

RARE DISORDERS WITHOUT BORDERS

AN ALL-IRELAND MEETING OF PATIENTS' ORGANISATIONS,
SCIENCE AND INDUSTRY TO MARK INTERNATIONAL
RARE DISEASE DAY AND THE IRISH PRESIDENCY
OF THE COUNCIL OF THE EUROPEAN UNION.

OUTCOME REPORT





BACKGROUND

An all-Ireland conference entitled Rare Disorders without Borders took place in Dublin City Hall on the 28th of February 2013 to mark Rare Disease Day, the internationally recognised day for rare diseases.

The conference was coordinated by the Rare Disease Task Force in Ireland, which brings together the Genetic and Rare Disorders Organisation (GRDO), the Irish Platform for Patients' Organisations, Science and Industry (IPPOSI) and the Medical Research Charity Group (MRCG) along with the Northern Ireland Rare Disease Partnership (NIRDP) and Rare Disease UK.

In line with the theme 'Rare Disorders Without Borders', the conference promoted collaborative and innovative ways of working between the Republic of Ireland and Northern Ireland, the rest of the UK and across the EU in the field of rare diseases.

The conference coincided with the Irish Presidency of the Council of the EU, which was fitting given this year's end of year deadline for the development of national plans for rare diseases in the EU, and the deadline of October 25th for all EU countries to implement the EU Cross Border Healthcare Directive.

Essentially, the day provided an opportunity for patients, industry and academic representatives to come together to look at better ways to improve not only the length, but also the quality of life of those with a rare disease, and to ensure equality of access to the best possible care for all these patients.

Executive summary

It is estimated that 6-8% of the EU population has a rare disease so approximately one in 12 Irish people are affected by a rare disease at some point in their lifetime.

The conference heard from a number of rare disease experts across Ireland and the UK that early diagnosis and treatment in centres of excellence offers the best outcomes for rare disease patients.

However, some diseases are so rare it is not possible for smaller countries to provide specialised services and patients need to travel; hence the need for increased cross border collaboration and information on travelling for care across Europe.

Currently many rare disease patients face years of delay in accessing an accurate diagnosis, which can have a significant negative outcome on their prognosis and quality of life.

The conference also heard there is a lack of expertise in Ireland for many rare diseases and a lack of specialised knowledge among many healthcare staff about the latest treatments and developments in individual rare diseases internationally.

The conference heard that Irish patients with rare diseases are also experiencing increasing difficulties in accessing treatment in other countries under the HSE's treatment abroad scheme. This is a

negative development as many medical treatments for rare disease patients are only available abroad and patients have previously been able to access them. Such a move also contravenes Ireland's responsibilities under the EU Cross Border Healthcare Directive.

The conference also heard that Irish funding is being cut for rare disease services, such as genetic testing and support services at the National Centre for Medical Genetics, Crumlin, despite the fact that there is increasing demand for genetic testing due to ongoing findings about genetic links to rare diseases.

The spectre of the recession and declining health budgets are hard to escape from but the consensus at the conference was that we need to move away from rationing rare diseases healthcare and research, as such moves can lead to more healthcare and economic costs in the long term, not to mention the huge human cost. Investment in diagnostics and research, both basic and translational, is vital and will pay off. Thanks to investment in these areas to date many rare disease patients who previously didn't live beyond childhood now have much longer life expectancies and improved quality of life.

While the recession throws up many challenges it has also created an opportunity for more focused thinking on how to maximise resources and use more innovative ways to provide the best healthcare and support for rare disease patients. Cost effective solutions

include increased co-operation across the EU to avoid duplication of services, collaboration on research and increased use of online technology and telemedicine to improve service access and support for rare disease patients.

Conference learning points

Challenges for rare disease patients

- Funding challenges due to the recession.
- Lack of adult health services for some rare diseases that were traditionally very life limiting.
- Many rare diseases are so rare that each country cannot have a centre of expertise for that particular disease.
- Patients with rare diseases are at a higher risk of isolation, discrimination, depression and poverty.
- Many rare disease patients find it difficult to travel, both from a mobility and cost point of view.
- Difficulties in accessing orphan drugs. Currently 76 orphan drugs have gained European marketing approval but not all are being accessed in every country, often

due to lack of knowledge.

