

## CONFERENCE ABSTRACT

### The Irish National Rare Disease Office (NRDO): A national step towards improving access to health and care services for individuals and families living with Rare Diseases

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**Introduction:** In Europe a 'rare disease' (RD) is defined as a life-threatening or chronically debilitating disease affecting no more than 5/10,000 people. There are an estimated 6-8,000 known RD 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. As chronic conditions, many of which are multisystemic, accurate and timely diagnosis and access to treatment is essential for empowering patients and Health Care Providers to manage care effectively. It is recognised that poorly coordinated care and delays in diagnosis are major issues for patients and families affected by rare diseases which are noted to be more problematic in smaller countries with more limited highly specialised expertise, (Rare Diseases UK, 2015 and Irish HSE 2012 'Have your say' Rare Diseases Public Consultation) and as emphasised in the recent EC Expert Group on Rare Diseases 'Recommendations to support the incorporation of rare diseases into social services and policies' (April 2016).

**Practice change and implementation:** In 2014 Ireland published its first National Plan for RD (2014-2018). Establishing a National Rare Diseases Office (NRDO) was a central recommendation of the plan to improve access to reliable information and empower patients and HCPs to make informed decisions about the management of RD.

**Aims and theory of change:** The aim of the NRDO (established in June 2015 by the Irish HSE) is to provide current and reliable information about genetic and RD to patients, families and health professionals. The functions of the office include:

Centralisation of Irish RD information through Orphanet Ireland; an international online portal for RD information and resources encompassing > 6,000 RD. ([www.orpha.net](http://www.orpha.net))

Staffing a RD Information Line to provide patients, families and HCPs with information and supports relating to RD.

Hosting a website with information and links to relevant RD services and organisations around Ireland and Europe [www.rarediseases.ie](http://www.rarediseases.ie)

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Engaging with clinicians, expert centres and researchers to map-out Irish RD resources to act as a 'hub' for integrating National Centres of Expertise in the emerging European Reference Networks.

**Initial outcomes:** In its first year of functioning, greater than 50% of calls to the information line were from patients and families, most commonly requesting signposting of medical specialists for specific RD.

**Conclusions and Outlook:** Over time, it is proposed that the NRDO will liaise with all national RD Centres of Expertise, with the Irish national integrated care programmes, and Rare Diseases European Reference Networks to progress improved integrated care pathways for RD patients and function as a national 'hub' within the Irish and European contexts, in line with the EC Expert Group on Rare Diseases April 2016 Recommendations.

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**Keywords:** rare diseases; integrated; patients innovation

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