Even though genetic information is available, doctors may be ignoring important clinical clues

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Credit: AI-generated image (disclaimer)

With the availability of home genetic testing kits from companies such as "23andMe" and "Ancestry DNA," more people will be getting information about their genetic lineage and what races and ethnicities of the world are included in their DNA.

Geneticists, meanwhile, are also getting more tailored information about disease risk and prevalence as genetic testing in medical research centers continues.

Physicians accept that cystic fibrosis, for example, is much more common in people with Northern European ancestry and that sickle cell disease occurs dramatically more often in people with African origins. These commonly accepted racial and ethnic differences in disease prevalence are just the tip of the iceberg when looking at clinical differences that vary based on genetics.

But there's a problem, a recent study from the National Institutes of Health found. Many physicians and other providers are uncomfortable discussing race with their patients, and also reticent to connect race or ethnicity to genetics and clinical decision-making, the study suggested.

Overall, physician focus groups "asserted that genetics has a limited role in explaining racial differences in health," the authors added.

As a primary care physician who teaches urban health to medical students and as a state minority health commissioner who advocates for health equity, I see this as a problem that health care systems, and their providers, need to address.

The state of the science

Commercial DNA tests, such as those provided by 23andMe, not only give people their racial and ethnic lineage but also can provide a weighted risk for diabetes, stomach ulcers, cancer and many other diseases. In April, the FDA granted approval to 23andMe to sell reports to consumers that tell them whether they may be at heightened risk.

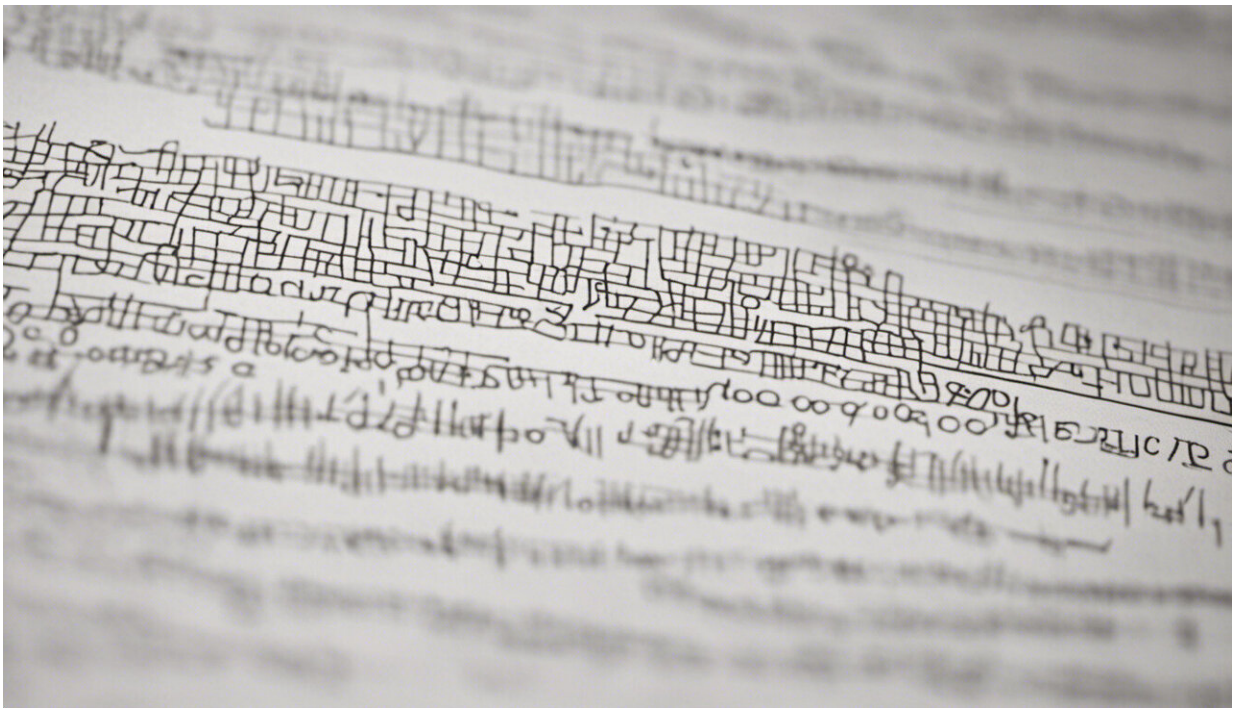
These companies already have the data that describe the risks for health

problems based on the percentage of their ancestry composition. Those differences have been published and known in academic circles for many years. With the widespread availability of DNA tests, patients will now know their increased individual risks.