- Eligibility issues and difficulties in knowing what patients are entitled to and lack of centralised information on how to access specialised care.
- There is still a lack of awareness about many rare diseases in the medical profession and among the general public.
- Shortage of geneticists and other rare disease health staff in Ireland and decreased funding for genetic testing despite increased demand.
- Many current diagnostic tests are not accurate or sensitive enough to enable a proper, timely diagnosis.

Solutions and opportunities identified during the conference

- Transposing the EU Directive should dramatically increase cross border collaboration on healthcare services for rare disease patients.
- Increased cross border collaboration will reap serious benefits both in improved quality of care and financial savings and will avoid duplication of services/ research.

- The approval of national plans on rare disease care offers fantastic opportunities to improve and standardise services going forward.
- the rare disease national plans for the UK and Ireland should have specific commitments on transnational cooperation within Ireland, North and South and between Ireland and the UK
- The creation of European reference networks and centres of expertise also offer huge potential to improve and expand rare disease services.
- Better education of healthcare staff about rare diseases is vital, as is easily accessible, up to date and accurate information for rare disease patients, their families and the general public.
- There is an urgent need for registries of patient numbers and classification of rare disease patients, and centralised information on what services are available and what research is ongoing in each EU country.
- Increased use of technology-e-health, telemedicine, Skype, etc-will bring services closer to the patient without excessive costs.
- There is a real need for holistic care. It is not just about treating the disease.
- The voice of patients must be heard. Patients representative groups and associations need to be supported and utilised by policymakers and the medical community to place the patient at the centre of decision-making about their care and to listen to what their experiences are.
- The International Rare Diseases Research Consortium (IRDiRC), which fosters international collaboration in rare diseases research, offers great opportunities.

Sarah Louise Leonard, Jamie O'Brien and Sarah Ivers who participated in the Patient's Perspective video which is now available on YouTube.

A photograph of three people standing in front of a large stone building. In the center, a man with a red mohawk and glasses has his arms crossed on top of a large blue sign. To his left, a woman with blonde hair smiles. To his right, a woman with glasses and a black leather jacket looks forward. The sign they are holding has a blue background with a geometric pattern and the text 'RARE DISORDERS WITHOUT BORDERS' in white and pink.

**RARE
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Challenges facing those with rare diseases

The conference heard from a number of different speakers and audience members about the various difficulties that currently face those with rare diseases: Lack of access to specialised services and treatments, difficulties in travelling, delays in diagnosis, lack of understanding of the realities of life for rare disease patients, lack of support services, as well as the financial, emotional and physical strains on the families of rare disease patients were issues that were referenced again and again.

A number of audience members pointed out that there is a lack of clarity on what patients are actually entitled to in Ireland and that sometimes this can change or differ in regions for no apparent reason.



Ms Avril Daly, Vice President EURODIS and Chair of GRDO, reminded delegates that while there are huge differences in the types of

rare diseases that affect people right across the spectrum of physical, sensory and intellectual disabilities, a lot of the issues experienced are the same.

Prof Kate Bushby, Professor of Neuromuscular Genetics, Newcastle, UK, and Vice-Chair of the EU Committee of Experts on Rare Diseases, spoke about the challenges currently facing rare disease diagnosis and management, and said the establishment of the EU Committee has been a great step forward in identifying the relevant issues and potential solutions for this cohort.

Issues identified by the Committee include problems with coding and classification of rare diseases, which means they can be more easily ignored by health systems; a lack of evidenced-based guidelines, which are common in more standard diseases and lead to improved outcomes; and the fact that there has been less incentive traditionally for pharmaceutical companies to develop drugs for these patients given the small market.

While acknowledging the challenging financial times we live in, Prof Bushby reminded delegates that not investing in rare disease services and research coupled with delayed diagnosis and inappropriate care, creates many, often invisible, long term financial costs for health services and of course severe human costs for the patients involved.



