Hello

What does living with an inherited retinal dystrophy mean for you?

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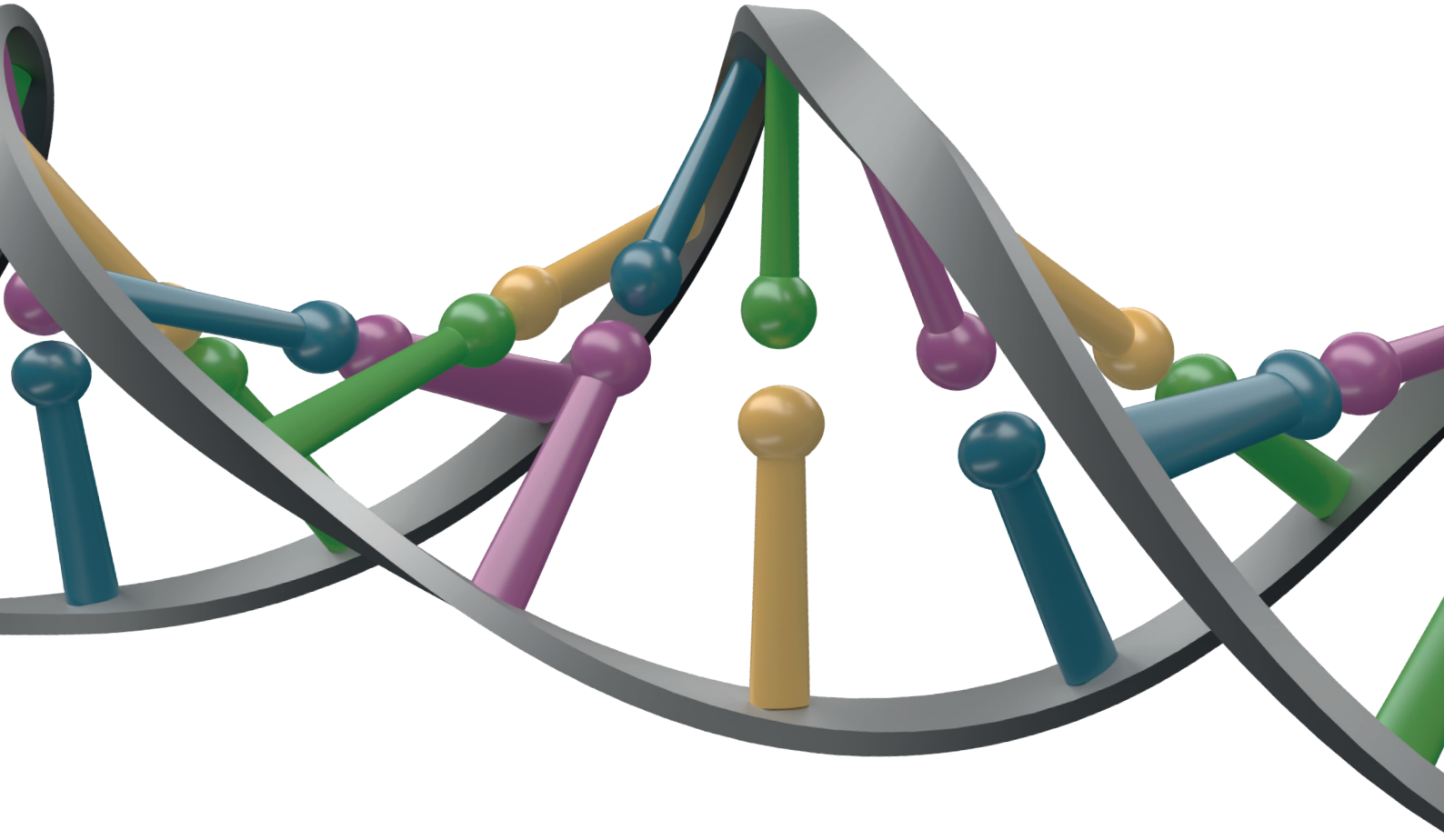
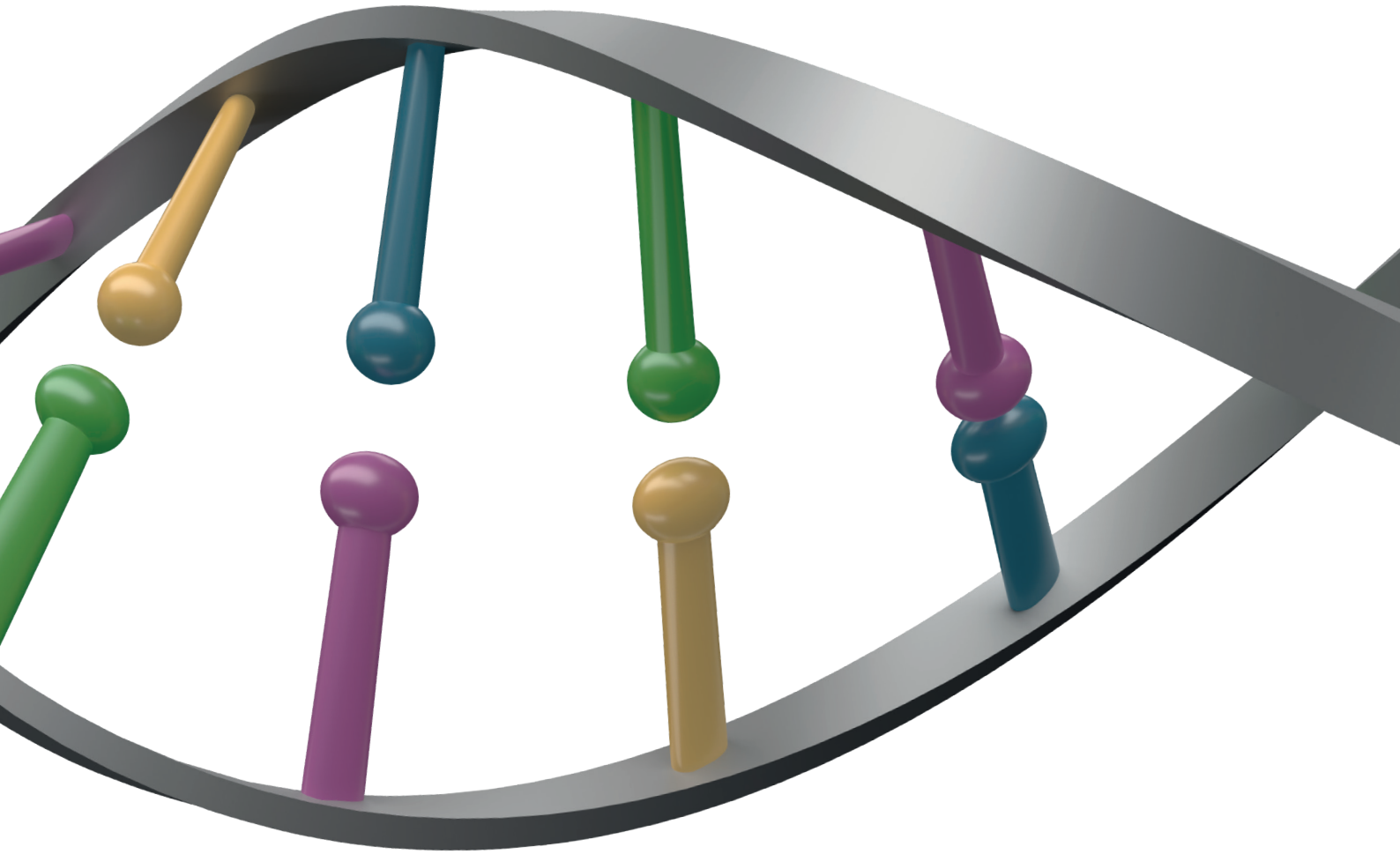
Contents

What is IRD?



