



## NUgene Funded to Join Genomics Network

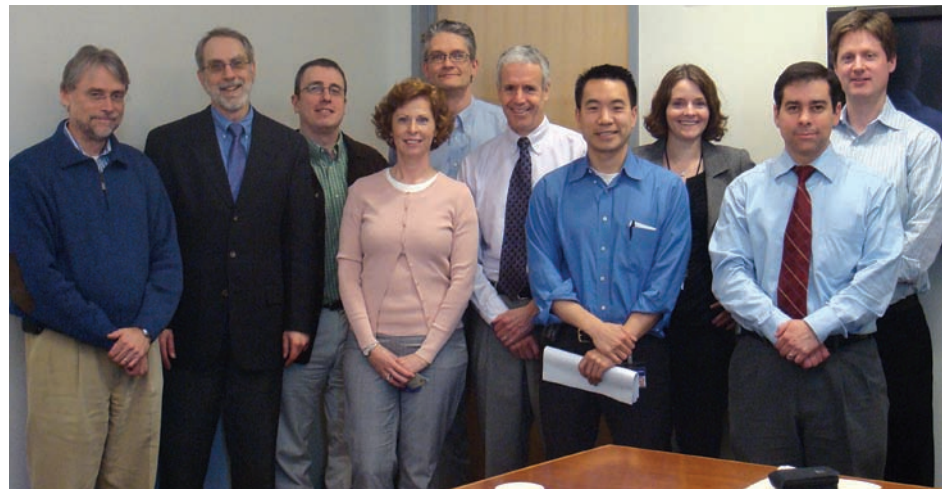
**N**Ugene is excited to announce a \$2.4 million award from the National Institutes of Health to participate in the national Electronic Medical Records and Genomics Network (eMERGE), which integrates two areas key to personalized medicine: informatics and genetics.

The network of five institutions, collectively awarded \$20 million by the National Human Genome Research Institute, a division of NIH, will evaluate whether data from electronic medical records (EMRs) can be used for large-scale genetic research. The other participants are Marshfield Clinic in Wisconsin, Mayo Clinic, Vanderbilt University, and the Seattle-based Group Health Cooperative.

The award marks an important milestone in the short history of NUgene, which was launched by CGM in 2002 as a biorepository of DNA samples and health-care information from participating Northwestern patients. The biobank now facilitates scientific investigations of genetic contributions to diseases and their treatments. It has logged 7,800 samples that are being used in eight studies.

Awardees of the NIH funding will study genetic variation of particular traits through a genome-wide association approach, using samples and data from their institutional biobanks. As these studies are conducted, network institutions will also evaluate the viability of using biobank-coupled EMR data for research, which would be significantly more efficient than the time-consuming, expensive process of soliciting new participants for each project.

Rex Chisholm, dean for research in the Feinberg School, former director of CGM, and



founder of NUgene, will lead Northwestern's activities under the four-year award.

The Northwestern research team will conduct genome-wide association studies to determine which gene variants might be related to type 2 diabetes and asthma, while assessing the suitability of EMR data to identify cases and controls for both conditions. The team will also determine best practices for community engagement and data sharing, taking into account the ethical concerns associated with privacy and confidentiality.

In addition, eMERGE network institutions are considering a variety of cross-institutional studies that could only be performed across a large patient population, with the ultimate goal of more rapidly identifying gene variants related to disease and therapeutic efficacy. Several sites expressed interest in collaborative efforts on type 2 diabetes and statin adverse effects. If these projects move forward, cases would be ascertained across multiple sites.

Given the interdisciplinary nature of this work, Northwestern has assembled a

**Members of the NU eMERGE team, from left to right: principal investigator Rex Chisholm, coinvestigators Phil Greenland, Geoff Hayes, Maureen Smith, Warren Kibbe, Bill Lowe, Abel Kho, Wendy Wolf, and Pedro Avila, and data architect Andrew Winter. Dave Baker and Amy Lemke were not present for the photograph.**

collaborative research group from the Feinberg School of Medicine with wide-ranging expertise. The project's coinvestigators include William Lowe, professor, and M. Geoffrey Hayes, assistant professor, both of endocrinology; Pedro Avila, associate professor, allergy-immunology; David Baker, professor, and Abel Kho, assistant professor, both of general internal medicine; Philip Greenland, professor, preventive medicine; Amy Lemke, research assistant professor, Center for Genetic Medicine; and Maureen Smith, instructor, and Wendy Wolf, research assistant professor, both from the NUgene project.

