



Retest because science has advanced

Steven R. shares his “why retest” story



“In order to confirm my diagnosis, I had to be tested twice. The first time I was tested, I received an inconclusive result. When whole genome sequencing became more affordable, I asked my eye specialist if we could test again.

In retesting, I was able to confirm the very rare variant in my family called *KIZ*, which is a rod-cone dystrophy.

For me, getting retested represented the hope that one day there will be clinical trials for my family to enroll in. To have a chance at benefiting from emerging treatments, I knew I needed to take action to find the genetic cause of my vision loss.”



Steven R.



“Negative” or “inconclusive” may not be the final answer

In the past 10 years, more than 100 new genes related to inherited retinal diseases have been identified. With the evolution of genetic testing technology, creation of more labs, and improved interpretation of results, the genetic cause of vision loss or impairment can now be determined in even more people.



Scan to connect with communities who understand your journey at EyesOnGenes.com.

Now, let's talk about getting retested

Before we start, let's prep. Check off the questions you would like to discuss with your eye specialist or genetic counselor:



Where to begin:

- Am I or my loved one a good candidate for retesting?
- If a previous result was positive, is retesting required?
- What are the steps to getting retested?
- Should we reconnect with a genetic counselor? If so, when should this be done?



How retesting works:

- Which genetic test would you recommend now that science has advanced?
- Will the test be more accurate than the first one that was taken?
- Should additional tests like electroretinography (ERG)* be taken to better confirm the results?



What to expect:

- What are the possible results?
- If an inherited retinal disease diagnosis is confirmed, what will we do next?

If you have any other questions, make sure to note them in the space below:

At your next appointment, be sure to **share this list and discuss the questions you've checked off.**

*A test to measure the electrical response of the eye's light-sensitive cells.