

HEALTHY LIVING

Will you inherit macular degeneration?

For those at-risk, saliva test can help gauge chances of developing the disease

BY BILL CORNWELL

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A sophisticated new genetic test that can help gauge the risk of developing Age-Related Macular Degeneration — the leading cause of legal blindness in the United States — and possibly lead to early, sight-saving treatment is now available in Naples and Fort Myers.

Dr. Ashish Sharma of Retina Consultants of Southwest Florida announced last week that the practice is offering the Macula Risk test at all five of its offices. In addition to Naples and Fort Myers, Retina Consultants maintains offices in Bonita Springs, Cape Coral and Port Charlotte.

Dr. Sunil Malkani of Eye Centers of Florida said his group became the first medical facility in the United States to offer Macula Risk when it began testing patients last March at its main office in Fort Myers.



Dr. Ashish Sharma of Retina Consultants of Southwest Florida.

Dr. Malkani, who serves on Macula Risk's physician advisory board, said there are plans to expand the testing to the group's other offices. Although he did not have exact figures, he estimated that more than 30 patients have availed themselves

of the test in Fort Myers.

Both physicians said Southwest Florida experiences high rates of AMD, principally because of the region's concentration of older residents. It is estimated that some 20 million American suffer from AMD, according to Retina Consultants.

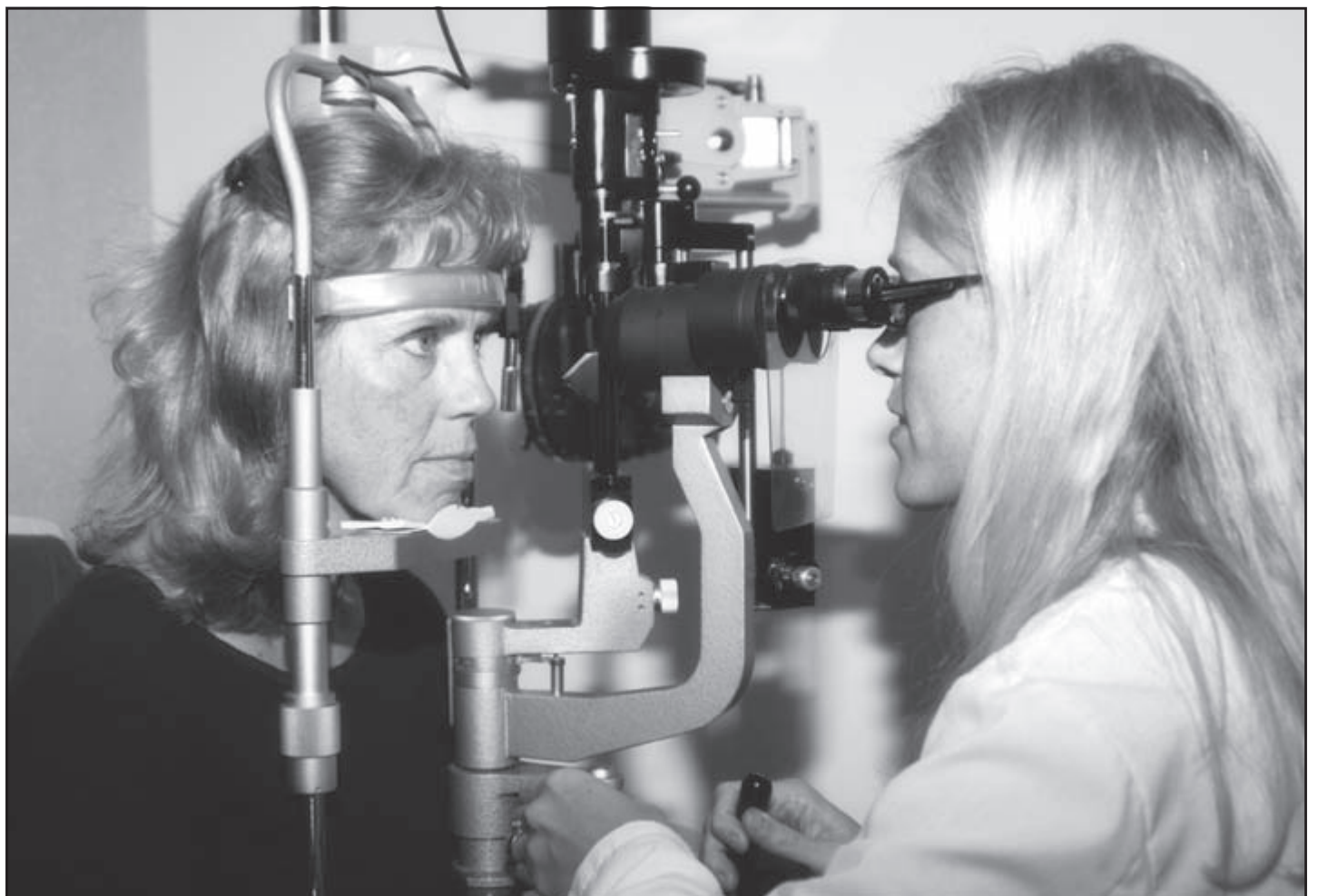
"Because of the large elderly population in Florida, we tend to see much more AMD than other areas do," said Dr. Sharma.