For example, Ashkenazi Jews, a specific Jewish ethnic population originating from Central and Eastern Europe, are known for having a disproportionate occurrence of a number of diseases, including Tay-Sachs disease, amyloidosis, breast cancer, colon cancer and many more.

The BRCA1/2 gene mutation greatly increases the propensity for breast and colon cancer and occurs in 1 in 40 people of Ashkenazi Jewish heritage, whereas 1 in 800 Americans in general carry that mutation. This 20-fold increased risk should prompt more aggressive screening for the gene, and more frequent and earlier mammography and colonoscopies in Ashkenazi Jews compared to the general population.

Relatively higher rates of these cancers occur in certain populations, such as Ashkenazi Jews, and demonstrates the need for more nuanced care based on data that is already available. But this information is too infrequently accessed by providers.



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Genetics knowledge growing fast

African-Americans are another group with higher rates of certain genetically driven diseases. African-American men have an increased occurrence of prostate cancer, kidney failure, stroke and other health problems. Prostate cancer in African-American men, for example, grows faster and metastasizes four times as often than in European-Americans.

But despite this increased risk for prostate cancer, doctors' use of the PSA (prostate specific antigen), a test that works well with identifying prostate cancer in African-Americans, has steadily decreased due to recommendations aimed at majority patients who come from European-related heritage. In European-Americans, prostate cancer can be more indolent and occurs at a lower rate than African-Americans.

Also, certain types of blood pressure medications – ACE inhibitors, for example – lead to worse outcomes in African-Americans when used singularly as first-line therapy for high blood pressure, yet these medications work very well in Americans of European decent, a large study of hypertension therapy found.

A follow-up study that looked at subsequent clinical practices – which was done in response to changed recommendations based on race – showed nearly a third of African-American hypertensive patients continued to be prescribed medications that cause worse outcomes.

African-Americans also have a four-fold increased risk for renal disease leading to dialysis. Geneticists suspect that they have identified the gene that drives this difference yet most clinicians do not have the resources to test for this gene and identify the 30 percent of African-Americans that carry it.

And a gene that greatly increases the risk for Alzheimer's disease, APOE-4, has also been identified and occurs disproportionately higher in European-Americans yet is almost nonexistent in African-Americans and is inconsistent in Hispanic-Americans. Great controversy exists surrounding the testing for this gene, given the devastating impact it could have on a patient or family. (Hispanic and African-Americans still have a very significant risk for Alzheimer's disease, but it is not driven by this gene).

Genetically different responses to medications

Patient response to medications vary according to the presence or absence of genetic variants, which can impact the dose and the effect of many pharmaceuticals. Some of these differences can be anticipated based on race or ethnicity. For example, Warfarin is a commonly used medication in the treatment of a number of cardiovascular disorders

including atrial fibrillation, deep vein thrombosis and heart valve replacement. It shows wide variations in dosing, with Americans of Asian descent requiring less medication and African-Americans requiring more to achieve equal effects. European-Americans have a variant gene that make having a major bleed on Warfarin much higher.

A popular cholesterol-lowering medication, Rosuvastatin, better known as trade name Crestor, is twice as powerful in patients of Asian descent, and their manufacturing label indicates starting at a much lower dose in this population. In fact, the highest manufactured pill dose of Crestor is "contraindicated in Asian patients."

Patient-centered care is the key

Because of the "patient-centered" movement in hospitals, clinics and insurance plans, providers are now feeling increased pressure to improve the quality of care provided to individual patients. Many outcomes and patient cost of care are now tracked by providers. And countless well-designed studies have validated verified differences in the clinical care of a number of pervasive diseases based on ancestry.

Providers need to educate themselves about the important differences that exist in their patient populations. Health disparities, while driven by a number of social factors, are also the result of some clinicians not applying known nuances in the care of special populations.

As home genetic testing grows, patients will be bringing their results to physicians for reaction and response. Physicians will need to be proactively prepared.

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