DÁIL ÉIREANN

AN COMHCHOISTE UM SHLÁINTE AGUS LEANAÍ

JOINT COMMITTEE ON HEALTH AND CHILDREN

Déardaoin, 26 Feabhra 2015

Thursday, 26 February 2015

The Joint Committee met at 9.30 a.m.

MEMBERS PRESENT:

Deputy Robert Dowds,	Senator Colm Burke,
Deputy Peter Fitzpatrick,	Senator John Crown,
Deputy Seamus Healy,	Senator Jillian van Turnhout.
Deputy Billy Kelleher,	
Deputy Sandra McLellan,	
Deputy Caoimhghín Ó Caoláin,	

DEPUTY JERRY BUTTIMER IN THE CHAIR.

The joint committee met in private session until 10 a.m.

Implementation of National Rare Disease Plan 2014-18: Discussion

Chairman: I welcome everybody to the meeting. I remind members, witnesses and the people seated in the Gallery that mobile phones should be switched off or left in airplane mode as they interfere with the broadcasting of proceedings. Failure to do so may mean a presentation is not covered further afield which would do everyone a disservice but we must also be fair to the staff who must wear headphones.

Next Saturday, 28 February, is national rare disease day. As we have done in the past, at the invitation of Deputy Ó Caoláin, this committee held a meeting on rare diseases which was a good and successful venture. We feel it is important that the committee marks national rare disease day with a meeting to discuss the subject. Last year, the committee discussed the launch of the national rare disease plan for Ireland. A year on, the committee wishes to take stock of progress made with the implementation of the national rare disease plan and to discuss the next steps to take with the key stakeholders that are responsible for its implementation.

I welcome the following: Dr. John Devlin, chair of the national steering group on rare diseases; Mr. Liam McCormack, Department of Health; Ms Helen Byrne, assistant national director, acute hospital services, HSE; Dr. Colm Henry, national clinical adviser and group leader for acute hospitals, HSE; Mr. Tony Heffernan, founder of the Saoirse Foundation; Ms Avril Daly, chair, Genetic and Rare Disorders Organisation; Ms Eibhlin Mulroe, CEO, Irish Platform for Patients' Organisations, Science and Industry; and Dr. Avril Kennan, member of the rare diseases task force. Apologies have been received from Mr. Philip Watt, CEO, Cystic Fibrosis Ireland, who has been a key player with us on this committee. He was to attend today but unfortunately he cannot do so for personal reasons. I am sure many of the speakers will cover the points that he might have wished to raise.

By virtue of section 17(2)(*l*) of the Defamation Act 2009, witnesses are protected by absolute privilege in respect of their evidence to the committee. If, however, they are directed by it to cease giving evidence on a particular matter and continue to so do, they are entitled thereafter only to qualified privilege in respect of their evidence. They are directed that only evidence connected with the subject matter of these proceedings is to be given and asked to respect the parliamentary practice to the effect that, where possible, they should not criticise or make charges against any person or an entity by name or in such a way as to make him, her or it identifiable.

Members are reminded of the long-standing parliamentary practice to the effect that they should not comment on, criticise or make charges against a person outside the Houses or an official, either by name or in such a way as to make him or her identifiable.

I invite Dr. Devlin to make his opening remarks.

Dr. John Devlin: On behalf of the Department of Health, I thank the Oireachtas Joint Committee on Health and Children for providing us with the first opportunity to discuss our response to tackling rare diseases, particularly in the context of the upcoming national rare disease day.

I recall that when we started work on the national plan for rare diseases, which is only a couple of years ago, a number of issues stood out, namely, the diagnostic odyssey or the journey

through no-man's land that patients and families had endured, and access to appropriate medicines and technologies. We have tried to keep these issues at the centre of our plan.

Ireland is no different from many other countries when it comes to rare diseases. The original European Council Decision 2009/C15/02 on rare diseases recommended that countries like Ireland should develop national plans and estimated that there were between 5,000 and 8,000 rare diseases. That meant there was often a limited number of patients with a specific disease and, probably more importantly, a scarcity of relevant knowledge and expertise. We know that rare diseases, when added together are relatively common and affect between 6% and 8% of the population. We also know that many of these conditions are complex, severe and debilitating.

In terms of developing a national plan, we held two national conferences as well as online public consultation to discuss what might feed into the development of a plan. It proved helpful to us and highlighted issues we were not particularly aware of at the time. For example, the role of carers who play a vital part in the overall care provided. I thank the patient organisations for all of their help, not just in their contributions they made but also in helping us to plan these days and for the feedback they provided.

I am chair of the national steering group set up by the Department to develop the plan. I wish to acknowledge the help and input from the patient groups that are members of the national steering group. Many of them are here today and will talk later.

The plan was published in July 2014 and we have made copies available for the joint committee. The plan is meant to operate over five years and will define our priority actions. In all, there are 48 recommendations and I shall outline some of them and the progress we are making to the committee.

The relevant areas include the identification of centres of expertise, access to medication and technology, research and access to clinical trials and the empowerment of patient organisations. In European terms, Ireland is a relatively small country. We have significant capacity and a good number of treatment reference centres. We will also have to co-ordinate with other countries as there are up to 8,000 conditions of which many are very uncommon. At EU level, work has started on developing European reference networks and countries like Ireland are playing a key role in this work already. Countries will have to nominate their references centre which will form part of these ERNs. If it is clinically needed, patient care could be shared across reference centres so patients or expertise may travel.

