

Guide to essential care for IRDs

DISCLAIMER: This Guide is intended to prompt you to ask questions about your care following diagnosis of IRD. It is not intended to act as a clinical guideline for your care. You should consult with your Eye Care Professional (ECP) and local patient support organisations with regards to your individual care, as they have the best knowledge about clinical practice and availability of supports in your local environment.

1. If you (or your family doctor) suspect you or your child have any form of IRD you should have an appointment with an optometrist at your local optician's practice or be referred to an ophthalmologist at a hospital. You should be referred to an ophthalmologist if your optometrist or family doctor believes a specialist eye doctor needs to confirm the diagnosis and/or to monitor disease progression.

2. An important step in the care pathway can be genetic testing to give a genetic diagnosis. A genetic diagnosis is necessary for enrolment in many clinical trials and new treatments that are gene based. However, access to genetic testing varies from country to country. In some regions, a genetic test may be only requested by a medical geneticist while in others the ophthalmologist submits the request. Self-referral is rare for genetic testing. The amount of time that it takes for this process can vary widely from region to region. Please see our Red Alert! Toolkit on genetic testing for more information on the importance of access to genetic testing <http://www.retina-international.org/toolkit-redalert>

Depending on the health care system of your region, clinical grade genetic testing can be paid for by public health care, self-pay or health insurance companies. For testing to determine suitability for clinical trials, the pharmaceutical companies involved in the trial would fund the specific genetic testing. This is known as research grade testing.

Genetic testing is recognised as of utmost importance by researchers and medical professionals alike. In this video, Prof John Flannery speaks about the importance of genetic testing for gene therapy:

<https://www.youtube.com/watch?v=WnTLJBAU1VA>

3. Genetic counselling is an integral part of the genetic testing pathway as many different issues, thoughts and possibilities can arise, from the initial decision to have a genetic test up to what the results can potentially mean. Self-referral is common for genetic counselling in many countries. Your local blindness charity is a good resource for how to access genetic counselling.

4. If you are diagnosed with IRD, you should be advised:

On how to protect your eyes from uncomfortable glare, for example by the use of anti-glare glasses or by adjusting the lighting in your home.

About tools to monitor your vision in case it changes. Any sudden deterioration in vision should be notified to your family doctor or Eye Care Professional immediately.

Of the possibility of seeing visual images known as Charles Bonnet Syndrome. These are called visual hallucinations and affect many people with sight loss as their brain adjusts to reduced visual input. They are not a sign of a mental illness.

5. If you are distressed by the diagnosis, you should be offered emotional support or signposted to a service which can help, for example a patient support group. Please see the member charities of Retina International for a local sight loss charity near you <http://www.retina-international.org/our-members> . You can find out more about this in our '[Coping with a Diagnosis](#)' section.

6. You should be referred to a low vision service for assessment to find out what you need to help support you in your daily life both at home, at school and when you are outdoors. This is known as visual rehabilitation. This should happen before you lose a lot of vision as it is much easier to learn how to make the most of your sight while it is still reasonably good. (In some places you will need to be referred to the low vision service by your ophthalmologist.). You can find out more about this in our '[Coping with a Diagnosis](#)' section.

7. If your sight is poor you should seek to be registered as sight impaired or severely sight impaired. This may entitle you to additional benefits and services that you are not otherwise entitled to avail of.

8. You should continue to have regular eye tests at your optometrist to check your eye health.