For more information on NUgene, please visit [www.nugene.org](http://www.nugene.org).

# Interim Director Peter Kopp Plans New CGM Initiatives



**Peter Kopp**

**N**ew interim CGM director Peter Kopp, associate professor of medicine in the Feinberg School division of endocrinology, metabolism, and molecular medicine, looks to draw on the center's strengths and the experience of its division directors to improve current programs and start new initiatives. He succeeds Rex Chisholm, who has moved into a new role as dean for research in Feinberg.

Kopp's plans include expanding the services provided by the center's Transgenic and Targeted Mutagenesis Laboratory for the cryopreservation of mouse embryos, hiring an assistant director for the Graduate Program in Genetic Counseling, redesigning that program's curriculum, promoting CGM's many outreach and educational initiatives, and continuing administrative support of the NUgene project. He will also maintain the center's role in recruiting new faculty to broaden the University's genetics expertise.

Enhancements are already evident in CGM's Genomics Core Facility, which provides a variety of DNA sequencing and genotyping services. A generous gift from Northwestern trustee and Feinberg School benefactor Ann Lurie enabled the core to purchase a high-throughput DNA sequencer, the first of its kind in the Chicago area, which will be available for use later this month. Kopp and core director Nadereh Jafari are also working toward automating many aspects of the facility's services, allowing it to handle larger projects and provide more efficient and reliable service. Finally, they also hope to expand the available bioinformatics analysis by hiring personnel with this skill set. More information on the core's new instrumentation can be found on page 5.

In addition to building on the center's current efforts, Kopp plans to initiate several others. In collaboration with Greg Beitel, the center's new associate director and associate professor of biochemistry, molecular biology, and cell biology in the Judd A. and Marjorie Weinberg College of Arts and Sciences, Kopp is planning a new graduate program in genetics. He also hopes to hire a clinical deputy director as the first step toward establishing a clinical arm in CGM. Both of these initiatives are in the early planning stages.

As a Feinberg faculty member and a Northwestern Memorial Faculty Foundation consultant, Kopp has been a center member since 2002. He brings both clinical and research expertise to the directorship. He earned his MD at the University of Berne, Switzerland, where he was an attending physician and researcher until moving to the United States in 1993. He started his career at Northwestern that same year as a research fellow in the lab of J. Larry Jameson, now Lewis Landsberg Dean and vice president for medical affairs at Feinberg. Kopp became an assistant professor of medicine in the division of endocrinology, metabolism, and molecular medicine in 1997 and was awarded tenure in 2006.

His research concentrates on congenital hypothyroidism and Pendred syndrome, a genetic disorder marked by congenital deafness and thyroid abnormalities. Pendred syndrome is caused by a mutation in the gene that encodes pendrin, an anion transporter. Kopp's work focuses on the function and structure of pendrin in thyroid cells. His lab also studies the genetics of diabetes insipidus, a condition that prevents the necessary retention of water, and the effects of genetic variants on androgen levels in young men, which is important for understanding aspects of future prostate cancer development.

Kopp's affiliations include the American Thyroid Association, where he is a member and former chair of the program committee and current chair of the continuing medical education and guidelines committee. He is an associate editor of the journal *Thyroid* and is on the editorial board of the journal *Endocrinology*.



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## Nobel Laureate to Deliver Scott Lecture

**N**obel laureate Craig C. Mello, Blais University Chair in Molecular Medicine at the University of Massachusetts Medical School and an investigator with the Howard Hughes Medical Institute, will deliver the 2008 Richard A. Scott Lecture on May 13.

Mello is widely known for his groundbreaking work in the area of RNA interference (RNAi). With colleague Andrew Fire, currently at Stanford University School of Medicine, he demonstrated in 1998 that double-stranded RNA was capable of silencing gene expression in a sequence-specific manner. The ability of RNA to regulate gene expression has since been shown to be a normal part of the developmental



Craig C. Mello

process for a wide array of organisms, including humans. The discovery of RNAi has proved to be not only an extremely powerful research tool for studying gene expression in model organisms but also a potential therapeutic avenue for treating cancer, heart disease, and viral infections.

Mello and Fire's discovery arose from their interest in studying genes involved in embryonic development. The Mello lab is interested in understanding the early spatial and temporal events involved in cell fate decisions. Working with the model organism *Caenorhabditis elegans* (*C. elegans*), Mello demonstrated that cell position in the embryo is an important determinant of cell fate, and he has identified numerous genes involved in directing cell-fate decisions. Recently his lab has provided important insights into the mechanisms by which double-stranded RNA is processed and into the protein networks involved in RNA-mediated gene silencing.

