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## **EUROPLAN National Conferences**

### **CONFERENCE FINAL REPORT - IRELAND**

#### **I. General information**

On July 21, 2009 the Irish Minister for Health and Children Ms. Mary Harney TD and the Deputy Chief Medical Officer responsible for Public Health Dr. John Devlin met with representatives from the Genetic and Rare Disorders Organisation (GRDO), which is an umbrella group of volunteers representing the voice of patients living with Rare Diseases in Ireland, to discuss the Irish government's involvement in the Europlan Initiative.

The Minister and Dr. Devlin agreed to support the initiative and saw it as an important step towards the development of a National Strategy to manage the care of over 150,000 Irish people living with a rare disease.

Since the first International Rare Disease Day on February 29, 2008, GRDO has worked closely with partner organisations the Medical Research Charities Group (MRCG) and the Irish Platform for Patients' Organisations Science and Industry (IPPOSI) on maximising opportunities for advocacy and publicity on the day through the organisation of conferences, briefings and press events.

As GRDO consists of a group of volunteers, the support of professional secretariats such as the MRCG and IPPOSI has not only strengthened the patients' voice but has also broadened the stake holding group to include scientists, medical professionals and industry representatives concerned with the various issues relating to rare diseases. Therefore GRDO felt very strongly that the Europlan Conference like the Rare Disease Day should be coordinated by the three groups.

**GRDO:** The Genetic and Rare Disorders Organisation (GRDO) is a non governmental organisation acting as a national alliance for voluntary groups representing the views and concerns of people affected by or at risk of developing genetic and rare disorders.

The mission of the GRDO is to provide a strong voice for voluntary groups representing people with or at risk of developing genetic or other rare disorders in order to achieve better support and services. The GRDO acts as a watchdog in relation to legislation concerning disability to ensure that the rights of its members are protected.

The GRDO was incorporated in 1988 and its first achievement was to successfully advocate for a dedicated national centre for medical genetics (NMCG) which was established at Our Lady's Children's Hospital in Dublin in 1994.

GRDO receives no state funding and is run on a voluntary basis.

**MRCG:** The Medical Research Charities Group (MRCG) is an umbrella group of medical research and patient support charities, which originally formed to represent the joint interests of charities specialising in restoring health through medical research, diagnosis and treatment and, where possible, the prevention of disease. A core belief of the group is that today's health research is tomorrow's healthcare.

The MRCG's mission is to generate a dynamic medical research environment in Ireland. This will be achieved by partnership with external organisations; advocacy to enable change; a forum to promote education and integration; and the provision of direct services to members.

**IPPOSI:** The Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) is a unique partnership of Patient Groups/Charities, Science and Industry on the island of Ireland. As a patient led partnership, the platform provides a structured way of facilitating interaction between the three key membership groups (and where possible with State Agencies) on policy, legislation and regulation around the development of new medicines, products, devices and diagnostics for unmet medical needs in Ireland.

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The IPPOSI vision is one where state of the art innovations in health care are available at the earliest stages to patients in Ireland. They do this through expertise, dialoguing, consensus building and networking.

IPPOSI is not a lobbying organisation but via expertise, dialogue, consensus and networking they work to smooth the path in Ireland for new medicines and therapies to move from basic science in laboratories to the patients who need them.

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| <b>Country</b>   | Ireland  |
| <b>Date &amp; place of the National Conference</b>         | January 20, 2011, Dublin   |
| <b>Website</b>   | www.europlan.ie  |
| <b>Organisers</b>  | GRDO / MRCG / IPPOSI   |
| <b>Members of the Steering Committee</b>                   | <p><b>Margaret Webb</b> – Chair</p> <p><b>Avril Daly</b> – Advisor</p> <p><b>Anna Moran</b> – Fighting Blindness</p> <p><b>Eibhlin Mulroe</b> – IPPOSI</p> <p><b>Karen Pickering</b> – MDI</p> <p><b>Fred Doherty</b> – Genzyme Ireland</p> <p><b>Prof. Eileen Treacy</b> – Childrens University &amp; Mater Misericordiae University Hospitals Dublin</p> <p><b>Prof. David Barton</b> – Centre for Medical Genetics</p> <p><b>Dr. Seamas Donnelly</b> – University College Dublin</p>  |
| <b>Names and list of Workshops</b>                         | <p>Centres of Expertise</p> <p>Orphan Drugs</p> <p>Research</p> <p>Patient Empowerment</p>   |
| <b>Chairs and Rapporteurs of Workshops</b>                 | <p><b>Centres of Expertise</b></p> <ul style="list-style-type: none"> <li>▪ Chair – Prof. Eileen Treacy</li> <li>▪ Rapporteur – Prof. David Barton</li> </ul> <p><b>Orphan Drugs</b></p> <ul style="list-style-type: none"> <li>▪ Chair – Eibhlin Mulroe</li> <li>▪ Rapporteur – Vincent Barton</li> </ul> <p><b>Research</b></p> <ul style="list-style-type: none"> <li>▪ Chair – John McCormack</li> <li>▪ Rapporteur – Alica May</li> </ul> <p><b>Patient Empowerment</b></p> <ul style="list-style-type: none"> <li>▪ Chair Avril Daly</li> <li>▪ Rapporteur – Patricia Towey</li> </ul> |
| <b>Attachments (programme, list of participants, etc.)</b> | <p>1. Conference Programme</p> <p>2. List of Attendees</p>   |

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## II. Main Report

On April 14 2010 the first meeting of the Europlan Organising committee was held in Dublin. At the meeting Ms. Margaret Webb a patient representative and former CEO of Debra Ireland was appointed chair. Ms. Avril Daly, a patient representative and CEO of Fighting Blindness / Chair of the GRDO acted as Europlan advisor.

