

IRELAND

EUROPLAN NATIONAL CONFERENCE

FINAL REPORT

7 September 2015, Dublin

FOREWORD

The EUROPLAN National conferences are aimed at fostering the development of a comprehensive National Plan or Strategy for Rare Diseases addressing the unmet needs of patients living with a rare disease in Europe.

These national plans and strategies are intended to implement concrete national measures in key areas from research to codification of rare diseases, diagnosis, care and treatments as well as adapted social services for rare disease patients while integrating EU policies.

The EUROPLAN National conferences are jointly organised in each country by a National Alliance of rare disease patients' organisations and EURORDIS – the European Organisation for Rare Diseases. For this purpose, EURORDIS nominated 10 EURORDIS-EUROPLAN Advisors - all being from a National Alliance - specifically in charge of advising two to three National Alliances.

EUROPLAN National conferences share the same philosophy, objectives, format and content guidelines. They involve all stakeholders relevant for developing a plan/strategy for rare diseases. According to the national situation of each country and its most pressing needs, the content can be adjusted.

During the period 2008-2011, a first set of 15 EUROPLAN National Conferences were organised within the European project EUROPLAN. Following the success of these conferences, a second round of up to 24 EUROPLAN National Conferences is taking place in the broader context of the Joint Action of the European Committee of Experts on Rare Diseases (EUCERD) over the period March 2012 until August 2015.

The EUROPLAN National Conferences present the European rare disease policies as well as the EUCERD Recommendations adopted between 2010 and 2013. They are organised around common themes based on the Recommendation of the Council of the European Union on an action in the field of rare diseases:

1. Methodology and Governance of a National Plan;
2. Definition, codification and inventorying of RD; Information and Training;
3. Research on RD;
4. Care - Centres of Expertise / European Reference Networks/Cross Border Health Care;
5. Orphan Drugs;
6. Social Services for RD.

The themes “Patient Empowerment”, “Gathering expertise at the European level” and “Sustainability” are transversal along the conference.

I. GENERAL INFORMATION

Country	Ireland
Date & place of the National Conference	7 September 2015, Alexander Hotel Dublin
Website	www.grdo.ie
Organisers	GRDO: Genetic and Rare Disorders Organisation
Members of the Steering Committee	<ul style="list-style-type: none"> • Ms Avril Daly, CEO Fighting Blindness, Chair GRDO • Mr Philip Watt, CEO Cystic Fibrosis, Ireland Chair of the Rare Disease Task Force • Mr Ken Rogan, IPPOSI • Ms Marie Downes, Medical Research Charities Group, MRCG • Ms Siobhan Collins, Fighting Blindness • Ms Anne Lawlor, GRDO • Dr Avril Kennan, Head of Research Debra Ireland
Names and list of Workshops	<p>1 - Methodology, Governance and Monitoring of the National Plan</p> <p>2 - Definition, codification and inventorying of rare diseases</p> <p>3 - Research on rare diseases</p> <p>4 - Care for RDs: Centres of Expertise and European Reference Networks for Rare Diseases</p> <p>5 – Orphan Medicinal Products</p> <p>6 –Social Services for Rare Diseases & Patient Empowerment</p>
Speakers	<ul style="list-style-type: none"> • Ms Avril Daly, CEO Fighting Blindness, Chair GRDO • Dr John Devlin, Deputy Chief Medical Officer, Department of Health • Professor Eileen Treacy, National Clinical Programme for Rare Diseases • Dr Derick Mitchell, Chief Executive, IPPOSI • Mr Philip Watt, CEO CF Ireland • Dr Avril Kennan, Head of Research, DEBRA Ireland • Mr Tony Heffernan, CEO & Founder Saoirse Foundation
Annexes :	<p>I. Programme</p> <p>II. Attendees</p>

II. MAIN REPORT

Introduction

The EUROPLAN National Conferences are aimed at fostering the development of comprehensive National Plans or Strategies for Rare Diseases addressing the unmet needs of patients living with a rare disease and integrating current European policies and recommendations in this field.