Patients and carers focus

To reinforce the many issues faced by patients living with rare diseases, a pre-recorded patient testimonial was played during the conference. The video was introduced by Ms

Meanwhile, Ms Katie Rigg, Specialist Nurse, Multiple System Atrophy Trust, said healthcare professionals need to admit when they do not know enough about a specific rare disease and be open about approaching those who do.

She reiterated the need for more education for healthcare staff on rare diseases and also highlighted the need for translational material on rare diseases as not everyone in Ireland, for example, has English as their first language.

Ms Rosemary Arbutnot, Director, Northern Ireland Rare Disease Partnership (NIRDP), meanwhile, said the strain on patient carers isn't taken into consideration and that they need support too, which isn't appreciated despite the huge savings they provide in relation to care.



Catherine Martin, a specialist Huntington's disease youth worker, who reminded the conference that people with rare diseases deserve the same level of care as people with common diseases.

The impactful video told the stories of a number of rare disease patients and featured them and their family members talking about how their condition impacted on their lives; the frustration and despair they feel due to the struggle to get accurate diagnosis and appropriate care as well as the impact on their quality of life and the feeling of isolation due to the lack of understanding of their conditions.

One member of the audience who has motor neurone disease reminded the conference that he is a person, not a disease, and this was something that resonated with all those present.

Seeking solutions for rare diseases

The conference was not just about raising problems but about seeking solutions and learning from our neighbours, with the aim of deepening the resolve to improve co-operation about rare diseases, both north and south in Ireland but also with the rest of the UK. The conference also provided an opportunity to stock take, and highlight the developments to date in improving medical treatments and services for rare disease patients.



Mr Philip Watt, Chairperson Rare Disease Taskforce and Medical Research Charities Group, said if some people wonder about the “point’ of investing in rare diseases given the shorter life spans of some patients with certain diseases they should look at the case of cystic fibrosis,. While this remains a very challenging disease in marked contrast to earlier years, people with CF are now expected to live into their 30’s and beyond thanks to improved specialised centres of care, multidisciplinary clinical teams, innovative therapies and earlier diagnosis. He emphasised the potential impact on all rare diseases through improved north/south and

trans-European cooperation on clinical trials, consensus in standards of care and public policy interventions on organ donation and transplantation. Within Ireland we need to move from the mindset of ‘backs to borders’ in the provision of health services in general and the need for greater pooling of resources in respect of rare diseases in particular.

Many recommendations have been formulated by the EU Committee of Experts on Rare Diseases, Prof Bushby noted. For example, the Committee has identified the creation of patient registries as a priority, so rare disease patients can be properly monitored and linked to appropriate care, as well as benefit from research, along with the creation of centres of expertise which provide holistic care. The creation of European reference networks should also significantly improve care for rare disease patients across Europe in terms of sharing expertise, she maintained.



Prof Bushby also pointed to the positive work to date in rare disease research funded by a number of EU

research funding platforms, saying this research has highlighted what can be achieved; that new therapies can be successfully developed and that collaboration is a key part of that. In addition, she promoted the importance of patient organisations as a voice for rare disease patients.

Ms Rigg also highlighted the benefits of video conferencing and telemedicine in bringing support and actual medical services to isolated patients.

Ms Daly said that the Irish national plan on rare diseases is eagerly waited by all stakeholders and it should allow a real platform from which to go forward regarding how rare diseases are treated in Ireland.

Examples of cross border healthcare collaboration

The conference heard details about a number of positive Irish cross border healthcare initiatives, both in terms of rare diseases and standard healthcare services. These initiatives provide very useful learning as well as actual resources to help further cross border healthcare services for rare disease patients.

Ms Paddie Blaney, All Ireland Institute of Hospice and Palliative Care, explained how high quality education and research and better patient experiences are coming about from the work of her Institute. She acknowledged getting cross border collaboration up and running can be quite difficult but said much



groundwork has now been done in the area and with determination and hard work much is possible and the outcomes are worth it.