Mello received a bachelor's degree in biochemistry from Brown University and a PhD in cellular and developmental biology from Harvard. After a postdoctoral fellowship at the Fred Hutchinson Cancer Research Center, he joined the faculty at the University of Massachusetts Medical School in 1995. In addition to the 2006 Nobel Prize, Mello's honors include the 2006 Paul Ehrlich and Ludwig Darmstaedter Prize, membership in the National Academy of Sciences in 2005, the Wiley Prize in Biomedical Sciences from Rockefeller University in 2003, and the National Academy of Sciences Award in Molecular Biology in 2003.

**Richard A. Scott, MD,  
2008 Lecture  
Tuesday, May 13, 4 p.m.  
Hughes Auditorium  
Robert H. Lurie Medical  
Research Center  
303 East Superior Street  
Chicago Campus  
Reception to follow in  
the Lurie Atrium**

The Scott Lecture Series was established in 2001 to enable CGM to bring a leader in the field of genetics to Northwestern. It is funded by a generous bequest from Richard A. Scott, MD, a graduate of the Northwestern University Medical School. Scott's subsequent medical career focused on cardiovascular disease and pathology. After he passed away in August 2000, his wife, Anne Lesak Scott, and family established the CGM lecture series to honor his lifelong interest in research.

Event details can be found on the CGM web site, [www.cgm.northwestern.edu/scott.htm](http://www.cgm.northwestern.edu/scott.htm).

## Hinchcliff Receives NMF Fellowship in Genetic Medicine

**M**onique Hinchcliff, clinical instructor of medicine in the division of rheumatology at Feinberg, was awarded the 2007 Northwestern Memorial Foundation MD-Scientist Fellowship in Genetic Medicine for her project “The Role

of TGF- $\beta$  Genetic Variants in the Pathogenesis of Scleroderma.”

Hinchcliff’s innovative research builds on the resources of the Northwestern Scleroderma Program, one of few scleroderma translational research centers in the United States, and the genetic capabilities of the NUGene project.

Scleroderma (SSc) is a chronic autoimmune disease affecting 100,000 Americans, primarily young and middle-aged women. It can cause scar tissue to form on skin and internal organs, leading to the thickening or hardening of the affected areas. While mild in some cases, SSc has the highest mortality among connective tissue

diseases, and there is no known cure. The pathogenesis of scleroderma is complex, involving interplay of three seemingly diverse processes: autoimmunity, fibrosis, and vasculopathy.

No existing biomarkers adequately predict the subtype or course of the disease or its response to various treatments. Because SSc is a clinically heterogeneous disease, with some patients following a very indolent course and others rapidly progressing to severe disease, better methods of predicting outcome and response to treatment are needed.

Investigators at Northwestern have already discovered that the Transforming Growth Factor-beta (TGF- $\beta$ ) plays a central role in the initiation and propagation of fibrosis in scleroderma and other diseases. When TGF- $\beta$  binds to its cognate receptor Transforming Growth Factor-beta receptor 2 (TGFBR2), it activates TGFBR1, Transforming Growth Factor-beta receptor 1. This initiates the SMAD cascade, a group of intracellular signaling molecules, and appears to activate sev-

eral profibrotic intracellular signaling cascades that ultimately lead to the excess deposition of collagen and other extracellular matrix proteins.

Northwestern’s current translational genetic research project tests the novel hypothesis that certain common and functionally relevant variants of TGF- $\beta$  and TGFBR1 predispose individuals to developing SSc. Hinchcliff is working with Boris Pasche, associate professor of medicine in the division of hematology and oncology, and John Varga, professor of medicine in the division of rheumatology, to understand whether genetic polymorphisms in the TGF- $\beta$  pathway contribute to SSc disease heterogeneity. Using NUGene repository DNA samples collected from patients seen in the Northwestern Scleroderma Program over the last three years, Hinchcliff is testing whether individuals with different subtypes of SSc possess distinct genetic polymorphisms in TGF- $\beta$  and TGFBR1. She is also investigating whether certain polymorphisms can predict disease subtype, progression of skin thickening, and internal organ involvement.

