

THE SCHEIE LOOKING GLASS

Volume 2, Number 3

Fall, 2001

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BREAKTHROUGH OFFERS HOPE FOR CHILDREN WITH GENETIC BLINDNESS

One of the greatest advances in research on retinal degenerative diseases occurred at the **Scheie Eye Institute, University of Pennsylvania**, when scientists at Scheie's **F.M. Kirby Center for Molecular Ophthalmology**, in collaboration with investigators at **Cornell University** and the **University of Florida College of Medicine at Gainesville**, successfully restored sight to dogs that had been born blind. The blindness was caused by a disease similar to the childhood blindness known as Leber Congenital Amaurosis (LCA). The findings were

that had a form of LCA caused by a mutation of the RPE65 gene. This gene is responsible for regulating the critical biochemical process that allows vision to recycle after light is used by the eye to see. The defective gene interrupts this cycle, leading to blindness. Children with this blinding condition have a defect in the same gene. Dr. Aguirre's laboratory found the gene by the painstaking process of matching up the gene responsible for the disease from hundreds of possible candidate genes.

University of Florida scientist **William Hauswirth, Ph.D.**, developed the vector, whose job is to carry the normal gene into diseased cells. In this case, the vector was a simple, adeno-associated virus that has not been shown to be toxic. Dr. Hauswirth used a normal version of the RPE65 gene, provided by the Cornell investigators, to produce this vector.

At Scheie's **F.M. Kirby Center for Molecular Ophthalmology**, senior scientist **Jean Bennett, M.D., Ph.D.**, developed procedures for delivering the corrective gene in collaboration with **Albert M. Maguire, M.D.**, clinician and scientist, and Dr. Acland. They treated the dogs by injecting the gene-vector combination into one eye in each of the affected dogs. **Samuel G. Jacobson, M.D., Ph.D.**, **Artur V. Cideciyan Ph.D.**, and **Tomas S. Aleman, M.D.**, examined the dogs both before and after the treatment to assess the integrity of the retina and the quality of vision. Dr. Jacobson stated that the success of the experiment "builds on decades of stellar work by talented scientific and clinical people all pulling together to help understand and solve this devastating visual problem." Dr. Aguirre agreed, "the results were wonderful and the teamwork inspiring."

The study was conducted more than a year ago on four dogs that were blind from birth. Several weeks after the treatment, testing showed the amazing result of restored vision. The dogs have continued to do well. One of the dogs, Lancelot,



Jean Bennett, M.D., Ph.D., Albert M. Maguire, M.D., Yong Zeng, M.D., and Vibha Anand, Ph.D., at the F.M. Kirby Center for Molecular Ophthalmology.

published in the May 2001 issue of *Nature Genetics*. The dramatic result could be considered miraculous because the treatment restored vision, rather than merely stopping progressive vision loss, by delivering needed genes to the retina.

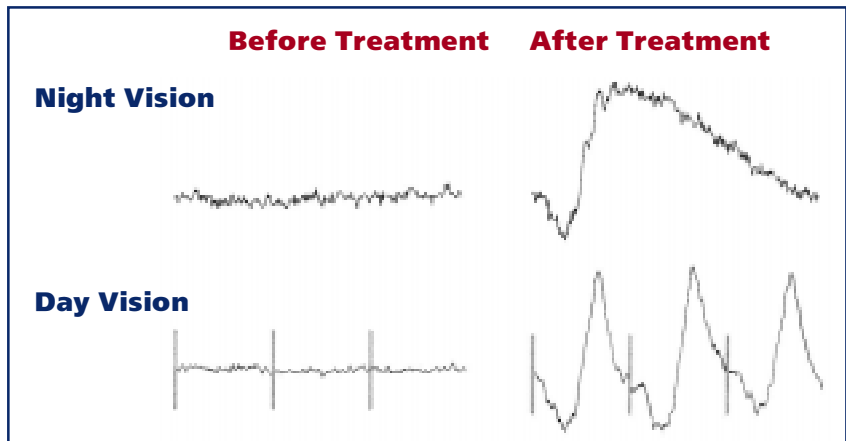
The collaboration of scientific teams from the three universities was a critical ingredient in the successful outcome. Cornell veterinary clinician-scientists **Gustavo D. Aguirre, V.M.D.**, and **Gregory M. Acland, V.M.D.**, are internationally recognized for their expertise in veterinary ophthalmology and canine genetics. Drs. Acland and Aguirre identified dogs of the Briard breed