This work does not happen in a vacuum. We need to develop care pathways, greater use of protocols, better access to new technologies and, most importantly and what the initiative will deliver, better outcomes for patients and families. Access to clinical trials is also very important as is the need for a supportive environment for research and to promote clinical trials in Ireland. Our report also acknowledges the key role of information and disease registers.

The plan has also recommended the development of a national office for rare diseases. Its purpose would be to facilitate the co-ordination and timely access to centres of expertise for rare diseases, both nationally and internationally. The office will also act as a national point of reference on services, diagnostics, care pathways and on information regarding rare diseases. The HSE has commenced work on establishing the office. Half-year funding for 2015 of €100,000 with €200,000, on a recurring basis in 2016, has been provided with a €2,000 once-off start-up non-pay cost. Staffing will include an information scientist, genetic counsellor and administrative support.

As the committee will know, the HSE has also established a national clinical programme for rare diseases. The programme aims to improve and standardise patient care for individuals affected with rare diseases in Ireland, by increasing detection and prevention, facilitating early timely diagnosis, co-ordination of care and providing better information and support.

The report also acknowledges the key role of patient organisations and what we need to do more of to empower patients and their families. As I said earlier, there is also a big EU dimension to this work. Recently, the EU rare disease committee has been reconstituted. It is setting new priorities for best practice guidelines, information, research and the establishment of the ERNs. All of this has a bearing on Irish centres of expertise and we are happy to talk about this later.

We also have a good working relationship with our colleagues in Northern Ireland. Prior to the publication of the plan, and subsequently, we have met our colleagues from Northern Ireland on a number of occasions, to discuss practical ways to provide better care and the sharing of information and promotion of research.

The launch of the plan completes this stage of our work and the HSE is now working towards its implementation. There is a lot more we need to do over the next five years. We are happy to answer any questions the committee has on our work.

Chairman: I thank Dr. Devlin and call on Mr. Tony Heffernan to make his opening remarks.

Mr. Tony Heffernan: I am Tony Heffernan and I am founder of the Saoirse Foundation. It a national charity which focuses on proactive advocacy, is an investor in Batten's disease and rare disease research and focuses on delivering positive practical supports for those affected by rare conditions.

The Saoirse Foundation is also actively involved in the development of Liam's Lodge which is a national respite and palliative care facility for children affected by rare, genetic and life limiting conditions. In addition, the foundation provides specialised transportation services for children via its BUMBLEance service which provides an ambulance that has been specifically designed for children. It is a service that is accessed weekly by children affected by rare and life limiting conditions.

During my time as the patient support and empowerment representative on the previous Minister for Health's national steering group for rare disease, I was also chairperson of the patient and support subgroup. The subgroup worked with the additional steering group and task force members on the development of our first plan which was launched in July 2014.

Personally, my wife Mary and I know the full impact a rare disorder can have on a family. As the parents of two children, Saoirse and Liam, both of whom were diagnosed with Batten disease, an ultra-rare genetic neurological condition, we know only too well the challenges of going through the system with a rare disorder in the family. The costs to us, personally, have been immense and cannot be measured, as we laid Saoirse to rest in January 2011 at the age of five and our boy, Liam, joined his sister in May last year when he was also five.

The national rare disease plan for Ireland will only ever be as good as its implementation. Each year, we celebrate international rare disease day on the last day of February, and this day should and will also need to focus on the community affected here in Ireland and in bringing more public and more state awareness on rare disease. As has been mentioned, this affects between 6% and 8% of the population during their lifetime. Such a significant number of the

population has not received the appropriate acknowledgement and focused attention it deserves within the health service, social care and education systems of this country. Individually each rare condition is, by definition, rare but, collectively, it is very common to have a rare condition.

As my colleagues from the rare disease task force will acknowledge, it is important to have a co-ordinated approach and strategy into developing research, technologies, therapies and collaboration for rare disorders. However, we must not forget the practical elements which need to improve and support those living with the conditions right now. Timely prevention and diagnosis of rare disorders is essential. Without accurate diagnosis, appropriate screening programmes and targeting of diagnostic tests, patients and families cannot access effective treatment and therapy or manage the condition and symptoms appropriately. That would give such people empowerment.

Even with the better-known conditions, there is a serious impact on the quality of life on the family unit, with increased stress, anxiety and financial burdens imposed just because a loved one has a rare disorder. This has serious implications for a person's life expectancy and quality of life and often results in an inefficient use of HSE resources. We need early diagnosis, with critical eyes from GPs and other care providers. We also need awareness of guidelines with regard to genetic conditions; more genetic counselling; empathetic diagnosis and follow-on support for the family unit that is currently lacking; automatic provision of information on conditions and symptoms, with specific referrals for rare disease support groups if available; access to information about treatments, research, therapy and third-party support, which may include clinical trials; and acknowledgement of the patient's and carer's expertise, which deserves respect and recognition from professionals.

The psychological issues encountered by those with a rare disorder range from the need for early assessment or educational assessment, as well as psychological interventions at different points in life. People and families with a rare condition can often feel isolated. Mental health issues can arise, affecting the patient, the family and carers. There are strong arguments to initiate an automatic procedure for access to a psychologist for the family and the person when a rare disorder is diagnosed. More effective use of the Internet is required, with the development of a credible source of information specifically needed. Online support groups or phone contacts can also be essential for those who experience social isolation due to lack of transport or carers, and for those who do not have a specific rare disorder support group. Our research with Trinity College, Dublin, indicates that the Internet offers people with the same rare disorder, for which there may not be a representative patient organisation, the possibility of accessing credible information and in establishing online support networks. However, this process needs to be managed correctly, with good oversight and governance.

