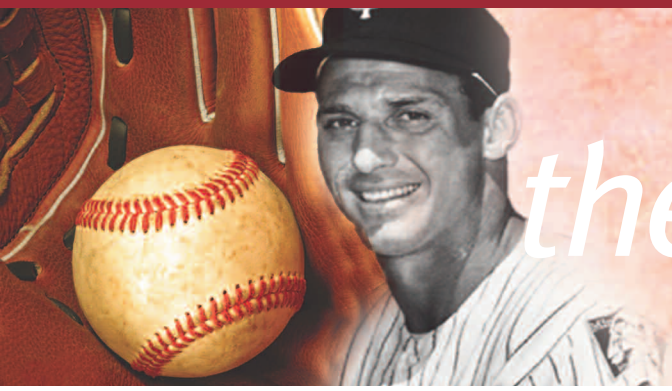
Spring 2009



the LINE UP

The **Bob Allison Ataxia Research Center (BAARC)** at the University of Minnesota provides vital support for research on diseases of the nervous system, such as ataxia and related disorders. BAARC is an affiliate of the Minnesota Medical Foundation.

BOB ALLISON ATAXIA RESEARCH CENTER

Researchers hone in on markers for AOA2

Since it was identified through genetic testing in the early 2000s, ataxia with oculomotor apraxia type 2 (AOA2) has become the second most commonly diagnosed form of recessive ataxia. But while more individuals are being diagnosed with AOA2, research on the disease remains scant. That paucity in data shouldn't last long, however, thanks to a team of researchers at the University of Minnesota's world-renowned Center for Magnetic Resonance Research (CMRR).

Under the direction of ataxia researcher Gülin Öz, Ph.D., radiology research associate Isabelle Iltis, Ph.D., is collecting information from people with AOA2 as well as healthy volunteers about the neurochemicals and brain regions involved in AOA2.

Using magnetic resonance spectroscopy, a scan that can measure neurochemical defects within the brain, Iltis hopes to identify which neurochemicals are playing a role in the disease and which brain regions are most affected by the defects. Study participants also receive a neurological exam and undergo a lumbar puncture for spinal fluid analysis.

"Every type of ataxia has its own neurochemical 'fingerprint,' so to speak," explains Iltis. "With the MR spectroscopy and other data, we can associate specific neurochemical defects with this specific form of ataxia."

Since May 2006, Iltis—together with CMRR research coordinator Diane Hutter, R.N., and collaborators Khalaf Bushara, M.D., and Christopher Gomez, M.D., Ph.D.—has recruited eight patients with AOA2 and about 23 healthy volunteers to participate in the study. In less than three years, she has learned that the two regions of the brain most impaired by AOA2 are the vermis and the cerebellar hemisphere, both located in the cerebellum.

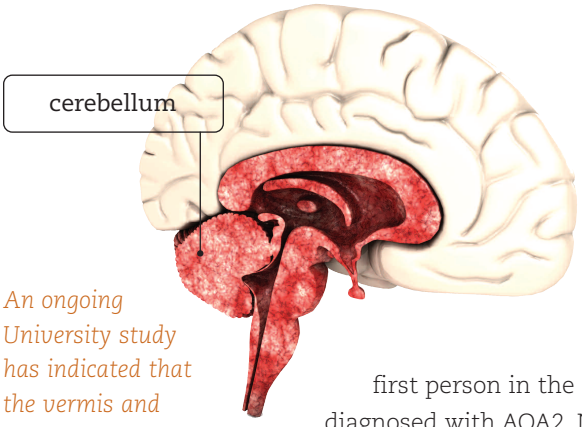
"We now have a neurochemical profile of AOA2 within these two regions of the brain, and it's distinctly different from what we see with other types of ataxia," she explains.

continued on page 2

Photo by Tim Rummelhoff

Gülin Öz, Ph.D., Isabelle Iltis, Ph.D., and Diane Hutter, R.N., are using magnetic resonance spectroscopy to study how ataxia with oculomotor apraxia type 2 affects the brain.