THE DANIEL MATZKIN RESEARCH FUND

Seventeen years ago, when Jack and Irene Brier learned that their grandson, Daniel Matzkin was severely visually impaired from Leber Congenital Amaurosis (LCA), they were heartbroken. Wanting only the best for Daniel, they brought him to see Dr. Samuel G. Jacobson, who has made it his life's work to find a cure. Mr. Brier, pictured below, established the Daniel Matzkin Research Fund to support Dr. Jacobson's research in hereditary retinal degenerations and specifically LCA. The major progress toward treatment of LCA reported in this month's Scheie Looking Glass was spurred on by the generous research funding of the Brier family.



Tomas S. Aleman, MD., Samuel G. Jacobson, MD., Ph.D., and Artur V. Cideciyan Ph.D., in the Center for Hereditary Retinal Degenerations Laboratory.



Night and day vision responses from the eye (electroretinogram or ERG) before treatment are electrically silent, as shown by flat lines (left). After gene therapy, there are large electrical waveforms indicating restoration of vision under both night and daytime conditions (right).

was present for a recent Congressional hearing where scientists from the three teams presented their findings. Lancelot, obviously behaving as a sighted animal, stole the show. On the impact of the findings to the field of gene therapy, Dr. Bennett feels that "the success in the dog study has a tremendous effect on the field in terms of increasing excitement about the potential for treating human retinal diseases for which there are currently no treatments."

The drama of the outcome for the dogs and the subsequent excitement at the Congressional hearing has not obscured all that needs to be accomplished before human trials can

begin. A pediatric evaluation and treatment facility must be created at Scheie. Dr. Jacobson, the F.M. Kirby Professor of Ophthalmology at Scheie's Center for Hereditary Retinal Degenerations insists that every possible precaution will be taken to insure that the methods are safe. As a result of Dr. Jacobson's careful screening of patients, recruitment of suitable candidates is already underway. The great success with the dogs has only strengthened the resolve of the scientists to push ahead to the ultimate success — a once-blind child seeing the smile on his parents' faces and smiling back.

By Ann Sacks

127TH ANNIVERSARY MEETING



*Mary Esther Dasenbrock, Mel Feldenheimer,
Henry Dasenbrock, Janet Steinberg and
Peggy Feldenheimer*



*Janey Wiggs, Stuart L. Fine and
Travis Meredith*



Oscar and Vivian Lasko



Stuart L. Fine and Melvin L. Rubin

On May 11 and 12, 2001, a scientific meeting was held at the Institute in celebration of the Department's 127th Anniversary. Invited guest speakers were Neil R. Miller, M.D. from Johns Hopkins and Joan O'Brien, M.D. from University of California, San Francisco. Dr. Melvin Rubin, Professor and Chair, Emeritus, University of Florida, delivered the 25th Annual Francis Heed Adler Lecture. On Friday evening, May 11th a dinner dance was held at The Four Seasons Hotel in celebration of the Department's 127th Anniversary and of the 10th Anniversary of Stuart Fine's chairmanship of the Department.



*Sep Rousta, Layla Kamoun,
Rebecca Walker, Scott Goldstein,
Nasreen Syed, Rob Stoltz and Damon Chandler*



*Ellen Peskin, Lea Bramnick,
Jim Tonascia, Maureen Maguire and
Renee Rees*



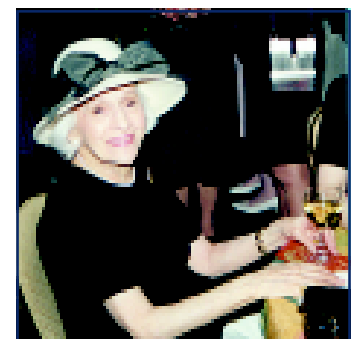
*Sep Rousta, Mina Massaro and
Nasreen Syed*



*Bob and Hannah Spatola, Eric Scheie,
Lois and Barry Hamilton*



*Gerry Klauer, Nancy Nolan and
Ernie Welde*



Miss Evelyn Monte

COMPREHENSIVE OPHTHALMOLOGISTS SEE IT ALL

I have the privilege of working with the Scheie Eye Institute faculty on the development program, an experience that has provided a unique opportunity to learn much about the department's research and education initiatives. The one area that has eluded me, by choice, is Scheie's patient service. To be honest, I don't like going to the ophthalmologist. I have a phobia about having anyone get near my eyes. So, for many years, I have avoided a trip to the ophthalmologist. However, my constant squinting to read signs and persistent headaches told me that it was time I to overcome my fear and I am glad I did.

