# Untangling Ethical Issues in Genetic Research

## DATA ADVANCES PRESENT FRESH CLINICAL OPPORTUNITIES — AND CHALLENGES

Imagine sitting in your physician's office and receiving the results of a genetics test. The results indicate that you have a high risk for Alzheimer's disease. Right now, there is no cure. What would you do with that information? What could your doctor do?

According to **Maureen Smith,** director of Northwestern's NUgene Project, this scenario illustrates some of the major ethical issues currently associated with genetics: the problems of returning research results to patients and preparing physicians and other healthcare providers with information and decision-support tools to help patients understand the results, as well as the need for much more research into the basis of and potential therapies for disease.

For Northwestern researchers pursuing such investigations into human medical conditions, a robust resource is available: NUgene, a biorepository of human DNA tied to the electronic medical records of approximately 12,000 patients. Investigators can use the DNA samples themselves or data aggregated from these patients. The NUgene Project is a long-term research enterprise sponsored by the Center for Genetic Medicine at the Feinberg School of Medicine.

NUgene researchers have collected these DNA samples from patient volunteers at Northwestern's medical affiliates, which provide the electronic medical records. The participants have granted consent for their samples and records to be used for medical research; they will not be identified by name in the research, nor will they receive the results of the particular investigation using their samples. They are kept informed of Northwestern's genomic research progress through the NUgene newsletter, which provides aggregate results in certain studies.

"None of this would be possible without the nearly 12,000 participants who've agreed to give of themselves to NUgene," says Smith. "We couldn't do any of this without people being altruistic and coming to Northwestern knowing that they are going to participate in research. So we're incredibly grateful to them."

One example of research using NUgene data is an ongoing study by the Northwestern Scleroderma Team led by **John Varga, MD,** John and Nancy Hughes Distinguished Professor, director of Northwestern's Scleroderma Program, and a member of the Robert H. Lurie Comprehensive Cancer Center of Northwestern University. His team is investigating the genetic basis of scleroderma, a rare, chronic autoimmune disease that causes skin and connective tissue to harden and tighten throughout the body. These genetic studies seek to identify potential new targets for classifying and treating the disease.

Smith and her colleagues are considering the question of returning results to individual participants, an issue that she describes as a rather recent development in biobanking research. Genomic research's sequencing technology can provide information about disease risk that participants have not anticipated, and sometimes

Photo used courtesy of NUgene Project

NUgene staff, clockwise from left: Tony Miqueli, Sharon Aufox, Oana Popescu, Maureen Smith, Michael Heathcote, and Jennifer Pacheco

the information is about diseases outside the scope of the original study. It's a complicated issue, one that involves respecting participants by providing information about research results as well as considering clinicians' role in interpreting those results.

"The way that medicine works now is if someone has a physical illness, they come to the doctor, and the doctor figures out what's causing it and how to treat it," says Smith, a board-certified genetic counselor. "If we are providing information before a person is sick, information about the risk to develop a condition before they have any signs, we are turning the medical paradigm on its head. The results could show a very high risk or it could be a very low risk. We don't know whether and in what instances that's going to be helpful for patients and their doctors."

The NUgene Project is currently conducting a pilot study in which researchers are examining genes for a predisposition to three different conditions and returning results to participants before they have signs of those conditions.

"We are learning all the time from these various studies that we are conducting," says Smith. "So we are trying to do them both with patients and with physicians in tandem to understand the downstream effects."

— *Joan Naper*