Inherited retinal dystrophy (IRD) is a collection of rare eye problems that are usually passed down from parents to children. Sometimes the name differs; IRD can also be called inherited retinal disease or inherited retinal degeneration.[1]



Some of the most common types of IRD are Retinitis Pigmentosa (RP), Usher Syndrome, Leber Congenital Amaurosis and Stargardt Disease.[2] But there are also many others and your doctor should give you more information on which type you have.

What does an IRD diagnosis mean?

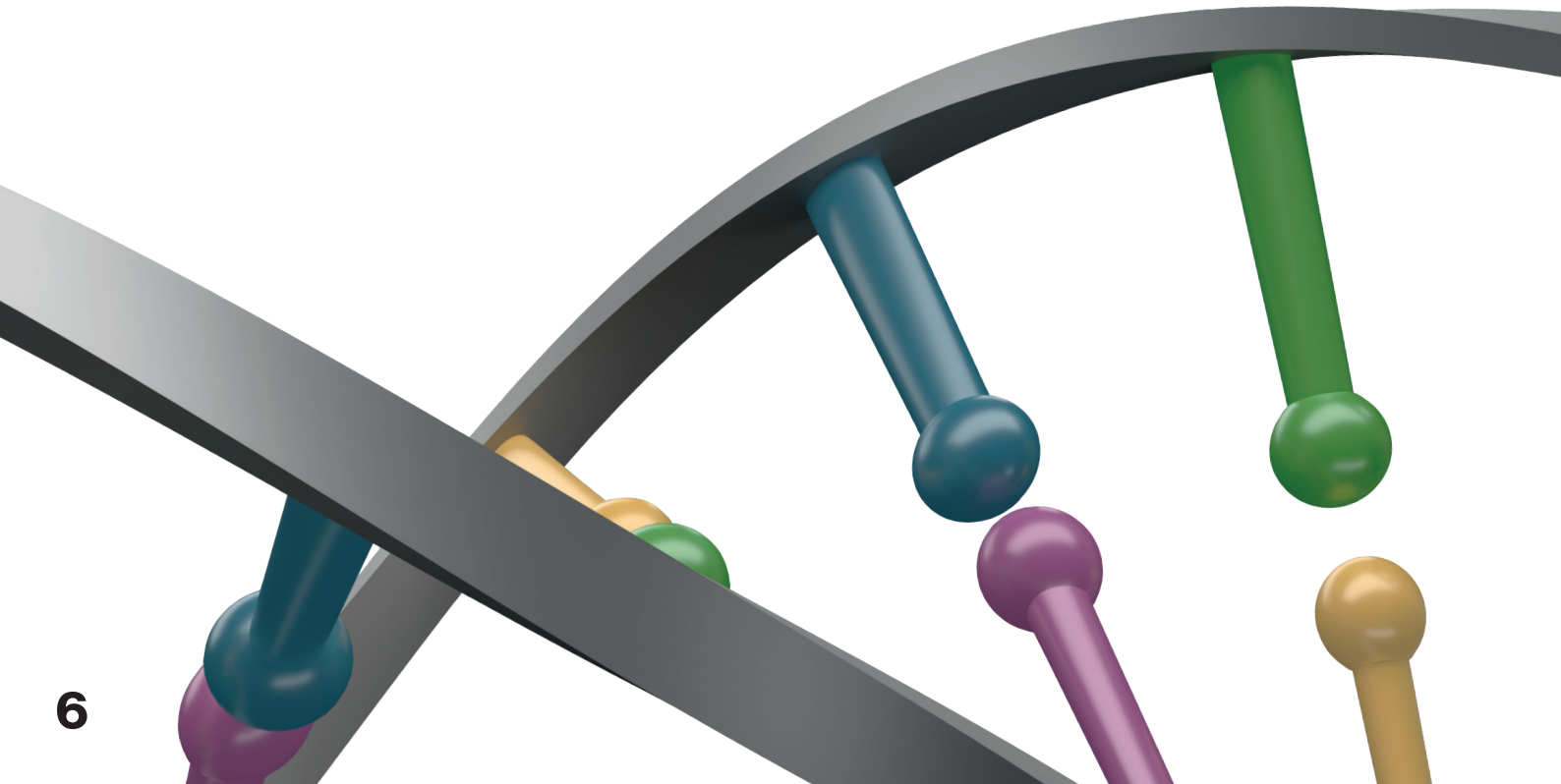
Your healthcare professional has given you this leaflet because you have been diagnosed with a form of inherited retinal dystrophy, or IRD.