“It is challenging. You need leadership to do this and you need the integrity of realising you may not get everything you want but that there is a central focus we can all agree on and that is worthwhile going after,” Ms Blaney stated.



Ms Sadie Bergin, of the long established cross border health and social care partnership, Co-operation and Working Together (CAWT), highlighted how cross border healthcare services are working in practice along the border counties of Ireland.

For example, Donegal patients can access radiotherapy in Belfast, while there has been a number of CAWT-supported north south health

conferences and research initiatives.

Ms Bergin said feedback from clients has been positive to date on the cross border services provided. While overcoming the difficulties of dealing with two completely different health systems is not easy, she said there is already a lot of groundwork now done, and CAWT's existing models of cross border health services are well established, validated and target orientated.

Mr Philip Watt, who is also CEO of Cystic Fibrosis Ireland, told the conference that he has seen great north south collaboration on CF research, including on a clinical trial for the new CF drug Kalydeco, which has ensured fast-tracking of the drug to eligible Irish patients. However, Mr Watt pointed out there is a need for increased north south collaboration in terms of CF patients in Donegal who currently have to travel to Dublin to access specialist CF services, when it would be much easier for them to travel to Belfast.



Mr Alastair Kent, Chairperson, Rare Disease UK, stressed the importance of engaging with patient organisations and utilising their knowledge, and added it is vital that

the medical profession realise the value of informed and engaged patients in getting the best outcomes.

Audience participation - Making cross border collaboration work

The conference hosted a specific question and answer session where audience members could ask the panel questions on making cross border collaboration work for rare disease patients.



Ms Eibhlin Mulroe, CEO, IPPOSI, chaired the session and the panel included Mr Rowan, Prof Bushby, Prof Eileen Treacy, National Centre for Inherited Metabolic Disorders; Dr Sally Ann Lynch, National Centre for Medical Genetics; Ms Fiona McLaughlin; Progressive Supra Nuclear Palsy Association; and Ms Rosie O Shea, GRDO.

Ms Mulroe noted that Ireland's steering group for the creation of the national plan on rare diseases has four patients on it (appointed by the Minister in 2011), which has not happened in other European countries. "This has been a long

journey but we are nearly there in terms of the plan. We are listening to our members and it is very important their stories are heard," she commented.

Ms Mulroe also added that patients and representative organisations have been communicating and collaborating across the border in Ireland for many years, and it is now a matter of our health systems catching up with that.

A common theme among the numerous questions raised were what timeframes are in place for Ireland to create centres of excellence for rare disease patients, where will they be based, and what level of cross border collaboration will there be.

In response, Ms Daly pointed out a number of units with expertise on particular conditions already exist in Ireland, and that centres in different parts of the country should be encouraged and supported to develop their individual services perhaps, as opposed to having one centralised centre for all rare diseases. However, discussions tackling these types of questions are ongoing and are being discussed in the formulation of Ireland's national plan for rare diseases.

Another audience member, who had a child with Duchenne's, asked about the scope of expertise travelling rather than patents and the scope of e-health in helping bring vital health services to rare disease patients. Prof Bushby said

the development of reference networks would help greatly and that e-health solutions would certainly be utilised. Another audience member mentioned there are virtual health care services in existence already where patients can Skype in and asked the panel how do we convince health providers that this is the way to go for patients who find it hard to travel.



Answering questions about her development of Irish centres of expertise, Prof Treacy reiterated that there already exists a lot of expertise on certain diseases within some larger teaching hospitals, for example in the case of the cancer services the most rare cancers are treated in specialised, centralised centres. She too agreed that there would never be a critical mass of expertise for some extremely rare diseases in Ireland given our small size. However, Prof Treacy also acknowledged the difficulties of patients who have serious disabilities travel abroad and stressed the importance creating a reference network to get a handle on the issue. Dr Lynch pointed out the Republic has an extremely low level of geneticists per head of

