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2012 REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

STATE OF THE ART OF RARE DISEASE ACTIVITIES IN IRELAND

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More information on the European Union Committee of Experts on Rare Diseases can be found at www.eucerd.eu.

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ACRONYMS

General

CAT - Committee for Advanced Therapies at EMA
CHMP - Committee for Medicinal Products for Human Use at EMA
COMP - Committee on Orphan Medicinal Products at EMA
DG - Directorate General
DG Enterprise - European Commission Directorate General Enterprise and Industry
DG Research - European Commission Directorate General Research
DG Sanco - European Commission Directorate General Health and Consumers
EC - European Commission
ECRD - European Conference on Rare Diseases
EEA - European Economic Area (Iceland, Switzerland, Norway)
EMA - European Medicines Agency
ERN - European reference network
EU - European Union
EUCERD - European Union Committee of Experts on Rare Diseases
EUROCAT - European surveillance of congenital anomalies
EUROPLAN - European Project for Rare Diseases National Plans Development
EURORDIS - European Organisation for Rare Diseases
FDA - US Food and Drug Administration
HLG - High Level Group for Health Services and Medical Care
HTA - Health Technology Assessment
IRDiRC – International Rare Diseases Research Consortium
JA - Joint Action
MA - Market Authorisation
MoH - Ministry of Health
MS - Member State
NBS - New born screening
NCA - National Competent Authorities
NHS - National Health System
PDCO - Paediatric Committee at EMA
RDTF - EC Rare Disease Task Force
WG - Working Group
WHO - World Health Organization

Pilot European Reference Networks

Dyscerne - European network of centres of expertise for dysmorphology
ECORN-CF - European centres of reference network for cystic fibrosis
Paediatric Hodgkin Lymphoma Network - Europe-wide organisation of quality controlled treatment
NEUROPED - European network of reference for rare paediatric neurological diseases
EUROHISTIONET - A reference network for Langerhans cell histiocytosis and associated syndrome in EU)
TAG - Together Against Genodermatoses – improving healthcare and social support for patients and families affected by severe genodermatoses
PAAIR - Patients' Association and Alpha-1 International Registry Network
EPNET - European Porphyria Network - providing better healthcare for patients and their families
EN-RBD -European Network of Rare Bleeding Disorders
CARE-NMD -Dissemination and Implementation of the Standards of Care for Duchenne Muscular Dystrophy in Europe project
ENERCA - European network for rare and congenital anaemia – Stage 3

GENERAL INTRODUCTION TO THE REPORT ON THE STATE OF THE ART OF RARE DISEASE ACTIVITIES IN EUROPE OF THE EUROPEAN UNION COMMITTEE OF EXPERTS ON RARE DISEASES

The 2012 Report on the State of the Art of Rare Disease Activities in Europe was produced by the Scientific Secretariat of the European Union Committee of Experts on Rare Diseases (EUCERD), through the EUCERD Joint Action: Working for Rare Diseases (N° 2011 22 01), which covers a three year period (March 2012 – February 2015).

The report aims to provide an informative and descriptive overview of rare disease activities at European Union (EU) and Member State (MS) level in the field of rare diseases and orphan medicinal products up to the end of 2011. A range of stakeholders in each Member State/country have been consulted during the elaboration of the report, which has been validated as an accurate representation of activities at national level, to the best of their knowledge, by the Member State/country representatives of the European Union Committee of Experts on Rare Diseases. The reader, however, should bear in mind that the information provided is not exhaustive and is not an official position of either the European Commission, its Agencies or national health authorities.

The report is split into five parts:

Part I: Overview of rare disease activities in Europe
Part II: Key developments in the field of rare diseases in 2011
Part III: European Commission activities in the field of rare diseases
Part IV: European Medicines Agency activities and other European activities in the field of rare diseases
Part V: Activities in EU Member States and other European countries in the field of rare diseases

Each part contains a description of the methodology, sources and validation process of the entire report, and concludes with a selected bibliography and list of persons having contributed to the report.

The present document contains the information from Parts II and V of the report concerning Ireland. A list of contributors to the report and selected sources are in annex of this document. For more information about the elaboration and validation procedure for the report, please refer to the general introduction of the main report[1].

[1] http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf

RARE DISEASE ACTIVITIES IN IRELAND

Definition of a rare disease

Stakeholders in Ireland accept the European Regulation on Orphan Medicinal Products definition of a prevalence of no more than 5 in 10'000 individuals.