My patient experience at Scheie could not have been more pleasant. From the moment I checked in at patient services to the conclusion of my exam that culminated in a visit to the Contact Lens Service and Optical Shop, I was treated with courtesy and respect. Before I even saw the ophthalmologist, I was greeted warmly by a registrar who quickly and efficiently whipped through the forms to prepare me for my appointment. No sooner was I seated for what I thought would be a lengthy wait for the doctor, than a kindly technician named Mary Ann whisked me off to an office to start my examination with a series of simple tests to check for vision disorders.

I then had the pleasure of being examined by **Jeffrey Wick, M.D.**, who, after conferring with Mary Ann, began a comprehensive eye exam. Dr. Wick not only took the time to explain the purpose for each step of the examination as it was being done, but also had the patience to answer my many questions. I later found out that Dr. Wick is the newest member of the Comprehensive Ophthalmology team and that he has an expertise in informatics, the science of information management. He is currently working on several projects to improve how SEI patient and research information is stored, handled and retrieved. He already has made improvements to the SEI Intranet to allow sharing of physician schedules, and he is working on implementing an electronic medical record.

Dr. Mina Massaro, whose research focuses on how specific problems in the conjunctiva and cornea affect the tear film, is seen inserting punctal plugs in a patient to treat dry eyes.



Dr. David Kozart is the Director of the Comprehensive Ophthalmology Service and the Department's Vice Chair for Administration. He is seen here performing a slit lamp examination assisted by his ophthalmic technician, Denise McGinn.



Dr. Jane Portnoy's focus is on patient care in the community setting. She provides consultations in comprehensive ophthalmology and neuro-ophthalmology at Scheie's satellite facilities at Penn Medicine at Radnor and Mercy Fitzgerald Hospital.

So what goes into the front lines of Scheie's Comprehensive Ophthalmology Service that makes this Department one of the best in the country? I decided to do a little behind the scenes investigation.

The Comprehensive Ophthalmology team at Scheie is the first line of defense in the early detection and treatment of eye disorders and disease. I was amazed to discover that a routine eye examination may uncover so many systemic diseases such as diabetes, hypertension and a variety of neurological and rheumatological disorders. And there are many dedicated professionals who play a key role in the identification, research and treatment of the most common eye ailments to devastating eye disorders which can lead to severe vision impairment.



Dr. Charles Nichols (right) is the cornerstone of the ophthalmology practice at HUP. A great deal of his clinical time is spent teaching medical students and residents. Above, Dr. Nichols oversees a senior resident performing cataract surgery.



Besides practicing comprehensive ophthalmology, Dr. Jeffrey Wick holds a Master's degree in Medical Informatics. He is seen here designing Scheie's new electronic communication infrastructure, including the website.

The comprehensive ophthalmologists possess a wide variety of skills, training and interests beyond their daily practice that lend to the overall patient experience. The Comprehensive Ophthalmology Unit is built on a strong foundation of knowledge, experience and wisdom. **David Kozart, M.D.**, Vice Chair of the Department and **Charles Nichols, M.D.**, Deputy Chief of the HUP Ophthalmology services, are key building blocks to this foundation, each with over 30 years in practice. Dr. Kozart, who completed his residency at Scheie, specializes in cataract surgery. Dr. Nichols, who studied pharmacology before entering ophthalmology, studies the effects that diseases such as AIDS and sarcoidosis have on the eye. He is a member of the NIH-funded Study of the Optical Complications of AIDS (SOCA) which evaluates new treatments for CMV retinitis in patients with HIV infection.

