National Rare Disease Plan for Ireland: An Interim Report on Implementation

Background
A National Rare Disease Plan for Ireland was launched by the Minister in July 2014. This is a generic policy framework for rare diseases. The scope of the plan is broad given that there are 8,000 rare diseases approximately affecting millions of EU citizens; and consequently, there can be a dearth of expertise and knowledge about some rare diseases, simply because they are so rare. The national plan derived from an EU Council Recommendation in 2009; it declared that the specificities of rare diseases are a limited number of patients and a scarcity of relevant knowledge and expertise; and that progress can be achieved through gathering of national expertise on them, which is scattered throughout Member States. The Council recommendation proposed that Member States develop a national rare disease plan by 2013 and, in particular, the Council called for concerted action at EU and national level in order to:

- enhance research in the field of rare diseases;
- identify centres of expertise by the end of 2013 and foster their participation in European Reference Networks;
- support the pooling of expertise at European level;
- share assessments on the clinical added value of orphan drugs;
- foster patients’ empowerment by involving patients and their representatives at all stages of the decision-making process;
- ensure the sustainability of infrastructures developed for rare diseases.

The National Plan represents Ireland’s response to the European Council Recommendation by providing a framework for action at national level.

Establishment of the Rare Diseases Steering Group
The Group was established by the Minister in 2011. Its purpose was to develop a policy framework for the prevention, detection and treatment of rare diseases based on the principles of high quality care, equity and to be patient centred. The policy is being operated over a 5 year period.
Composition of the Steering Group
The Steering Group was composed of stakeholders from diverse patient organisations, State agencies (Irish Medicines Board, Health Research Board), the Health Service Executive and the Department of Health.

The National Plan
The report consists of 7 chapters exploring a range of subject matters pertinent to the diagnosis, treatment and care of patients with rare diseases and their carers. A brief summary of each of these areas is as follows:

Why a plan is needed
The chapter dedicated to this subject defined rare diseases and estimated the level of these in the Irish population. It set out the background in Ireland and in the European Union context, and included the vision and underlying principles of this first National Rare Disease Plan for Ireland, covering the years 2014-2018.

How the National Rare Disease Plan was developed
The report described the national Steering Group that formulated the Rare Disease Plan and how it was developed with key stakeholders. In particular, it referred to the EUROPLAN Conference 2011 that led to the establishment of the Steering Group, the national consultation conference and the online public consultation, all of which played a key role in the development of the plan. A report on the public consultation process was published alongside the National Rare Disease Plan for Ireland.

Recognition of rare disease – Information and research
A specific chapter is dedicated to dealing with the recognition of rare diseases and the availability of information and research on them. It began with defining ‘a rare disease’ and then proceeded to cover pertinent areas on these topics, such as epidemiology, registers and computer information systems, exploring the strengths and weaknesses of each. It went on to examine the research dimension of rare diseases, covering such areas as funding of research in Ireland, networks of researchers, participation in international initiatives and infrastructure. The final section explored the opportunities and challenges in the area.

Prevention, diagnosis and care
The report explored issues around the prevention, diagnosis and treatment of rare diseases from an Irish context. Screening services, in the guise of the National Newborn Screening Programme, along with primary prevention measures, received attention at the start of the chapter, with the former assessed for its strengths and weaknesses. Genetic testing services in Ireland were examined as part of the diagnostic element of dealing with rare diseases,
followed by treatment and care services available. The context for services for EU patients and the question of improving care and care services, such as respite and palliative care, were also discussed.

**Enhancing access to appropriate drugs and technologies**

The challenges associated with drugs and technologies were examined in the report. It explored the various paths existing for the designation of orphan drugs and technologies, both at European and Irish level; the associated avenues for making these available to patients through community and hospital drug schemes; and accompanying budgets for funding such access.

**Empowering, protecting and supporting rare disease patients and carers**

A chapter in the report provided the opportunity to draw on the issues of rare disease from the perspectives of the patient and the carer. It was about empowering both through various means such as protecting their rights, preserving equity and facilitating access to accurate and timely information. The benefits of providing holistic care packages were examined, together with the impetus for developing the effectiveness of patients’ organisations.

**Implementation, monitoring and review of the National Rare Disease Plan**

It was acknowledged that the National Rare Disease Plan was being published at a time of significant health reform in the areas of primary care and acute hospital care. The report set out implementation and monitoring arrangements through the service planning process, in addition to planning for the next National Rare Disease Plan in 2019.

**Brief Summary of Progress**

The National Rare Disease Plan recommended that an Oversight Implementation Group of relevant stakeholders, including patients' groups, be established to oversee and monitor implementation of the plan's recommendations. This group was established by the Department of Health in 2015 and it has met on a number of occasions. One of the principal recommendations in the Plan was the establishment of a National Clinical Programme for Rare Diseases. This programme was to be responsible for, over time and among other functions, assisting with mapping and developing care pathways for rare diseases; facilitating timely access to centres of expertise – nationally and internationally; and developing care pathways with European Reference Centres for those ultra-rare disorders where there may not be sufficient expertise in Ireland.