Hinchcliff has several long-term goals for her work, including laying the foundation for multiinstitutional studies that can examine larger numbers of SSc patients. She also hopes to continue bringing discoveries made in the laboratory to the clinic, where she can help patients and their families better understand SSc, while identifying better methods for predicting disease severity and response to treatments. As a fellow at Northwestern, she witnessed firsthand the disconnect between basic scleroderma researchers and clinicians caring for patients with this rare disease. Several clinical trials are under way through Northwestern’s Scleroderma Program to determine whether recent laboratory discoveries can directly benefit patients with SSc.

The Northwestern Memorial Foundation MD-Scientist Fellowship in Genetic Medicine is provided by the generous support of the Northwestern Memorial Foundation. The award is designed to allow a highly qualified senior fellow the necessary time and support to develop and conduct a pilot research project, making him or her competitive for a career development award. The 2008 winner, Malek Muayed, will be profiled in the next issue.



**Monique Hinchcliff**

## New Instruments Keep Genomics Core at Forefront

**T**he Genomics Core unveils two new instruments this spring, ensuring that the facility remains at the forefront of DNA sequencing technology.

The first is a SOLiD™ System, a next-generation sequencer, acquired from Applied Biosystems through a generous donation from Northwestern trustee and Feinberg School benefactor Ann Lurie. According to Nadereh Jafari, Genomics Core director, next-generation sequencing will provide “another level of understanding of our genome” and make DNA sequencing faster, more comprehensive, and much less expensive.

The SOLiD System is a genetic analysis platform that enables massively parallel sequencing of clonally amplified DNA fragments linked to beads. The sequencing methodology is based on sequential ligation with dye-labeled oligonucleotides. The system is a “short reader,” meaning it can read between 25 and 35 bases at a time and sequence four gigabases per run.

Mate-paired sequence analysis enables detection and resolution of sequence variation, including SNPs (single nucleotide polymorphisms), gene CNVs (copy number variations), duplications, inversions, insertions, and deletions. Mate-paired libraries made with large insert sizes are required for the analysis of complex genomes such as human, mouse, and other model organisms. The SOLiD System is ideal for tag-based applications such as gene expression and ChIP sequencing, where a large number of reads are required.

The Genomics Core will dedicate a technician to the new sequencer to ensure the timeliness and quality of service. It also plans to hire a data manager and a bioinformaticist to offer high-level analysis. The system will be available for use this summer.

The facility has also acquired a Biomek ArrayPlex NX automated workstation, purchased with grant funds awarded by the vice president for research's Shared Facilities Program. The workstation will automate the setup of sequencing reactions, which, Jafari notes, will minimize mistakes and increase productivity. This instrument should be up and running later this spring.



**Genomics Core staff member Isabel Angulo prepares the new Biomek ArrayPlex NX automated workstation for use later this spring.**

### 10 Graduate from Genetic Counseling Program

CGM congratulates the 2008 graduates of the Graduate Program in Genetic Counseling. They are listed with their thesis titles:

**Angela Darnell**, “Deafness Curriculum in Clinical Genetics Training Programs”

**Kate Durda**, “Measurement of the Downstream Revenue Generated by a Clinical Cancer Genetics Program”

**Jessica Hartley**, “Identification of Colonoscopy Patients at High Risk for Familial Cancer: Physicians’ Opinions about Referral to a Cancer Genetics Program”

**Megan Latchaw**, “Health Insurance Coverage of Genetic Services in Illinois”

**Ann Lee**, “An Assessment of Billing Practices of Genetic Service Providers in Illinois”

**Nikita Mehta**, “Delineation of Patients with 9p Interstitial Deletion: A New Syndrome Mediated by Repetitive DNA in Proximal 9p”

**Christina Seelaus**, “Attitudes of Maternal-Fetal Medicine Specialists about Counseling Prenatal Patients on Soft Markers Seen on Ultrasound”

**Emily Smith**, “Reasons for Nonparticipation in a Hospital-Based Biobank”

**Ashley VanDenBoom**, “Common Practice among Obstetricians in Screening for Fetal Chromosome Abnormalities”

**Jessica Wehr**, “Pathologists’ Attitudes and Knowledge Regarding the Routine Screening of Lynch Syndrome-Related Tumors with MSI and IHC”

## CGM Member Profile: M. Geoffrey Hayes

**R**ecruited to Northwestern in 2007 by CGM and Feinberg's Department of Medicine, M. Geoffrey Hayes is an assistant professor of medicine in the division of endocrinology, metabolism, and molecular medicine and a member of the center; he is also an adjunct professor of anthropology in the Judd A. and Marjorie Weinberg College of Arts and Sciences.



