MEMBER STORY

by Best Doctors®

NAMES:
Steven, Daniel and Marie
AGES:
10, 15 and 46

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REASON MOTHER CONSULTED BEST DOCTORS:
worried that her son had an inherited eye disorder

BEST DOCTORS EXPERT SPECIALTY:
ophthalmology with sub specialty in medical retinal diseases

A plan of action for a family facing a genetic disorder

Marie had a family history of retinitis pigmentosa (RP), a genetic eye disorder that causes degeneration of the retina and can lead to severe vision impairment and even total blindness. She had therefore always been vigilant about any sign of a problem in her two sons, Daniel and Steven. So when 15 year old Daniel mentioned having blurry vision while playing football, she was immediately apprehensive. She had taken him for a visual acuity scan (ERG) when he was 9, and had been told it was inconclusive. However, while he had an aversion to glare, she had not noticed anything of particular concern, until now.

Marie thought of her relatives, going back at least four generations, who had lost their vision and quality of life to RP. If there were any chance that her son was affected, she was determined to act quickly and make sure that she received the best advice possible.

> **"When I learned that I was able to access Best Doctors through my insurance policy, I didn't think twice about it. For me and my family, it's one of the main benefits of being with my insurer."**

Best Doctors contacted one of the world's top specialists in retinal diseases. Reviewing Daniel's test results and symptoms, he first of all reassured Marie that it was highly unlikely that her son had RP. In his opinion, the ERG at 9 years old had actually been normal and, moreover, Daniel did not suffer from night blindness, typically the earliest symptom which usually appears at adolescence.

The expert was fascinated by the singularity of the case: with such a strong family history, why wasn't Marie a carrier? Suspecting that she did in fact carry the RP gene without suffering from the disease itself (a rare situation known as "reduced penetrance") he was convinced that the answers could be found in genetic testing. Only then could Marie and her family understand their risks, plan accordingly, and be eligible for potential clinical trials. Testing should be first performed on living relatives afflicted by RP, which would allow for the identification of specific genes and make it easier and less expensive to then testMarie, Daniel and Steven. Marie also received practical advice on routine testing, diet, supplements and other things she could do to ensure both sons' optimum eye health.

> **"The Best Doctors report gave us information we had not known about before including my own risk. It enabled us to talk knowledgeably about this genetic condition with our family GP, leading to a referral to a genetic specialist and ophthalmologist. We will begin genetic testing soon."**

Reassurance, clarity and a huge burden lifted

> **"I am so glad we went through this process, it has been awesome. We have lived with this disease hanging over our heads for so long."**