The second Irish EUROPLAN meeting took place in Dublin on September 7 2015 and was attended by 100 delegates from the field of rare diseases. Stakeholders from a wide variety of backgrounds were present, including: Industry; charity; patient groups; clinicians and healthcare professionals; academic researchers; and policymakers.

The event focused on the inherent challenges involved in implementing the National Rare Disease Plan and turning a cohesive framework into an everyday reality so that the lives of people with rare diseases are positively impacted. The main areas of focus were:

1. Centres of Expertise
2. Orphan Drugs
3. Research
4. Patient Engagement

Format:

Ms Avril Daly, CEO of Fighting Blindness, opened the meeting by outlining the vast progress made in the area of rare diseases in Europe since the establishment of Eurordis in 1997. Ireland has made significant advancements in very recent times, and the National Rare Disease Plan for Ireland 2014-2018 was published in June 2014. One of the main purposes of the event was to assess the achievements made during the first year of the plan and evaluate its sustainability as it progresses and is implemented further.

Theme 1 - Methodology, Governance and Monitoring of the National Plan

Dr John Devlin, Deputy Chief Medical Officer at the Department of Health

Dr Devlin opened the morning's proceedings by discussing the progress that has been made since the publication of the National Plan, while also outlining priorities for the near and long-term future.

He explained that a cohesive effort had been made to bring the issue of rare diseases to this point. Difficulties in accessing diagnosis/treatment, and indeed general expertise on rare diseases are still common in Ireland, however, but this is a problem we share with many other European countries.

The 48 recommendations contained within the National Rare Disease Plan cover the spectrum of rare diseases, from prevention, diagnosis, treatment, research, access to orphan drugs etc.

One of the recommendations of the National Rare Disease Plan was to establish a Health Service Executive (HSE) Clinical Programme for Rare Diseases, and this was realised in December 2013.

The programme will function as a means of implementing the Plan, said Dr Devlin; for example, identifying Irish Centres of Expertise, as well as providing assistance with applications for European supports if considered appropriate.

The National Rare Disease Office was established in the Mater Hospital, Dublin, in June, on foot of another recommendation from the national plan.

While these milestones are hugely significant, there is a significant amount of work to do if the blueprint for rare diseases as outlined in the national plan is to be realised.

Dr Devlin explained that a National Oversight Group has now been established by the Department of Health and its role will be as a departmental advisory group that provides oversight on the implementation of the recommendations of the plan. Terms of reference for the group are currently being discussed in advance of their publication. This will map out how the group can monitor progress of the implementation of the plan via key performance indicators (as mandated by EUCERD). These will also help the group identify priority areas for action in the coming years.

The purpose of the National Rare Diseases Office will be to facilitate patients in obtaining access to the best available treatment, via Centres of Expertise in Ireland or elsewhere in Europe. The Office will also function as a source of up-to-date information for patients on practical matters, as well as

clinical trials and new treatments. The Rare Diseases Clinical Programme will support the Office as is feasible for these functions.

In addition, the Office now hosts Orphanet Ireland, the online portal for rare diseases, orphan drugs and expert services. This service was previously hosted in Manchester, UK but Dr Devlin said there is now an Irish-based team to coordinate local Orphanet activity and ensure it is up to date with accurate information about current clinical trials and available treatments. The Irish Orphanet website is currently live (www.orpha.net).

The Office will support a helpline function over time it is proposed that it will also develop a role in rare disease surveillance. The work of the Office will be crucial in building a national picture of rare diseases as well as informing the Department of Health on future policy decisions.

The office received funding from the HSE this year and has been included in the estimates for Health in the upcoming Budget, he added.

Concerns were expressed during the meeting as to whether the Office is receiving sufficient funding; Ms Daly told delegates that the area of rare diseases is on a “funding trajectory” and is working towards a particular point.

Potential North-South collaboration is being explored, and suitable projects are currently being identified that could possibly be developed on a cross-border basis.

