific community do its best to make these issues clear to the public. The public, in turn, has a duty to pay attention to scientists if they're going to take ancestry testing seriously.

But scientists can do a lot more. The report recommends that standards for genetic ancestry testing be developed, and mechanisms put in place to ensure that the testing industry be held accountable by them. Researchers should also work to ensure that the population surveys they perform are made publicly available and used to keep our estimates of the linkage between DNA and ancestry up-to-date. Finally, the should work with scholars in other fields, like anthropology and sociology, to gain a full appreciation of how their work will be interpreted by the broader public.

Further reading:

The ASHG Statement (PDF).

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