Some ultra-rare conditions do not have access to any specialist centre or specific support group so the difficulties of lack of information, expertise and support are heightened. Overall, there is a lack of support for rare disorder patients with their medical and non-medical issues. Additionally, patient support services will need to have the ability to react urgently for some rare diseases, such as life-limiting, progressive degenerative and neurological conditions, which can cause drastic changes in the patient's well-being and care needs. I witnessed this twice with my two children, and the system does not react fast enough. Specialised social and support services for many sectors of society affected by more common health conditions have proven to be very successful and beneficial in improving the quality of life for those affected. Within the rare disease community, the overall availability of such services is severely limited, with less than a handful of the more common rare conditions receiving a similar level service.

Cystic fibrosis is an example.

The national rare disease plan identifies the need for the development of specialised social support services on a national and community level for those affected by rare conditions, which includes patients, family members and carers. However, the strategy on how to deliver these services is not known at present. Residential respite care facilities are currently limited in capacity and under enormous pressure, and there is a significant urgency in invest in new strategically located infrastructure across Ireland to provide respite care, appropriate long-term care and, where necessary, palliative care services. One of the most important and critical purposes of respite care services is to give family members time and temporary relief from the stress they experience while providing extra care for a family member affected by a rare disorder.

Therapeutic recreational programmes encourage social and personal development. In some instances the combination of a number of therapeutic programmes with respite programmes can bring added benefit for the patient and the family. Many rare disorders are life-limiting, as we have mentioned, and can lead to many young patients not making it to adulthood. Approximately 80% of all rare disorders are of a genetic nature, with 60% of those affected by rare disorders having a significantly reduced life expectancy. The majority of those affected by rare disorders are children, and 30% of children do not reach past the age of five years. When palliative care services are required for children, it is already and emotional time for the family and carers alike, and additional support services should be provided during this time. The development of procedures and policies, consistent with best practices, should be improved for children affected by rare disorders. I urge the committee, the Government and the State agencies to support the development of new initiatives, such as our own Liam's Lodge facility in Kerry, which will serve Munster and the mid-west, and be a national centre for respite.

For those affected by rare disorders, particularly those who have the possibility of longer life, social integration, including educational development, should seamlessly integrate into the patient's daily life and support their psychological and educational development. The systems must revolve around the patient and not the other way around. Those diagnosed with a rare disorder, their parents, carers and relatives are very frequently left in difficult positions with regard to employment and financial security. This is primarily down to no legal framework protecting the rights of those individuals to maintain employment or benefits due to delays in access or availability of services and support from the State. Some social frameworks exist around educational and early intervention services but access to these services is severely limited for persons with rare disorders, and particularly within the educational system. I acknowledge that this is outside the primary focus of this committee but it is members' responsibility and that of their colleagues that proactive steps are taken, as the correct result will have a positive impact on the health of the patients and family members.

The first national plan for rare disease for Ireland is an important step in the right direction. The successful implementation of the plan will require a well-structured strategy and a set of integrated, comprehensive policy actions to be developed, so as to reach the objectives and 48 recommendations outlined. It will also require a clear focus on best practice guidelines when considering clearly defined policies and protocols; a well-declared timeframe, with declared objectives and goals; new and effective changes to legislation; defined stakeholders and responsibilities; and constant monitoring and evaluation of the implementation process. I suggest this should be supervised by an oversight committee. There should also be allocation of appropriate instruments and resources; identification of opportunities and appropriate European or international partners in development, collaboration and co-operation with respect to cross-border

responses to some rare disorders; and the creation of centres of expertise.

A clearly defined management of change process is highly recommended and thorough analysis is also required to identify present resources and existing shortfalls. The services of the State should be benchmarked with our European counterparts. It is also of the utmost importance to ensure that those rare disorder patients currently receiving care, services and support are provided for during the transition from where we are now to where we must be before the second plan is published in 2019. As Dr. Devlin indicated, it is a five-year plan. The stakeholders of the oversight committee must include the previous members of the steering group at a minimum.

Ms Avril Daly: I am the chief executive of Fighting Blindness, a patient-led organisation founded on 25 February 32 years ago, funding research and treatment for conditions causing rare forms of sight loss. I also volunteer as the chair of the Genetic and Rare Disease Organisation, GRDO, the Irish National Alliance for Rare Diseases on the national steering committee developing plans for rare diseases. I am also a person living with a rare form of sight loss. Today I also represent the National Rare Disease Task Force. This voluntary forum was formed in 2011 by patient groups to support their representatives on the steering group. I will present to members the vision and underlying principles of The National Plan for Rare Diseases 2014-2018.

The recognition of the complexities of rare diseases, the provision of information to allow for developments in research leading to improved, efficient and prompt services and better outcomes for patients were critical areas of concern when we commenced our work on the steering group. As we have heard earlier, there are over 6,000 rare diseases that we know of, with more being continuously described. This means there is greater need for a dedicated facility to co-ordinate the information flow, the provision of expertise locally, regionally and internationally through a national co-ordinating centre. This centre or office will be responsible for developing appropriate pathways of care and would undertake epidemiological work, be equipped with the appropriate IT infrastructures and co-ordinated information systems to allow for the development of registers that will enable better planning of services and facilitate the development of research infrastructures.

