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New book ponders ethical issues of genetic testing

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A patient who tested positive for the gene that leads to Huntington's disease wrestled with a host of questions. Should she have children with her husband, knowing that each baby has a 50-50 chance of inheriting the mutation that causes the degenerative neurological illness? Should she have an abortion if prenatal testing showed the fetus had the mutation, or should she not have biological children at all?

Another patient with breast cancer who just learned that she has a genetic mutation associated with the disease asked psychiatrist and bioethicist Robert Klitzman, "Am I my genes?"

Klitzman, professor of clinical psychiatry at the College of Physicians and Surgeons and the Mailman School of Public Health, wasn't sure how to answer these or any of the myriad questions that come up when people learn they have a marker for inherited disease. And that led him to write his new book, *"Am I My Genes? Confronting Fate and Family Secrets in the Age of Genetic Testing,"* published by Oxford University Press.

With a new genome decoder hitting the market that will allow people to have their entire genetic makeup analyzed quickly and relatively cheaply, the challenges for patients and their doctors will get only more complex.

"We are going to be barraged by information," says Klitzman, who also directs the master's program in bioethics at Columbia's School of

Continuing Education. “There are many social, familial, ethical and legal questions the information brings up. How should we understand it? How should it affect our choices about who we marry, whether we screen embryos, how we see ourselves?”

Klitzman specializes in the intersection of medical theory and practice. His last book, When Doctors Become Patients, was influenced by his personal experiences after his sister was killed in the attacks on the World Trade Center.

In Am I My Genes? he interviews 64 patients grappling with one of three diseases, each of which has biomarkers for which they can be tested.

With breast cancer, the BRCA1 and BRCA2 mutations are responsible for between 5 and 10 percent of breast and ovarian cancers. While having the mutation dramatically increases the likelihood a woman will have cancer, not everyone with it will develop the disease.

Huntington’s disease often begins with psychiatric or movement-related symptoms when patients are in their 30s or 40s before it progresses to dementia. There is no effective treatment.

And alpha-1 antitrypsin deficiency is a disorder that can cause lung and liver disease. Although good treatment exists for it, many patients are misdiagnosed and told they have a more common pulmonary disease.

“Genetic information forces people to embark on journeys for which they are often unprepared. A lot of people are not used to thinking about this,” Klitzman says.

The diseases generate different sets of questions, and patients respond differently based on their cultural, religious and family perspective, as

well as life experiences. Looming large in the debate about genetic testing is the question of screening embryos for sex or possibly, in the future, other genetic markers correlated with desirable traits such as intelligence and athletic ability. They also can involve complicated privacy issues.

“Science is now giving us all this information,” Klitzman says. “Genetics can offer tremendous good, potentially. On the other hand, it can be misused.”

Major health care facilities are now engaged in biobanking—taking a sample of each patient’s genetic material and storing it. What would happen if law enforcement wanted to use material stored in a biobank to find a suspected criminal?

“Do we like biobanking because it will help the police find out who a rapist is? What if they decide they’re going to look at a mutation associated with being a rebel—will they use it in court? Markers may soon be identified that are associated with violence or impulsivity. Should that information be shared with a judge and a jury?”

Klitzman says the medical community, ethicists and government have not seriously grappled with the answers to these questions, but they should.

“I am interested in mapping out this brave new world that we are now entering so that we can be prepared to deal with it,” he says.“It turns out that genetics are much more complex than we thought. Genetics is both a lens into ourselves and a mirror back. It would be great to say, ‘Let’s not deal with it.’ But it’s here, and it’s here to stay.”

Provided by Columbia University