AOA2 continued from cover



An ongoing University study has indicated that the vermis and cerebellar hemisphere, both located in the cerebellum, are the brain regions most impaired by AOA2.

People with AOA2 have come from all over North America to participate in this study. Kory Tabor, a resident of Madison, Wisconsin, is one of them. Tabor was the first person in the United States to be diagnosed with AOA2. Now 31 years old, she stays positive by participating in research.

"The way I deal with it is to realize that I can do something about this disease, that I can be involved in finding new things that will help people who may not yet know they have AOA2," she says.

In developing a neurochemical profile of AOA2, Iltis hopes that one day degeneration in the cerebellum can be detected earlier—even before symptoms appear. With earlier detection, there's a better chance of intervening with a treatment before symptoms become debilitating, she says.

And for now, Iltis is pleased to have the opportunity to spend time with a dedicated group of research volunteers.

"I am really impressed with the people who are participating in this study," she says. "They are really amazing, motivated individuals. Whenever a patient is coming in for a scan, I know I am going to meet a really incredible, enthusiastic person that day. It's a nice way to work."

A home run for research

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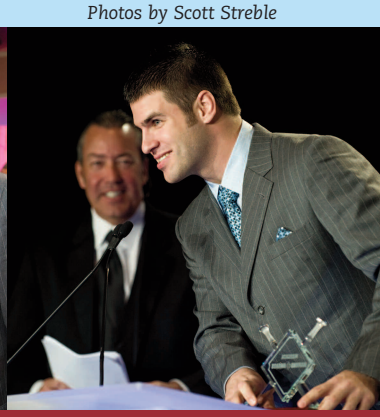
Photos by Scott Streble

Diamond Awards guests bid on dozens of exciting silent auction items.



Twins catcher Joe Mauer posed for a photo with fan Joe Peller at the VIP reception. Mauer later accepted two awards at the event from emcee Dick Bremer.

The fourth annual Diamond Awards, held January 22 at the Minneapolis Marriott City Center, celebrated the Minnesota Twins' 2008 baseball season and raised more than $263,000 to advance leading-edge research and patient care for neurologic illnesses at the University of Minnesota.

Because of continued support from loyal event sponsors and attendees, the University can continue to build an all-star team of world-renowned doctors and researchers working to strike out devastating neurodegenerative and neuromuscular diseases such as ataxia, muscular dystrophy, multiple sclerosis, Parkinson's disease, and ALS (Lou Gehrig's disease).

A partnership of the Bob Allison Ataxia Research Center, Minnesota Medical Foundation, Baseball Writers Association of America's Twin Cities chapter, and Minnesota Twins, this event has raised about $2 million in its history.

Karen's Hope Ataxia Benefit

Monday, June 15, 2009
Oak Marsh Golf Course, Oakdale

This golf event benefits the Bob Allison Ataxia Research Center at the University of Minnesota. For more information, contact Valerie Petermann at 612-624-4444 or v.petermann@mmf.umn.edu, or visit www.karens-hope.org.

'A sense of hope'

Couple's commitments to Diamond Awards show confidence in the University's neurodegenerative disease research

Photo by Scott Streble

Patrick Bradley and Patty Carney-Bradley

Five years ago, ataxia was an unfamiliar disease to Patrick Bradley and Patty Carney-Bradley. They knew little about it, but they had heard that Minnesota Twins legend Bob Allison had it and eventually died from its complications.

"If you grew up in Minnesota, you knew Bob Allison because he was one of the great baseball heroes," says Bradley, who grew up in Austin, Minnesota, as did Carney-Bradley. "I used to get my first sunburn of the year at a Twins game," he says.

Today Bradley and Carney-Bradley not only know what ataxia is, but they've become advocates for research into this condition and other neurodegenerative diseases through their support of Diamond Awards.