population compared to many European countries, including Northern Ireland. (see Appendix 1) She said this issue and the shortages of other key staff and funding at the National Centre for Medical Genetics has been highlighted to the Government. As a result of the policy not to replace staff who have retired, resigned or who are on maternity leave the centre has seen a 24% drop in whole-time equivalents since 2008. Despite finally obtaining a consultant training programme at the Centre after seven years, Dr Lynch said the Centre's service would continue to deteriorate given the amount of time it takes to complete training and build up services. "So we have a Tsunami of referrals with extreme staffing shortages meaning we couldn't possibly implement what Prof Bushby has been talking about. We could not take on providing the multidisciplinary care recommended by Prof Bushby. We haven't a hope as we can just about provide a diagnostic and genetic counselling service. For all those on our waiting list, I apologise, we are doing our best, having the most efficient appointments system in Crumlin Hospital" Dr Lynch told delegates. Questions were also asked on when the new Clinical Lead for rare diseases would be appointed. However there was no Government official on the panel who could answer these questions.

Government input

The conference featured presentations from two Ministers, one from Northern Ireland and one from the Republic. Minister Edwin Poots, Social Services and Public Services and Public Safety, Northern Ireland, confirmed he strongly supports increased collaboration with the Republic on the provision of cross border health services for rare disease patients, and noted that "rare diseases do not respect borders".

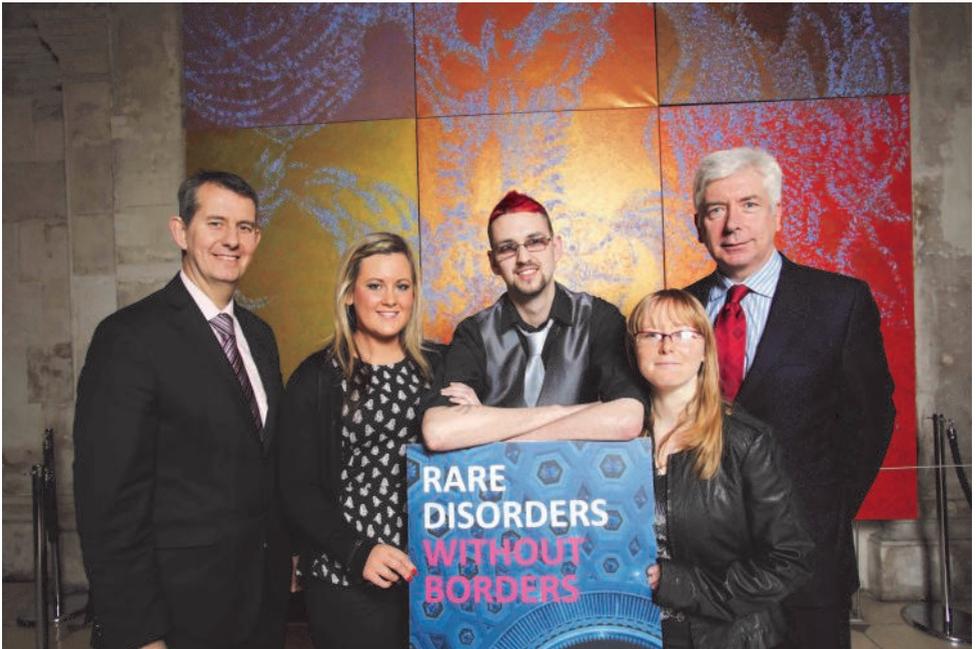
"I always have the mantra 'the right care by the right people at the right time' and that's the advocacy we should have when it comes to rare diseases," he commented. Minister Poots agreed with other speakers



that the right care can be expensive, but not providing it is also expensive. Ensuring equality of access for patients in Northern Ireland, given the geographic difficulties, emerged as a key issue during the UK's consultation process on its national plan for rare diseases. Minister Poots said it has been consequently acknowledged that developing clinical networks



The conference was attended by more than 200 delegates and was jointly opened by Edwin Poots, Minister of the Department of Health, Social Services and Public Safety in the Northern Ireland Executive and Alex White, Minister of State for Primary Care in the Irish Government



with the Republic could help support sustainable service models for rare disease patients in Northern Ireland, and that flexibility and collaboration are in the best interest of this cohort. The implementation of the EU Cross Border Directive on Healthcare should also help clarify and increase this collaboration, he concluded.