Receiving a diagnosis can be extremely overwhelming.

You may go through a range of emotions and have a lot of questions. Hopefully this leaflet, will answer some of them.

It's important to remember that IRD is a progressive disease, meaning that your vision may get worse over time.

Your diagnosis doesn't change who you are.



What causes IRD?

IRD is caused by a mutation in one or more of the genes responsible for the function or the development of the light-sensitive cells in the retina at the back of your eye. These cells either do not develop properly or begin to degrade and stop working, which leads to gradual vision loss in affected people.[1]

You may wonder how you came to have an IRD. Different types of IRD have different inheritance patterns depending on the type of mutation and the gene(s) affected.

To date, over 260 genes linked to IRD have been identified.[3]

How rare is IRD?

While IRD is rare, you are certainly not alone. IRD is estimated to affect more than **2 million people worldwide** (about one in every 2,000 people).[4]

People of any age can be affected by IRD but it is most commonly diagnosed in children, and is the main cause of vision loss for people aged 15–45 years.[4]

How is IRD diagnosed?

There are two types of diagnosis for IRD; clinical and genetic.

You should have received the clinical diagnosis from your doctor, telling you which type of IRD you have. Reaching a clinical diagnosis involves observing the signs and symptoms and running some tests to check your eyes, such as taking photographs of the retina at the back of the eye.

A clinical diagnosis should give you information about which type of IRD you have and what the likely course of your disease will be.

The next step after a clinical diagnosis is a **genetic diagnosis,** which is needed to **identify the specific gene mutation causing your IRD and help identify the next steps.**

A genetic diagnosis can only be confirmed after genetic testing. For your test, a specialist laboratory will look at your DNA, usually from a blood or saliva sample. They will then identify the gene mutation that is causing your IRD and confirm your clinical diagnosis. To date, over 260 gene mutations linked to IRD have been identified.[3]

