

Link found between gene, macular degeneration

Scientists at the University of Michigan Kellogg Eye Center have come a step closer to understanding the genetics of macular degeneration, a disease of the retina that affects 13 million Americans, and causes the loss of central vision.

Research scientist Radha Ayayngari has found that a gene associated with one retinal disease, retinitis pigmentosa, also causes a form of macular degeneration.

In the August issue of *Genomics*, Ayayngari reports that the gene RPE65 has a direct link to a form of early-onset macular degeneration that primarily affects males.

The finding is important because it will ultimately help scientists understand how macular degeneration progresses, and it raises an intriguing question: How can a single gene cause two very different eye diseases, each affecting a different

segment of vision?

Ayayngari's study marks the first time any scientist has mapped a macular degeneration gene to the X chromosome.

Macular degeneration causes the loss of central vision that is needed for such activities as reading and driving. The peripheral or side vision is generally not affected. Retinitis pigmentosa (RP) first affects peripheral and night vision and eventually leads to total blindness. There is no cure for either disease.

"Our best findings are exciting because they will help scientists understand how this gene works and how a single mutation causes macular degeneration," says Ayayngari.

She said that other scientists, including Kellogg's Anand Swarop, have studied the RPE65 gene extensively and have discovered a large number of mutations

that lead to retinal diseases, including RP.

Now, with the new discovery, researchers may gain a more complete understanding of the mechanisms that protect or destroy central and peripheral vision.

STUDY UNCOVERS LINK

Ayayngari discovered the gene's role in macular degeneration in her study of 10 males, all from the same family, who had the early-onset form of the disease. Only one of the 10 had some damage to peripheral vision (in addition to having macular degeneration) that is associated with RP. The men were affected at an early age—teens or young adults—rather than at the typical age of 60 or older.

According to Ayayngari, "As we learn more about early-onset macular degeneration, we

are certain to understand more about age-related macular degeneration.

She explains that by studying the course by the disease in families with different ages of onset, researchers may find common features or genetic clues that apply to both forms of macular degeneration.

Ayayngari notes that the patients in the study had a form of macular degeneration that is similar to the most prevalent form of the disease: dry age-related macular degeneration.

The Kellogg scientist also describes the form of macular degeneration in the study as X-linked, meaning the gene is carried on the X chromosome. Because men have only one X chromosome (paired with a Y), they have a 50 percent chance of inheriting any such mutation carried by their mothers.

Because women have two X chromosomes, one defective

gene is not sufficient to cause the disease; instead women serve as carriers, passing the bad gene to sons, who then may develop macular degeneration.

In the Kellogg study, 12 female family members known to carry the RPE65 gene were examined. None of the women had any abnormal loss of visual acuity.

Ayayngari points out that not all forms of macular degeneration are X-linked; in the general population women are as likely as men to be affected.

The study also revealed that the mutation for macular degeneration in the study RPE65 gene may have one of two characteristics: the defective gene may be shorter than the healthy gene, or it may generate an entirely new protein.

Ayayngari's next steps are to learn more about the nature of the mutation, and then to design studies that could one

day lead to treatments for macular degeneration. For example, if the gene is found to create a new protein, scientists could look for ways to block it from interfering with other proteins in the retina, or, alternatively, to promote the action of necessary proteins.

Ayayngari is the primary author of the paper published in *Genomics*. Among the other authors is Paul A. Sieving, M.D., Ph.D., Director of the National Eye Institute (NEI). Before his appointment to the NEI, Sieving was a physician and researcher at the U-M Kellogg Eye Center.

For the study, he characterized the clinical characteristics of the family over a ten-year period.

In addition, Kellogg associate professor Julia E. Richards, played a key role in mapping the gene during the early part of the study.