The organisation committee consisted of four patient representatives including Mrs. Karen Pickering of Muscular Dystrophy Ireland, Mrs. Anna Moran of Fighting Blindness as well as Ms. Webb and Ms. Daly. The Medical and Scientific communities were represented by Prof. Eileen Treacy from the Children's University and Mater Misericordiae University Hospitals Dublin, Prof. David Barton of the National Centre for Medical Genetics at Crumlin Hospital and Dr. Seamas Donnelly of University College Dublin. Industry was represented by Mr. Fred Doherty of Genzyme Ireland.

The Health Ministry assured the group of their support and provided Farmleigh House, in the Phoenix Park in Dublin, which is one of the most prestigious state buildings in the country as the venue for the conference which was due to take place on Friday December 3, 2010. Due to extreme weather conditions resulting in the closure of the airports, roads and indeed the venue itself the Europlan conference was postponed at short notice to take place on Thursday, January 20, 2011. Fortunately the conference programme remained unchanged by the postponement.

GRDO have been advocating for better care pathways and services for people affected by Rare Diseases since its inception in 1988. The GRDO advocated for and were successful in bringing about the establishment of the National Centre for Medical Genetics (NMCG) in 1994 to address an unmet need in the health service. Until recently Ireland has been a genetic isolate and evidence has shown a higher instance of rare disease here than in other member states, however the concerns of people living with rare diseases had not been prioritised and were viewed as a disability issue and not as a public health priority.

In 1997 with the emergence of organisations like Eurordis, patients began to become more empowered and educated on the myriad of issues associated with RDs. Members of The GRDO and other voluntary and charitable groups established the Medical Research Charities Group in 1998 to give a unified voice to organisations seeking funding to develop patient led medical research.

In 2001 members of the same group along with scientific, medical and industry representatives established the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) as a platform to build consensus on issues relating to the development and access to therapies for unmet medical needs in Ireland. So for ten years now these three groups have worked for the needs of stakeholders in the field of RDs, through open discussion, task forces and conferences, gaining consensus that have in turn educated and empowered patients.

On February 25<sup>th</sup> 2010 a conference organised by the GRDO, MRCG and IPPOSI was held in Dublin to mark international rare disease day. Minister Aine Brady, a junior minister for health at the Department of Health and Children reinforced the Irish Government's commitment to the development of a national strategy to tackle the issue of rare diseases despite the complexities and the substantial challenges they present to health care systems.

Minister Brady acknowledged "the need for integrated planning to give effect to better diagnosis and treatment for these conditions". She also confirmed that many of the elements of a national plan for rare diseases have commenced such as the Health Information Bill which is expected in 2011 which would be a requirement to develop appropriate information systems and registries for rare diseases. Equally, the ongoing work within the Health Service Executive (HSE) on restructuring will contribute to the organisation of reference centres and networks for the management of rare diseases. New policies on screening are being developed and an example is the ongoing work on screening for Cystic Fibrosis which has been underway since 2009.

The Irish government has recognised the value of the European Project for Rare Diseases National Plans Development (EUROPLAN) and has appointed Dr. John Devlin, Deputy Chief

Medical Officer to the recently established Expert Group on Rare Diseases, with Mr. Charlie Hardy acting as alternate.

Monthly meetings were held by the organising committee and separate meetings and discussions were held to feed this information into the office of the Chief Medical Officer at the Department of Health and Children (DoH&C) the policy arm of the Health Ministry. The process also allowed for engagement with representatives from the Health Service Executive (HSE) who are the service delivery arm of the Ministry. A meeting was held with the Director of Clinical Care Directorates, HSE, for Ireland, Dr. Barry White around the establishment of a clinically led centre for Rare Diseases. This discussion is on going and the indications are positive.

A general election will be held on Friday February 25. The GRDO, MRCG and IPPOSI are hopeful that any incoming government will continue to work on the establishment of a national plan and are happy to report that the three main parties have included the issue of Rare Disease in their election manifestos.

As this was the first interactive event to be held that would cover diverse issues relating to RDs, it was felt by the organising committee that the conference should commence with a 'scene setting' plenary session. This would allow for the Europlan initiative to be fully explained before discussion commenced.

Ms. Margaret Webb, Chair of the committee opened the event with an overview of the issues and the importance of the Europlan initiative. The European context was highlighted by Mr. Yann Le Cam, CEO of Eurordis. Dr. John Devlin gave the Irish Government's perspective and Dr. Alvin Shih, Head of Genetic & Orphan Drugs for Pfizer gave the industry perspective on the issue.

To reinforce the many issues faced by patients living with rare diseases, a pre-recorded patient testimonial was played before the plenary session and each workshop.

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As the meeting was only feasible as a one day event, the organising committee decided that the main issues that are of particular concern currently to stakeholders should be prioritised and addressed in a workshop format. These were:

- 1 – Centres of Expertise
- 2 – Orphan Drugs & Access to Treatments
- 3 – Research for Rare Diseases
- 4 – Patient Empowerment & Support

Issues relating to Governance, Codification and Sustainability were all captured in the four main workshops.

As well as a chair and co-chair, each workshop was addressed by relevant speakers who presented delegates with the issues relating to each subject from the perspective of the patient, the policy makers, and the medical/scientific professionals. While this did take time away from the general discussion it was a vital aspect of the meeting as many delegates were unfamiliar with all the aspects of the various subjects. This expert involvement allowed for a more informed and stimulating discussion.

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## **Main Themes**

### **Theme 1 - Methodology and Governance of a National Plan / Strategy (NP)**

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Whilst there was not a specific workshop focusing on methodology and governance of a national strategy, it was a recurring theme of the day in both plenary sessions and the four workshops.

It was clear to delegates that structures already exist in Ireland in the areas of Centres of Expertise, Patient Registries, Research, Patient Empowerment and Orphan Drugs.

