

Retina International Press Release

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DUBLIN, February 28th, 2022- This Rare Disease Day, the members of Retina International are raising awareness for the importance and benefits of genetic testing for Inherited Retinal Degenerations (IRDs).

Rare Disease Day, taking place on February 28th, is a day to demand equity and inclusion for the 300 million people around the globe living with a rare disease. Rare eye diseases do not only affect vision, but they also affect the individual's wellbeing and inclusion in work and education. Retina International highlights the need for equitable access to services, including genetic testing and counselling for those living with an IRD.

IRDs are a diverse group of rare eye diseases that cause progressive degeneration of the retina and often result in loss of vision. These rare eye diseases impact the affected individual, their families, and their caregivers in a number of ways. Retina International's IRD Counts Study conducted in 2019 found that people with an IRD in the USA and Canada were 28.8% and 24.4% less likely to be in paid employment than the general population, respectively.

Getting a genetic test is the only way to definitively diagnose an IRD. A genetic test makes it possible for the individual to participate in appropriate clinical trials for relevant treatments, or to join a patient registry which helps populate clinical trials in the future. Yet, there is a need for greater awareness and training for healthcare professionals on the benefits of genetic testing and counselling for IRDs. Retina International recently undertook a two-phase study to investigate the current genetic testing and counselling landscape. The preliminary data shows that 36% of eyecare professionals were either not aware of genetic testing for IRDs, remained neutral on affected individuals going through the process, or did not encourage it.

Franz Badura, Chair of Retina International, states that, "There is a lack of awareness among both eye care professionals and patients on the importance of genetic testing for IRDs. Because IRDs are rare diseases, some ophthalmologists may see just one case of an IRD in a year, and they are not

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aware where to send the patient, or what further steps to take. Then there are the patients who were diagnosed 20 years ago but have not visited the ophthalmologist since. It's important to educate the community on not only why genetic testing is important, but also on the process of receiving that genetic diagnosis. Awareness is the first step!"

Receiving a genetic diagnosis and counselling is a long and difficult process and the burden is largely on the individual to navigate complex care pathways. In the same RI study, 42% of respondents did not receive genetic counselling prior to genetic testing and 34% did not receive genetic counselling after receiving a genetic test. Genetic counsellors play a fundamental role in the process, equipping the individual with the understanding and information to adapt to a genetic diagnosis. A best practice model on access to genetic testing and counselling services for IRDs is needed urgently.

Dr. Juliana Sallum MD, PhD., expresses that "There is a need for medical education on the importance of going deep on the diagnosis. The clinical diagnosis is not enough, patients need to know their genetic diagnosis. We need to include genetic tests in healthcare providers' list of covered tests and make sure patients are receiving genetic counselling along with the test. In a genetic counselling consultation, the patient has the opportunity to learn about the specific type of IRD they have and the implications for them and their family, regarding the risk of disease for other family members and for future babies in the family. The good news is that there are many labs prepared to provide good quality genetic tests, and the cost is progressively decreasing. Telemedicine also allows equitable access to genetic counselling in big countries with geographical challenges. However, we need to call attention to this matter because there is still a lot to be improved."

This Rare Disease Day, Retina International is calling for equitable, affordable, and timely access to genetic testing services for those living with IRDs. Equitable access to and reimbursement of genetic testing and counselling services will:

1. Improve care and wellbeing of people living with an IRD,
2. Aid decision making,
3. Increase access to clinical trials and therapies (where available), and

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4. Expedite medical research and innovation

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Retina International is a global umbrella organization for patient-led charities and foundations that support research into rare, genetically inherited and age related retinal conditions.

RI believes that education leading to participation is a key driver in successful innovation.

Retina International strives to educate its community who in turn will be the educators of their peers, of all relevant stakeholders and decision makers and will utilize the understanding to Participate as informed actors in development of research, through the provision of advice, and perspective to all stakeholders and decision makers allowing the community to better Innovate to support and promote research for unmet need.

Retina South Africa is a founder member of Retina International

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