Irish Minister of State for Primary Care Alex White expressed the Government's commitment to ensuring appropriate services and fair access to treatment for rare diseases, whatever the challenges. He welcomed the Directive and said it provides a "coherent and uniform set of rules for patients throughout the EU that will start a new phase of co-operation" between member States' health systems.

Minister White echoed many of the speakers at the conference by saying that many rare diseases are so rare it is not possible for every country to have sufficient expertise on them and patients will have to travel to obtain the best services; hence the need for greater cross border collaboration across the EU.

He also confirmed that greater cross border collaboration is already being explored with Northern Ireland.

"However, at EU level it is agreed, including in the 2009 Council recommendation on rare diseases, that where possible expertise should travel, rather than patients themselves," Minister White told the conference.

While noting the HSE has a limited budget, he confirmed it is in the process of setting up a dedicated clinical programme for rare diseases and is appointing a Clinical Lead for rare diseases, though it is uncertain if the post will have additional resources according to other speakers at the conference.

Confirming that work on Ireland's national plan for rare diseases is progressing well, Minister White revealed that just under 500 submissions were received during the online consultation process last year.

Minister White also praised patient advocates and the conference itself for keeping pressure on politicians and ensuring the issue of rare diseases remains in the spotlight.

Detailing the work in this area, the Minister said that following on from the first EUROPLAN conference in January 2011, the important work of developing the national plan for rare diseases has been continuing under the steering group established by Minister Reilly in April 2011. The

remit of the group is to develop a policy framework for the prevention detection and treatment of rare diseases based on high quality care, equity and the requirement to be patient centred. He said the national steering group has identified a number of areas relating to rare diseases which it will consider. These include centres of expertise, access, medication and technology, research and information, patient empowerment and support.

Minister White said that a range of initiatives are under consideration and he understood that it is planned to complete and publish the rare diseases plan in the latter half of 2013.

How the EU Directive will work

Despite its long lead in time, many are confused about how exactly the EU Cross Border Healthcare Directive will work.

Mr John Rowan, Healthcare Systems Unit, European Commission, gave an enlightening overview of its application with

specific reference to rare disease patients at the conference. He said if a patient is entitled to a treatment at home they are entitled to reimbursement for that treatment abroad, and the only reason they should be turned away is if there is no capacity. For some treatments and specialised care (e.g. hospital care), health systems may require patients to seek prior authorisation. Authorisation may be refused if there is no "undue delay" in the patient's home country but the authorisation processes must be reasonable, and properly reasoned, he stressed. Each country will have to have at least one national contact point where patients can receive details on the Directive and how to get treatment in another EU country.

Many diagnostic services are likely to fall outside prior authorisation so reimbursement will be possible under the Directive and they should be easier to access because of it, he said. While it is hard to predict the impact of the Directive, Mr Rowan said a key benefit should be greater information will be available across the EU about health entitlements, quality and safety standards, and treatments offered.



Conclusion

The conference succeeded in highlighting the significant challenges that rare disease patients face, but also the fantastic opportunities and innovative solutions that can be utilised to dramatically improve their lives.