MEDICAL DATEBOOK

SEPTEMBER

Blood drive

Garden City Hospital will hold a blood drive 7 a.m. to 5 p.m. Monday, Sept. 16, in the Alan Breakie Medical Office Building on the hospital campus at 6245 Inkster Road. The Red Cross is operating on less than a one-day supply of blood. Call Terry Carroll at (734) 458-4267.

Arthritis Self-Help

The Arthritis Foundation, Michigan Chapter will offer an "Arthritis Self-Help Course" 1:30 p.m. Sept. 18 and 25 at St. Mary Mercy Hospital, 36475 Five Mile Road, Livonia. Call (800) 494-1650.

Osteoporosis

Dr. George Leach, a Livonia-based endocrinologist, will offer free bone density screenings of the heel 8-11 a.m. Saturday, Sept. 21, at Clarensville Middle School, 20210 Middlebelt Road, Livonia. Seminar includes question and answer session. Call (248) 476-1210.

Menopause

Dr. Pam Smith of the Center for Health Living & Longevity will discuss "Nutrition, Weight, Memory & Menopause" 1:30 p.m. Wednesday, Sept. 25, at Health Dimensions, 32995 Hamilton Court (south off 12 Mile Road between Farmington and Orchard Lake roads), Farmington Hills. Cost is \$15. Call (248) 489-1573.

Fibromyalgia

The CFS/Fibromyalgia Support Group meets 7 p.m. the second Thursday of the month at the Livonia Civic Center Library, 32777 Five Mile Road. The next meeting is Sept. 12. Call (248) 346-3164.

Prostate screening
Garden City Hospital will offer a prostate screening day Sunday, Sept. 15, at the Allen Breakie Medical Office Building. Screening includes PSA blood test. Cost: \$5. Call (734) 458-4330 to schedule an appointment.

Loss of baby

"Parents Supporting Parents," a support group for parents coping with the loss of an infant through miscarriage, stillbirth or newborn death, meets 7 p.m. the third Monday of each month at Providence Hospital-Southfield, 16001 W. Nine Mile, Southfield. Next meeting Sept. 16. Call (248) 849-5342.

Alzheimer's/Dementia care
Lutheran Home Livonia, 28910 Plymouth (between Inkster and Middlebelt), Livonia, will offer

"Communicating & Connecting with the Alzheimer's/Dementia Resident," a one-hour program presented 6:30 p.m. Tuesday, Sept. 17, 9 p.m. Wednesday, Sept. 25, at Health Dimensions, 32995 Hamilton Court (south off 12 Mile Road between Farmington and Orchard Lake roads), Farmington Hills. Cost is \$15. Call (248) 489-1573.

Birth
Dr. Daniel Laframboise will present on workshop on "Birth Trauma" 7 p.m.

Thursday, Sept. 12, at Andan Chiropractic Clinic, 18444 Farmington Road, Livonia. Call (248) 474-5252.

Health care

Learn about the types of services home care provides, how to choose a home care provider and payment options. St. Mary Mercy Hospital will host a lecture on "Health Care Options in Your Home" 1:30 p.m. Wednesday, Sept. 18, in the Auditorium. All who attend will receive a free booklet on "A Consumer's Guide to Home Health Care" No fee. Pre-registration is required. Call (734) 655-8940.

ANA Walk

The American Heart Association's Metro Detroit American Heart Walk will take place Saturday, Sept. 21, on the grounds of the Meadowbrook Festival at Oakland University, Rochester.

Walkers can choose a 1 mile or 2.5-mile course. Call (800) 968-1193 or visit www.heartwalk.kintera.org/detroit.

Think Trim

"Think Trim," an alternative to dieting, will be presented at Schoolcraft College, 18600 Haggerty Road, Livonia, 9 a.m. to 4:30 p.m. Saturday, Sept. 21. The jewelry workshop costs \$57. To register, call (734) 462-4413.

Children's clothing
Eliminate the stress of shopping. The Tri-County Mothers of Multiples will hold a children's clothing and equip-

ment sale 10 a.m. to 12:30 p.m. Saturday, Sept. 21, at the William Costick Activities Center, Shannon Hall, 28600 11 Mile Road, Farmington Hills. Call (248) 446-9717.

