Since the beginning of scientific exploration people have been trying to figure out what makes us who we are. How much of our physical and personality traits are determined by our genetic makeup versus the environment? The same question can be asked of our propensity towards certain diseases. For example, we have all been warned that a high fat diet (environment) is a major risk factor for stroke. But now, a gene (probably one of many) has been found that strongly influences who is at risk for stroke. Such discoveries alter the thinking about what really causes disease.

Dwight Stambolian, M.D., Ph.D., physician-scientist at our F. M. Kirby Center for Molecular Ophthalmology, believes that gene identification is the key to common diseases such as myopia (nearsightedness) and age-related macular degeneration (AMD). AMD is the major cause of severe and irreversible vision loss in the U.S. and developed world. Myopia is the 6th leading cause of blindness and severe vision loss in the world. Severe myopia can lead to retinal detachment, glaucoma, and a form of macular degeneration. AMD robs the sight of older adults by destroying the macula, the area responsible for straight-ahead vision. Most research to date has been directed towards controlling the manifestations of the disease, either by identifying some of the environmental causes or devising treatments that thwart eyeball growth (in myopia) or blood vessel growth (in AMD). If the genes responsible for these diseases are found, it will open exciting new avenues for treatments and hopefully prevention.

The process of finding genes responsible for any disease is complicated and painstaking work. “It’s like trying to find the street address of a house somewhere in the world with only a photo to go by,” states Stambolian. When searching for a single gene causing a single rare disorder, the clues provided by the patient’s well-documented pedigree (family history) give scientists a head
In 1988, the late David Cogan, M.D., convened several dozen ophthalmologists in Bethesda, MD to discuss our ophthalmic heritage. The group initially was known as the Ophthalmic History Society. It was renamed the Cogan Ophthalmic History Society after Dr. Cogan's passing in 1993.

I attended the 15th meeting of the society which was held on the Averell Harriman estate in Harriman, New York, in April 2002. Approximately 80 people attended. The talks were eclectic, covering a wide range of topics from Rembrandt’s depiction of a blind man in the Bible to snow blindness in Antarctica to the possibility that Theodore Roosevelt had a blind eye from an unoperated retinal detachment.

Is there a place in our current education program for ophthalmic history? I am convinced that the answer is a resounding yes. The fame of our heroes in ophthalmology is short-lived. Recently I confirmed this lack of awareness. Only a few of our residents and faculty could correctly match a list of achievements with the names of those who contributed so much of the knowledge that we employ in our daily practice.

Beginning this year, our department will inaugurate an ophthalmic heritage lecture series. Bill Frayer, an early member of the Cogan Ophthalmic History Society, and Nick Volpe, our residency program director, will organize the curriculum. The goal for the series is to inform as well as to pay respect to those whose discoveries and inventions continue to contribute so richly to our current practice.

By Stuart L. Fine

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**Scheie Welcomes New Fellows**

**MADHURA TAMHANKAR, M.D.**
Fellow in Neuro-Ophthalmology
Madhura was graduated from the University of Bombay, India where she also completed her internship year and a residency in ophthalmology. She did research at Wills Eye Hospital for two years before completing an internship in surgery at Yale University. Madhura enjoys river rafting with her husband and two children and rumor has it that her culinary skills are superb.

**FEMIDA KHERANI, M.D.**
Fellow in Oculoplastics
The University of Alberta in Edmonton, Alberta, Canada was home to Femida from college through her residency in ophthalmology. She plays volleyball and loves to travel. Femida looks forward to skiing in her hometown of Calgary and rejoining her husband, a family practitioner, in Vancouver.

**ANDREW MOHAMMED, M.D.**
Fellow in Glaucoma
A graduate of Virginia Commonwealth University, Andrew attended Medical College of Virginia where he also completed his residency in ophthalmology. In his sparse, spare time, Andrew enjoys S.C.U.B.A. diving, reading, and hiking. We welcome him north of the Mason Dixon line!