For example, there are good models of disease specific centres of expertise that could be drawn on when discussing the development of a particular Centre of Expertise for RDs. It was agreed a multidisciplinary coordinated approach needs to be applied to the development of a Centre of Expertise in Ireland. There were some delegates who believed that a COE of RDs may not necessarily be one centre under one roof. As Ireland is a small country some felt that better communication tools and methodologies could allow existing centres to collaborate more effectively.

There were also inputs from patient representatives on a perceived reluctance on the part of some medical professionals not to collaborate and consult. These issues were deemed a priority and need to be addressed urgently as this is seen to prolong the diagnostic journey of the patient and reduce effective access to treatment.

Indeed the diagnostic journey and the lack of information were the main concerns of the patients and medical representatives on the day. This was articulated by most speakers that the urgent need for access to information, a centre of expertise on RD and the development a care pathway were of paramount importance when developing a strategy for rare diseases.

The Irish Health ministry has committed to establishing a group of stakeholders to work on the development of a national plan. This will start with a mapping exercise and will focus on the structure, governance and monitoring of a national strategy as well as issues of sustainability. The group will be convened in spring 2011.

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## **Theme 2 - Definition, codification and inventorying of RD**

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Whilst the subject of Definition Codification and Inventorying of Rare Disease was not focused on specifically at the Irish conference it was discussed in workshops and in plenary.

- **Definition of RD**

In Ireland the definition of rare diseases reflects the European Commission recommendation that an RD is classified as affecting no more than 5 in 10,000 people.

- **Classification and traceability of RDs in the national health system**

At the Centres of Expertise workshop and at the Patient Empowerment workshop the frustration of patients who, despite having a diagnosis, have not had their disease categorised was raised. The current system does not allow for the adequate classification or traceability of diseases. Many parents who have young children with a rare disease can not access state benefit through the medical card scheme as their child's condition is not listed.

Ireland must adopt international classifications for rare diseases, incorporate ICD-11 when available and must also learn from abroad regarding the most appropriate registries and databases required which can be used to plan and manage services in Ireland.

- **Inventories, registries and lists**

There are some patient registries in existence in Ireland many of which were established through public and charitable partnerships spearheaded by patient led organisations.

Delegates emphasised the importance of patient registries in patient care providing valuable information on the safety and efficacy of treatments, enhancing knowledge and understanding and facilitating clinical trials.

Their importance in the future planning of more effective service delivery and for health funding accountability was highlighted.

Data collection, legal and ethical issues, financial/funding problems, logistical and political challenges were identified as areas which need to be addressed. Further, issues of ownership,

control and sharing of patient data need consideration due to the reticence, fear and mistrust arising from a possible misuse, theft or loss of patients' health information and needs.

The Health Information Bill is expected to be published in 2011 and it is hoped will address these issues.

- **Information and training**

Again this was a recurring theme at the conference.

Patients see themselves as educators in the field of rare diseases. Many parents noted that as the providers of day to day care of their children they were in many cases more expert in the intricacies of dealing with medical care than professionals.

In the case of a more common rare condition there are patient representative groups providing information to medics, scientists and to the general public through media and events. With ultra rare diseases there were less support groups and charitable organisations and so the provision of information was more difficult.

During the workshops many patients commented on the length of time it had taken them to receive diagnosis. All cited a lack of information as the main cause of this delay followed by a reluctance of healthcare professionals to gain more understanding – an area that could be looked at in education and training of Rare Diseases.

In the case of ultra rare diseases there were examples of patients linking in with groups in Europe and in the United States to gain more access to information and in turn educate the medical professionals and other patients at home.

The support of organisations like Eurordis and information generated through Orphanet were seen as invaluable and resources that should be utilised more by Irish patient groups.

The fact that the rare disease group supporting patients in Ireland, GRDO, is not funded and is run by volunteers was discussed. It was suggested that there should be an effort to fund this organisation so that it can provide the information so desperately needed by patients but also be a repository of information for medical practitioners throughout the health care system.

GRDO have undertaken a programme whereby they will speak to medical students about Rare Conditions. IPPOSI and the MRCG are also engaging in educational programmes for medical professionals and for scientists and policy makers.

Irish Rare disease patients suffering from conditions where multiple systems are affected, are exasperated by constantly explaining to different specialists how the condition affects them. A book on the condition for professionals would help specialists understand the interaction between the various systems and the condition and, with the support and input of patients suffering from the condition, the book could accurately reflect the lived reality. This is hugely beneficial to patients and professionals.

- **How to improve information on available care for RDs in general, for different audiences**

The improvement of information flow is contingent on funding and support. Patient led charities and national support groups are currently providing general information and this will need to be sustained.

Centres of Expertise in specific disease areas provide excellent information but this is of little benefit to patients with ultra rare disorders. This will need to be addressed within the plan.

- **How to improve access to quality information on RDs**

Coordinated IT systems linking with international groups and better utilisation of communications platforms such as the web and new media were all highlighted as the most cost effective way of improving access to information. Accreditation for websites detailing particular conditions was suggested. The continuation of collaboration with Eurordis on Rare Disease Day and the growth of the day itself internationally were also mentioned as an enabler to improve communications around the issue.

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- **How to ensure adequate training of healthcare professionals on RDs**

The system of training for healthcare professionals must be addressed through their governing bodies with the support of all stake holding groups. Patient groups are already taking a lead in this by producing information pamphlets, books and giving lectures to medical students.

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### Theme 3 - Research on RD

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- **Needs and priorities for research in the field of RDs**

The consensus of the group was that basic and translational research needs to be linked and centres of expertise need to be formed. Treatment centres need to become involved in research to improve health service delivery.

The Head of Research Infrastructure & Special Initiatives Unit at the Health Research Board (HRB) Dr. Anne Cody provided a brief overview of their funding portfolio, and addressed why research was so important for improving health outcomes. She also highlighted the importance of establishing patient needs when embarking on research.