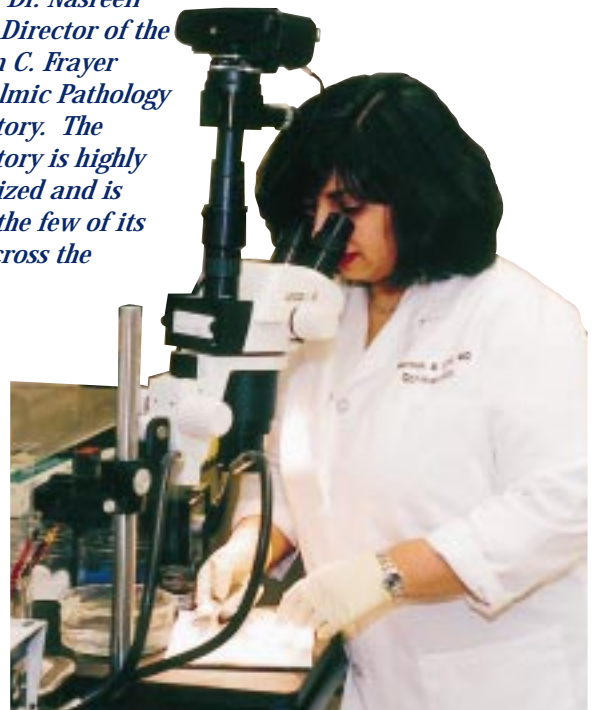
This Unit boasts four additional comprehensive ophthalmologists who provide the mainstay for treatment and care of Scheie patients at its four clinical sites.

Mina Massaro, M.D., a graduate of the Scheie residency program, specializes in dry eyes, blepharitis (inflammation of the eyelid) and ocular surface disease, as well as computer vision syndrome. In addition to performing both refractive and cataract surgery, Dr. Massaro's research supported by the McCabe Fund focuses on testing the conjunctiva (the skin on the surface of the white part of the eye) for the presence of a certain protein associated with eye inflammation. **Nasreen Syed, M.D.**, Director of the Ophthalmic Pathology Laboratory completed her residency at the Medical College of Wisconsin and completed her fellowships in ophthalmic pathology and uveitis at the University of Wisconsin. Rounding out the team are **Jane Portnoy, M.D.**, and **Anna Singh, M.D.** Dr. Portnoy, a specialist in neuro-ophthalmology, divides her practice between Mercy Fitzgerald Hospital and Penn Medicine at Radnor, while Dr. Singh practices at Mercy Fitzgerald and SEI and specializes in glaucoma.

The final phase of my patient experience at Scheie came with a fitting by the Contact Lens Service as well as ordering a much-needed new pair of spectacles at the Scheie Optical Shop. Professionals at both areas could not have been more helpful. Now, I am completely equipped until my next visit to Scheie, which I shall fear no longer.

By Fran Manning Smith

In addition to her clinical practice in comprehensive ophthalmology and her subspecialty practice in uveitis, Dr. Nasreen Syed is Director of the William C. Frayer Ophthalmic Pathology Laboratory. The Laboratory is highly specialized and is one of the few of its kind across the nation.



SEI WELCOMES NEW RESIDENTS

Gabrielle Bonhomme, M.D.

Gabrielle was graduated from Penn Medical School and completed the Transitional Program at Presbyterian Medical Center. She hails from Washington, D.C. and was graduated from George Washington University. She enjoys running and playing the piano.



Damon Chandler, M.D.

Damon is familiar to SEI faculty and staff having spent last year at SEI as a research and clinical fellow. He spent 8 years at Duke as an undergraduate and medical student followed by a General Surgery internship at Massachusetts General Hospital. Art history, classical music, and racketball keep Damon busy when not at Scheie.



Len Feiner, M.D., Ph.D.

A graduate of Princeton University, Len was graduated from Penn Medical School where he also earned a Ph.D. degree in neuroscience, studying axon guidance receptors. Len spent last year as an intern in the Presbyterian Transitional Program and when time permitted, he could also be found in his laboratory at our department's **F. M. Kirby Center for Molecular Ophthalmology**. He is married to Shoshana Feiner, M.D. and enjoys spending time with daughter Ella, age 2.



Leila Mokhartarzadeh, M.D.

Leila was graduated from Princeton University and received her medical degree from University of Michigan where she also completed her internship. She is an avid marathon runner, so look for her along the Kelly and West River Drives. She plans to be married in April, 2002 to Bob Khazaeni, M.D. who will complete his residency in Emergency Medicine at the University of Michigan this year.



Michael Tracy, M.D.

Mike spent last year in La Jolla, CA as an ophthalmic technician and photographer after having completed his internship at St. Luke's-Roosevelt Hospital in NY. He was graduated from UC-San Diego and earned his medical degree at Columbia College of Physicians and Surgeons. Mike performs magic and writes short fiction stories in his spare time.