Dr Devlin concluded by saying that a certain amount of progress has been made in the first year of the plan, with attitudes within the Irish health service having changed significantly. First steps have been taken but a significant amount of work remains to be carried out.

“We have moved from a system which was entrenched to one of growth and expansion and that is very welcome. The important thing is to prioritise the future.”

The meeting heard, however, that more information and clarity for patients is needed in terms of these achievements since the publication of the National Rare Disease Plan.

Theme 2 - Definition, codification and inventorying of rare diseases

The importance of the accurate definition of rare diseases, as well as endeavours to commence a new coding and inventorying system cropped up on several occasions throughout the meeting.

Professor Eileen Treacy explained that a complex disease or condition is a disease or disorder that combines a number of symptoms or signs that require well planned organisation of services over time, because it implies one or more of the following circumstances: A large number of possible diagnosis/management options or co-morbidities; difficult interpretation of clinical and diagnostic test data; high risk of complications or morbidity or mortality related to the problem and its management.

There has been progress in the coding of rare diseases but this has been slow, explained Dr. Avril Kennan. The International Classification of Diseases (ICD) is the global health information standard for mortality and morbidity statistics and the latest version, ICD 11, is due to be released in 2018.

The meeting heard that the World Health Organisation has been working with ORPHANET on this and it is envisaged that a greater representation of rare diseases will be evident in this updated version.

Dr Devlin also told the meeting that coding for rare diseases will deliver a more comprehensive picture of the true pattern of rare diseases in Ireland – current coding systems are too generic for the specificity of rare diseases and do not provide an accurate picture of what is happening in hospitals and clinics nationwide.

There have been significant problems in initiating and sustaining rare disease research registries. Dr Kennan said that there have been positive moves in this area, however; for example, the HSE is about to introduce individual health identifiers, which will be critical to the establishment of registries.

The design of registries is also key and the particular software employed is a hugely important aspect, as this will enable registries to be live and “move with the times” as care pathways change, as well as enabling the sharing of data internationally.

Registries are becoming increasingly important but the challenges experienced heretofore in terms of sustainability will become less of a problem over time as the use of registries becomes more embedded. Registries support clinical care and allow patient engagement, the meeting heard.

It is envisaged that future registries will have patient portals where patients can personally input data and also retrieve it where appropriate.

Theme 3 - Research on rare diseases

Dr Avril Kennan, DEBRA Ireland

Research is a critical component of the National Rare Disease Plan but it is also a very broad area, and straddles all aspects of rare diseases contained within the plan, explained Dr Kennan. Clinical research will form the basis of how rare diseases are cared for into the future.

Most people are aware of basic research, which elucidates the underlying causes of a particular disease, and eventually leads to better diagnosis, and also applied research, into potential therapies and cures. The less obvious aspects to research are also important, however.

One issue in terms of rare disease research is that significant tranches of funding, e.g., from pharmaceutical companies, may not always be available. Therefore, the rare disease community needs to work together to speed the progress of research and also smooth the path to the development of new therapies.

There is a huge unmet need in rare disease, but barriers to participation in clinical trials still remain and understanding these is key.

Dr Kennan's own organisation DEBRA Ireland have led the development of a series of clinical practice guidelines for epidermolysis bullosa (EB). This helps translate the relevant research findings into everyday clinical practice – something that doesn't always happen.

There are four key themes for research with the National Rare Disease Plan. One of the overarching aims was to establish a network of rare disease researchers. Work will be done on this in conjunction with the Irish Orphanet team, which will provide a basis for the development of the network and enhance international visibility for Irish researchers in this space.

In parallel with this, tying in with other established mechanisms could be helpful – even something as basic as the creation of a group on LinkedIn. This could provide an easy forum for discussions and importantly could include all stakeholders.

Patient involvement in research is crucial, and the concept of patient-public-involvement is gaining traction across Europe.