**DAVID YOUNG, M.D.**
Fellow in Pediatric Ophthalmology
David was graduated from the University of San Diego and the University of Hawaii Medical School. He headed east to Providence, RI to complete a residency in ophthalmology at Brown University Medical School. When not at CHOP, he spends every available moment with his wife and son.

**ROBERT A. STOLTZ, M.D, PH.D.**
Fellow in Surgical Retina
Rob is no stranger to Scheie Eye Institute having completed his residency here. In July, 2003, he will join the Scheie faculty on the Retina Service. Congratulations to Rob and his wife, Christine, who recently became parents of Gillian.

By Sue Hess
Outreach Program Tops 1,000 Patients

Evelina DiFranco and Sydrick Rabusa with 4Sight patients in West Philadelphia.

It makes no sense. Laser treatment for diabetic retinopathy is one of the best treatments available in all of medicine. Yet, diabetic retinopathy is still one of the leading causes of legal blindness in the United States. Why?

Timing of laser treatment is crucial in achieving optimal results. Ophthalmologists and internists who care for patients with diabetes recommend annual dilated eye examinations to make sure treatment is performed when it is needed. However, some patients have never had their eyes checked for signs of diabetic eye disease and many others do not return for years after having a single examination. Scheie’s 4Sight Program is aimed at getting more of its West Philadelphia neighbors into the habit of annual examinations.

Veteran 4Sight caseworkers Sydrick Rabusa and newly-hired Lori O’Brien call UPHS diabetic patients who live in West Philadelphia and invite them to participate in the program. All who choose to join receive educational information (over the telephone and printed material), assistance in scheduling appointments, and appointment reminders. Those who have no way to travel to Scheie receive taxi vouchers. 4Sight has a fund for uninsured members to cover the costs of their examinations and treatment but most members (more than 90%) have insurance to cover the costs.

Program Director Evelina DiFranco proudly notes that the 1,000th patient will be enrolled by the end of the year. After investigating many ways of reaching patients with diabetes, such as health fairs and mail solicitations, the one-on-one approach of telephone contact emerged as the most efficient and productive way of enrolling patients in the program. Scheie provides the 4Sight office facilities and salaries for the staff. However, with the additional patient examinations and treatments for diabetic eye disease from 1000 participants, as well as some funding from local philanthropic foundations, the program is close to breaking even.

The lessons learned in developing 4Sight have been spread through presentations at meetings of the American Public Health Association (APHA) and the Association for Research in Vision in Ophthalmology (ARVO). Researchers and practitioners from communities across the country have been eager to learn how 4Sight operates. The 4Sight program can serve as a blueprint for other academic medical centers to create a sustainable service for their neighboring communities.

Researchers and practitioners from across the country have been eager to learn how 4Sight operates.

The 4Sight program continues to investigate ways to improve the eyecare delivered to the residents of West Philadelphia. Last year, Evelina DiFranco explored the feasibility of screening patients with diabetes at UPHS-associated kidney dialysis centers. She arranged for a loan of a portable fundus camera from Canon, received training on the camera, and traveled to 3 dialysis centers. However, only a third of the 121 patients approached consented to the fundus photography; many stated that they were already under the care of an ophthalmologist or that they were not interested in the program. Three patients had signs of treatable disease, but after a full examination at Scheie by a retinal specialist, none required treatment. Evelina concluded that the considerable effort to implement screening within the dialysis centers was not as efficient as the standard approach of contacting patients by telephone.

Even though contacting patients one-by-one by telephone is laborious, 4Sight will continue to do so because it works better than any other approach yet examined. Many times patients have told Sydrick Rabusa that they just have not gotten around to making an appointment; Sydrick then suggests that they turn their call into a 3-way call with Scheie appointment schedulers and the task is quickly accomplished!

By Maureen Maguire, Ph.D.

Macula with diabetic retinopathy (left) and normal macula (right).
When you think of taking a vacation, what comes to mind? Perhaps you think of exploring another country, learning about new people, places and things. Well, that’s exactly what five Scheie physicians have done in the past year, but with a twist: they used their vacation time to travel to another country in order to teach.

