

The logo for the National Rare Diseases Office (NRDO) features the letters 'NRDO' in a bold, sans-serif font. The 'N' is blue, the 'R' is purple, the 'D' is magenta, and the 'O' is pink. The letters are arranged horizontally and are the primary visual element of the logo.

**NRDO**

National Rare  
Diseases Office

The National Rare Diseases Office (NRDO) was established by HSE and officially opened by Minister for Health, Dr Leo Varadkar in June 2015.

The office is staffed by information scientists and supported by 2 Consultants all of whom have significant experience working with individuals and families affected by rare disorders

One of the functions of the office is to provide **current and reliable information about rare diseases to people with rare diseases and their families as well as health care providers and researchers.**

Dissemination of information about our office, Freephone information line; contact email and web addresses is ongoing through attendance by staff at conferences; educational meetings; engaging with patient groups, print advertising and social media.



# NRDO

## National Rare Diseases Office



Between June 2015 and March 2016 staff responded to 55 direct information requests on the following conditions:

Amyloidosis?

22q11.2 deletion syndrome

Alkaptonuria

Autoimmune epilepsy

Bannayan Riley Rubalcava

Bernard Soulier Syndrome

Chromosome 2 deletion/full exome Testing

Cockayne Syndrome

DDX3X

Complaint

Dwarfism

EDS

EDS 3

Emory dreifuss muscular  
dystrophy

Fabry/Anderson

Family

G6PD

Gary Lyme disease

Giant Cell Temporal Arthritis

GNe myopathy

Hereditary Spastic

Paraperises

Huntington's Disease

Icthyosis

Loeys- Dietz syndrome

Menieres Disease

Multifocal motor neuropathy

Multiple Sulfatase

Multiple Systems Atrophy

Rare Disease prevalence

Oligastrocytoma

Osteogenesis Imperfecta

PKU

Rare disease

Rare neurological conditions

Stickler

Susac syndrome

SWANS

TMAU

Treatment Abroad vs CBD

TUBB2B - polymicrogyria

tuberous sclerosis

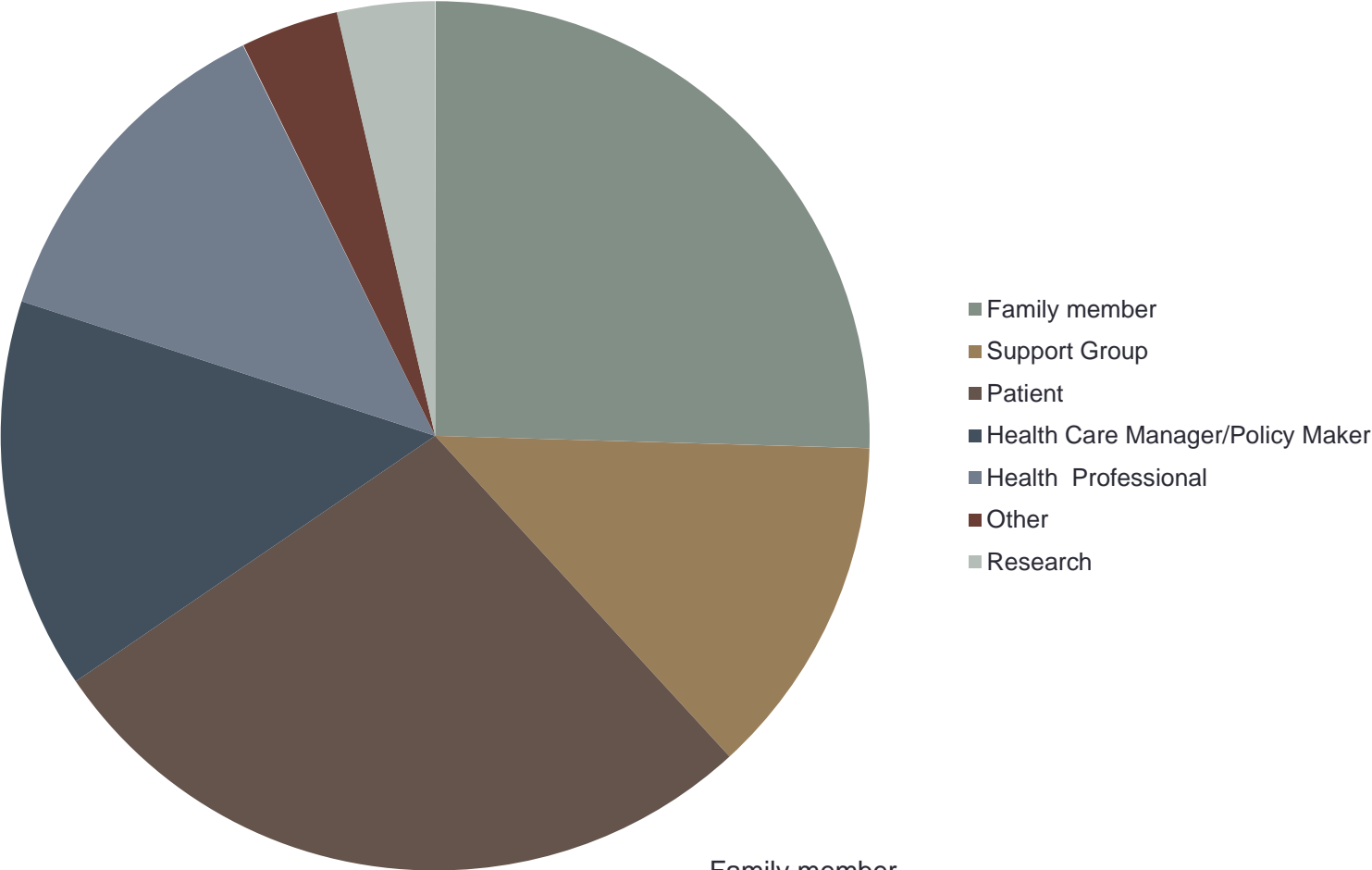
undiagnosed hypertextensibility

Undiagnosed Neurological condition

unknown

Wiskott-Sldrich Syndrome (WAS)