Another issue highlighted within the plan is the importance of supporting research within the health service. The Centres for Expertise must have the capacity to undertake research and this should ideally be clinician-led as they have a unique knowledge of a particular condition that a research scientist won't. Clinicians must be supported to undertake research within their post. This would have the additional benefit of increasing patient access to clinical trials.

While there are pockets of excellence, research time is not protected and many clinicians are unable to even find the time to apply for membership of a clinical trial. Lack of experience in clinical trial participation is also an issue. A suggestion that late stage clinical trials should be attracted here was made – these would be more appropriate for clinicians and researchers lacking experience.

Research cannot be concentrated solely within the Centres for Expertise, the relatively new Hospital Groups must also be considered and research must be a pillar of their activities.

There have been breakthroughs via the Health Research Board and Molecular Medicine Institute in establishing improved clinical research infrastructure, and the Clinical Research Centres also represent a significant development.

Ireland clearly does not have the capacity to study every rare disease but there are pockets of excellence and some form of rare disease research is being undertaken in every institution in the country. While a dedicated funding stream would be ideal, this does not look to be a proposition realistic in the short to medium-term. Without this, a way must be found to incentivise rare disease research so that true innovation can be realised in the area, Dr Kennan asserted.

Registries are also central to impactful clinical research and efforts being made to establish live registries will aid future research efforts and participation in clinical trials.

A representative from the Health Research Board explained that many research activities the Board is involved in may in fact help rare diseases, despite not being originally designed to do so, and the recently improved research infrastructure will also benefit the area.

Theme 4 – Care for RDs: Centres of Expertise and European Reference Networks for Rare Diseases

Professor Eileen Treacy provided clarity on what the proposed Centres for Expertise and European Reference Networks (ERN) would comprise.

She explained that the definition of a centre of expertise is “a highly specialised healthcare provider for a complex disease or condition, designated through a formal assessment process”.

ERNs are expected to improve care for a particular disease by delivering highly specialised healthcare for complex diseases or conditions, and according to the European guidelines, these will comprise a group of highly specialised healthcare providers that are in compliance with a list of criteria and conditions, as set out by the European Commission.

The ERNs ultimately improve access to diagnosis, treatment and the provision of high quality healthcare to individuals who have rare conditions, particularly rare diseases or disorders, requiring the particular concentration of pooled resources or expertise. ERNs will also function as focal points for medical training and research, as well as information dissemination and evaluation. Stringent criteria for recognition as a CE or ERN have been developed by EUCERD.

Article 12 of the 2011 Cross Border Care Directive stated that the Commission would support member States in the development of ERNs between healthcare providers and Centres of Expertise, in particular in the area of rare disease. The Networks are based on voluntary, not obligatory participation, and will remain open to new membership on an ongoing basis provided that the healthcare providers fulfil all the required conditions and criteria, Professor Treacy pointed out.

The expectations regarding ERNs are the provision of improved access to shared databases, shared care pathways, guidelines, teaching and training, as well as infrastructure and funding resources into the future.

Recommendation No. 23 of the National Rare Disease Plan outlined that the Department of Health and the Health Service Executive in Ireland should encourage and support its national centres of expertise, once so designated, to seek recognition as EU-designated centres of expertise or as national members of ERNs. Recommendation No21 stated that the Health Service Executive should map out Centres of Expertise and healthcare pathways in accordance with what EUCERD have

outlined in its excellence and quality criteria. These criteria form the basis of applications for membership of ERNs.

One of those criteria centres on capacity – does the proposed Centre of Expertise have the capacity to manage patients and provide expert advice, provide patient access to multidisciplinary team of experts, integrating medical, paramedical and psycho-social care needs? The Centres must also have demonstrated expertise in care by evidence of referrals, publications and research activities etc. Evidence of continuity of care between children, adolescent and adult care must also be demonstrated.

It was previously recommended during the combined national EUROPLAN I meetings that prior to the establishment of Centres of Expertise, there must be a mapping out of expertise nationally. Professor Treacy explained that for smaller countries, where Centres of Expertise for all rare diseases may not be possible or feasible, then networking between medical experts nationally should be considered, while also promoting cross-border care cooperation. A central network coordinator will be required in this instance.