The research dimension in this field is an area of concern for patients and other stakeholders as funding dedicated to research on rare diseases remains limited. However, the value of fundamental rare disease research cannot be underestimated on moral, scientific and economic arguments. Rare diseases represent a huge burden on the individual, the community and the State, resulting in an unacceptable unmet clinical need for thousands of Irish patients. Scientifically, rare diseases often serve as models for more common diseases and the complexity of rare disease often requires multidisciplinary approaches. Basic, fundamental research is the engine of discovery; it generates new knowledge, drives innovation, and underlies all past and future breakthroughs.

The establishment of the Orphan Drug Acts in both the United States and Europe has generated incentives for the translation of basic research towards product development, and led directly to the growth of the biotech industry as we know it today. Put simply, the medicine of tomorrow depends on sustained investment in basic research today. We are concerned that in an environment where the State prioritises research leading to immediate economic impact we neglect the development of our pipeline for future discovery. This impacts not only the provision of better medical care in the years to come but on our position as leading educators and our place in global innovation.

An area that requires more structure and sustained support is the development of rare disease registries, which are crucial building blocks for sound policy on rare disease. Where well-implemented registries exist, there is better understanding of prevalence, impact and the likelihood of developing a treatment for the rare disease in question is increased.

Furthermore, the collection of patient data creates better standards of care and dramatically improves patient outcomes and life expectancy. The development of an all-Ireland network of rare disease registries covering the island of Ireland should be developed to meet this objective, but it must be resourced correctly. This network will enhance and standardise rare disease registries in line with the Health Information and Quality Authority, HIQA, draft guidelines, data protection legislation and international best practice, facilitating the interoperability and harmonisation between international rare disease patient registries. This function needs to be supported by the new national office for rare diseases.

Such infrastructure will enable more Irish success in European research grants and more involvement with international networks such as E-Rare, and facilitate engagement in the rare diseases aspects of BBMRI-ERIC and the ECRIN-ERIC. Systems must be put in place to enhance the utility of data held in relevant health service-based information systems, including hospital records, laboratory cytogenetic and molecular genetics data.

We believe that the development of any future information systems must provide for a rare disease code in a patient record so that all people with rare diseases may be easily identified. This was singled out as a patient priority in the consultation on the national plan. The development of a rare disease ID card linked to a person's PPSN should be explored once the provisions of the proposed information Bill have been enacted and promulgated.

With respect to pregnancy, where family members are known to be at risk of being carriers of genes for rare diseases they have appropriate access to pre-conception genetic testing and counselling, which can inform them about the risks involved in becoming pregnant. The Health Service Executive Governance Committee/Group on Newborn Screening within the integrated services directorate needs to consider the population benefits of newborn screening, including whether programmes need to be expanded or modified, and the need for carrier screening. The Department of Health needs to also provide a policy framework for population-based screening programmes.

Patients are currently very concerned about the governance and clinical guidelines for send out tests, which should be audited yearly on the quality and diagnostic yield of tests sent out from non-hospital sources in order to minimise wastage. We are also concerned that Ireland no longer has a national centre for medical genetics.

The national clinical programme for rare diseases, through the soon to be opened national office for rare diseases, must be supported in the development of the clinical and organisational governance framework that will underpin care pathways and access to treatment for rare disease patients, particularly in the context of the transition from paediatric care to adult care. It is important to remember that some children do not grow out of conditions; they grow with them.

National centres of expertise in Ireland need to be identified for groupings of rare conditions, based on clinical need and built on foundations already established. It is also important that broader clinical guidelines take account of the requirements of rare diseases. The potential for co-operation on an all-Ireland basis must be realised. Centres of expertise must be integrated into national funding planning, with provision for adequate staffing for multidisciplinary

care, as well as sustainable research infrastructure for clinical investigation in addition to competitive research.

The role of the designated centre of expertise in Ireland should include research relevant to rare disease, in particular with regard to registries, as already mentioned, health service and translational research. The capacity of Ireland's five clinical research facilities to engage in rare disease research nationally or in collaboration with international collaborative research must be enhanced.

As a steering group we recommend that guidelines, monitored by HIQA, be developed on coding and recording of rare diseases within relevant Irish health data systems that are consistent with European and global recommendations. The implementation of the Health identifiers Act is urgent, and the forthcoming health information Bill is vital to that.

Centres of expertise should be supported in seeking recognition as EU designated centres of expertise or associated national centres in European reference networks for rare diseases, RDERNs, according to the timeframe, framework and standards currently being developed at European level through the complementary work of the European Union Committee of Experts on Rare Diseases, EUCERD, and the EU cross-border healthcare directive.

Guidelines need to be developed in palliative care provision to address the complex and multi-systemic nature of many rare life-limiting conditions, and account must be taken of the particular needs of children with rare disease in this ongoing programme of work. In order to improve and understand rare diseases among the next generation of medical professionals, appropriate modules relating to rare diseases need to feature within all undergraduate and postgraduate programmes. This could also be addressed through professional bodies with the support of all stakeholder groups, including patients and their families.

We are concerned about the lack of development of strategies concerning the provision of high technology and orphan therapies. The HSE has committed to the development a working group to bring forward appropriate decision criteria for the reimbursement of orphan medicines and technologies, but this has not yet been enacted.