M. Geoffrey Hayes

Hayes came to Northwestern eager to work with investigators interested in developing large-scale genome-wide association studies, to which he brings statistical analysis expertise and an evolutionary genetics perspective. His interests lie in using genetics to find out not just how people get sick but also why.

By using evolutionary genetic methods to study the natural history of a disease, for example, he notes that researchers may be able to determine why genes that increase risk for disease, such as those underlying type 2 diabetes susceptibility, remain so prevalent in human populations today.

He is currently involved in two genome-wide association studies (GWAS) at the University. The first project, "GWA Mapping: Maternal Metabolism-Birth Weight Interactions," is headed by William Lowe, professor of medicine in the division of endocrinology, metabolism, and molecular medicine, and funded by the National Institutes of Health. Using a subset of the DNA samples collected by the Hyperglycemia and Adverse Pregnancy Outcome (HAPO) Study, Lowe, Hayes, and their colleagues will map the genes that shape birth size by investigating the interaction between glucose levels in pregnant women and genetic variation in both mothers and babies. This study is part of the Gene Environment Association (GENEVA), the first project funded by NIH's Genes, Environment, and Health Initiative (GEI). GENEVA comprises seven other genome-wide association studies, two genotyping centers, and 30 environmental technology projects.

Hayes will also provide statistical analysis for the studies conducted by Northwestern's NIH-funded Electronic Medical Records and Genomics

Network (eMERGE) team. (See page 1.) Led by Rex Chisholm, dean for research in the Feinberg School, former director of CGM, and founder of NUgene, the team will test the use of biobanks — DNA samples tied to electronic medical records — to conduct GWAS. They will look at two phenotypes: type 2 diabetes, a disease commonly studied in GWAS, and asthma. The latter may prove challenging using the biobank method, Hayes notes, since determining cases and controls from medical records may be difficult. There are a number of different diagnoses for asthma symptoms recorded in the electronic medical records, and researchers must rely on these, billing records, and insurance codes rather than hallmark diagnostic tests such as a methacholine challenge.

Finally, Hayes will revisit his graduate anthropological work in a project recently funded by the National Science Foundation as part of its International Polar Year initiative. In collaboration with Dennis O'Rourke at the University of Utah, Hayes will study the genetic variation of the Inupiat population of Alaska's northern coastline (the North Slope). Starting in the village of Barrow this summer and continuing the next three summers in another seven villages, Hayes and his lab will collect DNA from volunteers to get a sense of the genetic variation across the region today. Coupled with the genetic information from prehistoric human remains collected by O'Rourke, the data will show genetic variation across space and time. Eventually, the data will also allow researchers to determine ancestor-descendant relationships of the current Inupiat people and members of earlier populations in the region, analyze migrations, and study replacement versus admixing colonization models.

As a graduate student at the University of Utah, Hayes used ancient DNA methods to test migration models posed by the archaeological record in the East Canadian Arctic and the Aleutian Islands. He earned his PhD in anthropology in 2002. He completed his postdoctoral work at the University of Chicago, where he spent the last few years working on a GWAS of type 2 diabetes in a Mexican-American population in Starr County, Texas, the first to be conducted in a highly admixed population. His work has been published in *Diabetes*, *Human Biology*, *Molecular Genetics and Metabolism*, and the *Proceedings of the National Academy of Sciences*, and he currently serves on the advisory board of CGM's Genomics Core Facility.

## Travel Fellowship Winners Present Research at Scholarly Meetings

The CGM Travel Fellowship competition awards outstanding graduate students and postdoctoral fellows a \$500 travel stipend to present the results of their genetics-based research at scientific conferences across the country. CGM covers some of the costs of traveling to these meetings, where researchers network and share their findings with other scientific professionals. Below are the fall 2007 CGM Travel Fellowship recipients:



**Ryan P. Hobbs,**  
“PKC $\beta$ -dependent Modulation of Desmoplakin-Intermediate Filament Interactions through Plakophilin-2 Promotes Desmoplakin Assembly into Desmosomes,”

presented as a part of the intermediate filament subsection at the 47th annual meeting of the American Society for Cell Biology (ASCB) December 1–5 in Washington, D.C. Adviser/mentor: Kathy J. Green, professor of pathology and dermatology, Feinberg School of Medicine.