In November 2014, the National Cancer Strategy published information regarding its designation of national centres of expertise for cancer, which includes rare disease cancers. There are also 13 designated cystic fibrosis centres of expertise, and the National Centre for Hereditary Coagulation Disorders has a statutory designation governed by the National Haemophilia Council.

The 2012 HIQA National Standards aim to provide a roadmap for quality safety and reliability of healthcare and it is now proposed that the Authority will establish a system of hospital licensing to be implemented.

Over the past 18 months, the National Clinical Programme for Rare Diseases has developed a preliminary template of expertise nationally based on 350 of the more common rare diseases which have been outlined in the ORPHANET prevalence document for rare diseases. (This does not include cystic fibrosis or rare cancers which are governed by separate clinical programmes.)

As recommended and supported by the Acute Hospital Division, the Clinical Programme Working Group and Clinical Advisory Group, with the input of the Rare Disease Alliance representative and rare disease patient representative who sit on the working group, have developed a process for self-assessment of existing national expert centres or networks for expertise in Ireland via a self-

assessment template. The template has been designed based on 16 core recommendations of the EUCERD expert criteria in line with other Member State approaches.

The Clinical Programme has already assisted with the self-assessment of a number of the larger relevant health care providers this year. The self-assessment application, when completed by a health care provider is then reviewed by a sub-group of experts from the RCPI National Clinical Programme for Rare Diseases clinical advisory group who are not involved in the particular programme to be assessed. After this assessment, the application is then forwarded to the HSE Acute Hospitals Division and the Department of Health for nomination (if appropriate) for external review by the ERN-PACE external accreditation team at whichever level is appropriate for that programme.

The first call for ERNs will take place in early 2016, and an external accreditation procedure will take place, carried out by the approved external accreditation team, explained Professor Treacy, reiterating that there will be yearly calls for membership.

Emerging European partners have already approached noted Irish leaders in the rare disease field to consider joining ERN formations.

The Acute Hospital Division has sent out call for interest for HCPs to consider self-assessment for national Centre of Expertise designation with a view to possible entry into ERNs to the Hospital Group CEOs and HSE Clinical Programme Leads. The call is for sole providers or potential networks to nominate a co-ordinator to apply for the self-assessment process, with the first call for interest due in mid-September 2015. Further advice in relation to this process will be obtained at the October EC ERN meeting in Portugal.

Professor Treacy concluded by saying the priority now is to disseminate information on ERN membership to all healthcare providers. Many centres and Health care leaders are already enthusiastic and organised to comply with the process of self-assessment. She said there is now a major role for the Rare Disease Alliance and the patient support groups to identify Rare Disease Providers and Groups areas that could contribute to, or benefit from greater European collaboration and called for the Rare Disease Alliance to disseminate the relevant information and Information Sheet (provided).

Ms Daly reiterated Professor's Treacy assertion that a cohesive effort is being made within the entire rare disease community of stakeholders to inform and encourage healthcare providers to take part in the process of applying for assessment.

One parent at the meeting suggested that patient advocacy be incorporated into the Centres of Excellence, as patient and parents are self-educated very strongly on the particular rare disease and would be of great benefit.

Ms Daly expressed her belief that the area of rare disease research needs a "champion" but acknowledged that the medical profession have become increasingly active as stakeholders.

Theme 5 – Orphan Medicinal Products

Dr Derick Mitchell, Chief Executive, IPPOSI

Dr Mitchell began by explaining to delegates that he is a member of the implementation group for the implementation of the National Rare Disease Plan, and rare diseases will continue to be a priority for IPPOSI.

Unfortunately, the area of access to orphan drugs has seen little progress, and Dr Mitchell explained that while work is being carried out behind the scenes in terms of implementing the recommendations pertaining to orphan drugs, this has been slow to make an impact.