Grief support

Angela Hospice in Livonia offers ongoing grief support groups every second and fourth Tuesday of the month. The next meeting is 1 p.m. and 6:30 p.m. Tuesday, Sept. 24. Call Bereavement Coordinator Ruth Favor at (734) 441-7810.

College

"Saving for College," a one-session class sponsored by Beaumont Hospital's award-winning Parenting Program and taught by a certified financial planner is offered 7-9 p.m. Wednesday, Sept. 25. Class meets in the hospital's Administration Building, Classroom 1, 3601 W. Thirteen Mile, Royal Oak. Call (800) 613-7377.

Midwifery Institute Fund-raiser
Family Fun Day noon to 4 p.m.

Saturday, Sept. 28, West Park Band Shell, Ann Arbor, live music, games, face painting, karate demonstration, complimentary refreshments, scavenger hunt. The midwifery Midwifery Arts Center 722 Brooks St. Call (734) 663-1523 or visit www.holisticmidwifery.org for details.

OCTOBER

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Senior fest

The Center for Lifelong Learning at Henry Ford Community College will hold an Autumn Senior Fest 10:30 a.m. to 1:30 p.m. Friday, Oct. 4, at 22586 Ann Arbor Trail in Dearborn Heights. Sarah Talley from the Social Security Administration will discuss the Social Security Trust Fund extension, employment income after retirement, what every woman needs to know about Social Security, and related issues. Cost is \$10 per person and includes a buffet lunch, Big Band entertainment, exhibits, and door prizes. To register, mail a check (payable to Henry Ford Community College) to Senior Fest, Center for Lifelong Learning, 22596 Ann Arbor Trail, Dearborn Heights, MI 48127 by Sept. 15. Call the Center for Lifelong Learning at (313) 317-1500.

Healthy heart

Beaumont Hospital's Sharing & Caring program will offer "Keeping a Healthy Heart" 7-9 p.m. Thursday, Oct. 3, in the hospital's 1st Floor Conference Room, Beaumont Cancer Center, 3577 W. 13 Mile Road, Royal Oak. Call (248) 551-559. Call (877) 345-5500.

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2002 FATHER'S CONFERENCE

BY DADS... FOR DADS... ABOUT DADS

Dads Empowered & Birmingham Youth Assistance, presents the 3rd Annual Michigan Fathers Conference



SATURDAY, OCTOBER 5, 2002

8:00 AM-12:30 PM

\$22 (includes continental breakfast & childcare)

380 SOUTH BATES ST • BIRMINGHAM

CALL (248) 644-5832 TO REGISTER!

KEYNOTE SPEAKER

JAMES LEVINE, Ed.D.

DIRECTOR, THE FATHERHOOD PROJECT

Daddy Stress/Daddy Success: New Strategies for Balancing Work & Family. How dads can reduce the conflict they feel between work and family life and increase their connection with their children.

Workshops Include:

- Dads & Daughters • Parenting Your ADHD Child • Co-Parenting
- Father Talk • Media's Messages to Dads • Dads and Sons
- Husband, Father, Man: Setting Priorities • Secrets of an At-Home Dad

(Additional workshops available)

Speakers Include:

- Chris Edwards, FOX 2 News, chief meteorologist
- Tom FitzPatrick, therapist & co-director of the Fathers Forum
- Roger McPhail, director of the Fathers Resource Center
- Chick Moorman, creator of Parent Talk
- James Windell, M.A. clinical psychology
- Drew Yanke, co-director of the Fathers Forum

WORKSHOP FOR MOTHERS:

How Mothers Can Promote Successful Fathering in Today's Family

Saturday, October 5, 2002 • 5-7pm

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Location: Oakland Schools

3800 Lake Road • Waterford

Call to register: 248-542-4882

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