The point was made that key stakeholders (including patient groups, health professionals, HSE, policy makers, translational researchers, industry) must be consulted when developing a National Strategy on Research. It was also pointed out that clinical/translational research should be performed at an international level, if possible, to achieve statistical significance, especially for rare/ultra rare disease populations. Dr Cody stated that there has to be a broad range of research into Rare Diseases to underpin clinical decisions. This will open up new avenues for prevention, diagnosis and treatment.

Tony Ward, a patient representing Fighting Blindness a patient led research charity, is affected by a rare retinal disease. He commented that research is an expensive process and that it often takes a long time before patients see the benefits from it; patient led organisations can help to get the ball rolling in this respect by their commitment to funding research.

Early diagnosis is essential in rare diseases but we heard from some patients for whom diagnosis took up to 21 years. This is a good reason to invest in the greater understanding of rare conditions, recognise them when they present and to be aware of the research taking place with orphan conditions.

Industry will choose research sites carefully - if the infrastructure, patients and appropriately qualified and experienced personnel are not available, industry will seek to conduct its trials in

more favourable locations where conditions are right. European legislation exists to encourage the development of treatments for rare disease and has proven to be effective. There also needs to be alignment for funding agencies if research is to go beyond being solely beneficial to trial patients.

The issue of patient registries was also discussed in this workshop. An examination of individual privacy issues vs. public benefit needs to be considered in the development of patient registries for rare diseases. The difficulties of getting registries up and running were highlighted; the barriers with regional ethical committees were mentioned.

- **Fostering interest and participation of national laboratories and researchers, patients and patient organisations in RD research projects**

There is no *dedicated* state funded rare disease programme in Ireland. However, the MRCG/HRB Joint Funding Scheme does accept applications from members of the MRCG funding research in the RD area. The HRB grant schemes are also open to *other* diseases areas – applications are awarded based on excellence not rarity. If successful a project receives 50% of its funding from the HRB and 50% from the charity member.

The HRB/MRCG Joint funding scheme – despite being open to all disease areas – has been successful in developing research in the area of rare diseases. Many small patient groups with no experience of grant calls and proposals have been supported by the MRCG in their endeavours to understand not only the funding mechanisms to research projects but the review process and other issues. This has resulted in projects being undertaken that otherwise would not have received funding. It has also increased interest in rare disease research.

Individual patient groups also foster the development of rare disease research through fundraising advocacy and their international partnerships.

Research Clinicians mentioned that when applying for grants they ‘play down’ the rarity of the condition they are hoping to research and ‘play up’ its relationship to other more common diseases as they are more likely to be funded.

- **Sustainability of research on RD**

A strong, appropriately resourced research infrastructure must be developed as a priority so that it is ready to engage in transnational clinical trials.

Researchers must endeavour to try and link rare disease research to more common disease to optimise research funding potential. E.g. if it can be shown that the research can be applicable to more common forms of disease this is a good argument with which to draw in funds. E.g. what is done in Retinitis Pigmentosa (RP) research is relevant for example to what will be done in Age-related Macular Degeneration (AMD) research.

The discussion called for recognition of the value of the contribution biobanking can make in the field of rare disease, where statistical significance is a real issue. Where genetic conditions are poorly understood, biobanking can play an important role in the research process.

Funding for research into rare diseases, patient information and registries were deemed a priority. A consensus amongst the group was that we must work together; a network of professionals must be established to pool information, both nationally and internationally.

- **EU collaboration on research on RD**

All delegates agreed particularly in the case of very rare conditions. European collaboration and networking is essential when dealing with small patient populations. The involvement of research driven patient organisations in EU funding networks hasn't been leveraged.

E-Rare was mentioned as a possibility to look at as a support infrastructure for researchers in rare diseases to network; Ireland is not currently signed up to this

EU funding mechanisms should be used to support and sustain research networks. The group also thought it was important to point out that the sustainability of research projects in rare diseases will be encouraged by the implementation of Europlan.

Dr Carmel Mullaney, a Specialist in Public Health Medicine at the Department of Public Health, Health Service Executive - South East, spoke about Eurocat.

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Eurocat is a European network of population-based registries for the epidemiologic surveillance of congenital anomalies. It started in 1979 and more than 1.5 million births are surveyed per year in Europe. There are three registries in Ireland linked to Eurocat, and these cover approx 60% of the population.

Dr Mullaney discussed how Eurocat works in terms of data collection and data sources and highlighted how Eurocat contributes to the field of Rare Diseases, for example, it provides information on structural defects and chromosomal abnormalities, and epidemiology and prevalence rates.

She also spoke about why European collaboration is important – to facilitate the pooling of data, to enable comparison of data and to be able to share expertise.

A representative from Orphanet strongly encouraged funders and patient organisations to get more involved. Orphanet is a database of information on rare diseases and orphan drugs for all publics. Its aim is to contribute to the improvement of the diagnosis, care and treatment of patients with rare diseases.

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## **Theme 4 - Standards of care for RDs - Centres of Expertise (CoE)/ European Reference Networks (ERN)**

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**Professor Eileen Treacy:** Consultant Paediatric and Adult Metabolic Physician at the National Centre for Inherited Metabolic Disorders (Children's University and Mater Misericordiae University Hospitals) and Clinical Professor of Inborn Errors of Metabolism (Paediatrics) at Trinity College Dublin. Prof. Treacy provided a background to the theme and expectations for Centres of Expertise.

**Ms Josie Godfrey:** Head of Policy for the National Specialised Commissioning Team in England. Works with the Advisory Group for National Specialised Services. Ms. Godfrey discussed commissioning for rare disorders in the UK

**Professor Andrew Green:** Professor of Medical Genetics at University College Dublin, clinical geneticist and Director of the National Centre for Medical Genetics in Our Lady's Children's Hospital. Prof Andrew Green elaborated on working Irish examples of Centres of Excellence.