National plan/strategy for rare diseases and related actions

On 20 January 2011 the Genetic and Rare Disorders Organisation, Irish Platform for Patients' Organisations, Science and Industry IPPOSI and Medical Research Charities Group, MRCG in collaboration with EURORDIS organised a National Conference on Rare Diseases[2] in the scope of the Europlan project (see section "National rare disease events"). The conference welcomed over 160 participants from all stakeholder groups. The Conference was "an important milestone" in the development of a national health strategy for rare disease patients.

There is currently no national plan/strategy for rare diseases in Ireland, but the first steps have been taken to elaborate a plan. A National Steering Group of stakeholders has been established (Spring 2011) under the aegis of the Department of Health and Children to work on the development of a five-year national plan, starting with a mapping exercise and focusing on the structure, governance and monitoring of a national strategy. The Minister for Health appointed four patient representatives from GRDO, IPPOSI and MRCG to the Steering Group: the Steering Group held their first meeting in April 2011 and meets every 1-2 months with the aim of completing the first plan in the second half of 2012. A national consultation on the national plan and second national conference on rare diseases are planned for 2012.

Centres of expertise

The Health Service Executive (HSE) does recognise that particular centres have particular expertise, and would give specific funds to support those specialist services. The HSE is responsible for these services and supports centres of expertise and laboratories, including 8 cancer centres, the National Centre for Medical Genetics which provides a service for patients (both adults and children) affected by or at risk of a genetic disorder, and the National Centre for Inherited Metabolic Disorders, a tertiary care referral centre for the investigation and treatment of patients suspected of having a metabolic genetic diseases, linked to the newborn screening programme.

A policy concerning centres of expertise is under development as part of the national plan for rare diseases.

Pilot European Reference Networks

Ireland participates, or has participated, in the following European Reference Networks for rare diseases: Dyscerne, EPNET, EPI, Care-NMD, EN-RBD and the Paediatric Hodgkin Lymphoma Network.

Registries

There are 9 patient registries for rare diseases registered with Orphanet: four of these contribute to the EUROCAT registry. The Medical Research Charities Group (MRCG) created a Steering Group in 2008 involving the MRCG, Health Services Executive (HSE), Health Research Board (HRB) and the Health, Information and Quality Authority (HIQA) to oversee research into the area of patient registries in Ireland. The aim was to identify existing patient registries in Ireland, to describe these in detail (functions, methodologies, standards, funding mechanisms) and also to identify best practice and guidelines for quality standards in this area. The research was presented at an IPPOSI/MRCG run event in October 2011. The outcome report from that event entitled "Towards a National Strategy for Patient Registries in Ireland, considerations for Government[3]" was launched in 2011. Part of a national strategy on Patient Registries in Ireland is the mainstreaming of the role and work of registries into existing and forthcoming policy. The immediate priority is the inclusion of a stronger focus on Patient Registries in: the programme of work of the Quality and Clinical Care Directorate of the HSE, including the development of clinical standards in specific areas of policy and the appointment of clinical leads in particular areas of policy; the work of the Health Information Quality Authority; and the Health Information

[2] http://www.europlan.ie/ and http://download.EURORDIS.org/europlan/2_EUROPLAN_Guidance_Documents_for_the_National_Conference/final_report_ireland.pdf

[3] http://ipposi.ie/index.php/information-centre/patient-registries/179-towards-a-national-strategy-for-patients-registries-report

Bill. The Health Information Bill is expected to be published in 2012 and will address ethical and legal issues concerning data collection and sharing patient data.

Ireland also contributes to other European registries, such as EUROCARE CF and EUROCAT.

Neonatal screening policy

Neonatal screening is in place for galactosaemia, hypothyroidism, phenylketonuria, homocystinuria and maple syrup urine disease. Neonatal screening for cystic fibrosis started as of 1 July 2011[4]. New governance arrangements are being developed for screening.

Genetic testing

Genetic testing in the Republic of Ireland is available through the National Centre for Medical Genetics (NCMG)[5], Our Lady's Children's Hospital, Crumlin, which processes approximately 13,000 cytogenetic and molecular genetic tests annually. The cytogenetic and molecular genetics laboratories are externally accredited by CPA (UK). The National Centre for Medical Genetics is funded via Our Lady's Children's Hospital, Crumlin, which in turn is funded by the Irish Health Service Executive. The National Centre for Medical Genetics is publicly funded via the Irish Health Service Executive. When a genetic test is not available from a laboratory in Ireland, and is clinically indicated, DNA samples are sent to specialised laboratories abroad.