They have made gifts to the annual event, which benefits the Bob Allison Ataxia Research Center (BAARC), since 2004, increasing their gift size each year. They became Home Run–level sponsors of the 2009 event with a $7,500 gift.

"It's an event around a great American pastime for a great cause," Bradley says. "How could you *not* want to go?"

The couple were introduced to BAARC by John Marcotte, a colleague of Bradley's and a member of BAARC's board of directors. They were impressed as they talked to the scientists about their work, but they became even more engaged when they learned that University ataxia researchers were collaborating with investigators studying other brain disorders to help find solutions for all of those diseases faster.

"With my mother having had Alzheimer's, my interest was piqued," says Bradley, who is now a BAARC board member himself. "It's communication breakthroughs that allow scientific breakthroughs."

Today he and Carney-Bradley say they're happy to spread the good word about the neurodegenerative and neuromuscular disease research happening at the University of Minnesota, their alma mater. And a fun, upbeat event like Diamond Awards makes that easy, Carney-Bradley says.

"There's such a sense of hope, which I think is so important when you're talking about such serious illnesses," she says.

To learn more about how you can sponsor next year's Diamond Awards, contact Valerie Petermann at 612-624-4444 or v.petermann@mmf.umn.edu.

University of Minnesota

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Advancing a cure, one gene at a time

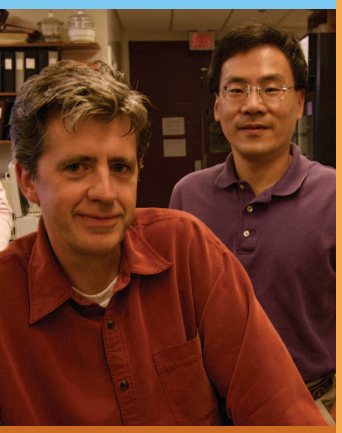

Photo by Richard Anderson

Michael Koob, Ph.D., and Young Yoon, Ph.D.

Although Friedreich's ataxia is the most common type of ataxia, an effective treatment remains elusive. But thanks to the efforts of Michael Koob, Ph.D., and his laboratory team, the path to a cure could be getting shorter.

Aided by a one-year, $75,000 seed grant from the Bob Allison Ataxia Research Center (BAARC) in 2008, Koob and his colleague Young Yoon, Ph.D., are working to develop a novel gene therapy for Friedreich's ataxia.

This form of ataxia is caused by a lack of a protein called frataxin, which works in the cell's "power house," the mitochondrion. In devising an effective gene therapy to treat the disease, the Koob lab has engineered new ways to get this missing protein into ataxia-stricken cells.

The group began this diligent work with yeast cells. First, they created a yeast gene to compensate for the lack of the frataxin in a diseased cell. Then they inserted that gene into the mitochondria of yeast cells in the lab.

It's a process that had never been successfully achieved anywhere in the world.

By injecting the yeast's frataxin gene into the cell's mitochondrion instead of its nucleus, Koob and his team found that the gene could be more easily replicated. From there, they found that they could transfer those mitochondria from one cell to other cells that didn't have the frataxin protein. As a result, those cells were rescued.

"Basically, we've shown that this type of engineering can cure Friedreich's ataxia in yeast," says Koob, an associate professor in the University of Minnesota's Institute of Human Genetics. "Now we need to take the next step toward making this work beyond the lab."

Long term, Koob's work could help pave the way for a successful gene therapy for people with ataxia and other neurodegenerative diseases.

The promise of this research has been recognized by several organizations: This year, Koob's lab has received more than $1 million collectively from the Friedreich Ataxia Research Association, the Muscular Dystrophy Association, the State of Minnesota, and the Minnesota Partnership for Biotechnology and Medical Genomics to design and test the gene therapy in mouse models.

"The BAARC grant allowed us to get the data we needed to get additional funding," Koob says. "The payback has been tenfold."

the **LINE UP** **Spring 2009**

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To make a clinic appointment at the Ataxia Center, call 612-626-6688 or visit www.ataxiacenter.umn.edu.

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