Of the key performance indicators for the National Rare Disease Plan, in terms of its implementation and monitoring, two of these directly relate to orphan drugs. One relates to the number of orphan drugs that are made available in Ireland, having obtained European Marketing Authorisation and then priced and reimbursed by the HSE. Dr Mitchell told delegates that in 2014, a total of 17 orphan drugs received marketing authorisation in the EU, which was a record number. He ascertained that this has received a mixed response among the rare disease community – should this be welcomed or is there much more that can be done in terms of increasing this number?

In Ireland, the National Centre for Pharmacoeconomics (NCPE) carried out health and technology assessments (HTAs) on nine of these drugs, of which the vast majority were reimbursed and made

available. Preliminary figures for 2015 suggest a similar number of drugs will be approved for reimbursement.

There positives and negatives to this tally, explained Dr Mitchell; if the majority of orphan drugs being assessed are then approved, this suggests that criteria other than cost-effectiveness are being considered. Lack of transparency, however, means that these other possible criteria remain unknown.

This relates to the recommendation within the National Rare Disease Plan that the HSE develop a working group to establish and bring forward criteria for the reimbursement of orphan drugs in Ireland. This proposed Group has not yet been established.

Transparency is vital, so that both patients and providers know which drugs are being reimbursed, the values involved in any economic evaluation, and what other information, if any, is being considered as part of that process. This will lead to equity and consistency with respect to accuracy across the entire system.

The second involves the establishment of a Governmental system for the compassionate use of medicines, and Dr Mitchell said there has been significant debate about the potential framework of such a system. There are a number of similar models in operation throughout Europe upon which a potential Irish system could be based, as well as Irish examples.

One is the Haemophilia Product Selection & Monitoring Advisory Board (HPSMAB) – in the 10 years since its establishment, it has achieved a significant reduction in the price of drugs for the treatment of haemophilia. These have decreased from 26 per cent above the EU average to just under 36 per cent of the EU average currently.

Dr Mitchell expressed his regret that there has not been more progress in the area of orphan drugs. Only a fraction of rare conditions have orphan drugs capable of treating them, and even where there are available therapies, there are myriad barriers to gaining access, including cost. As the understanding of the underlying biology of rare diseases increases, the definition of rare diseases will be subject to change. This will lead to the development of more orphan drugs.

He recommended that the area of approval and access to orphan drugs be treated as a priority in the short to longer term, as patients find it difficult to navigate the various routes to gaining access

to suitable therapies. Patient education in this regard is also key, and patients as advocates are extremely powerful.

Dr Mitchell further advised that in the case of an orphan drug being considered “exceptional”, IPPOSI will enter into informal discussions with the NCPE on the health economics impact, but the bearing of this on the final decision is unclear.

Ms Daly added that this is one of the more complex areas within the National Plan, and said that patients need more clarity and information on this topic.

One representative from industry explained that there is no mention of orphan drugs in the HSE’s annual service plan, with no ring-fenced budget; this may lend itself to the slow progress in the area.

The point was also made that we cannot access the list of reimbursed drugs from HSE.

Theme 6 –Social Services for Rare Diseases & Patient Empowerment

Tony Heffernan, CEO and founder of the Saoirse Foundation

Tony Heffernan established the Saoirse Foundation in 2009, and the charity works with the medical profession, researchers and other international charities providing patient support, advocacy and invest in research into Batten Disease.

He spoke of his personal experiences in relation to patient empowerment, having lost his two children to Batten Disease.

Access to clinical trials is of paramount importance, as is access and support that reaches organisations. While more funding is required for rare diseases, Tony said he does not believe in ring-fenced budget – he believes that this would lead to unavoidable constraints in funding, with people missing out on vital therapies if the budget has been spent. The concept of a ring-fenced budget for rare disease therapies and service was one that was endorsed by several of the day’s participants, however.

As a father of children with a rare disease, Tony spoke of his personal difficulties in accessing proper care, diagnostics, and clinical trials. He believes there is an element of stigma attached to rare disease.