Diagnostic tests are registered as available in Ireland for 19 genes and an estimated 21 diseases in the Orphanet database[6].

The Disability Act Part IV, passed by the Oireachtas and signed into law in 2005 states that genetic testing shall not be carried out unless the consent of the person has been obtained. In addition, genetic tests cannot be used in relation to employment, insurance, pensions or mortgages.

National alliances of patient organisations and patient representation

The Genetic and Rare Disorders Organisation (GRDO) is a non-governmental organisation created in 1988 which acts as an umbrella group for rare disease patient organisations. GRDO was initially founded in 1988 with a view to lobbying for the establishment a National Centre for Medical Genetics. In 1992 this Centre was established by Government. Until October 2011, the organisation was run by volunteers and has since 1988 acted as an advocate for the voluntary sector concerned with genetics. This has been achieved by creating awareness and providing information on genetic disorders to policy makers and health officials in order to achieve a high quality of services for those directly affected by genetic conditions and their families. GRDO also acts as a watchdog in relation to legislation concerning disability to ensure that the rights of people with genetic conditions are protected: the organisation was involved in the consultation process for the Disability Act, 2005 resulting in the inclusion in the Act of provisions regarding genetic tests. Since October 2011, a part-time employee has been hired by GRDO to facilitate the development of the organisation.

In 2011, GRDO, together with the Medical Research Charities Group (MRCG) and the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) and other patient organisations grouped together to form the Rare Diseases Towards 2013 Task Force which will support this National Steering Group charged with the development of a national plan for rare diseases and provide input from the appropriate stakeholders. At the end of 2011 GRDO launched a survey to gather information relating to patient support and advocacy organisations operating in Ireland for people with rare conditions. This information will be used to assist the Taskforce to engage with the Rare Diseases Steering Committee of the Department of Health.

The Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) has a special interest in the rare disease area given that one of its strategic objectives is to address together with key stakeholders (patients' organisations, scientists and industry (and where possible with State Agencies) policy, legislation and regulation around the development of new medicines, products, devices and diagnostics for unmet medical needs. As a non-lobbying organisation, a unique partnership of patient groups/medical charities, science and industry, IPPOSI works to smooth the path in Ireland for new medicines and therapies to move from basic science in laboratories to the patients who need them. This is achieved through expertise, dialogue, consensus building, networking etc. Since its establishment in 2001 the organisation has been involved in a number of conferences relating directly and indirectly to the rare disease area and to therapy development for unmet medical need including Orphan Medicinal Products Regulation of the EU; the Commercialisation of Health Research, the EU Clinical Trials Directive, Clinical Research Infrastructure in Ireland, Access to Medicines and

[4] http://www.hse.ie/eng/services/healthpromotion/newbornscreening/

[5] http://www.genetics.ie/

[6] Information extracted from the Orphanet database (September 2011).

New Medical Technologies in the Era of Health Technology Assessments in Ireland, Patient Registries in Ireland etc. IPPOSI have a place on the Ministry for Health Steering Group developing a strategy for Rare Diseases in Ireland.

Sources of information on rare diseases and national help lines

Orphanet activities in Ireland

Since 2004, there is a dedicated Orphanet team for Ireland and the UK, hosted by the University of Manchester in the UK. This team is in charge of collecting data on rare disease related services (specialised clinics, medical laboratories, ongoing research, registries, clinical trials and patient organisations) in Ireland and the UK for entry into the Orphanet database. There is also an Orphanet Ireland national website[7]. Under the establishment of the National Steering Group, it is hoped that a plan can be developed to run an Orphanet team for Ireland in an accredited centre in this country.

Official information centre for rare diseases

There is no official information centre for rare diseases in Ireland other than Orphanet. However, GRDO operates as a conduit to information on rare diseases and it is hoped that the National Plan for Rare Diseases currently in development will prioritise the establishment of a national information centre for rare diseases.

Help line

There is currently no help line dedicated to rare diseases in general, but some disease specific help lines exist and are funded through public/private partnerships.

Other information on rare diseases

Public information about rare diseases is also provided by patient organisations and GRDO. The MRCG supports patient groups and charitable organisations in securing research funding for rare diseases. IPPOSI provides web-based information and policy support to patient groups. Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) and MRCG are funded partly by the government and membership fees.

