DNA, race, and a new era in medicine

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The Human Genome Project, a 13-year international research effort, ended in 2003 with the complete mapping of the human genetic sequence. The US Department of Energy and the National Institutes of Health sponsored this project, which may be described as a "gateway" through which an architectural paradigm of the biology of human essence is provided. "Identifying all human DNA," "determining sequencing of 3 billion base pairs," "storing this information into databases," "improving tools for analysis," "transferring the data to the private sector," and "addressing ethical, legal, and social issues" were the project goals.[1]

The technology developed out of this endeavor has been revolutionary, and many professions and industries have been affected. Genomic medicine has produced remarkable advances, but it has also fueled widespread apprehension and trepidation, as well as zealous commercialism and entrepreneurship, particularly in the biotechnology arena. Research scientists, physicians, software giants, pharmaceutical companies, entrepreneurs, bioethicists, clergy, policy makers, attorneys, politicians, and others are engaged in a war over issues involving DNA testing, stem cell therapy, cloning, and commercial human biobanking. More and more, these issues are coming to affect us as PAs.

Personalized medicine for our patients?

Completing the Human Genome Project made it evident that the DNA sequence of any two people is 99.9% identical. The 0.1% variability is what makes us different from one another and is the focus of much of the efforts to develop genomic medicine. How will genomics change our clinical practice? Are we ready for the changes it will bring? What demands will it make of us as providers? Will it enhance our ability to treat patients?

Since all diseases have a genetic component, the Human Genome Project has opened the door for researchers to isolate genetic errors that cause disease.[2] Such advances will have a profound impact on the way diseases are predicted, diagnosed, treated, and prevented.

For instance, gene therapy is an exciting application. Diseases for which gene therapies are being developed and tested include cystic fibrosis, AIDS, sickle cell anemia, cancer, heart disease, and diabetes. A new generation of designer drugs is also fast approaching now that medicines based on genetic sequencing have become commercialized. Genes can tell us how people may respond to specific drugs, and personalized drugs can be developed to work effectively with a patient's genetic makeup.[3] This emerging field is called pharmacogenetics, and it now includes medicines for conditions such as diabetes, Alzheimer's disease, and heart disease.

The AAPA has policy on many of the issues discussed in Sounding Boards. To read the AAPA policy paper Genetic Testing in Clinical Practice, go to www.aapa.org/policy/PB5genetic-testing.html.

Race, ethnicity, and ancestry

The genomics revolution is affecting medicine in yet another way. Historically, race has been one of several factors used to guide treatment choices for some medical conditions, such as hypertension and heart disease. We were taught as clinicians to use race as an indicator for certain screening tests. Medical residents learned to include race when they presented patients to attending physicians. Scientists now say, however, that data do not support using race to govern treatment decisions.

The human genome originated in Africa, as most people now know, and most human populations share a mix of African, Asian, and European genes. During the 2005 American Society of Human Genetics Annual Conference in Salt Lake City, Utah, genomic and social scientific data on human genetic variation were presented. This presentation laid the groundwork for a robust debate on whether ancestry as opposed to race and ethnicity should be used when describing populations and selecting treatment modalities.

Traditionally, race and ethnicity are terms used to describe people who are united on the basis of physical characteristics that are genetically transmitted. Definitions may consider commonalities such as nationality, region of origin, religion, ancestry, culture, and language. A panel at the 2005 meeting stated that large numbers of people do not fall into only one category. During the discussion, scientists urged alternatives to using race as a population description and argued that because race is

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not congruent with observed human genetic variation, a new vocabulary is needed to describe human populations.

The panel spoke about the need to change not only how populations are described but also how research is done. Ethnicity, social demographics, ancestry, ethnoancestry, geographic ancestry, and biogeographic ancestry are the new terminologies recommended. Because genes provide a better predictor of how people respond to drugs and other treatments, using such terms would make discrimination and stereotyping less likely to influence assessment, treatment, and plans of care.[4]

Individual variations in the human genome can now provide a blueprint for treatment. Scientists are moving from treatments based on population genetics to those personalized to the individual. Experts like Georgia Dunston, PhD, Director of the Human Genome Resource Center at Howard University, are also urging pharmaceutical companies to tease out biological pathways and to avoid marketing solely on the basis of populations.

Some of the downside

As new genomic technology and its implications become more widely known, concerns grow about its possible misuse. DNA testing provides one example. On one hand, prospective parents seeking genetic information with which to plan a pregnancy, or wanting to learn whether an unborn child has a genetic defect, can benefit from genetic counseling that includes DNA testing. On the other hand, consider an identification card of the type that China is experimenting with. This card has a chip that contains 18-digit markers coding for the DNA of the cardholder and also includes nationality, gender, birth date, and parental information. According to experts, no 2 people in 10 billion have the same DNA—except in the case of identical twins. Because people may share a name and a birth date, and have similar facial features and even voices, experts in China argue that such a "gene" card would assist law officials since the DNA information it would carry would be unique to one person and would never change.[5]

With the headlines regularly carrying news of identity theft and continuing concerns about privacy, people are once again being confronted with the potential negative impact DNA testing may have on society. As a result, many may develop negative attitudes toward genetic research and hesitate to undergo genetic testing that could prove to be valuable if not lifesaving.

The role of PAs

As vital members of the health care community, PAs must enhance our knowledge, especially of available applications and of medical geneticists and genetic counselors. We must be sensitive to individual and community attitudes toward genetic testing. And we must continue to make patient privacy a high priority. Revealing genetic information about patients and their families may have profound emotional, financial, and psychological effects, especially in situations where discrimination can arise.

As we become educated, we must also educate and empower our communities. One thing we can do is to organize open community forum panel discussions designed to engage in interactive dialogue regarding the implications of genetic research for personal health, health care, and effects on society. An excellent resource is The National Human Genome Research Institute, which funds, supports, and develops research strategies. Resources available in association with National DNA Day (April 25th), which commemorates the completion of the Human Genome Project and the discovery of the double helix, can also be helpful. Over the past 3 years, the American Society of Human Genetics (ashg.org), the Genetic Alliance (geneticalliance.org), and the National Society of Genetic Counselors (nsgc.org) have sponsored this day. Available educational tools include Webcasts, chat rooms, and even mentorship to provide genetic experts for school classrooms. Resources, downloadable lectures, games, and lesson plans are available online.

Many people are fascinated with DNA research and how it has led to the discovery of genetic markers that can identify ancestral migration patterns. DNA tests may determine the percentage of a person's Native American, European, East Asian, or African ancestry. Identifying ancestral origin is made possible by genetic testing.[6] As more people learn about the Human Genome Project, DNA testing, and their impact on society, we can help to ensure that the promise this technology holds for us will be balanced with efforts to use it in a way that maintains human dignity.[7] □

JAAPA welcomes commentary about issues of significant concern to the PA profession. Authors should e-mail manuscripts to jaapa@aapa.org.

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