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Alan M. Laties, M.D.
Richard A. Stone, M.D.

Center for Preventive Ophthalmology and Biostatistics

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Mary Brightwell-Arnold, B.A.
Stuart L. Fine, M.D.
Bojidar Madjarov, M.D.
Maureen G. Maguire, Ph.D.
Ellen Peskin, M.A.
Renee C. Rees, Ph.D.

Center for Hereditary Retinal Degenerations

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Artur V. Cideciyan, Ph.D.
Samuel G. Jacobson, M.D., Ph.D.

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Mina Massaro-Giordano, M.D.
Charles W. Nichols, M.D.
Stephen E. Orlin, M.D.
Jane Z. Portnoy, M.D.
Anna Singh, M.D.
Michael E. Sulewski, M.D.
Nasreen A. Syed, M.D.
Jeffrey P. Wick, M.D.

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Janice Petner
Cynthia Silvestri
Fran Ward

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Stephen E. Orlin, M.D.
Michael E. Sulewski, M.D.

F.M. Kirby Center for Molecular Ophthalmology

Jean Bennett, M.D., Ph.D.
Joshua L. Dunaief, M.D., Ph.D.
Kristen Huang, Ph.D.
Edward N. Pugh, Ph.D.
Eric A. Pierce, M.D., Ph.D.
Dwight E. Stambolian, M.D., Ph.D.
Michael J. Tolentino, M.D.

Genetics

Dwight E. Stambolian, M.D., Ph.D.

Glaucoma

Jody R. Piltz-Seymour, M.D.
Anna Singh, M.D.
Richard A. Stone, M.D.

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Ranjoo Prasad, O.D.

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Joshua L. Dunaief, M.D., Ph.D.
Stuart L. Fine, M.D.
Juan E. Grunwald, M.D.
Albert M. Maguire, M.D.
Michael J. Tolentino, M.D.

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Rong Wen, M.D., Ph.D.

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Steven L. Galetta, M.D.
Grant T. Liu, M.D.
Nicholas J. Volpe, M.D.

Ocular Vascular Research Laboratory

Joan Dupont
Juan E. Grunwald, M.D.
Charles Riva, D.Sc. (Adjunct)

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James A. Katowitz, M.D.

Director Ophthalmic Technicians

Michele Sheehan, COMT

Optical Shop

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Pathology

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Pediatric

Oculoplastic Surgery
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Strabismus-Adult

Nicholas J. Volpe, M.D.

Ultrasound

Kym Gendron

Uveitis

Nasreen A. Syed, M.D.

Vitreoretinal Surgery

Alexander J. Brucker, M.D.
Albert M. Maguire, M.D.
Michael J. Tolentino, M.D.

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Ann Sacks

Education Activities

Sue Hess
Jenny Bartelle

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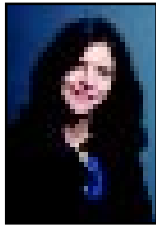
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Stuart L. Fine, M.D.
Chairman and Director



Laura J. Balcer, M.D.
Neuro-Ophthalmology
Epidemiology



Jean Bennett, M.D., Ph.D.
Retinal Degeneration
Genetics Research



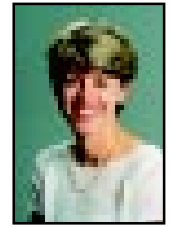
Alexander J. Brucker, M.D.
Retina & Vitreous



Artur V. Cideciyan, Ph.D.
Retina Research



Joshua L. Dunaief, M.D., Ph.D.
Medical Retina
Retina Research



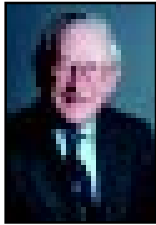
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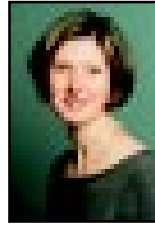
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Retina Research



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Degeneration



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Pediatric Oculoplastic
Surgery



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Comprehensive Ophthalmology



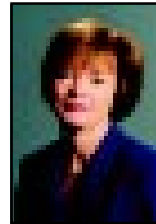
Alan M. Laties, M.D.
Retinal Degeneration
Research