While patient information is becoming more accessible, with the establishment of Orphanet and the National Rare Disease Office, he believes that it is not only patients that need education but also health professionals and clinicians.

Obtaining useful and accurate information is key and all stakeholders including industry, patient organisations, health workers and patients must engage in a bid to facilitate this.

In a bid to progress this, Tony is now in direct discussions with the HSE's National Lead for Patient Advocacy, and is also engaging in one-on-one meetings with acute hospitals, community services, and palliative care. He told the meeting that everyone has a right to know how their child will be cared for.

On this point, the meeting heard that more people need to become involved in efforts to improve the treatment of rare disease – patients and families must be encouraged to join in in order to drive progress into the future. While progress is slow and frustrating,

Misdiagnosis or failure to diagnose remains a huge problem – Tony explained that he was told that his son did not have Batten Disease on three separate occasions. This was echoed by several audience members, who expressed frustration with their inability to obtain a diagnosis in a timely fashion.

The psychological and social needs of people and children with rare diseases are not always met - access to services such as genetic counselling is very poor. Without access to these basic supports, life can be intolerable for patients and their families.

Spend within the health service in the area of rare disease should be examined closely, as there may be potential for cost-savings, Tony advised.

European involvement and collaboration was an ongoing theme of the meeting. The on-going European Patients' Academy on Therapeutic Innovation (EUPATI) project was also referred to as a positive development. This aims to empower patients by providing them with scientifically reliable, objective, comprehensive information to patients on medicines research and development. In addition, EURORDIS is campaigning for a European Year for Rare Disease to be held in 2019, which would help with wider awareness.

Parents of children with rare diseases present at the meeting told of their need to obtain diagnosis abroad, and subsequently being told in Ireland that the appropriate care was not possible because of lack of resources. Concern was expressed that recent cuts to health services here have led to a lack of equipment and other resources that are negatively affecting the care of patients with rare diseases.

Medical cards also cropped up as an issue; people with rare diseases are not automatically entitled to a medical card and may rely on the discretionary medical card system. One delegate told of how her son lost his medical card for a time only for it to be returned.

Other patients present expressed their frustration at the difficulty in obtaining access to medical records – as a rare disease may have many manifestations, the person may be under the care of several specialists and this a complete set of medical records is hard to obtain. The meeting heard that strides in terms of electronic referrals and individual health identifiers will undoubtedly aid in facilitating this. Similarly, these initiatives will also facilitate continuity of care and help patients who may have to see a number of specialists before receiving a definitive diagnosis.

Patients were urged by Philip Watt, CEO of CF Ireland, to “remain the awkward voice” and feel free and capable of expressing frustrations at the failure of their needs being met.

III. ANNEXES

Annexe 1: Programme



AGENDA

This event focuses on challenges experienced during the adoption and implementation of national rare disease plans and on how to turn national plan elements into a reality that positively impacts the everyday lives of people living with a rare disease.

<p>9.00am</p>	<p>Registration</p>
<p>9.30am</p>	<p>SESSION 1</p> <p>Chairperson: Ms Avril Daly, CEO Fighting Blindness (opening remarks)</p> <p>Dr John Devlin, Deputy Chief Medical Officer, Department of Health</p> <p>Professor Eileen Treacy, National Clinical Programme for Rare Diseases</p> <p>Dr Derick Mitchell, Chief Executive, IPPOSI</p> <p>Q&A</p>
<p>10.45am</p>	<p>SESSION 2</p> <p>Chairperson: Mr Philip Watt, CEO CF Ireland</p> <p>Dr Avril Kennan, DEBRA Ireland</p> <p>Mr Tony Heffernan, CEO & Founder Saoirse Foundation</p> <p>Q&A</p>
<p>12.00pm</p>	<p>Event closes</p>



Annexe 2: Attendees

<p>Attendees Registered:</p> <p>Total Registered:</p>	<ul style="list-style-type: none">• Patient representatives 50• Policy makers 6• Medical Professionals 20• Research 11• Industry 10• Other 3 <ul style="list-style-type: none">• 100
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