**Drs. James Katowitz, Scott Goldstein and Bill Katowitz** traveled to Nairobi, Kenya to consult with local physicians on oculoplastics issues and to teach local ophthalmologists how to care for these patients. Their trip was coordinated by **Mr. Kuku Sharma**, an optician in Kenya who is the father of **Dr. Atul Sharma**, Scheie’s 2001-2002 Co-Chief Resident.

Mr. Sharma has ties to many missionary eye hospitals in Africa, Asia and Central America, and arranged for the three Scheie physicians to spend a week at the Eye Unit at Kikuyu Hospital, 30 Km outside of Nairobi. Kikuyu is a Protestant-run missionary hospital consisting of eye, medical, surgical and pediatric units. Each unit is a self-contained, one-story building, and the buildings are scattered across a few acres of the Kenyan countryside.

On their first day at Kikuyu, over 60 patients were examined and over the next four days, 45 operations were performed. During the first two days, surgeries were performed by the Scheie physicians, with the Kikuyu physicians assisting. On the last two days, the roles were reversed.

According to Dr. Goldstein, two of the most gratifying cases involved siblings with neurofibromatosis. In the case of the siblings, the disease had caused significant eyelid disfigurement to the point where the teens were ostracized by their family and village, and sent to an orphanage. After extensive reconstructive surgery and recovery, the teens were allowed to return to their village, where a celebration took place in honor of their suitability to now marry and have families!

As a third-generation Indian born in Kenya, choosing to teach in Africa was a natural choice for Dr. Atul Sharma. He spent six...
weeks in a missionary hospital in Dar-es-Salaam, Tanzania. The facility, considered the most medically-advanced in the country, is one of many eye hospitals run by Christoffel-Blindenmission, the largest non-governmental organization in the world dedicated to providing ophthalmic care to medically-underserved populations worldwide.

While Sharma was able to teach local ophthalmologists some of the newer techniques in cataract extraction, he also learned a tremendous amount about efficiency in providing patient care. He and the local ophthalmologists performed 15-25 cataract surgeries per day. Most of the patients traveled long distances from remote villages in hopes of finding a cure for their blindness, and most were amazed to discover that a simple operation to remove cataracts could restore their eyesight.

Most recently, Dr. Ranjoo Prasad also chose to spend her vacation in another country. She was invited as a volunteer consultant to visit the Low Vision Project Jordan, which is based in the capital city of Amman. The project, which was initiated by Christoffel-Blindenmission in 1999, trains local optometrists and optometry students and is responsible for the evaluation of visually-impaired children at schools for the blind. Along with giving patient care, Prasad, an optometrist herself, visited the program at the Jordan University of Science & Technology in Irbid, Jordan. She also visited schools for the blind, low vision programs in university ophthalmology departments, and met with government officials interested in supporting further expansion of the project.

All of these doctors devoted their time, energy and talent to broadening the scope of eye care on a global level and, in the words of Atul Sharma, they “learned the value of my profession and the effect it can have on people in a way I never imagined.”
American Academy of Ophthalmology
Scheie Eye Institute Alumni Reception at the Peabody Hotel, Orlando, Florida, October 21, 2002

Sandy Brucker, Stuart Fine and Judy Alexander

Lisa Schocket, Mike Ibarra, Lisa Pritchard, Christina and Ken Shindler

Harvey Lincoff from Cornell, Ellie Fine and Ingrid Kreissig from Germany

Nick Volpe and Stuart Fine

Jen Thorne from Johns Hopkins and Terry Brucker

Ursula Schmidt-Erfurth from Germany, Ingrid Adamsons and Sandy Brucker

Stuart Fine and Jackie Kaiser

Larry Rand from Boston and Edward McManus, Director, National Alliance for Eye and Vision Research
Scheie Physician-scientist Searches for His Field of Dreams . . . Continued from page 1

Start. This is not true for common diseases caused by more than one gene, where a heterogeneous gene pool and diverse environmental factors present daunting obstacles. In addition, the location of these genes and the proximity of other specific genes also may play a role in disease expression. “Multiple genes interact with each other to cause disease. Where they are and how they interact is the key.”