**Milan Joksimovic,**  
“Neurogenic Potential of the Rostral Floor Plate,” presented at the 37th annual Society for Neuroscience Conference November 3–7 in San Diego. His

research showed *in vivo* how the dynamic interplay of canonical Wnt signaling and Shh may orchestrate floor-plate neurogenesis and a production of dopamine neurons. Adviser/mentor: Raj Awatramani, assistant professor of neurology, Feinberg.



**Minjong Park,**  
“Rab32 Regulates Melanosome Transport in *Xenopus* Melanophores by Protein Kinase A (C $\beta$ /R11 $\beta$ ) Recruitment,” also presented at the December ASCB meet-

ing. Park received feedback from other labs that are trying to elucidate how organelle transport is regulated. Adviser/mentor: Vladimir Gelfand, professor of cell and molecular biology, Feinberg.

CGM also congratulates the spring 2008 Travel Fellowship winners, **Aparna Ramachandran** and **Rachel Lander**. Their work will be featured in the fall 2008 newsletter.

Travel Fellowship winners from summer 2007 through spring 2008 will be honored at the Robert H. Lurie Comprehensive Cancer Center of Northwestern University’s Annual Posters and Wine Session on June 25 in the Lurie Atrium. Winners will display their research posters for the Northwestern community.

All graduate students and postdoctoral fellows who are participating in genetics-based research at the University are encouraged to apply for upcoming Travel Fellowship awards. The next deadline is Friday, July 11. For more information, please visit [www.cgm.northwestern.edu/travel\\_app.htm](http://www.cgm.northwestern.edu/travel_app.htm).

## Transgenic Lab Moves to Olson Pavilion

The Transgenic and Targeted Mutagenesis Laboratory main lab has moved into its recently renovated suite, 8402, within Robert H. Lurie Comprehensive Cancer Center space on the eighth floor of the Olson Pavilion, 710 North Fairbanks Court. The embryonic stem cell lab and laboratory director Lynn Doglio’s office are now in this location. An open house will be held in the near future. For more information, contact Doglio at 312-503-0088 or stop by the lab.

## Honors and Awards for CGM Members



From left: Erwin Goldberg, Kasturi Haldar, Stephen Miller, Hank Seifert, Paula Stern, Robert Vassar, and Teresa Woodruff

**Erwin Goldberg**, professor of biochemistry, molecular biology, and cell biology in Weinberg College, has been elected to a three-year term as treasurer of the American Society of Andrology. ASA promotes collaboration among many disciplines to advance knowledge of the male reproductive system. Goldberg is using genetics and molecular genetic approaches to understand the regulation of male spermatogenesis. The two main approaches at present involve transgenes for ectopic expression of stage-specific genes and targeted disruption of testis-specific genes. Goldberg is also collaborating on HIV research with scientists at Yale University and the Southwest Foundation for Biological Research in San Antonio.

**Kasturi Haldar**, Charles E. and Emma H. Morrison Professor of Pathology and professor of microbiology-immunology at Feinberg, has been named editor in chief of *PLoS Pathogens*. Published by the Public Library of Science, the journal is an open-access publication of peer-reviewed research related to pathogen biology. Haldar's research focuses on understanding the regulation of pathogenic vacuoles and their interaction with the secretory pathway responsible for delivering the proteins and lipids necessary for membrane growth in organelles of eukaryotic cells.

**Stephen Miller** has been named Judy Gugenheim Research Professor in Microbiology-Immunology at Feinberg. Miller is internationally recognized for his research on pathogenesis and regulation of autoimmune diseases. His work has significantly enhanced the understanding of immune inflammatory processes underlying chronic autoimmune disease by employing animal models of multiple sclerosis and, more recently, type 1 diabetes.

**Hank Seifert**, professor of microbiology-immunology at Feinberg, has been elected a fellow of the American Association for the Advancement of Science. Fellows are elected by AAAS peers in recognition of their contribution to advancing science and its applications. Seifert's lab studies the

molecular mechanisms used by the strict human pathogen *Neisseria gonorrhoeae* to evade innate and adaptive immune responses to cause the sexually transmitted disease gonorrhea.