- **Identification of national or regional CoE all through the national territory by 2013**

Appropriate Centres of Expertise for Rare Diseases and suitable healthcare pathways must be identified to eliminate delays in the diagnostic journey so that patients and professionals understand where the most appropriate care can be provided. Centres must be multi-disciplinary and should have the ability to manage as well as diagnose rare disorders to include genetic counselling and testing, treatment, access to clinical trials and must link in with European counterparts on all issues including healthcare professional training.

As discussed at the meeting Re: High level Group on Health Services and Medical Care, EU Rare Diseases Task Force, 2006 (now called European Union Committee of Experts on Rare diseases) there should be sufficient capacity and experience, evidence by accreditation and by peer review of having the appropriate clinical expertise to diagnose and safely manage these patients with enough staff in a timely fashion and also to have the scientific experience in the

team and with evidence of outcome publication showing good outcomes. There should be access to multidisciplinary teams of experts and the appropriate psychosocial care. As recommended, the Centre of Expertise would combine research with care. It is important that the Centres would be involved in education of not only their staff but outreach and shared care model and that there was a way to transfer this information.

It is clear that although there is no one centre of expertise for RDs in Ireland, there are many centres of expertise in specific areas e.g. neurology, haematology among others. These are models that could be looked at when formulating a national strategy.

Two centres focused genetic and rare disorders are:

**The National Centre for Medical Genetics (NCMG)** which provides a comprehensive service for all patients and families in the Republic of Ireland affected by or at risk of a genetic disorder. The Centre is based at Our Lady's Children's Hospital, Crumlin, Dublin and comprises three integrated units, a Clinical Genetics service, a Cytogenetics laboratory and a Molecular Genetics laboratory. The Centre provides a service for both children and adults. There is a strong research element to the unit; the University College Dublin Department of Medical Genetics is based in the Centre.

*And*

**The National Centre for Inherited Metabolic Disorders** located at the Children's University Hospital, Temple St., Dublin. It is the tertiary care referral centre for the investigation and treatment of individuals suspected of having a metabolic genetic disease in Ireland and is linked to the Newborn Screening Programme. The National Centre plays a major role in preventing and treating disability for the Irish population. The National Centre screens for conditions such as PKU, Homocystinuria, Maple Syrup Urine Disease, Galactosaemia and provides a programme for the investigation, counseling and treatment for patients (mainly children) with metabolic diseases, detected by newborn screening and family screening.

The Unit investigates suspected cases of metabolic disorders referred from hospitals throughout the country and provides novel treatments for patients with these rare genetic metabolic disorders. A holistic and family centered approach is used by the team, composed of input from medical, nursing, dietetic, psychology, social workers and laboratory staff.

Although Ireland still only performs newborn screening for a limited number of conditions, the current Director of the National Newborn Screening Programme is working with the Department of Health and Children to expand newborn screening and is in consultation with a European Group, the Executive Agency for Health and Consumers of European Commission Task Force in their evaluation of population newborn screening for specific rare disorders in member states of the European Union.

- **Sustainability of CoE**

Centres of Excellences must be supported with mechanisms that ensure long-term funding and stability. This requires ring-fenced budgets protected from the vagaries of local financial issues. While central government funding will always be the main mechanism for the establishment and maintenance of most Centres of Excellence, innovative solutions including co-funding by patient organisations and EU network funding should also be exploited, especially for ultra-rare diseases.

- **Participation in ERN**

It was agreed that there is strong collaboration in this area among the scientific communities and the patient groups with European groups. There is some participation among clinicians in ERNs but this is again not well coordinated and better in some disease areas than others. This will need to be improved as its value is unquestionable and very necessary particularly in the field of Rare Diseases and Ultra Rare Diseases.

Ireland has to seek recognition of its proposed Centres of Excellence as Centres of Expertise in ERNs according to internationally accepted standards in this area (High level Group on Health Services and Medical Care, EU Rare Diseases Task Force, 2006, now called European Union Committee of Experts on Rare diseases).

The recently approved European Parliament Directive on Cross-Border Care makes particular provision for patients with rare disorders and hopefully will improve access to information across European sites particularly for ultra-rare disorders (with a prevalence of less than 1: 100, 000) whereby there may not be adequate expertise and a Centre of Expertise in Ireland by facilitating health experts across Europe in the exchange of best practices and innovation in health technology and eHealth.

- **How to shorten the route to diagnosis**

A central office for Rare Disease information should be established, to act as a national point of reference for enquiries regarding services, diagnostics, information regarding clinical trials, links to established databases such as “Orphanet” and expertise for all rare diseases. This could be established immediately and should not wait for 2013.

Patient and carer contributors emphasised their frustration with the current system including the lack of coordination, lack of continuity of care and the difficulty in accessing services. These real-life experiences highlight the urgent need for coordination of services for Rare Disease patients.

Population screening and advances in therapies for Rare Diseases will emphasise the need for agreed care plans and pathways which provide life-long care.

The EU Charter of Patients’ Rights and the newly-established Directive on Cross Border Care will give Irish patients access to diagnosis anywhere in the EU as a right, funded by the Irish Exchequer. Ireland will therefore be paying for these services one way or another – it is clearly in the best interests of patients for their care to be delivered in Ireland.

Prof Kieran Murphy, president of the Irish Medical Council addressed concerns from patients about a perceived lack of understanding by medical professionals by mentioning the Irish Medical Councils work and the Ethical guidelines “An Introduction to the Guide for Professional Conduct and Ethics for Registered Medical Practitioners” available on their website: This may be of use to patients in these situations.

- **How to offer suitable care and organise adequate healthcare pathways for RD patients**

Care for patients with Rare Diseases, while coordinated from Centres of Excellence, should be delivered as close to the patient as possible.

Centres of Excellence must have expertise and competence in all areas of patient care, including social care; all Centre of Excellence activities must be patient-focused.

- **How to ensure in CoE multidisciplinary approaches and integration between medical and social levels**

Information Technology, in particular a national electronic healthcare record, was seen by patients as a key need: we heard that an average of eight specialities are involved in treating each rare disease patient.

