Genomics pioneer says ophthalmology leads the way in genetic research

New techniques, long-term follow-up and research highlighted the featured lectures at this year's AAO.

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CHICAGO – The study of genomics has reached a level in ophthalmology that is still only a distant hope for other medical specialties, said an expert in genomics speaking here

"Your field of ophthalmology is on the cutting edge of my topic, which is to emphasize how our understanding of the genome is beginning to impact the practice of medicine in dramatic ways," said Francis S. Collins, MD. "Ophthalmology is leading that charge."

Dr. Collins, the director of the National Institute of Health's National Human Genome Research Institute, delivered the keynote speech at the opening session of the American Academy of Ophthalmology meeting here.

He described an effort, known as the International HapMap Project, which aims to identify and catalog genetic similarities and differences and place all the data in a public database. The six-country project, which has been under way for 3 years, is to complete its work within weeks, he said.

This project's first success story involved age-related macular degeneration, according to Dr. Collins. Using phase 1 of the HapMap data, investigators at Yale University were able to identify that a variant in a particular gene, complement factor H, is responsible for roughly half the genetic susceptibility to AMD.

"No one expected that. It set off an earthquake in the field of genetics that a single gene that nobody predicted would be involved in a single disease, and that a single variant of the gene, carried that much ... risk," Dr. Collins said.

"In the space of just a few months, utilizing the HapMap approach, a disease that many weren't sure was particularly genetic has been broken wide open by this approach," he said.

Dr. Collins predicted that this same process will occur for many diseases in the future, in ophthalmology as well as other specialties.

"This will enable you to move into the ability to make diagnostic predictions about who is at risk [for a disease]. This is most interesting if you have a prevention to offer," he noted.

Dr. Collins said that a new project of the National Eye Institute is creating a genotyping network to enable ophthalmologists to have access to certain research laboratories to obtain precise molecular diagnoses of particular eye diseases. The aim is to have this project operational by February of 2006, he said.

Other named and featured lectures at this year's AAO meeting are recapped below. These items originally appeared in daily coverage of the AAO meeting on OSNSuperSite.com.

Kelman Lecturer lauds bimanual MICS

The impact of bimanual microincision cataract surgery can be equated with that of phacoemulsification, according to the inaugural presenter of a new named lecture.

Bimanual microincision cataract surgery (MICS) is a development that "alters cataract surgery as physicians currently know it," said I. Howard Fine, MD. Dr. Fine delivered the first Charles D. Kelman Lecture at the AAO meeting.

Introducing Dr. Fine, David F. Chang, MD, called him "perhaps the best-known cataract surgeon in the world." Dr. Fine said he was "humbled" to be named the first Kelman Lecturer. He said Dr. Kelman's invention of phacoemulsification was "ahead of its time."

While 2-mm coaxial phaco "is an improvement worthy of note," Dr. Fine said, bimanual MICS is better. He said the separation of the infusion and aspiration functions in bimanual MICS is "important in challenging cataract cases."

This picture of I. Howard Fine, MD, was shown during the introduction to his inaugural Charles D. Kelman Memorial Lecture at the AAO meeting. This "king of phaco" picture was taken on Dr. Fine's birthday in April, according to sources close to him who provided the picture to the AAO.

Bimanual MICS lets the surgeon have complete control, he said.

"The enormous advantage of bimanual MICS is evident," he said.

Using bimanual MICS "takes your wrist out of the equation," Dr. Fine said. "You're limited to just finger use. It's like holding a pen, where you set your wrist and let your fingers do all the work."

The Charles D. Kelman Lecture, which has been endowed with $1 million by the AAO, will honor outstanding innovators in the cataract field. The lecture will be presented annually at the AAO meeting.

Barraquer Lecture: H-LASIK

With follow-up of 5 to 8 years, LASIK correction of hyperopia and hyperopic astigmatism was accurate and stable, according to this year's Barraquer Lecturer.

Delivering the lecture named for her father, José Barraquer, Carmen Barraquer, MD, focused on the challenges refractive surgeons face in correcting hyperopia.

In introducing Dr. Carmen Barraquer, James J. Salz, MD, said she has "made a significant contribution in refractive surgery and exemplifies the character of Dr. José Barraquer."

Dr. Barraquer reminded attendees how much her father contributed to the field of ophthalmology, and she said she was deeply honored to deliver the lecture named for him.

“My father developed more than 20 original techniques for refractive surgery. He designed more than 30 instruments and wrote three books on keratophakia and keratomileusis,” she said.

The advent of LASIK “has allowed us to make a keratome evolution,” she said.

Dr. Barraquer discussed the results of a retrospective study, with follow-up of 5 to 8 years, of hyperopia and hyperopia with astigmatism in young patients. The study included 74 eyes of 41 patients who ranged in age from 8 to 26 years, she said.

The Schwind excimer laser was used in the study, with an optical zone size of 6.5 mm to 7 mm for all eyes.

The average preoperative refractive error was +5.46 D, and average postoperative refractive error was +0.82 D, Dr. Barraquer said. She noted that these results were stable over the course of follow-up.