So where does a scientist whose first love was baseball begin the search for genes? Stambolian believes that the best way to do this is to select populations that are not only homogeneous in their heritage, but also in their environment. He has identified several such populations descended from a small number of ancestors or “founder” families who have married and stayed within the group.

The Amish in Lancaster, PA are descended from several founder families who live in the same environment and have many common habits such as working outdoors and not smoking. Within this group, the incidence of myopia is low, but the frequency of AMD is significant. The Orthodox Ashkenazi Jews in Monsey, NY and Lakewood, NJ are another relatively homogeneous population whose immediate ancestors come from Brooklyn, NY. They too tend to marry within the ethnic group and have similar habits. They have a high incidence of severe myopia, which also starts around age five.

Homogeneous populations by their nature are closed. Once identified, gaining access to the families for eye exams, blood samples and personal details can be challenging. Persistence and patience by Stambolian finally convinced a rabbi in the Jewish community and a trusted physician in the Amish community to promote the study to the people. After access, the single most important factor contributing to the success of the study was Stambolian’s insistence that his team go out to the participants as opposed to asking them to come to Scheie.

His team of four research coordinators does just that — taking lensometers, blood sampling equipment and eye charts to homes or clinics in the communities. Since the Amish do not use phones, a nurse in the area facilitates the appointments. For the Jewish population whose lifestyles are regimented by religious and community activities, visits were scheduled to accommodate time constraints. Stambolian praises his research coordinators: “They are absolutely essential for keeping the enrollment high and the study on track.”

The myopia study looks at families in which one parent and at least two children have myopia. Then information and blood samples are collected on as many family members as possible. The genetic “information” is processed to try to find where the susceptibility to myopia lies. At this point, Stambolian’s team has sent 500 specimens from the Jewish community and 300 specimens from the Amish community to the Center for Inherited Disease Research (CIDR), an exclusive NIH resource laboratory that only accepts specimens from promising studies. CIDR will perform a genome-wide screen with 400 markers for myopia. This screen should find the location (like determining what state that house is in) where various myopia genes reside. The process will then continue with more markers to hone in on the locations of these genes.

As the mapping nears completion in the Jewish and Amish populations, the study will continue with additional populations including African Americans, Asian Americans and Caucasians in Philadelphia. Other Scheie physicians, including Drs. Lin Chen and Eydie Miller-Ellis, are helping in the acquisition of Asian and African American subjects, respectively.

Not content with keeping several balls in the air, Stambolian saw the opportunity to tackle another common yet devastating eye disease, age-related macular degeneration, and already has started collecting data on the Amish. There is potential for studying hundreds of common diseases in these populations using Stambolian’s ideas — a grand slam for this physician-scientist who once dreamed of becoming a big league ball player. It’s not baseball, but it’s certainly worthy of the Hall of Fame.

Stambolian’s laboratory team from left to right: Scarlett Geunes-Boyer, Chris Moy, Melissa Schlifka, Ken Wang, Lauren Zahn, Tim Baradet and Kristen Huang (missing is David Brooks) assists with the painstaking work of gene identification.
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Richard A. Stone, M.D.

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Mary Brightwell-Arnold, B.A.
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Lectures and Seminars
January – June 2003

Visiting Professor Lectures are on Thursdays:
7:45-8:30 AM (Scheie Eye Institute Auditorium - Lower Level)
Noon-1:00 PM (Thayer Conference Room, SEI 5th Floor)