(Fig 1) Profile of contacts

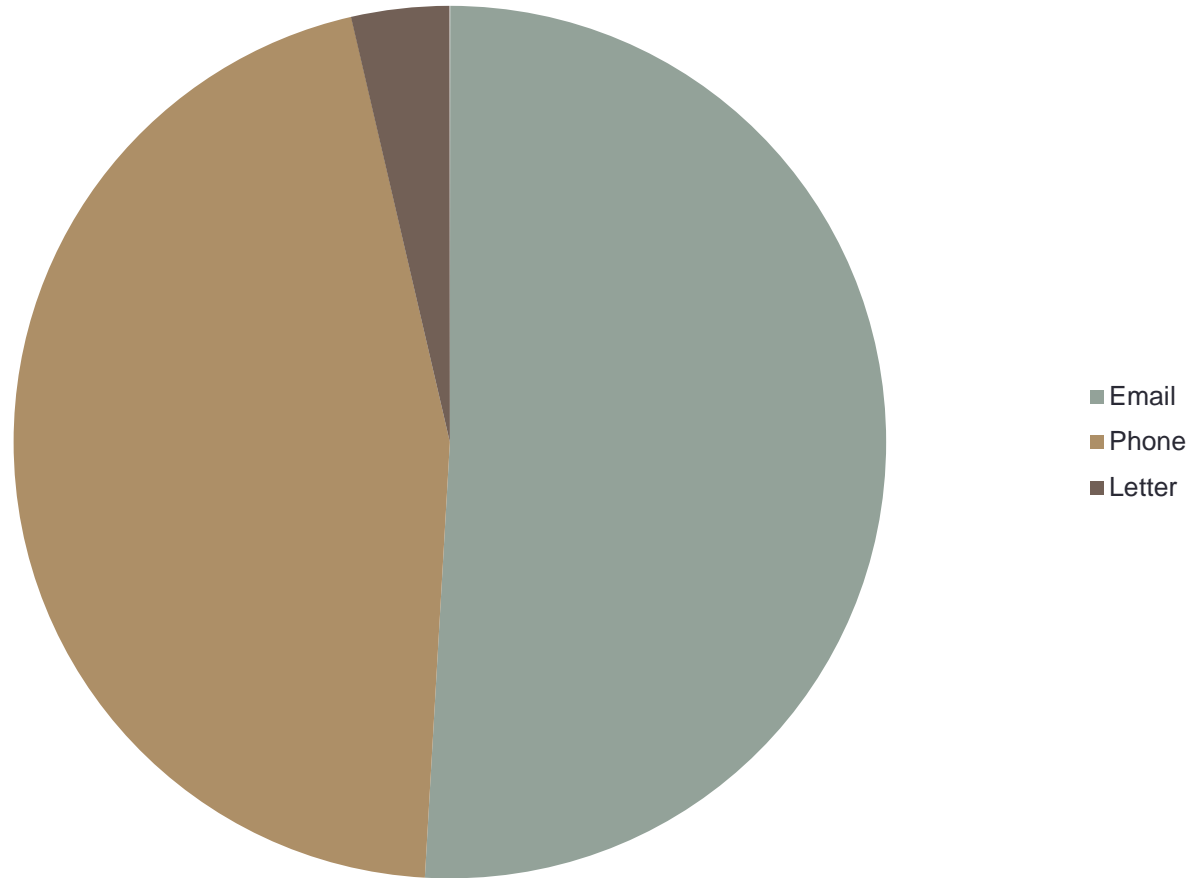


Family member	14
Support Group	7
Patient	15