We recommend the preliminary economic evaluation of current activity and costs for orphan medicines and technologies for rare disease patients across all hospitals settings. The approach should include an assessment system similar to that for cancer therapies established under the national cancer control programme and link with the clinical added value of orphan medicinal products, CAVOMP, at European level. Applications for the use of orphan medicines and technologies in hospitals should be dealt with in the context of a national budget, rather than through individual hospital budgets.

An annual report documenting the use of both existing and new-to-market orphan medicines and technologies in Ireland and a summary of applications received and decisions relating to those applications should be made publicly available. The existing horizon scanning between pharmaceutical companies and the HSE, including clinical value assessment authorities, should be enhanced to improve information available regarding orphan medicines in the pipeline and the future needs for these medicines.

We ask that the HSE applies a set of guidelines on the prescribing of orphan medicines and technologies in Ireland and evaluate clinical outcomes regarding use of orphan medicines. We recommend that early dialogue between the HSE and companies running clinical trials in

Ireland with Irish patients where licence approval is imminent. Sponsors could be offered an incentive to run trials in Ireland, increasing access to innovation for Irish patients.

The principles of patients' empowerment are integral to all aspects of the national rare disease plan for Ireland, both now and in the future, in recognition of the fact those patients and their carers require significant clinical and non-clinical support. We are calling for an oversight implementation group of relevant stakeholders, including patients groups, led by the HSE to be established to oversee and monitor implementation of the national rare disease plan's recommendations and associated key outputs. The HSE will report to the Department of Health using key performance indicators on a periodic basis in accordance with reporting requirements under the national plan. The European Union has mandated EUCERD's KPIs and that Ireland must report on these by the end of this year. There needs to be an overall review of the national rare disease plan prior to development of the next plan in 2019.

As patients and representatives of patients, we welcome the recent commitment to the establishment of a NRDO. This will facilitate the coordination and timely access to centres of expertise nationally and internationally, and will provide information regarding new treatments and management options for patients and medical professionals. The information outlined highlights how the office will be a cornerstone in the facilitation of many elements of this plan but it must be sustained and built on into the future.

We remain in a time of economic constraint. We are aware of this, but strongly believe that the name "rare" should not be understood to mean few. Investment in this plan will lead to considerable savings for Exchequer and not just in the long term but today and tomorrow. Patients on the diagnostic Odyssey are as we speak being moved from department to department in hospitals or between hospitals and between counties. They are lying on trollies, helpless and untreated. These are the people who will benefit from the implantation of the published plan.

EU structural and investment funds are instruments used to support economic social and territorial disparities that exist across the Union. They are designed to promote greater cohesion in its regions. They, therefore, form the basis of the EU cohesion policy, providing the investment framework to deliver the Europe 2020 objective of "smart sustainable and inclusive growth". A key area of highlighted in this instrument is "social inclusion". The amount to be invested between 2014 and 2020 is €352 billion, which represents one third of the total EU budget for the next seven years. Each member state is allocated a portion of the global amount. Why could these funds not be considered for a programme of this nature? The focus of this plan is to bring those patients and their families living with rare disease out of the shadows and margins of society by enabling them to become active citizens who will contribute, day by day, hand in hand, to a more inclusive, cohesive and productive society.

Ms Helen Byrne: I thank the committee for the invitation to attend the meeting to discuss tackling rare diseases. I am joined by my colleague, Dr. Colm Henry, national clinical advisor and group lead for acute hospitals. The Department of Health's first national rare disease plan, A Rare Disease Plan for Ireland 2014-2018, was published in July 2014 in line with European Council recommendations. The National Clinical Programme for Rare Diseases, NCPRD, in Ireland was initiated in December 2013 under the national clinical strategy and programmes division of the HSE in collaboration with the Royal College of Physicians Ireland with a view to improving and standardising the quality of care for patients with rare diseases in Ireland. Professor Eileen Tracey, clinical lead, and a programme manager were appointed for the programme. The key initial priorities for the clinical programme are to assist with the identification and designation of national centres of expertise, with a view to having the option

to join European reference networks, to facilitate the development of a NRDO in line with the European information portal for rare diseases, Orphanet, and the development of care pathways for the more common rare diseases.

The NCPRD developed a business model for a NRDO in 2014, and applied for entry to the EC rare diseases joint action. Funding for 2015 has been allocated for the following staff for the office: a full-time information scientist, half of which will be funded by EU for three years; a full-time genetic counsellor; a consultant geneticist, 0.3 whole-time equivalent; and administrative support, 0.5 whole-time equivalent. Recruitment is under way for the information scientist. The four main functions of the NRDO include building information on the availability of expertise in Ireland for rare diseases and making this information available on the Orphanet web site as a resource for both patients and clinicians; a helpline for patients with access to a genetic counsellor; supporting the HSE in the mapping and validation of centres of expertise in Ireland; and in the long term, a role in rare diseases surveillance.

The NRDO will be based in the Mater Misericordiae University Hospital pending the establishment of the new national children's hospital. It is hoped to launch the opening of the office in late in the second quarter of 2015. The core benefit to the population and the State of the establishment of a central rare diseases office will be the streamlining of access to diagnosis for patients and appropriate quality treatments with increased efficiencies in the delivery of care. Professor Eileen Tracey, clinical lead, is the designated co-ordinator for the island of Ireland and the designated person to participate in the EC Joint Action on Rare Diseases. Her work will include establishing and validating the Orphanet information and function in Ireland through the NRDO and the dissemination of information nationally for rare diseases. Her participation in this project is due to commence in May 2015.