JANUARY 9, 2003
7:45 AM
David G. Brooks, M.D., Ph.D.
Katherine L. Nathanson, M.D.
Reed E. Pyeritz, M.D., Ph.D.
Department of Genetics/Univ. of PA
Genetic Eye Diseases
Noon
Tonia Rex, Ph.D.
Scheie Eye Institute / F.M. Kirby Center for Molecular Ophthalmology /Univ. of PA
The Responses of Photoreceptor Cells to Experimental Retinal Detachment
JANUARY 16, 2003
Richard P. Millis, M.D., MPH
Glaucoma Consultants Northwest, Seattle, Washington
7:45 AM: Perspectives on Physician Competence
Noon: Retinal Nerve Fiber Layer 2003
JANUARY 23, 2003
7:45 AM
Ophthalmic Heritage Lecture
Moderators:
William C. Frayer, M.D. and Nicholas J. Volpe, M.D.
Noon
Peter Sterling, Ph.D., Univ. of PA
Efficient Circuits from Stochastic Synapses
JANUARY 25, 2003 (Saturday)
CME: Glaucoma Update, 2003
8:00 AM – Noon
Guest Speaker:
Wallace L. M. Alward, M.D.
University of Iowa
JANUARY 30, 2003
7:45 AM
Janet D. Steinberg, OD and Dawn Ciccarone, MS, OTR/L
Scheie Eye Institute / University of Pennsylvania
The Low Down on Low Vision
FEBRUARY 6, 2003
Noon
Alexander Dizhoor, Ph.D.
Pennsylvania College of Optometry
Study of Regulation of cGMP Synthesis in Photoreceptors
FEBRUARY 13, 2003
Susan R. Carter, M.D.
University of California / San Francisco
7:45 AM: The Ophthalmologist’s Role in the Management and Treatment of Graves' Disease
Noon: Use of the CO2 Laser in Aesthetic Eyelid Surgery
FEBRUARY 20, 2003
7:45 AM
Prithvi S. Sankar, M.D.
Scheie Eye Institute / University of PA
Counseling: A Travel Through History
Noon
Kathleen Battaglia, Ph.D.
Department of Biochemistry / University of Pennsylvania
Photoreceptor Cytoskeleton Proteins
MARCH 6, 2003
Jonathan M. Holmes, M.D.
Mayo Clinic School of Medicine
7:45 AM: Clinical Trials in Strabismus and Amblyopia
Noon
Kathleen Battaglia, Ph.D.
University of Pennsylvania
Retinopathy of Prematurity; What Can We Learn from the Lab?
MARCH 8, 2003 (Saturday)
Francis Heed Adler Lectureship
8:00 AM - Noon
Richard L. Abbott, M.D.
University of California / San Francisco
MARCH 27, 2003
Rudolf Guthoff, M.D.
University of Rostock, Germany
7:45 AM: Micromorphology of the Anterior Segment of the Eye: New Developments in Confocal Microscopy
APRIL 3, 2003
Louis A. Karp Lectureship
8:00 AM:
Robert D’Amato, M.D., Ph.D.
Harvard Medical School
Advances in Angiogenesis & Cancer Biology
Janey Lee Wiggs, M.D., Ph.D.
Harvard Medical School
Massachusetts Eye and Ear Infirmary
Genetics of Glaucoma
APRIL 24, 2003
Lawrence F. Jindra, M.D.
SUNY at Stony Brook
7:45 AM: Pharmacologic Management of Glaucoma
Noon: Advances in Glaucoma Surgery
MAY 22, 2003
Ophthalmic Heritage Lecture
7:45 AM
Moderators:
William C. Frayer, M.D. and Nicholas J. Volpe, M.D.
MAY 30-31, 2003 (Friday-Saturday)
129th Anniversary Meeting
129th Anniversary Meeting
Guest Speakers:
Michael X. Repka, M.D.
The Wilmer Institute / Johns Hopkins University
Michael Ip, M.D.
University of Wisconsin Medical School
JUNE 5, 2003
Ophthalmic Heritage Lecture
7:45 AM
Moderators:
William C. Frayer, M.D. and Nicholas J. Volpe, M.D.

For more information on lectures and seminars, call Sue Hess at 215-662-8020 or e-mail to sue.hess@uphs.upenn.edu