Health Care Manager/Policy Maker	8
Health Professional	7
Other	2
Research	2



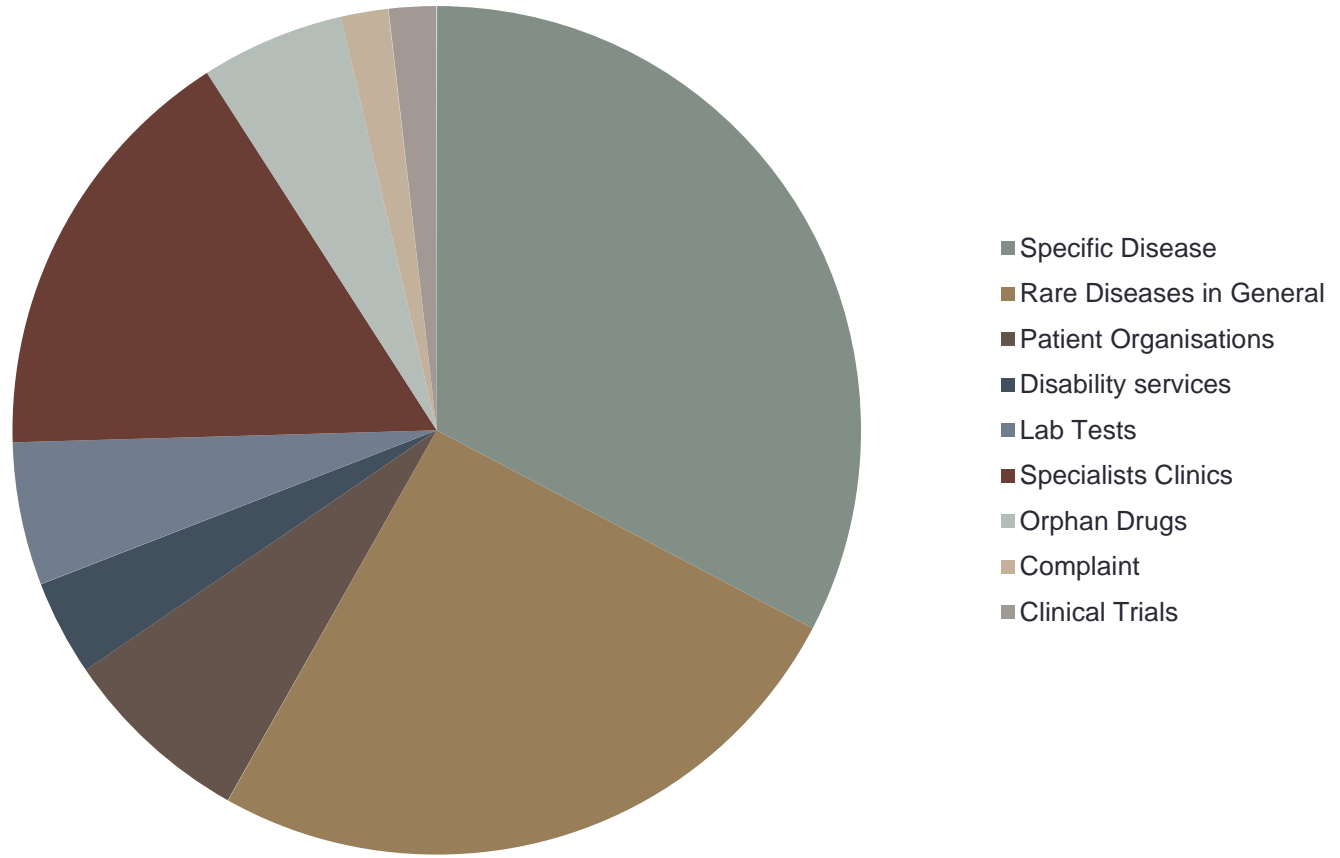
(Fig 2)