**Paula Stern**, professor and vice chair of molecular pharmacology and biological chemistry at Feinberg, has been recognized as the 2008 Distinguished Woman in Medicine and Science by the Northwestern Medical Women Faculty Organization. The award is given annually for model leadership in medicine and science. Stern's research investigates signal transduction pathways mediating the effects of hormones, cytokines, physical forces, and pharmacological agents on bone cells. Her recent work focuses on a parathyroid hormone-stimulated  $\text{Gal}\alpha 12/\text{Gal}\alpha 13\text{-RhoA-phospholipase D}$  signaling pathway identified in osteoblastic cells — those responsible for bone formation — and its involvement in cytoskeletal integrity and cell survival.

**Robert Vassar**, associate professor of cell and molecular biology at Feinberg, has received the MetLife Foundation Award for Medical Research in Alzheimer's disease. Vassar researches the role of  $\text{A}\beta$ , the beta-amyloid peptide that forms amyloid plaques, one of the types of brain lesions that characterize the pathology of Alzheimer's disease, and BACE1, which is required for the generation of all forms of  $\text{A}\beta$ .

**Teresa Woodruff**, Thomas J. Watkins Professor of Obstetrics and Gynecology and chief of Feinberg's Division of Fertility Preservation, received a National Institutes of Health Roadmap Grant for work in fertility preservation in female cancer patients. The grant funds the Oncofertility Consortium, a nationwide interdisciplinary group developed by Woodruff that is dedicated to the advancement of technologies that will provide improved fertility-preserving options to cancer patients. Woodruff's laboratory investigates the regulation of ovarian follicle growth during the mammalian reproductive cycle, a process essential for fertility that may be disrupted during cancer treatment.

# Mouse Genetics Group Holds Third Biannual Meeting

**K**eynote speaker David Valenzuela, vice president for functional genomics and chief of the VelociGene division of Regeneron Pharmaceuticals, discussed the impact of transgenic mouse research on the pharmaceutical industry at the spring 2008 Mouse Genetics Group meeting May 9.

During his seminar, “The New Mouse Transgenics: From Genetic Research to Drug Discovery and Development,” Valenzuela described new technologies that will bring drugs to clinical development more quickly and efficiently.

The mouse genome comprises an estimated 25,000 genes, only a small fraction of which have a known function. The most common approach to discovering the function of a gene is to examine the effects of inactivating or “knocking out” the gene. Producing knockout mice involves the genetic manipulation of embryonic stem (ES) cells and is notoriously time consuming and expensive. Valenzuela and his colleagues have developed new technologies that allow the rapid gene targeting in ES cells and the efficient generation of mice directly from the manipulated cells.

By combining new technologies with automation, Regeneron has dramatically increased the throughput and speed of knockout mouse production. As a result, the VelociGene division is one of two research groups chosen by the National Institutes of Health to spearhead the Knockout Mouse Project (KOMP), a federally funded initiative to knock out all known genes in the mouse. Valenzuela and his group plan to generate 3,500 knockout ES cell lines over a five-year period. Also, in what can only be described as a genetic tour de force, Valenzuela’s group has genetically engineered mice that can be used to produce

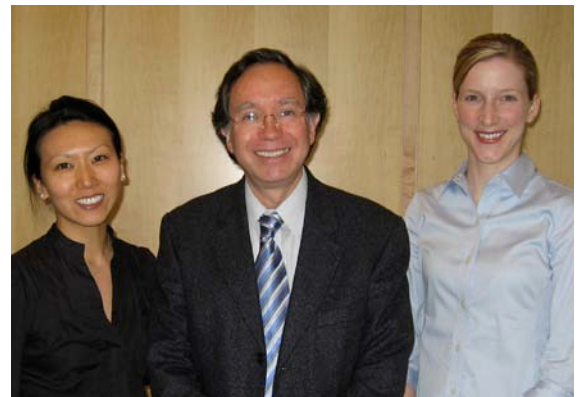


**At left: Presenters discuss their research at the poster competition. Below: David Valenzuela (center) with poster competition winners Caroline Ko (left) and Laurie Eldredge.**

“humanized” monoclonal antibodies that can be directed to recognize and inactivate proteins of therapeutic interest such as cytokines and growth factors. These antibodies can then be used as drug candidates to treat human diseases.

The third biannual event of the Mouse Genetics Group started with a seminar on gait analysis in animal models by Thomas Hampton from Mouse Specifics, Inc.; it was organized by mouse group member Craig Weiss, research associate professor of physiology in Feinberg. Afterwards members attended the group’s first poster competition, featuring more than 20 posters detailing the innovative mouse genetics research taking place at Northwestern and other area universities. Laurie Eldredge, an Integrated Graduate Program in the Life Sciences student, and Caroline Ko, a visiting graduate student, shared first prize.