AMD is a progressive disease associated with aging that erodes vision and can lead to a total loss of central vision, which is controlled by a small part of the retina known as the macula. Composed of light-sensitive cells at the center of the retina at the back of the eye, the macula discerns details of focused, straight-ahead vision. These details are essential to proper eyesight. If the macula is compromised, images become dim and "black holes" obscure or block normal sight. These sorts of sight disturbance are usually warning signs of AMD.

Anyone with a family history of AMD (which is usually defined by an occurrence of the disease in a parent or sibling) should be keenly aware of changes in their vision, even if they decide against genetic testing, Dr. Malkani said.

Left untreated, these visual disturbances can progress, resulting in varying degrees of blindness that can impair or eliminate the ability to read, drive, watch television or perform any sight-related task.

AMD carries a strong hereditary component. The Macula Risk test, which involves collecting a saliva sample, for the first time allows physicians to determine the likelihood of someone developing advanced AMD. The test examines a patient's DNA, which holds genetic markers that provide clues to the development of AMD. Not every at-risk patient will inherit the AMD markers, but genetic testing is the only way to determine who has the markers in advance of actual symptoms.



COURTESY PHOTOS

Age-Related Macular Degeneration is the leading cause of legal blindness in the United States.

In addition to the saliva sample, patient and family histories are taken at the time of the test, the physicians said.

Testing is strongly recommended for patients with early or intermediate AMD (the test can be predictive of the ultimate severity of the disease) and for individuals over who have a family history of the disorder, said Dr. Sharma. Testing is usually done on patients over the age of 50, since the disease is age-related and usually occurs after the half-century mark has been reached, but Dr. Malkani said some patients are requesting tests while in their 40s. The test need be taken only once in a patient's lifetime.

The test is essential for at-risk individuals, Dr. Sharma said, because early diagnosis and treatment often can prevent the worst consequences of the disease, namely blindness. Many AMD sufferers ignore subtle vision changes until substantial damage has occurred and treatment is ineffective.

Dr. Malkani said the saliva samples are sent to a laboratory that specializes in genetic testing; results are usually available in two to four weeks. The cost of the test, both physicians said, is \$399, and it is currently not covered by most insurance plans.

Dr. Malkani said the test initially cost \$750, but that ArcticDx Inc., the Canadian firm that created the test, agreed to drop the price to the current level in light of the reluctance of insurance companies to provide coverage. But he predicted that the price will rise again, and may eventually settle in the range of \$500.

ArcticDx is a private Canadian enterprise that describes itself on its Web site as a "molecular diagnostic company." ArcticDx also manufactures a test to assess the risk of colon cancer.

Dr. David Chow, a Toronto physician who is an advisor to ArcticDx, is quoted on the company's Web site as saying the Macula Risk "will identify patients at risk

so they can be diligently monitored."

One such patient is Pat Thiel, a 58-year-old nurse who lives in Punta Gorda and will soon be tested at the Retina Consultants office in Port Charlotte. She will be one of the first patients under the care of Retina Consultants to take the test. Her 81-year-old father, who lives in her home state of Minnesota, suffers from AMD.

"My father was diagnosed with AMD in both eyes when he was in his sixties," Mrs. Thiel explained. "It has progressed over the years, and for the last four years he has been totally blind. When he was being treated at the Mayo Clinic, the doctors told him to tell his children about this. They said the probability that it can pass on to a child is great."

Mrs. Thiel said the tragedy of her father's blindness is made more acute by the fact that he otherwise remains in splendid physical condition.

"He is still in wonderful shape," she said. "He could do what he wanted to do if he wasn't blind. But he is."

As a nurse who works with a family practice physician, Mrs. Thiel said she occasionally comes across patients who exhibit symptoms of AMD. "We refer them (to an ophthalmologist), of course, but it impresses on me that early detection is key," she said. "And there are a lot more treatment options than there were for my father."

Although Mrs. Thiel professed to be thankful that a test of this sort has been developed, she also acknowledged that the anxiety of learning if she is indeed at high risk for AMD is a bit unsettling.

She said she is mentally preparing herself for the results, whatever they may be.

"Yes, I am afraid of losing my vision," she said. "And I will be even more concerned if I find that the level of risk is higher than I had known. But I really have no choice. I've been telling (her father) that I'm going to do it, and I am. Still, I am very anxious."

Mrs. Thiel said her two sisters in Min-

IT'S NOT JUST IN THE GENES

While heredity plays a major role in AMD, it is not the only risk factor, and the developers of the genetic test note that many — but not all — of these risk factors are controllable. Besides age and genetics, risk factors include:

- ▶ Smoking
- ▶ Obesity
- ▶ Race (Caucasians have a higher risk of AMD)
- ▶ Gender (females are more susceptible)
- ▶ Hypertension
- ▶ High blood cholesterol
- ▶ Exposure to UV light (sunglasses can reduce this risk) ■

nesota probably will be tested since they all are above the age of 50 and, of course, share the same genetic risk.

There is no "cure" for AMD, and blindness once experienced cannot be reversed, but the early introduction of drug therapies can sometimes prevent or often arrest the advancement of the disease. Blindness is not inevitable, if the disease is detected and treated at an early stage, both physicians said.

For patients like Mrs. Thiel who are at genetic risk for AMD, the test could be one of the most important medical procedures she ever receives. Whatever the results, she wants to know what the future might hold and what she can do to keep her sight. She does not want to passively accept her fate.

Mrs. Thiel said she will take the test soon, meaning that in several weeks she should have the results in hand.

"I will be more concerned if I learn that I am at high risk," she said, "but I would be more concerned not knowing what that risk is." ■