

Study of genetics offers new realm of possibilities

Island research center work known worldwide

The study of genetics, the area of science that includes inherited traits that are both positive and negative, has become in the last 10 years one of the most exotic, sophisticated and debated topics in medicine.

Peering into microscopes and performing time-consuming chemical tests, researchers are attempting to find the keys to unlock what makes certain people susceptible to diseases, deformities and malfunctions in their bodies.

By identifying or diagnosing genetic diseases like Tay-Sachs, mongolism or a variety of anemias, physicians can attempt to treat them, according to Dr. Nicholas G. Beratis, chief of genetics in the Institute for Basic Research, Willowbrook.

One of the institute's jobs is to help diagnose genetic disorders, particularly in its biochemical laboratory on the third floor.

On counters dotted with microscopes, test tube holders and glassware of every size and shape, technicians prepare cultures and slides for study.

But before any analysis starts in the institute's modern laboratories, persons suspecting they have a genetic disorder are carefully screened by a person called a genetic counselor.

It's his or her job to meet with suspected carriers and trace their families' histories, listing any member family who definitely had the disease.

Take, for example, the case of a Jewish woman who has a history of Tay-Sach's disease in her family. The disease affects Jews from Northern Europe and is a form of lipidosis, or a deficiency in the way a cell converts fats.

If she or her husband is a carrier of the disease and they decide to have children, there is a 50 percent chance they will produce a child who also will carry the disease, without grave side effects.

If both are carriers, there is a 25 percent chance of their child having the disease.

When a woman with a genetic disease becomes pregnant, she may, after meeting with a genetic counselor, decide to have a procedure called amniocentesis.

First used in 1930, the process involves a physician drawing some fluid from the amniotic sac that surrounds a fetus.

Just 13 years ago, the first studies were done using amniotic fluid to detect potential diseases linked to chromosomal abnormalities, like mongolism.

The process is simple. A physician identifies where the fetus is located in the uterus by ultrasonography. Sound waves are bounced through a woman's abdomen and the reflected waves are transformed into an outline of the fetus, which appears on a screen.

A physician then takes a fine needle and guides it into the fluid surrounding the fetus and draws about a half ounce of fluid. The procedure is usually done in the second trimester, or between the 12th and 24th week of pregnancy.

The fluid is then sent to a laboratory to be analyzed.

Here's where the Institute for Basic Research gets involved.

Once it gets the fluid, it is put into a centrifuge and cells are separated from the fluid. The cells are then placed in a medium filled with nutrients and cultured for between two and three weeks, although in some cases, the process can be expedited.

After the cells have been growing, a technician prepares slides of the cells and they are analyzed under high-power microscopes in either the institute's chromosomal laboratory or its biochemical laboratory.

Results of the analysis are then returned to the physician or hospital that sent the fluid to the institute.

"A genetic counselor does not tell parents what to do," emphasized Dr. Beratis. "Our job is to give them all the facts."

Those facts may often include that their child has a high risk of being born deformed or will not live long after it is born.

Dr. Edmund C. Jenkins, head of the institute's cytogenetics laboratory, said that many people don't take advantage of genetic counseling "because they feel there's no alternative to abortion."

In rare incidents of genetic diseases, some treatment is available.

The Willowbrook resident, who has refined and modified some staining techniques to improve analysis of chromosomal studies, said that's just one reason why there's a need for more research into developmental diseases and abnormalities.

Under the state Department of Mental Hygiene, the institute serves the entire metropolitan area, accepts cases for study from all over the state, and occasionally, has studied cases from throughout the world.

—CHRIS OLERT