The HSE NCPRD-acute hospitals division is currently embarking on an exercise to identify and designate existing centres of expertise for diagnosis, multidisciplinary care, clinical research and training in rare diseases according to accepted European criteria. The initial sites to be reviewed for the three major services being considered for 2015 and early 2016 are clinical genetics in Our Lady's Children's Hospital, Crumlin; inherited metabolic disorders in Children's University Hospital, Temple Street; and hereditary coagulation disorders in St. James Hospital and Our Lady's Children Hospital, Crumlin. This work will also inform the identification of centres of expertise abroad for patients whose disorders cannot be fully served in Ireland in collaboration with European reference networks and the development of processes for patients to access this expertise for timely diagnosis and care.

As a member state, we have obligations under EC Directive 2011/24/EU, application of patients' rights in cross border health care. This directive is of particular relevance to patients with rare and ultra-rare diseases. This directive on the application of patients' rights in cross-border health care will strengthen co-operation between highly specialised healthcare providers across the Union by the establishment of a system of European reference networks. Establishing such networks of highly specialised healthcare providers will help to provide affordable, high-quality and cost-effective healthcare to Irish patients requiring a particular concentration of resources or expertise which may be available in Europe but not specifically in Ireland. European reference networks for rare diseases should serve as research and knowledge centres, treat patients from other EC countries and ensure the availability of subsequent treatment facilities where necessary. The HSE has established a national contact point to facilitate information regarding the treatment abroad scheme and the cross-border care directive. Further information is available on the HSE website. It is hoped that the national rare disease office will, in time, also

assist with and facilitate the national contact point for queries involving rare disease patients.

A recommendation of the national rare disease plan was for the national clinical programme for rare diseases through a national office for rare diseases develop the clinical and organisational governance framework that will underpin care pathways and access to treatment for rare disease patients, particularly in the context of the transition from paediatric care to adult care. The national clinical programme for rare diseases has now assembled a sub-group to develop a model of care for this work.

This concludes my opening statement and together with my colleague we will endeavour to answer any questions Members may have.

Deputy Billy Kelleher: On foot of reading the rare diseases report it is evident we have a lot to do as we are starting from a very low base. What is needed is a commitment that it should be done and resourcing of same to ensure it is done. That in itself will be a huge undertaking but it should not deter us from doing what we need to do and what is right. A few issues arise from the report and the presentations. We talk about rare diseases. They may be rare but, collectively, they are substantial in number. This is evident across the European Union also. I often wonder why there is not more transnational collaboration between national bodies and research institutes. Ireland has a population of less than 1% of the European population. Will we have enough critical mass to do our own research and the necessary support and capacity? I wonder if we are slow and reluctant as a nation to collaborate in a meaningful planned way through national governments and national agencies on a one to one basis. There is a great deal of work that could and should be done. That is not only in the context of also in the area of treatment and providing supports. Certainly the national rare diseases offices and its equivalents across Europe must come together in a more defined targeted way.

Research is scattered. However, there must be some way of pooling it together between industry, hospitals and universities coming together in a more collaborative way. We all understand that research is secretive in nature initially but there should be a better pooling of resources.

I spoke in the House last week on the national centre for medical genetics. I am still unclear about it. What has happened to it? Why has it gone? I would have assumed at this stage that we would have understood its importance, through the advances in medical science, embryology, stem cell research and so on and the assessment of genetics and chromosomes, and that we would be trying to enhance our national genetics office, not trying to undermine it. What is happening? When I raised it in the House, to be truthful I got a very poor answer. If we are serious and if there is a commitment to a national rare diseases office one of the singular largest elements of rare diseases is the whole issue of genetics. Therefore, genetics and genetics testing should be a central focal point. I would greatly appreciate answers on that issue.

I have been looking at the list of rare diseases. The vast majority of them, 80%, are genetic. I would appreciate some information on the reason the national rare diseases office has disappeared, was undermined or chastised. Something has happened to it as a centre and we should address it.

Many Deputies and Senators would have been on the journey with families from time to time where a family gets a diagnosis of a rare disease and the system is unable to say exactly what it is. There may have been some time before the diagnosis but when it comes the system cannot support them well initially and the family is left very much on its own, other than good-

will support. Often they trawl through the Internet and reams of research data to try to find answers. It is a cling to hope and it can be a very difficult journey. In that context while we have a proposal for a national rare diseases office, is there any way in which to humanise the bureaucracy through the system? I appreciate it is difficult to humanise bureaucracy. It can be harsh at times, not intentionally, because of its bureaucratic nature, particularly with families and individuals with rare diseases in practical areas, such as the kiting out of houses, and the willingness of a bureaucracy or the HSE to help the local general practitioner get that extra training to assist a patient who may be the only one he or she will have with that rare disease. We should look at the issue from the top down but also assist general practitioners and nurse specialists in communities from time to time when one patient or one family has a specific illness or disease. If that were done it might help alleviate some of the pressures on families from time to time.

Deputy Caoimhghín Ó Caoláin: I welcome not only the panellists but all our guests. It was a wonderful idea to dedicate a day in the year to international rare diseases because it has allowed us to have hook on which to hang this focus. The focus might not happen only but for it. I thank all my colleagues on the committee for the support annually in this address.