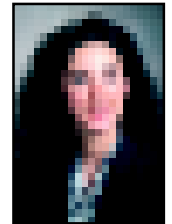
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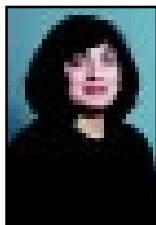
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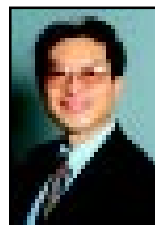
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Retina & Vitreous
Retina Research



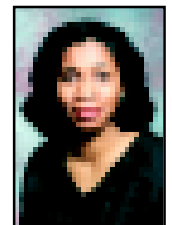
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Terri L. Young, M.D.
Pediatric Ophthalmology

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Bill Nyberg and Jim Berger

LECTURES AND SEMINARS **AUGUST 2001 – JUNE 2001**

Visiting Professor Lectures are scheduled on Thursdays from 7:45-8:30 AM in the Scheie Eye Institute Auditorium followed by a lunch seminar Noon-1:00 PM in the SEI 5th Floor Conference Room.

AUGUST 16, 2001

Neil M. Bressler, M.D.
The Wilmer Institute /
Johns Hopkins University

AUGUST 23, 2001

Martin A. Mainster, M.D., Ph.D.
University of Kansas at Kansas City

SEPTEMBER 20, 2001

Terry Kim, M.D.
Duke University Eye Center

OCTOBER 4, 2001

Maureen G. Maguire, Ph.D.
Scheie Eye Institute /
University of Pennsylvania
Graham Quinn, M.D.
University of Pennsylvania /
Children's Hospital of Philadelphia

OCTOBER 11, 2001

T. Michael Nork, M.D.
University of Wisconsin at Madison

OCTOBER 18, 2001

Arthur Caplan, Ph.D.
Center for Bioethics /
University of Pennsylvania

OCTOBER 25, 2001

Lloyd Paul Aiello, M.D., Ph.D.
Harvard Medical School / Joslin Clinic

NOVEMBER 1, 2001

Joseph Besharse, Ph.D.
Medical College of Wisconsin

NOVEMBER 4, 2001

Neuro-Ophthalmology Update/ CME
Scheie Eye Institute /
University of Pennsylvania

NOVEMBER 29, 2001

George Spaeth, M.D.
Wills Eye Hospital

DECEMBER 6, 2001

Michael E. Sulewski, M.D.
Scheie Eye Institute /
University of Pennsylvania
Eric Pierce, M.D., Ph.D.
Scheie Eye Institute /
University of Pennsylvania

DECEMBER 13, 2001

Carmen S. Puliafito, M.D., M.B.A.
Bascom Palmer Eye Institute /
University of Miami

DECEMBER 20, 2001

Elias Reichel, M.D.
New England Eye Center /
Tufts University

JANUARY 3, 2002

Marco Zarbin, M.D., Ph.D.
UMDNJ Medical School

JANUARY 10, 2002

David H. Abramson, M.D.
Cornell University-NY Hospital

JANUARY 17, 2002

M. Edward Wilson, Jr., M.D.
Storm Eye Institute / Medical
University of South Carolina

JANUARY 24, 2002

David L. Epstein, M.D.
Duke University

JANUARY 31, 2002

Douglas Frederick, M.D.
Dartmouth Medical School

MARCH 7, 2002

M. Bruce Shields, M.D.
Yale University School of Medicine

MARCH 21, 2002

Jason S. Slakter, M.D.
Columbia University College of
Physicians and Surgeons

APRIL 4, 2002

David Robert Guyer, M.D.
New York University School of
Medicine

APRIL 18, 2002

Josh Dunaief, M.D., Ph.D.
Scheie Eye Institute /
University of Pennsylvania
Rong Wen, M.D., Ph.D.
Scheie Eye Institute /
University of Pennsylvania

MAY 16, 2002

Stuart L. Fine, M.D.
Scheie Eye Institute /
University of Pennsylvania

MAY 17-18, 2002

128th Anniversary Meeting / CME
Scheie Eye Institute /
University of Pennsylvania

MAY 23, 2002

Julian J. Nussbaum, M.D.
Medical College of Georgia

MAY 30, 2002

Paul Lee, M.D., J.D.
Duke University Eye Center

JUNE 20, 2002

Terri L. Young, M.D.
University of Pennsylvania /
Children's Hospital of Philadelphia

JUNE 22, 2002

Francis Heed Adler Lecture
Scheie Eye Institute /
University of Pennsylvania

**For more information on lectures and seminars,
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