Rare diseases (RD) are a significant public health issue

- By definition RD are life-limiting or chronic illnesses
- 1/17 people affected by RD = 270,000 Irish RD patients
- Average 96 RD patients per GP in Ireland
- 75% of RD are paediatric onset
- 1/3 of deaths under age 5 due to RD

ISSUES for RD PATIENTS

- Obtaining a diagnosis, finding the expertise
- Obtaining accurate information
- Navigating the system/integration and coordination of multidisciplinary care

The National Rare Disease Office was set up as one of the key recommendations of the National Rare disease plan 2014-2018

- Educating and empowering patients through knowledge

Central Role: To Respond to RD Information queries

- Patients
- Health care professionals
- Agencies

www.rarediseases.ie
Website

Information Line
1 800 24 0 365

www.orpha.net
Rare Disease resource portal

Reliable RD Information

- Medical Signposting
- Treatment Abroad Scheme and Cross-Border Directive
- Disease specific information
- Clinical Trial and research links

National recognition of RD expert centres suitable to join European Reference Networks and ‘trial ready’ researchers

European Reference Networks (ERNs) for Rare Diseases are networks of clinical expert centres which will improve access to care and treatment for RD patients across Member States

NRDO National and European ongoing activities:

- Recruiting national expert centres to Orphanet to eventually join ERNs
- Developing a Transition model of care integrating national service providers
- Identifying expert centres’ clinical practice guidelines and coordinated care pathways
- Developing rare disease registries