Methods of contact with the office (N=55)



Email 28  
Telephone 25  
letter 2

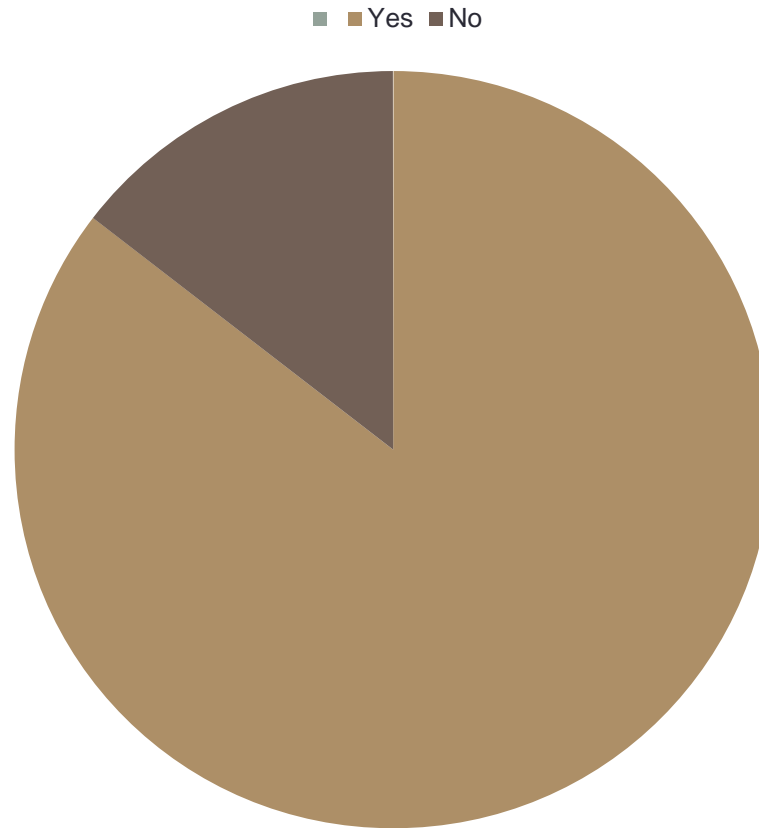
# Categories of Information Requested included



(Fig 3)

Specific Disease	18
Rare Diseases in General	14
Patient Organisations	4
Disability services	2
Lab Tests	3
Specialists Clinics	9
Orphan Drugs	3
Complaint	1
Clinical Trials	1