- **How to evaluate CoE**

Performance of Centres of Expertise must be monitored by audit of outcome measures – is the Centre improving patient outcomes and experience? This kind of health services audit and research requires dedicated resources. The current experience is that clinical and support staff are “overwhelmed by service commitments” and there should be a balance between research and care at Centres of Expertise.

#### 4.1. Orphan Drugs (OD)

**Chair:** Ms Eibhlin Mulroe, CEO IPPOSI

**Co-Chair:** Mr. John McCormack, Chair MRCG

- Mr Shaun Flanagan, HSE Corporate Pharmaceutical Unit
- Mr. Yann Le Cam, CEO EURORDIS - Rare Diseases Europe
- Ms Willis Hughes-Wilson, Senior Director, Health Policy Europe, Genzyme

##### ▪ **Future of OD**

Pharmaceutical companies are committed to the Irish economy and future pharmaceutical investment here is key to a competitive knowledge based economy and the enhancement of clinical research in Ireland. A vibrant pharmaceutical sector is also an essential precondition to ensure high levels of public health protection.

There is recognition that OD are costly and that development costs are considerable, irrespective of the number of patients to be treated. Overall national health spend, including pharmaceutical spend is likely to remain 'flat' for some time. There is a need to find long-term sustainable resourcing, irrespective of funding mechanism. The cost of doing nothing is also considerable at individual and societal level.

There are new mechanisms and approaches to drug assessment which can be exploited. Funding for OMPs needs to be ring-fenced, following the example of countries such as France. Current budgetary approaches are not adequate. There is a need for policymakers/funders to accept that at least some OMPs cannot be assessed within existing mechanisms

Industry should interact much earlier in the development and launch cycle with policymakers/funders.

We need to build information sets. Passing the Health Information Act is essential. The need to boost participation in registries was emphasised.

CAVOD (Clinical Added-Value in Orphan Drugs) can be applied to 'unique treatments of specific benefit'. Also the new EU programme on *Social Responsibility in the field of Pharmaceuticals* broadens the policy perspective in relation to ODs and needs to be taken into account by Irish policy makers and funders. Ireland should get involved in the CAVOD process and use it to shape reimbursement mechanisms.

- **Access of RD patients to orphan drugs Pricing and Reimbursement**

The process for assessing orphan drugs must be transparent. Rarity, disease severity, the availability of alternative therapies and the magnitude of new treatment impact all need to inform decision making. It is imperative that a national rare disease plan is supportive of research, the research environment and addresses the issue of reimbursement

Reimbursement systems need to be shaped by strong policy direction and societal guidance. Health Technology Assessment is just one element of the cost/benefit assessment. More transparency as to pricing rationale would be welcomed by funders. The inequality of access to treatment is a persistent problem. This occurs regionally, within Ireland, and also unfortunately across the island of Ireland and EU as a whole.

Information on OD conditions is fragmented and hard to get at. In Ireland the funding mechanism means that data in relation to hospital-based spending are aggregated and unhelpful, while community-administered spend is easily captured.

Inconsistencies in health policy mitigate against fair access to drugs. For example, the drive towards Centres for Excellence is not matched by ring-fenced funding of high-cost medication. As a consequence, the Centres of Excellence may feel 'penalised' financially.

The EU has done its work in setting out frameworks. Ireland now needs to take the issue forward itself in the shape of a National Strategy.

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- **Compassionate use and temporary approval of orphan drugs. Off label use**

Ireland needs to move ahead with its national plan. However, immediate measures are needed to help those who cannot wait until 2013 deadline.

Mrs. Margaret Whelan who is affected by Pompe disease told of her on-going struggle to gain access to an orphan drug that has proven to be an effective treatment prolonging life and independence of those who are currently receiving the treatment. Ireland is one of the few countries where the therapy is not reimbursed. It has been a life saving treatment to some patients.

All parties (patients, carers, industry, funders, policymakers, clinicians) need to be innovative and flexible in their approach.

There was a suggestion that Ireland should look at schemes such as Australia's "Life-Saving Drugs Programme" and other 'compassionate use' mechanisms.

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## Theme 5 - Patient Empowerment and Specialised Services

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- **Involvement of patients and their representatives in decision-making processes in the field of RDs**

Patient Organisations are central to promoting awareness the area of rare disease. They provide an essential support and information service to many rare disease patients nationally and internationally. Their role in fundraising, to provide vital seed funding for research and their engagement in driving forward human clinical trials is essential and, as important factors in the rare disease landscape their contribution must be recognised and their vital work supported.

In Ireland there are many volunteer and patient led as well as professional organisations providing support to and advocating for people living with rare diseases. There are 24 volunteer groups who are members of the support and patient rights group GRDO; there are over 30 patient led charitable and voluntary organisations who are members of the MRCG, the majority of whom have an interest in RD. Also the patient groups represented through GRDO and MRCG are active on the board and committees of IPPOSI.

These patient led charities and voluntary groups range from large organisations such as the Irish Cancer Society to mid sized organisations such as Cystic Fibrosis Ireland, to volunteer groups run by parents of children affected by rare diseases such as the Cystinosis Foundation and Bee for Battens.

In the majority of cases patient groups were founded by those affected by RDs and are funded solely through donations from the public and sponsorships.

MRCG has a mentoring programme where larger organisations provide information and support to smaller groups to enable them to become more educated and therefore more empowered patient advocates.

The need for further support for patients representing those affected by an ultra rare condition was highlighted; there are little supports for these groups individually but supports do exist through umbrella organisations.

Patient led advocacy is very common in Ireland. There is access to politicians and local representatives. Patients are working more often in multistakeholder forums and are very active in policy groups.

The issue remains that parents of children affected by rare disease as the primary care giver more often than not do not have the time to engage as advocates. This is an issue that will need to be addressed.