In her presentation - I am not making a distinction one from the other as they were all very informative - Ms Avril Daly talks about a dedicated facility. When she talks about a dedicated facility she describes a facility to co-ordinate the information flow, the provision of expertise locally, regionally and internationally to a national co-ordinating centre. The centre or office will be responsible for developing appropriate pathways of care and would undertake epidemiological work, be equipped with appropriate IT infrastructures and co-ordinated information systems, to allow for the development of registers that will enable better planning of services and facilitate the development of research infrastructures.

Does that equate with the national office for rare diseases? It should but I do not know from the outline. I ask Mr. John Devlin and Ms Helen Byrne as I am not certain that what Ms Avril Daly has said, and that I believe is needed, is what is actually in the plan. We received correspondence here and my colleague Senator Colm Burke referred to it a week ago that the national office is getting ready. It cannot happen soon enough.

The staff for the office would be one wholetime equivalent information scientist, one wholetime equivalent genetic counsellor, 0.3 wholetime equivalent consultant geneticist and 0.5 wholetime equivalent administrative support - I wonder what half of it would be? Given what is outlined here I hope Mr. John Devlin will be able to advise that this is just the initial commitment because it does not equate with what I believe - and what I believe all who are interested in a real and serious focus on rare diseases know - is required. I hope that what Ms Avril Daly has described is where we are going to get to. I would appreciate if he would advise what is the real plan relating to the national office because there are so many aspects of what is needed that are not signalled at this point.

The registries are vital. I strongly concur with an all-island approach given that I have spent my lifetime 4 miles from the Border. The people I know within my catchment area all bear the same names, face the same problems and are part of the same genetic make up as each and every one of us. Many of the representative organisations are structured on an all-island basis and it is very important that this co-operation is there or we will not get the full picture. We need to get an all-island picture. Will the national office help to develop these registries? I would expect that some of that work would require more than the scientist and the counsellor. The administrative staff would be taking on a lot of this work in respect of the IT requirements.

This database is a vital part of everything that is needed.

We no longer have a national centre for medical genetics. I understand that Our Lady's Children's Hospital, Crumlin has downgraded the national centre for medical genetics to a department within the hospital. Is this the case? Can we get information about its status? Given the fact that the greater number of rare diseases are genetically based, why are we seeing the downgrading, if not the elimination, of the only medical genetics department we have had up to now at a time when we are seeking to seriously address the deficiencies relating to rare diseases through the establishment of a national office?

There seems to be no oversight relating to the national rare disease plan for Ireland. There is no oversight committee. Is this under consideration? Will it be introduced? With all of the attention, why was there almost no referencing to national rare diseases, let alone the office in the HSE service plan for 2015? This baffles me and I am still at a loss as to why it would not have been a critical part of it if this was a serious attempt to address the issues surrounding rare diseases.

I thank Mr. Heffernan in respect of all the work that is being done. As a parent with direct experience, his testimony today is very important and striking. There are many points I would like to explore with him but there is one he referred to that is particular to him. It relates to what the diagnosis of a rare disease within a family means in so many ways. It is not only the terrible tragedy of the onset of that news but, as Mr. Heffernan says, the consequential difficulties that present relating to employment and financial security for him, his wife and his family. This area needs to be addressed and we need a joined-up response to situations of particular difficulty and hardship that occur when a rare disease presents in a family. I thank Mr. Heffernan for drawing attention to that this morning.

Senator Jillian van Turnhout: I welcome all the witnesses. The fact that we have a day each year to focus gives this committee an opportunity to focus on the issue, although we have other encounters during the year. In preparing for today, I looked back at the meeting of 15 January when the Huntington's Disease Association of Ireland appeared before us and raised some very important issues, as the witnesses today have done. Ms Daly's words about how rare does not mean few rang true. When one looks at the figures, one sees that they are significant and that it is a significant health issue. When I see the number of children who are directly affected due to the prevalence in childhood and the number of children who die before their fifth birthday, it makes me wonder where we should be going. This is where I come to Mr. Heffernan's presentation. I am a bit like him in the sense that I want to see implementation and action. It reminds me of the last time Ireland was before the United Nations Committee on the Rights of the Child. The committee said we were very good on paper but had issues about implementation. This is where I would like to focus. The Children's Rights Alliance produced a report at the time called From Rhetoric to Rights that dealt with how we ensure these rights are realised.

Mr. Heffernan raised a point about the oversight committee and said that the committee must include the previous members of the steering group as a minimum. Why is there a question over that? Perhaps there could be an explanation as to what is happening in the process of the oversight implementation group. It is very important that it would bring together the expertise so I would be interested in knowing more about that. In reading the plan, I assumed the implementation group was there because we need to move to the implementation phase. I would like to know what is happening because it is very important.

I raised the issue of genetics with the Huntington's Disease Association of Ireland. Is a

genetics unit planned within the new national children's hospital. My understanding is that there should be but I would like to know whether organisations individually or the HSE have been involved in the scoping relating to rare diseases and the needs assessment for the national children's hospital relating to rare diseases. This is not just in the location because I understand based on a visit by the committee to the hospital that it will have satellite and services linked to the hospital. I am looking at the wider remit of the national children's hospital and the co-ordinated supports it will provide rather than just the physical premises. Have organisations been involved in any scoping or needs assessment? It is critical that they are involved at this stage.