In 2008 a report funded by Ireland's Health Research Board discovered an urgent need for information and support resources for both patients and medical professionals encountering rare disease patients in their practice. The report entitled *An investigation into the social support needs of families who experience rare disorders on the island of Ireland*, was published by Rehab Care, a unit of the independent non-profit organisation Rehab Group[8]. Amongst the findings, were that 73% of general physicians admit to difficulties in providing patients and families with appropriate information; some 60% of physicians access rare disease information via the internet; and patients need a reliable resource that does not present a worst-case scenario leading to additional stress and worry. The report recommends developing a centre of excellence in Ireland dedicated to rare diseases that could support health professionals and also provide materials suitable for patients and their families. The authors recommend that Orphanet, as a freely-accessible information resource for professionals and patients, receive a high profile in Ireland, along with UK charity Contact a Family[9].

Good practice guidelines

Clinical guidelines exist for certain diseases.

Training and education initiatives

In 2011 IPPOSI and the School of Medicine at University College Dublin launched a Rare Disease Module for 3rd year medical students. IPPOSI/UCD planned the first module of its kind in Ireland to focus exclusively on rare diseases and the impact on patients. The lecturers on this module are scientists, clinicians and patients describing their own condition to students. The plan is to role this out to other medical schools in Ireland and Europe to bring patients and their patient organisations into the classroom.

National rare disease events in 2011

IPPOSI, the Irish Platform for Patients' Organisations, Science and Industry holds 2-3 conferences annually to tackle various questions in the field of rare diseases and orphan medicinal products.

[7] http://www.orpha.net/national/IE-EN/index/homepage/

[8] http://www.rehab.ie/index.aspx

[9] http://www.cafamily.org.uk/index.php?section=861

On 20 January 2011, the Irish Europlan conference in Dublin took place. The organising committee included members of GRDO (The Genetic and Rare Disease Organisation), IPPOSI, the Irish Platform for Patient Organisations, Science and Industry and the MRCG, the Medical Research Charities Group, Fighting Blindness, Muscular Dystrophy Ireland, EURORDIS, Genzyme Ireland, the National Centre for Inherited Metabolic Disorders, the National Centre for Medical Genetics and the University College of Dublin. The Europlan conference provided a wide range of views from the academic, clinical, private and patient organisation sectors under the topics "Centres of Expertise", "Orphan Drugs and Access to Treatment", "Research" and "Patient Empowerment and Support".

GRDO hosted an information/patient-focused discussion event on Monday 28 February to mark International Rare Disease Day 2011. Those present included people with rare conditions, patient advocates, scientists and clinicians. Topics address included Genetic Testing and Genetic Counselling, Orphan Drugs, Pre-implantation and Genetic Diagnosis.

Hosted rare disease events in 2011

Amongst the events hosted by Ireland in the field of rare diseases and announced by OrphaNews Europe where: The 40th ESPC Symposium on Clinical Pharmacy, which included a Workshop on Cross-Border Healthcare and Rare Diseases (19-21 October 2011, Dublin).

Research activities and E-Rare partnership

National research activities

The Medical Research Charities Group (MRCG) was formed in 1998 to inform and support charities in Ireland in the development of their medical research. As an alliance promoting medical research, the MRCG works to raise the profile of medical research, increase funding, and ultimately alleviate suffering and mortality caused by illness. Since 2006 the MRCG charities have been co-funding research projects with the Health Research Board (HRB). This is made possible by an allocation to the HRB from the Department of Health and Children. While the scheme does not focus solely on rare diseases a number of research projects in the area have been funded. Since the Scheme was put into action in 2006, 44 projects (covering rare and non rare conditions/diseases) have been supported. In this joint funding scheme the Department of Health and Children provides an ongoing annual allocation of €1 million to the HRB which is matched by the research charities. Total investment for the three years 2006, 2007, 2008 was €6 million of which €3 million was provided by the Department of Health.

In addition to the joint funding scheme activities, the MRCG also has a working group on rare diseases and has prepared a policy paper on rare diseases entitled "It's not rare to have a rare disease"[10].

Participation in European research projects

Ireland contributes, or has contributed, in European rare disease research projects including: AUTOROME, EPOKS, EURAPS, EUROPEAN LEUKEMIA NET, EVI-GENORET, GENESKIN, MANASP, MOLDIAG-PACA, NEUROPRION and NOVSEC-TB.

E-Rare

Ireland is not currently a partner of the E-Rare project.

IRDiRC

Irish funding agencies are not currently committed members of the IRDiRC.