The difficulty of managing an illness whether as a patient or carer and giving time to the decision making process was exemplified at the research workshop when Caroline Heffernan who has cystic fibrosis and acts Patient Advocate of the Cystic Fibrosis Association of Ireland spoke about what daily life is like with CF, from the physiotherapy she must do to the medications she must take, and the commitment involved in ‘managing’ this disease. Caroline also spoke about how the impact of CF on her lifestyle is mentally quite challenging; not only for her but also for her family, especially during times of hospitalisation. Ways to enable these patients who are in the health system to have a voice must be employed.

- **Support for activities performed by patient organisations**

Anne Lawlor, who is the mother of a 26 year old woman with 22q11, shared her own experience of being a parent on a journey through Diagnosis, Information and Empowerment.

Anne mentioned that many rare conditions have the same issues such as delay in diagnosis, lack of understanding of the complexities of rare conditions, lack of professional expertise and lack of services. She mentioned the need to manage care over the lifetime of her child and emphasised that children grow up, not out of their condition. She outlined the need for medical, social and emotional support.

Anne’s daughter, Aine was very frustrated knowing in young adulthood that she was different, but didn’t get a diagnosis until she was 16. Aine’s mother Anne was determined to find out more about the condition and eventually did so by attending an international conference where she met others who understood her experiences. Anne also met Prof. Kieran Murphy at the conference and he has been very supportive ever since, even undertaking a research programme on 22q11.

The example of Irish patients meeting with experts from their own country at international events was repeated throughout the session.

Anne believes Knowledge is Power and is hosting an international conference on 22q11 in Dublin in 2013.

Judy Windle, one of the founders of GRDO, raised the issue of funding for a national organisation to support people living with rare diseases through the provision of information and direction to organisations that may help them at home and abroad. She stressed the need for condition specific groups to consult umbrella organisations, particularly those affected by ultra rare conditions that have no support groups. For 23 years GRDO has advocated for people with rare diseases with absolutely no funding. She suggested if such a group was funded appropriately there would be a central information point for patients and medics and the group could work with stakeholders to educate professionals.

A parent with adult children at risk of developing Huntington's disease mentioned the unfairness of people being weighted by insurance companies for a family history of a condition even if they get a favourable predictive genetic test which can't be considered under current legislation. He has a voice through the Huntington's Association of Ireland.

Innovative ways to support patient group involvement must continually be explored and supported (e.g. joint patient/researcher conferences, research/patient rare disease reference materials).

There are many examples given of support for the activities of patient organisations by medical professionals and the scientific communities. Prof. Kieran Murphy, Head of Psychiatry Royal College of Surgeons and President of the Irish Medical Council discussed his interest in carrying out research into 22q11.

Prof. Murphy complemented patient representatives like Anne Lawlor for their inspirational and enthusiastic approach to being advocates. He stressed the benefit of the patient /professional relationship in conducting research and managing a condition. He discussed the need to share

knowledge and the benefit of shared conferences for professionals and people affected by a condition.

The Medical Research Charities Group (MRCG) co-fund research projects together with the Department of Health and many of the projects have involved rare disease research. In the last three years (2006-2009) €1 million euro has been allocated by the Department of Health; a sum which has been matched by the MRCG charities in the co-funding arrangement. This important work continues.

IPPOSI as a unique, non lobbying group in Ireland with members from patient organisations, science and the pharmaceutical sector, has been working systematically over the last three years providing its membership with up to the minute information on the latest initiatives, treatments, research, funding opportunities etc in the field of rare disease

- **Specialised social services: Respite Care Services; Therapeutic Recreational Programmes; Services aimed at the integration of patients in daily life**

Joe Mooney, who has Muscular Dystrophy, is the CEO of Muscular Dystrophy Ireland. Mr Mooney described how he benefitted from the services of the Centre for Independent Living and Personal Assistants Scheme. He succeeded in living independently through the support of family friends but also the vital services of voluntary and charitable organisations that have been established by patients and their families.

Kate Power, who is Respite Coordinator with Muscular Dystrophy Ireland, gave a presentation of their support and respite service. Main points included the preference for in-home respite care, the need for parents and children to have time to bond as parents and children (outside of the caring relationship), the need for peer support and fun for young people with MD and the need for others in the family to get a break.

Kate discussed the lack of appropriate respite care for people with a condition and their carers and the fact that MDI fund their respite provision solely through fundraising.

Carers mentioned their frustration at the lack of appropriate respite care and therapeutic services. Services already provided through both state and charitable funding are in a vulnerable position because of cuts to the health budget of the state. There is now a genuine

fear that essential services provided through charitable organisations will be jeopardised and parents will face the prospect of being unable to care for their children at home.

The point that children with rare diseases grow into adults with rare diseases was stressed in the context of respite care services.

Many people affected by rare conditions used this as an opportunity to highlight the difficulties they had in getting an initial diagnosis, seeing professionals who understand the complexities of their rare conditions, the lack of professional expertise in rare conditions and the lack of respite and support services.

The need for psychological support for family members at risk of rare genetic disorders was mentioned.

- **Help lines**

There is currently no funding for a state funded helpline for rare diseases. There are however examples of voluntary groups providing professional counselling services and peer support to patients. There are also disease specific help lines funded through public/private partnerships and through state support.

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## Horizontal Themes

### **Theme 6 – Sustainability**

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The issue of sustainability was an overarching theme throughout the day and was referred to in each workshop. The realities of current economic restrictions will mean that the stakeholders will need to be creative and dynamic in their approach to this.

Public/private partnerships are already in existence in some areas while charitable and publicly funded initiatives are also evident. An examples of a good Public/Charitable partnership was the Health Research Board/ Medical Research Charities Group joint funding scheme, whereby patient led research charities can apply for matched funding for their projects through the HRB.