## Rare Disease related Queries

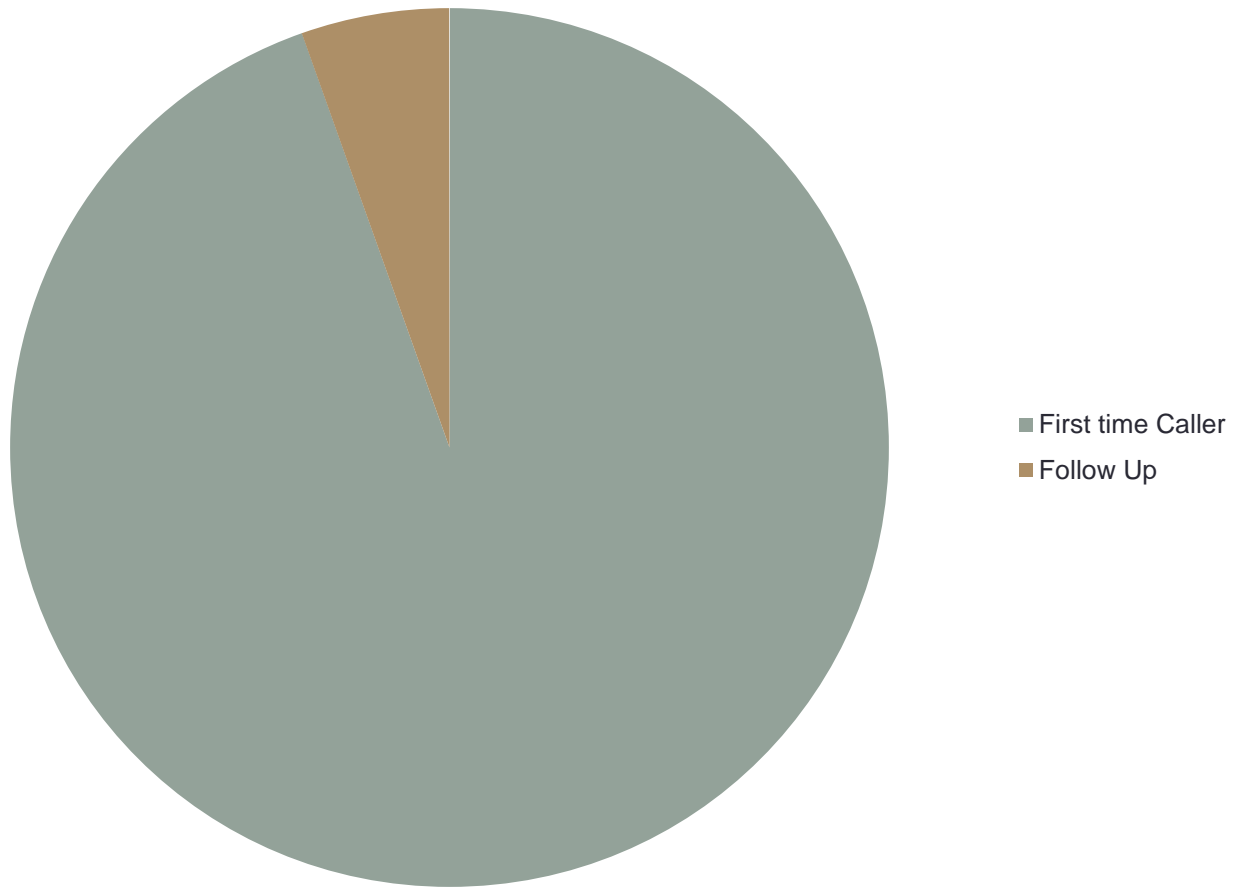


(Fig 4)  
Rare Disease Queries/? More appropriate  
to refer elsewhere

47 of the 55 queries responded to were related to a Rare Diseases; 8 were not rare disease specific and therefore were referred to an alternative more appropriate area



# Contact with Office was in the main first-time callers



First time 52  
Follow up 3

The NRDO maintains an information booth in the concourse area of the Mater Misericordiae University Hospital. This booth contains information leaflets on rare diseases and patient organisations  
Stocks regularly replenished

NRDO and RareConnect information leaflets are also available from the booth



Parent Information



Eolas d'Othair



RARE DISEASES



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## National Rare Disease Office

Share  

### What is a rare disease?

A 'rare disease' is defined in Europe as a life-threatening or chronically debilitating disease affecting no more than 5 people per 10,000. There are an estimated 6-8,000 known rare diseases affecting up to 6% of the total EU population, (at least 30 million Europeans), and perhaps up to 300,000 Irish people during their lives. This means that 6-8% of the Irish population or 1 person in 12 may have a rare disease at some stage in their lifetime. Approximately 70-80% of rare diseases are genetic and are present throughout the person's entire life.

Learn more about rare diseases with the [Irish Easy Guide to Rare Diseases 2013](#) which gives you more information and real-people stories.

### Who we are

The National Rare Diseases Office provides current and reliable information about genetic and rare diseases to patients, families and health professionals. It was established in June 2015 by the Health Services Executive (HSE). It is staffed by Information Scientists who have significant experience working with individuals and families affected by rare disorders. Find out [what we do](#).