In line with this recommendation, a National Clinical Programme for Rare Diseases was established in 2013 under the National Clinical Strategy and Programmes Division of the HSE. The programme is a joint initiative between the HSE and the Royal College of Physicians of Ireland. This clinical programme is the channel for assisting with the advancement of a number of
other recommendations in the plan, such as those on National Centres of Expertise for rare diseases; and in this vein, the programme designed a framework for the designation of Irish Centres of Expertise in cooperation with the HSE Acute Hospitals Division in line with the recommendations stipulated in the national plan.

The Rare Diseases National Plan also recommended that 'the National Clinical Programme for Rare Diseases through a National Office for Rare Diseases develop the clinical and organisational governance framework that would underpin care pathways and access to treatment for rare disease patients, particularly in the context of transition from paediatric to adult care'.

As such the National Clinical Programme has developed a draft transition model of care which is currently in the public consultation stage.

The National Clinical Programme for Rare Diseases and the Department of Health encouraged designated centres of expertise to participate in European Reference Networks (ERNs) for Rare Diseases in line with the national plan. ERNs are European networks connecting health care providers and centres of expertise so that international expertise on specific rare diseases may be pooled together for the benefit of patients. Any centre applying for membership of an ERN must have strategies in place to ensure that care is patient-centred; that patients' rights and preferences are respected; and must show a research component to their work.

Hence, the recommendations in the national plan that related to empowering and protecting patients and carers, and research on rare diseases will be fulfilled in part through this process. It is expected that ERNs will have a major structuring effect by linking thematic expert centres across the EU.

With the encouragement of the National Clinical Programme for Rare Diseases and the Department of Health, five centres of expertise were designated in June 2016 during the first round of calls from the European Commission for participation in European Reference Networks. Three of these designated centres applied for membership of ERNs and two were approved.

In 2014, the Minister for Health announced €850,000 for investment into charity-led research priorities, which particularly benefits rare disease research. Five charities were to provide matching funding bringing the total investment to €1.9 million. They were to share €850,000 in State funding to take part in international research into rare medical conditions. In 2016, the next cohort of projects was funded by the State with €1.686M, matched by charity funding of €1.224M. The total funding of €2.91M is shared between 11 charities. Six of the 15 projects with a total value of €1.1M address rare diseases. The next round of this joint funding initiative will open in autumn 2017. The EU commission now intends to explore the possibility to further strengthen the collaboration between Member States and the Commission in the area of research on rare diseases.
The establishment of a National Rare Disease Office (NRDO) featured prominently in the recommendations of the Rare Disease plan. The national office has now been established by the HSE. Its work is currently led by the NCPRD Clinical Lead, Prof Eileen Treacy, supported by an Information Scientist, part-time Administrative Officer, part-time Genetic Counsellor and by a 0.2FTE Consultant Geneticist.

It is, among other functions, providing up-to-date information regarding new treatment and management options, including clinical trials. In addition, the post of Information Scientist for the office is being funded jointly by the HSE and the EU Commission.

The NRDO has already performed a preliminary situation analysis of the existing rare disease registries and is in the process of assigning these known registries on our national Orphanet site, which is the international rare disease reference and information portal funded by the EU. The EU Commission has started the development of a European Platform on Rare Diseases Registration. Its principal goal is to enable sharing and use of rare diseases’ patient data across Europe, among the multitude of existing patient registries, within and across rare diseases. Thus the registration of all registries in Ireland will be encompassed by the work of the EU Commission with the participation of EU member states, including Ireland.

A number of recommendations about access to appropriate drugs and technologies were contained in the plan. One chief recommendation in this regard refers to the HSE developing a Working Group to bring forward appropriate decision criteria for the reimbursement of orphan medicines and technologies; and that the approach should include an assessment system similar to that for cancer therapies established under the National Cancer Control Programme. The HSE Acute Hospitals Division is developing the terms of reference, required membership and reporting relationship for this committee. This Committee will also be supported by the National Clinical Programme for Rare Diseases Clinical Advisory Group.

The national plan for rare diseases recommended that the Health Identifiers Bill and the Health & Patient Safety Bill be published. The former was published in 2013 and enacted in 2014. The Individual Health Identifier part of the project is now being implemented by the HSE. A revised and much expanded General Scheme of a Health Information and Patient Safety Bill was approved by the government in November 2015 and published on the Department's website. It is currently with the Attorney-General's Office for formal drafting and with the Oireachtas Committee on Health for Pre-Legislative Scrutiny.

Two recommendations referred to training in rare diseases for healthcare professionals. The Department of Health has contacted formally the various healthcare representative and professional bodies about implementing these recommendations.
Finally, rare diseases have already been tabled on the agenda for North-South meetings. Therefore, future work to deepen cooperation between both jurisdictions on rare diseases is anticipated.