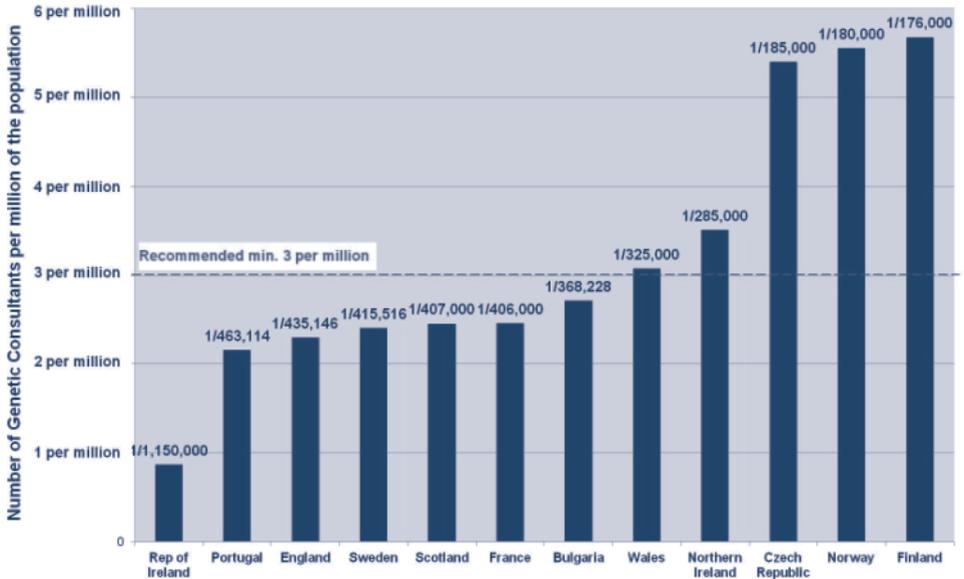
2013 is an incredibly important and exciting year for patients with rare diseases right across the EU. The Directive and the deadline for national plans mean real timeframes and actions to improve care for rare disease patients will now be put in place. It is vital now to engage all stakeholders in order to fully realise the potential of the Directive and Ireland's national plan, and ensure that EU cross border collaboration is fully maximised to the benefit of rare disease patients.



Ms Christine Collins, Chairperson, NIRD, closed the conference by saying she hoped the huge amount of learning that the day provided can be utilised and fed back to

policymakers and service providers. "We want this to be the start of the process, the start of a raft of work going forward, and we've got to get a bit more strategic about how we do that.... We are stronger together," she concluded.

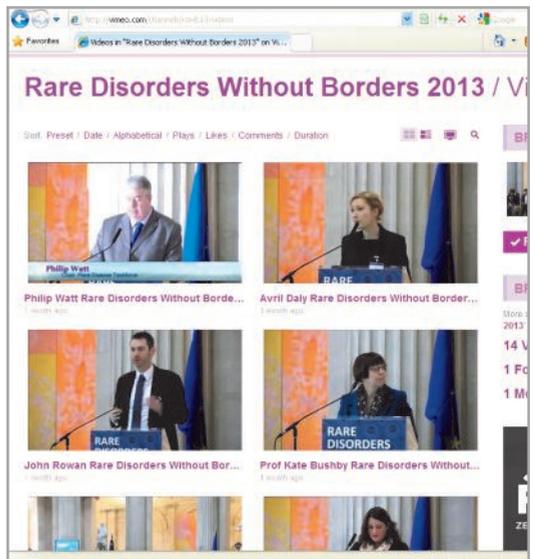
Appendix 1



Online resources

To fully appreciate all the contributions made at Rare Disorders Without Borders, it is possible to view the entire conference online via a recorded webcast at;

<http://vimeo.com/channels/rdwb13>,



NOTES:

- This report provides a summary of the presentations and discussions that took place at the Rare Disorders Without Borders conference on the 28th of February 2013.
- For full programme information on the conference and its speakers go to www.ipposi.ie.
- On the website you can also review the posters that were submitted to the poster call. The first prize went to Dr Paula Byrne UCD –Teaching Medical Students about Rare Disease. Second prize went to Motor Neurone Disease Association - New Patient Charter and the most engaging poster prize went to Michael Holden-Tripability.
- The Twitter hashtag for the conference was #RDWB13. To search for tweets on the conference enter the hashtag into the Twitter search box.
- You can view the photo gallery from the conference on a dedicated Flickr photo stream page (<http://www.flickr.com/photos/93672142@N04/>).
- Our sponsors had no editorial control of this outcome report which was independently compiled by Ms. Pricilla Lynch



Photographed at the launch of the conference programme were Philip Watt Chairperson of the Rare Disease Taskforce, Meadhbh Cleary who has been diagnosed with the rare condition Scleroderma and Eibhlin Mulroe CEO of IPPOSI



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