For relevant information click on one of the sections below:

- ▶ [Patients and Families](#)
- ▶ [Healthcare Providers](#)
- ▶ [Service Gaps and Complaints](#)
- ▶ [Disclaimer](#)



Find Hea



Conditio



Conne



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- Specialist Care** ▼
- ▶ [Addiction Services](#)
- ▶ [Sexual Health Services](#)
- ▶ [Public Health](#)
- Rare Diseases** ▶
- ▶ [Cancer Services](#)
- ▶ [Kerry Health Services](#)

## Find Specialist or Service

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### How to find a disease specialist in Ireland

For certain rare diseases where this specialist care may not be broadly known, centres in Ireland providing services for patients with rare diseases may be found listed within our national rare diseases database, [Orphanet](#). However, the specialist for your specific rare disease may not be listed in Orphanet. This does not necessarily mean that there is no expertise in Ireland for this rare disease – it may mean that the full range of services may not be available or that the service is in the process of being evaluated for inclusion in Orphanet. You can [contact us](#) for help finding an expert centre or rare disease specialist in Ireland.

Your GP may be able to direct you to the specialist expertise that is relevant to your condition and age in Ireland through [HSE Services](#).

### Appropriate services or treatments for my rare disease

Sometimes services are not available as soon as needed, taking account of a person's current state of health and the probable course of the disease. In cases such as these your specialist consultant doctor may recommend an appropriate treatment in another EU country. In these cases an application to [Treatment Abroad Scheme \(TAS\)](#) may be appropriate.

The [Cross Border Directive \(CBD\)](#) also facilitates Irish residents to access healthcare in other EU countries. This directive describes Irish patients' right to access the same healthcare in other EU countries that they would be entitled to within the public health system in Ireland. Referral for care under the CBD can be made by a GP, a hospital consultant and certain other HSE clinicians. [CBD office contact details](#)

### Find Health Services



- ▶ [by Topic](#)
- ▶ [by Location](#)

### Conditions & Treatments



- ▶ [Explore 600+ conditions and treatments](#)



**Need help?**  
Call our Infoline  
on **1850 24 1850**

### Connect with us







- Apply & Register
- Everyday Care
- Care when you need it
- Community and Social Care
- Specialist Care ▾
  - ▶ Addiction Services
  - ▶ Sexual Health Services
  - ▶ Public Health
  - ▶ Cancer Services
  - Rare Diseases ▶**
  - ▶ Kerry Health Services

## Patients and families

Share Print

Individuals and families who have received a new diagnosis of a rare disease can have many questions. If you are trying to find information about your rare disease or to understand what supports and services are available to you there is information on the links below that can help you. If you do not find the information that you are looking for, [contact us](#).

<a href="#">Difficulty Finding Information</a>	<a href="#">Find Specialist or Service</a>
<a href="#">Newly Diagnosed or Undiagnosed</a>	<a href="#">Benefits and Schemes</a>
<a href="#">Rare Disease Support Groups</a>	<a href="#">Disability Information</a>

Find Health Services




- ▶ [by Topic](#)
- ▶ [by Location](#)

Conditions & Treatments



- ▶ [Explore 600+ conditions and treatments](#)

**Need help?**  
Call our Infoline on **1850 24 1850**



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**Rare Diseases** ▶

## Healthcare Providers

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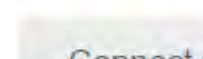
### Information Databases

The following links are recommended for clinicians or healthcare providers seeking reliable information on rare diseases. All are accessible free of charge.

[Orphanet](#) is a European reference portal for information on rare diseases and orphan drugs. This includes information on rare diseases expertise in Ireland.

The [US National Organization for Rare Disorders \(NORD\)](#), provides patient friendly information and an index of physician guides for a number of rare diseases. It is important to note that treatment information will be American focussed.

[Online Mendelian Inheritance in Man \(OMIM\)](#) is a catalogue of human genes and genetic disorders and is targeted at health care professionals and researchers.





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## Contact Us

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Address: National Rare Disease Office, Mater Misericordiae University Hospital, Eccles St., Dublin 7.

Email: [rare.disease@mater.ie](mailto:rare.disease@mater.ie)

Rare Disease Infoline: 01 854 5065 Mon-Thurs inclusive 9.30am -1.30pm

Office Telephone: 01 8097475

### Find Health Services



- ▶ by Topic
- ▶ by Location

### Conditions & Treatments



- ▶ Explore conditions and treatments



Neurology  
Call on 1850 22 22 22

### Connect with us



THANK YOU

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**NRDO**

National Rare  
Diseases Office



Ireland

