FIFTY YEARS OF HUMAN GENETICS AT MICHIGAN

DEPARTMENT'S ANNIVERSARY CELEBRATES THE PAST AND FUTURE OF THE GENETICS REVOLUTION

Spend some time in the Buhl Building, which houses the Department of Human Genetics, and you're likely to notice a palpable sense of anticipation. Sally Camper, Ph.D., the James V. Neel Collegiate Professor of Human Genetics, certainly does. Camper has chaired the department — the first human genetics department in the nation — since January 2005.

"There's a lot of excitement around here about understanding genetics at a completely different level now," says Camper. When the department was established in 1956, Watson and Crick had discovered the structure of DNA just three years before. By 2000, the human genome had been mapped. "So many areas for investigation have opened up," she says.

Microbial and mouse genetics played key roles in laying the department's foundation too, Camper notes. In 1964 Myron Levine, Ph.D., now professor emeritus of human genetics, published a landmark study describing how viruses alter bacterial DNA. His work paved the way for many genetic engineering techniques used today. One of Levine's post-doctoral fellows, Hamilton "Ham" Smith, went on to receive the Nobel Prize in 1977 for his work on DNA restriction enzymes, chemical 'scissors' that cut DNA.

Among the department's strengths, says Camper, are its contributions in the areas of gene identification for Mendelian disorders, caused by a single genetic mutation; developing animal models of disease; and the department's affiliation with such an outstanding hospital.

The strong link between clinical practice and the science of genetics was evident right from the start: in the department's very first year, Neel launched a series of lectures on genetics for medical students as part of their curriculum. Patients whose family history of colon and related cancers has been studied and documented since the late 1800s still regularly come to the Cancer Genetics Clinic today, notes Camper. The clan's records, which include data from 929 descendants over seven generations, comprise one of the largest medical histories on record. Countless other families have benefited from insights provided by counselors trained in the department's genetics counseling program. Established in 1979, the two-year, master's level program was reaccredited in 2005.

NEEL'S VISION REALIZED

The department was founded in 1956 by James V. Neel, M.D., Ph.D., a prominent population geneticist and visionary who studied the interaction between the environment and genetics and foresaw how genetics could improve the diagnosis and treatment of medical conditions. He served as chair for a quarter-century. Neel made seminal contributions to the field through his discovery of the genetic basis for Cooley's and sickle cell anemias, extensive work in Japan on the long-term effects of radiation, proposing the "thrifty gene" hypothesis (which explained such diseases as diabetes and obesity as genetic survival mechanisms during times of scarcity) and his work describing the genetic characteristics of isolated Amazonian Indian tribes. Neel died in 2000.

Photo: Gregory Fox

James Neel in 1998

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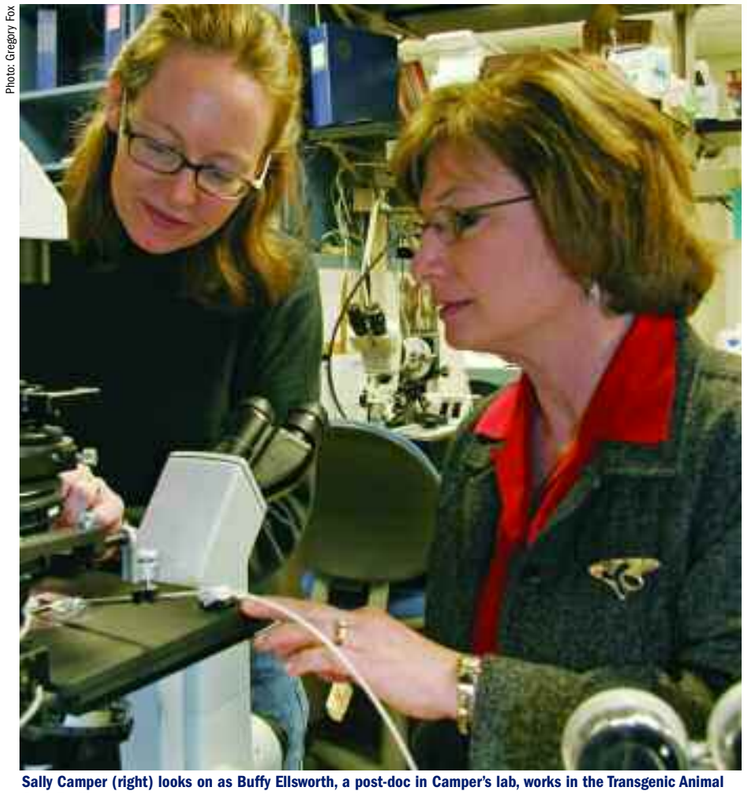

Sally Camper (right) looks on as Buffy Ellsworth, a post-doc in Camper's lab, works in the Transgenic Animal Core Facility.

THE PACE OF DISCOVERY QUICKENS

Thomas Gelehrter, M.D., professor of human genetics, who did groundbreaking work investigating the molecular mechanisms of gene function, became chair of the department in 1988. Chief among his accomplishments, says Camper, was recruiting faculty working in that area. "That level of understanding is so important when you're trying to identify the gene for something like cystic fibrosis," says Camper, "to understand how it mutates and causes disease."

Identify the gene for cystic fibrosis is precisely what Francis Collins, M.D., Ph.D., did the following year. He then discovered the genes for neurofibromatosis, a genetic disorder that causes tumors to grow on nerves, as well as Huntington's disease, a degenerative brain disorder. Together Collins and Gelehrter published the definitive medical genetics text, *Principles of Medical Genetics*, in 1990. Three years later Collins was tapped to head the National Human Genome Research Institute.

Over the next decade faculty research advances continued at a rapid pace: isolating and cloning genes for several neurological diseases for further study; discovering the effects of mutations in the genes that control body development; curing Duchenne muscular dystrophy in animal models. Much of this work was done using mice in the department's Transgenic Animal Core Facility, established by Camper in 1989 shortly after she joined the faculty. Camper and colleagues also identified the gene responsible for congenital deafness and are exploring gene therapy approaches to correct the mutation.

Work on gene therapy continues in the Center for Gene Therapy, established in 1997. The center's researchers have made strides toward understanding — and repairing — the genetic flaws that occur in conditions such as hemophilia B and epilepsy.

A FUTURE DRIVEN BY TECHNOLOGY

Camper is often asked about the future of genetics. "There's been an interesting evolution," she says. "Now that we have the whole human genome at our disposal, I see us entering a time where we discover more about complex diseases — diabetes, heart disease, asthma, cancer, Alzheimer's — in families and in larger populations."

And, of course, translating those insights to clinical care. In the spring of 2005, the University of Michigan Board of Regents approved a new Center for Genetic Health and Medicine. Its mission is to act as a catalyst for further collaboration among clinicians, geneticists and biostatisticians. Two postdoctoral researchers have already earned fellowships to continue work on programmed cell death, a process associated with cancer and other diseases, and the genetics of aging.

Faculty are pursuing many more promising areas that fuel Camper's enthusiasm and expectations: tailoring medical treatment to an individual's genetic makeup; better understanding the interplay of genetics and environmental factors; refining 'lab-on-a-chip' technology that makes high-throughput, cost-effective analysis of genetic material possible, for starters.

"Technological advances really do drive the science," she says. "We've gone from the arduous process of sequencing one genome to a time when we might sequence an individual's entire DNA. That's amazing, and it will really change what's feasible."

—Kim Roth