Orphan medicinal products

Orphan medicinal product committee

This will be addressed as part of the work of the National Rare Disease Steering Group at the Department of Health. Protocols for access to orphan medicinal products are under development.

Orphan medicinal product incentives

This will be covered in the National Rare Disease Plan.

[10] IPPOSI Information Document on Rare Diseases – 19 February 2009
http://www.ipposi.ie/index.php/information-centre/rare-diseases

Orphan medicinal product market availability situation
This will be covered in the National Rare Disease Plan.

Orphan medicinal product pricing policy
This will be covered in the National Rare Disease Plan.

Orphan medicinal product reimbursement policy
This will be covered in the National Rare Disease Plan. The reimbursement of medicines in general is provided for through a number of "community drug schemes" and "National High Tech Schemes".

Other initiatives to improve access to orphan medicinal products
This will be covered in the National Rare Disease Plan. There is no system at present which deals with pricing and reimbursement of orphan medicinal products. The process is the same for all new therapies and treatments in Ireland, all of which undergo a rapid HTA and may then undergo a full HTA. There is no special criteria for orphan medicinal products.

No formal derogation from these general reimbursement schemes exists but individual hospitals may decide to supply a patient with an expensive orphan medicinal product neither reimbursed under the community drugs schemes nor accessible via other schemes. Companies sometimes provide orphan medicinal products to patients free of charge on a compassionate use basis[11].

Orphan devices
This will be covered in the National Rare Disease Plan.

Specialised social services
Some non-rare disease specific social services exist in Ireland, such as those provided by the Centre for Independent Living and Personal Assistants Scheme. Other support services and respite care are provided by specific rare disease patient organisations.

DEVELOPMENT OF RARE DISEASE ACTIVITIES IN 2011 IN IRELAND

National plan/strategy for rare diseases and related actions
On 20 January 2011 the Genetic and Rare Disorders Organisation, Irish Platform for Patients' Organisations, Science and Industry IPPOSI and Medical Research Charities Group, MRCG in collaboration with EURORDIS organised a National Conference on Rare Diseases[12] in the scope of the Europlan project (see section "National rare disease events"). The conference welcomed over 160 participants from all stakeholder groups. The Conference was "an important milestone" in the development of a national health strategy for rare disease patients.

There is currently no national plan/strategy for rare diseases in Ireland, but the first steps have been taken to elaborate a plan. A National Steering Group of stakeholders has been established (Spring 2011) under the aegis of the Department of Health and Children to work on the development of a five-year national plan, starting with a mapping exercise and focusing on the structure, governance and monitoring of a national strategy. The Minister for Health appointed four patient representatives from GRDO, IPPOSI and MRCG to the Steering Group: the Steering Group held their first meeting in April 2011 and meets every 1-2 months with the

[11] *EMINET: Initial investigation to assess the feasibility of a coordinated system to access orphan medicines,* C. Habl, F. Bachner, May 2011 (pp52-53).
[12] http://www.europlan.ie/ and http://download.EURORDIS.org/europlan/2_EUROPLAN_Guidance_Documents_for_the_National_Conference/final_report_ireland.pdf

aim of completing the first plan in the second half of 2012. A national consultation on the national plan and second national conference on rare diseases are planned for 2012.

Neonatal screening policy

Neonatal screening for cystic fibrosis started as of 1 July 2011[13]. New governance arrangements are being developed for screening.

Registries

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National alliances of patient organisations and patient representation

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[13] http://www.hse.ie/eng/services/healthpromotion/newbornscreening/

[14] http://ipposi.ie/index.php/information-centre/patient-registries/179-towards-a-national-strategy-for-patients-registries-report

Research activities and E-Rare partnership

IRDiRC

Irish funding agencies are not currently committed members of the IRDiRC.

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  IPPOSI Information Document on Rare Diseases (19/02/09)
- http://www.ipposi.ie/index.php/information-centre/rare-diseases
  Rehab Care Report: An investigation into the social support needs of families who experience rare disorders on the island of Ireland
- http://www.rehab.ie/about/PDFS/July2008/RehabCare_RD_Report.pdf

[15] The contributors and validators of the report have contributed information which is accurate to the best of their knowledge. However, readers should take note that the contents of this report are illustrative and not exhaustive.
[16] All websites and documents were last accessed in May 2012. A more detailed list of sources is available in the full report: http://www.eucerd.eu/upload/file/Reports/2012ReportStateofArtRDActivities.pdf