To further analyze how hyperopes responded to the refractive laser treatment, Dr. Barraquer divided the patients into three groups: low, moderate and high hyperopia. The low hyperopia group included 19 eyes with a preoperative error of 1.25 D to 3 D. The moderate group included 38 eyes with errors of 3 D to 5.99 D. The high hyperopia group included 17 eyes with errors of 6 D to 8.25 D.

In the low hyperopia group, 90% were corrected to within 0.5 D of emmetropia after surgery. In the moderate group, 82% were within 0.5 D of emmetropia, and in the high hyperopia group, 30% were within 0.5 D of emmetropia. All patients were within 1 D of emmetropia, Dr. Barraquer said.

Almost all eyes in the moderate hyperopia group (97%) had unchanged uncorrected visual acuity postoperatively, while 95% of the low hyperopia group and 88% of the high hyperopia group had unchanged UCVA. Best corrected visual acuity remained unchanged in 95% of the low group, 94% of the moderate group and 65% of the high hyperopia group, Dr. Barraquer said.

She recommended the use of a 6.5- mm optical zone for young patients and 7 mm for adults.

She cautioned surgeons “not to mistake hyperopia for presbyopia” in adults.

Options for nerve sheath meningioma

While there is still no ideal treatment for optic nerve sheath meningioma, recent advances in radiation therapy have provided promising outcomes that bode well for management of these tumors in the future, according to Neil R. Miller, MD.

“There have been major shifts in the paradigms for diagnosis and management,” said Dr. Miller, delivering the William F. Hoyt Lecture.

Observation is still appropriate for certain patients, Dr. Miller said, provided there is no significant visual dysfunction, progression of visual loss or intracranial extension of the tumor.

While in most cases some action is called for, surgery is rarely a feasible option, Dr. Miller said.

“[The disease] is usually not surgically curable unless it is accompanied by the removal of the optic nerve,” he said.

Surgery is indicated only in very specific circumstances, such as when excision of primarily exophytic tumors can be performed without visual loss.

Radiation therapy has become the preferred intervention for many specialists, but radiation is not without side effects, Dr. Miller said. For conventional fractionated radiation therapy, side effects include radiation retinopathy and radiation optic neuropathy.

“Are we giving too much radiation?” Dr. Miller asked. “If we could decrease the dose, we could reduce the risks and still eliminate the progression of the tumor.”

A relatively new option is stereotactic or three-dimensional conformal fractionated radiation therapy, which is more focused on the tumors with the use of intensity-modulated beams, Dr. Miller said.

He noted that a number of reports have touted at least the short-term results of 3-D conformal fractionated radiation therapy.

It is hoped that medical therapies will become available in the future that will avoid the side effects of radiation therapy, but these have not appeared yet, Dr. Miller said.

“Right now there is no medical therapy as an alternative, but hopefully this will change,” he said. “There have been a tremendous number of advances in both the diagnosis and the treatment of optic nerve sheath meningiomas within the past several years, and hopefully we will see more in the future.”

Vision screening efforts

The first lady of Illinois is leading an effort in her state to improve vision screening, and she called on insurance companies to pay pediatricians for vision screening as part of well-child visits.

Patti Blagojevich, the wife of Illinois Governor Rod R. Blagojevich, said that better pediatric vision screening programs are needed to avoid preventable vision loss in children.

The Illinois Pediatric Vision and Amblyopia Campaign was launched in 2004, Mrs. Blagojevich said, after she became personally aware of the need for better screening efforts through the unfortunate experience of a staff member.

Speaking at the Pediatric Ophthalmology Subspecialty Day, Mrs. Blagojevich said the idea for the campaign was germinated 4 years ago when she learned that an associate's 7-year-old son had become blind in his left eye due to untreated amblyopia.

"How could this happen in this state and country to a child who kept up with his well visits?" Mrs. Blagojevich asked.

She said pediatricians "are overwhelmed" and that they do not know enough about amblyopia.

"It ends up in a heap of other problems," she said. "Vision screening should be a part of all well visits. And insurance must pay for these screenings if they're going to get done."

In an interview with *Ocular Surgery News*, Mrs. Blagojevich said that vision screening "needs to be put on the list of things to talk about with pediatricians" during well-child visits.

"In addition to talking about vaccines and weight gain, parents need to know to bring vision into the dialogue. Doctors need to screen these patients earlier to prevent vision problems," she said.

To date, through the Illinois Pediatric Vision and Amblyopia Campaign, nearly 1.5 million brochures on amblyopia and vision screening have been distributed to parents, teachers and caregivers, she said.

Mrs. Blagojevich also touted the Illinois governor's All Kids health insurance proposal, which will provide affordable health insurance to children in the state.

As part of her presentation at the meeting, Mrs. Blagojevich presented the Gov. Rod R. Blagojevich PATH (People are Today's Heroes) Award to Marilyn T. Miller, MD. Dr. Miller received the award in recognition of the care she has provided to children for the past 30 years, Mrs. Blagojevich said.