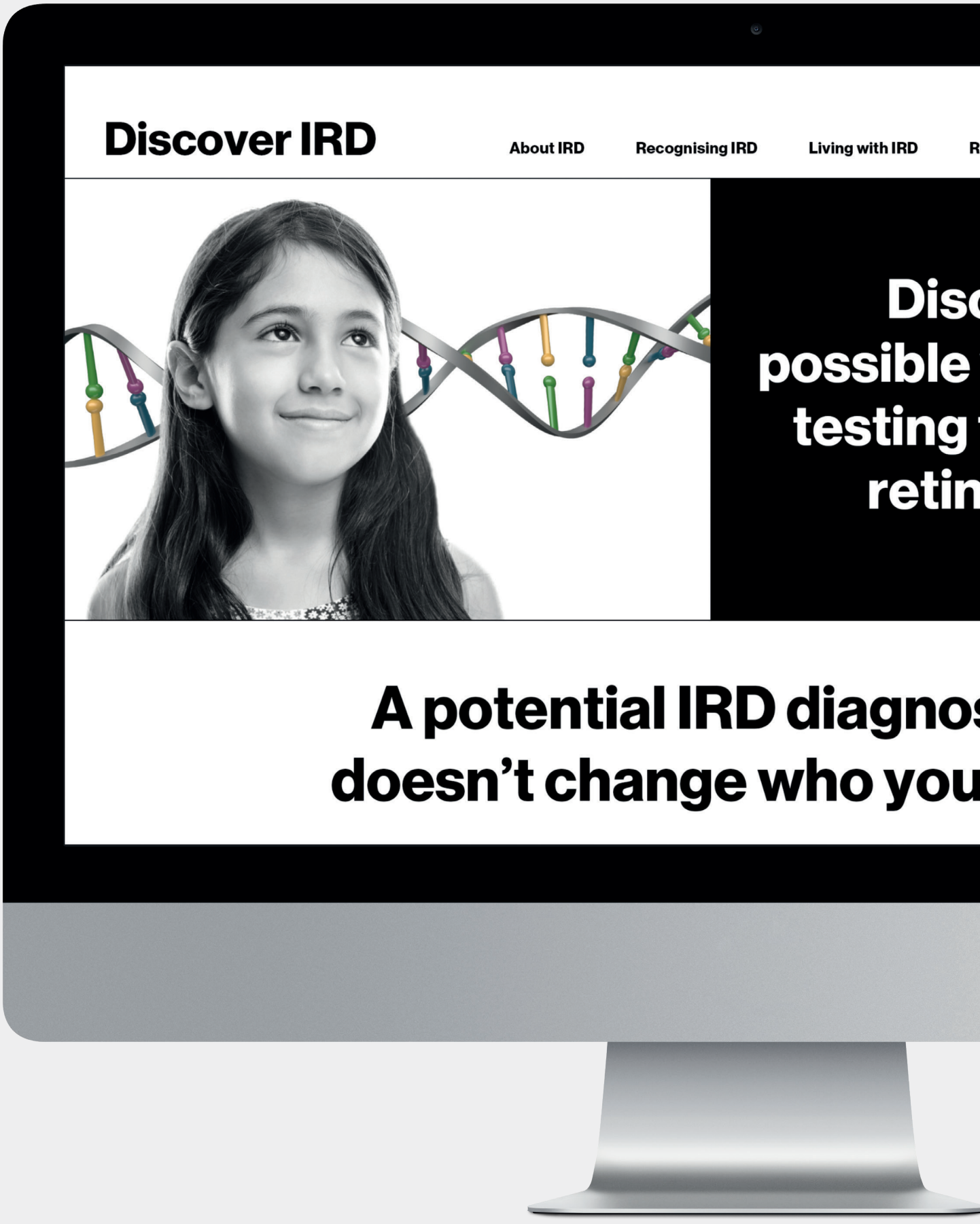
What can a genetic test offer?

A genetic diagnosis can give a better idea of how your disease may progress, which will help you better manage your condition.

A genetic diagnosis will clarify which gene mutation(s) is responsible for your condition and give more information about it. This may help you to feel reassured about your condition, your future, and the likelihood of passing on the disease to any children you might have. Receiving a genetic diagnosis can also identify if you might be a candidate for available treatments or for any clinical trials, where you can be part of research into the treatment of IRD. If you think a genetic test could be the right option for you, speak to your doctor about the possibility.

What can you expect after diagnosis?

After a genetic diagnosis, your doctor will talk to you about your options and may suggest genetic counselling. You may be eligible for treatment or enrolment in a clinical trial and you may want to reach out to one of the many support groups that exist for people living with an IRD.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About IRD
Recognising IRD
Living with IRD
Disc
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testing
retin
A potential IRD diagnos
doesn't change who you

Discover what's possible with genetic testing for inherited retinal dystrophy.

Find out more at

DiscoverIRD.com.au

References

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Novartis Pharmaceuticals Australia Pty Limited. ABN 18 004 244 160. 54 Waterloo Road, Macquarie Park, NSW 2113.

McCann Health NOLX20627M. AU-15225. January 2021.

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