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[Search](#)

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[Eye Information](#)

[Education and
Training](#)

[Research](#)

[Endowments](#)

[Giving to Iowa
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[News items](#)

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UI researchers identify mutated gene associated with form of blindness

IOWA CITY, Iowa -- A research team led by University of Iowa Health Care investigators has identified a gene that, when mutated, causes a hereditary form of blindness. Although the type of blindness is rare, the researchers hope that the methods and models they developed to study the condition will help in investigations of other forms of blindness.

In the February issue of *Nature Genetics*, the researchers report that 94 percent of the subjects they screened who have Enhanced S-cone Syndrome (ESCS) have a mutated NR2E3 gene located on chromosome 15. ESCS is a gradually deteriorating condition, initially involving loss of night and peripheral vision.

"This study has far-reaching biological implications," said Val Sheffield, M.D., Ph.D., UI professor of pediatrics and a Howard Hughes Medical Institute associate investigator.

Sheffield and his colleagues believe that they and other investigators can use the model for the ESCS investigation to study other forms of blindness and their possible genetic causes. This work may eventually help clinicians when diagnosing and then treating the conditions.

"By identifying specific genes, you can put people into potentially effective treatment categories," said Sheffield, explaining that possible treatments would depend on the type of blindness.

Sheffield and his co-investigators discovered the ESCS-NR2E3 connection while looking for the cause of Bardet-Biedl Syndrome, another disorder that results in blindness and that also includes abnormalities such as mental retardation, obesity and short stature. The researchers determined that the mutated gene on chromosome 15 was not involved in Bardet-Biedl Syndrome but was associated with the eye.

The researchers then looked at DNA samples from the laboratory of Edwin Stone, M.D., Ph.D., UI professor of ophthalmology and visual sciences. The researchers found the mutations on chromosome 15 in two patients' DNA. Both patients had been diagnosed with ESCS.

The researchers then went on to find an additional 27 individuals with ESCS who had mutations to the NR2E3 gene. That finding was made possible thanks to Samuel Jacobson of the University of Pennsylvania, who supplied the researchers with the subjects.

Sheffield said he is not sure of the exact role of NR2E3, but he believes that the gene may be involved in a molecular "switch" that tells a cell to become an S cone.

A person relies on two types of cells to see -- rods and cones. Rods allow an individual to distinguish between dark and light, but not colors. Rods help in night vision. Cones allow a person to see colors. There are three types of cones, based on wavelengths: short, medium and long. The short wavelength cone, or S cone, is usually the rarest type of cone. Typically, only about 5 percent of a person's cones are S cones.

The enhanced S cones production is what makes ESCS so interesting, Sheffield said. In most diseases that lead to blindness, photoreceptors (the rods and cones) die. In ESCS, some photoreceptors do die, but there are an increased number of S cones that make a person more sensitive to blue light.

The study was supported in part by Public Health Service research grants, the Foundation Fighting Blindness, the Grousbeck Family Foundation, the Carver Charitable Trust and the Horvitz Family Foundation.

University of Iowa Health Care describes the partnership between the UI College of Medicine and the UI Hospitals and Clinics and the patient care, medical education and research programs and services they provide.

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