

# Gene mutation linked to retinitis pigmentosa in Southwestern US Hispanic families

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Thirty-six percent of Hispanic families in the U.S. with a common form of retinitis pigmentosa got the disease because they carry a mutation of the arrestin-1 gene, according to a new study from researchers at The University of Texas Health Science Center at Houston (UTHealth) School of Public Health.

Retinitis pigmentosa is a group of rare, genetic eye disorders in which the retina of the eye slowly degenerates. The disease causes night blindness and progressive loss of peripheral vision, sometimes leading to complete blindness. According to Stephen P. Daiger, Ph.D., senior author of the study, an estimated 300,000 people in the U.S. suffer from the disease, which gets passed down through families.

In the study published recently in *Investigative Ophthalmology & Visual Science*, UTHealth researchers found that in a U.S. cohort of 300 families with [retinitis pigmentosa](#), 3 percent exhibited a mutation of the arrestin-1 gene. However, more than 36 percent of Hispanic families from the cohort exhibited the arrestin-1 mutation and they all came from areas in the Southwestern U.S., such as Texas, Arizona and Southern California.

"When I started studying retinitis pigmentosa in 1985, we set out to find the 'one' gene that causes the disease. Thirty-three years later, we've found that more than 70 genes are linked to retinitis pigmentosa," said Daiger, a professor in the Human Genetics Center and holder of the Thomas Stull Matney, Ph.D. Professorship in Environmental and Genetic Sciences at UTHealth School of Public Health.

Some of the genes that cause retinitis pigmentosa are recessive, which means two mutations are required, and some are dominant, which means you only need one mutation. Arrestin-1 piqued Daiger's interest because that particular mutation

is dominant while all previously found [mutations](#) in the gene are recessive. This unexpected finding shows that even a single mutation in the gene is sufficient to cause the disease.

Daiger and his team have identified the genetic cause of retinitis pigmentosa for 75 percent of families in their cohort. Possible treatments for some forms of retinitis pigmentosa are being tested but are still limited. However, the speed at which companies are developing gene therapies and small molecule therapies gives reason to hope, he said. Daiger and his collaborators have begun to connect some of the patients in the retinitis pigmentosa cohort to clinical trials that treat specific [genes](#).

"I want our cohort families to know that even if there is not an immediate cure for their specific gene mutation, at this rate it won't be long until a therapy becomes available," said Daiger, who also holds the Mary Farish Johnston Distinguished Chair in Ophthalmology at McGovern Medical School at UTHealth.

Support for the study, titled "A novel dominant mutation in SAG, the arrestin-1 gene, is a common cause of retinitis pigmentosa in Hispanic families in the Southwestern United States," was provided by the William Stamps Farish Fund and the Hermann Eye Fund.

**More information:** Lori S. Sullivan et al. A Novel Dominant Mutation in SAG, the Arrestin-1 Gene, Is a Common Cause of Retinitis Pigmentosa in Hispanic Families in the Southwestern United States, *Investigative Ophthalmology & Visual Science* (2017). [DOI: 10.1167/iovs.16-21341](https://doi.org/10.1167/iovs.16-21341)

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