There is a history of public funding agencies supporting the activities of voluntary groups with social care, respite and counselling and support services. There is now every likelihood that the public funding will be reduced significantly and charitable agencies providing an essential service will be unable to operate. This will mean that more public fundraising will need to be undertaken by charitable groups.

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## **Theme 7 - Gathering expertise at the EU level**

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The Europlan Conference highlighted the ongoing need for strong partnerships between all stakeholders nationally and internationally.

The issue of the gathering of expertise at EU level and further afield was raised at each workshop.

There was considerable evidence that patient groups are already engaging with their peers internationally. Certain patient groups are also fundraising to bring experts to Ireland for one off clinics in the hope that they will be able to make an accurate diagnosis. It was agreed that while this is a necessity for some ultra rare disorders, expertise must be gathered in a coordinated way through accredited centres.

Some patient groups have excellent relationships with international groups and particularly with research professionals internationally.

There was more than one example of Irish patients meeting Irish researchers at European events and discussing collaborations, having no previous knowledge of each other's work.

It is clear that the structures are in place to engage internationally but again this comes back to the need for accredited centres and a coordinated approach.

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## Conclusion of the Final Report

The Europlan initiative was seen by all stakeholders as an important and successful step towards the establishment of a National Strategy for Rare Diseases in Ireland by 2013

The organisation of the conference enabled patient, scientific, medical and industry representatives who have been working closely on the rare disease agenda for more than a decade to engage with relevant state agencies in a coordinated and productive way.

The conference itself provided the opportunity for patients and their representatives to attend a dedicated Rare Disease event and to engage in discussion on the realities of life with these conditions for the very first time.

The programme was ambitious for one day; some delegates were of the belief that certain recommendations were aspirational only and would never be addressed in time for 2013. There were questions that could not be answered articulately because of gaps in information at the time.

The information gap in certain aspects of the recommendation informed the approach taken by the Irish Europlan organising committee and this is why it differed in some aspects from other countries. It was not possible to address all of the Europlan recommendations in very great detail however, but it did allow for the stakeholders to consider what is in existence and what needs to be developed from scratch.

Presentations from experts helped delegates to have a better understanding of what is already in place, as well as the experiences of other countries.

The vulnerability and fear associated with the diagnostic odyssey for rare disease patients as well as the constant uncertainty and adjustments to ever changing realities were clearly articulated in all workshops. Not knowing where to go for specific information and the lack of

someone to talk to as well as the feeling of being ignored were recurring themes throughout the day.

The need for a Centre of Expertise for Rare Diseases in Ireland was the mantra of the day and was repeated in every workshop and every discussion. This gap is seen as the root of the problems of access to information and diagnosis for patients and for medical professionals alike.

There are some very good systems in place and excellent models of disease specific centres of expertise to refer to when establishing a National Centre for Rare Diseases.

In the last ten years great advances have been made in medical research and development. Scientists have more understanding of the complex processes involved in rare conditions and are identifying causes and potential therapies. This enhanced understanding is raising great hopes from scientific and therapeutic progress. This is hugely exciting for all stakeholders but there remains an underlying fear that the research being funded and developed over the years – in many cases by patient groups, will not be accessible to them when they become medicinal products.

The research workshop highlighted the need for good links with patients/patient groups and well trained, experienced scientists, researchers, healthcare personnel. Pharmaceutical commitment to financial investment is also another key factor given that research into rare diseases is a lengthy and expensive process.

In a dynamic and changing environment, health research in Ireland needs to be positioned where it can make its greatest contribution for patients, the health system and the economy - a fact readily acknowledged by the Department of Health and Children. Clinical research is an area of key importance. It is clear however that more translational research is required particularly on the management of rare disease. Translational research aims to reduce disease incidence, morbidity and mortality by bringing scientific discoveries from laboratory to clinical application. It is through practical application that processes can be enhanced to become more integrated and effective and this is essential to the management of rare conditions.

Patient organisations have been important drivers of research in Ireland and abroad. They have developed into highly successful and innovative partnerships with scientific leaders. Some research into RDs would not have happened without seed funding from patient groups. These committed groups are required to stimulate development of research projects and will be essential in driving forward human clinical trials.

Other patient groups are working throughout Ireland and across the globe providing essential support and information services to rare disease patients and their families.

Rare Disease Patient Support Groups are often created by patients and parents desperate for information on specific rare conditions and exhausted from complicated journeys from specialist to specialist. They have displayed an ability to grow strong networks of patients working closely with researchers to further knowledge of orphan diseases, and develop therapies. The Europlan conference highlighted once again that patients and parents of children with rare conditions are active in the support of R&D for these conditions.

The challenge of the never ending quest for information is one of the most significant themes from the Irish Europlan Conference. Patients are hungry for information. Joint conferences where patients attend, engage and, often present together with medical professionals and researchers is the most common way in which patients can keep abreast of the latest research and developments. It is also an important networking and support opportunity.

The Department of Health and Children in Ireland is committed to tackling rare diseases. Patient representatives and patients, the medical and scientific communities as well as industry representatives stand ready to engage. Dr. John Devlin, Deputy Chief Medical Officer at the Department of Health has demonstrated a willingness to learn from and work with all stakeholders in this important task.

Progress cannot happen without the active and willing engagement of informed patients. Knowledgeable patients have the best understanding of the complexities associated with rare conditions. The role of patient groups in providing support and information in this regard is

essential. Informed patients can make educated choices about whether or not to participate in research.

Patient groups have contributed their time, energy and commitment to the establishment of a national strategy for many years. There can be no progress without these vital partners and sustainable supports must be developed to ensure their engagement in any decision making process.

The Europlan conference has played a vital role in ensuring that Ireland develops, budgets for and implements, with clear vision, pace and ambition an appropriate national plan for rare diseases by deadline 2013.

Now with the commitment of the Department of Health and all stakeholders to work together to the establishment of a National Strategy on Rare Diseases the time is right for commitment to become action.

## II. Document history

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