The Mouse Genetics Group was started in 2007 by CGM’s director of research and educational programs, Michael Kennedy, and CGM faculty members Tom Bozza, assistant professor of neurobiology and physiology at Weinberg



College, and Raj Awatramani, assistant professor of neurology at Feinberg. The group was designed to provide a forum for researchers to share ideas, experimental approaches, and resources, while providing opportunities for labs wishing to develop transgenic mouse models to connect with the broader community. For more information about CGM’s Mouse Genetics Group, please contact Michael Kennedy at 312-503-5604 or [m-kennedy@northwestern.edu](mailto:m-kennedy@northwestern.edu).

## Silverstein Series to Preview Film *In the Family*

CGM will host a Chicago-area preview screening of Joanna Rudnick's film *In the Family* as part of its Silverstein Lecture Series this spring.

Coproduced by Kartemquin Films, the documentary follows Rudnick through her personal experience with a positive BRCA1 genetic test, a result that indicates significantly increased risks of developing breast and ovarian cancers. At age 32, Rudnick must choose between her fertility and preventing cancer through surgery. While charting her own story, Rudnick also explores the lives of other women either facing a BRCA1 genetic test or dealing with cancer that followed a positive result.

Screenings will be held on the Chicago campus on May 19 and the Evanston campus on May 21, with panel discussions following. Joining Rudnick on the Chicago panel will be

Cathy Wicklund, director of CGM's Graduate Program in Genetic Counseling, and Lee Shulman, professor of medicine and head of the section of reproductive genetics in Feinberg's

division of obstetrics and gynecology. The Evanston panel will also include Rudnick and Wicklund, along with Wendy Rubinstein, assistant professor of medicine in Feinberg and medical director of the Evanston Northwestern Healthcare Center for Medical Genetics. Details for both events can be found on the CGM website, [www.cgm.northwestern.edu/silverstein.htm](http://www.cgm.northwestern.edu/silverstein.htm).

Funding for the Silverstein Lecture Series is provided by the Herman M. and Bea L. Silverstein Medical Research Fund for Genetic Medicine. All Silverstein events are free, open to the public, and designed to provide forums where leading experts discuss the importance of developments in genetics research in terms understandable to nonscientists.

**Chicago event**  
**Monday, May 19, 7 p.m.**  
**Thorne Auditorium**  
**375 East Chicago Avenue**

**Evanston event**  
**Wednesday, May 21, 7 p.m.**  
**Ryan Family Auditorium**  
**Technological Institute**  
**2145 Sheridan Road**

## CGM Launches E-Magazine of Science Outreach

*Science in Society (SiS)*, an online magazine that provides a home for all of Northwestern's science outreach efforts, is CGM's latest effort toward public science education.

The magazine (<http://scienceinsociety.northwestern.edu>) launched in February 2008 with its winter issue, featuring the oncofertility research of Teresa Woodruff, Thomas J. Watkins Professor of Obstetrics and Gynecology and chief of Feinberg's Division of Fertility Preservation, and her collaborators, as well as a commentary on the ethics of reproductive technology by Laurie Zoloth, professor of medical humanities and bioethics and religion and director of the Center for Bioethics, Science, and Society. The site has logged well over 3,000 visitors since its launch and now lists more than 15 University departments, centers, and programs as outreach partners, including groups from nanotechnology, physics, and Northwestern Memorial Hospital.

Michael Kennedy, CGM's director of research and educational programs, conceived the University-wide outreach initiative in 2005 when he realized that people had to visit many different sites to gather disconnected information, leaving many of the

University's valuable public education resources underutilized. With the support of the Office for Research and the Northwestern University Advanced Media Production Studio, Kennedy developed a dedicated platform for Northwestern researchers to share their work with the public and comment on developments in the broader science community. The site features original articles by University faculty, fully illustrated with animations, slideshows, and static images; links to relevant science news from across the web; archived multimedia; and a list of outreach partners and their upcoming events.

The spring issue is being published in early May with an article on personalized medicine by Rex Chisholm, dean for research at Feinberg, and another exploring genetic testing by Cathy Wicklund, director of CGM's Graduate Program in Genetic Counseling. This summer *SiS* will begin publishing science writing by Medill graduate students and articles submitted by its outreach partners. Future plans include discussion boards and online chats, as well as a mentorship program with the Boys and Girls Club of Chicago leading to the eventual launch of *Science in Society for Kids*.