Another issue that was raised with me was the need for a credible source of information. What developments are happening on that side? We all turn to the Internet even though we know that we will not get a diagnosis but sometimes it is the only place where one may get answers. I know that when we visit medical services and medical services try to convey information to us, we do not always take in all the information so we go home and try to research and find out more. Rare diseases by their nature are rare so how do people find that information in a co-ordinated way? The witnesses have raised those issues. What steps are there for Irish parents because I see the pressures on the support groups and NGOs? We get testimony after testimony about the important work that is being done by advocates working in the area, usually parents who have been through the process and are, therefore, able to translate and understand what this parent or individual is going through.

Mr. Heffernan spoke about Liam's Lodge which is to be applauded. Has there been any State involvement or interest in what the Saoirse Foundation is trying to do in developing Liam's Lodge? Anybody I know sings the praises of BUMBLEance. We must all strive harder to provide this environment at what is potentially such a scary time for a child. We have raised with the national children's hospital the fact that it is an issue for children throughout their childhood. We are not just dealing with very young children. Teenagers will also have rare diseases.

Senator Colm Burke: I thank the witnesses for coming before the committee and commend them on their dedication and commitment. When I first came across this issue three and a half years ago, it was a case of everyone fighting for his or her own corner. It is important that we work together to help families coping with a rare disease.

Has the 2011 EU directive on cross-border health care been transposed? I understand it has not been transposed but I am open to correction. If it has not been transposed, is there any indication of when this will happen? In some cases treatment abroad presents no problems and it works out very well, but in others it can involve battles lasting for two or three months which leave families traumatised. The experience varies among HSE areas and from one medical condition to another. Have any measures been put in place to make the process more efficient and less time-consuming? Something needs to be done because the last thing a family in this situation needs is indecision. They have already come through a traumatic experience and it is important they get a decision on accessing care abroad.

Are Mr. Heffernan and Ms Daly happy with the availability of respite care based on their experience of dealing with families or can more be done to improve the availability of care? I have come across a number of families who are providing care on a 24-7 basis find they cannot access the supports they need. This is why respite care is fundamental to families.

Senator John Crown: I welcome the witnesses and beg their indulgence if I sound tired and cranky. It recently dawned on me that I am well into my 22nd year of being back in Ireland

and I am still trying to grapple with the issues arising in regard to the common disease of cancer. We are not doing very well with the common diseases and, while that does not mean we should not try to deal with the rare diseases, I caution the witnesses on a certain issue of prioritisation. What made the difference in cancer care in Ireland was not the bureaucracy called the national cancer control programme but the now Minister for Finance, Deputy Noonan, who said in his first speech as Minister for Health in 1995 that if one of his relatives had cancer he would not allow him or her to go to certain hospitals in this country. I gently remind him this was an ever so slight plagiarism of something I had said the previous year but I was honoured to be plagiarised in that manner. He also spoke about appointing cancer specialists. That is what made the difference. Cancer mortality rates started to improve, particularly in breast cancer, which was probably the area that was most deficient in terms of treatments not being made available in Ireland. The national cancer control programme had nothing to do with these improvements. It streamlined certain procedures but it did not materially affect their outcome.

I do not want the witnesses to take this the wrong way but somebody coming away from this meeting might get the message that the country with the smallest number of paediatricians, adult and paediatric neurologists or adult and paediatric dermatologists, nearly non-existent paediatric endocrinologists, despite our obesity epidemic, and very few adult endocrinologists needs to prioritise the management of uncommon diseases by setting up a new bureaucracy and a registry. We may need them but we are not going to fix the problem until we grapple with the fundamental structural problems of Irish health care, which are no closer to being fixed than when I first started to point them out more than two decades ago.

I have not formally studied which places are most successful in dealing with individual rare diseases. Typically in the case of cancer, a leading medical school-based centre develops an interest in a programme and has sufficient resources and personnel to allow sub-specialisation to develop. This is unlike the situation in Ireland, where we expect paediatricians to look after asthma in children and then provide a specialist service to those with uncommon or rare diseases. We must also grapple with the fundamentally irrational nature of having six medical schools in a small population. That is twice the European average and approximately two and a half times that of the United States on a *per capita* basis. None of them is remotely excellent. I can highlight individual cases of people who did brilliant jobs in difficult circumstances in terms of building internationally recognised units, but we do not approach the issue systematically.

I have to say something about the national centre for medical genetics because I have a dog in this fight. We use genetics services, primarily historic, for breast cancer patients where we notice a suspicious family history and want to find out if the patient has a genetic syndrome which would be relevant to the her health as well as to other members of her family in terms of early diagnosis or prevention of cancer. The reality is that we did not stop having a national genetics centre; we never had one. We had a very good academic centre that Professor Andrew Green, to his extraordinary credit, built mainly on the basis of research funding. It attracted very limited statutory funding. He and I had a gentle historical disagreement because we both applied for a research grant which ended up getting his unit up and running on a large scale. However, he has been providing a genetics service on a national basis without the kind of structure or ring-fenced budget that a national centre should have. When Crumlin was in its most recent paroxysm of financial turmoil, it re-examined the centre's catchment area. This has happened to me in St. Vincent's. I have been told over the years that I should not take patients from Galway because St. Vincent's was not a national centre for any of their cancers and they could instead go to their local hospitals. When I pointed out that the local hospital did not have an oncologist, the response I received was "tough". Doctors behaving ethically have had to over-

come very unethical administrative impediments to providing services to patients. While there is much to be said for proper registration procedures and connecting to international networks to tap into expertise, it is also necessary to advocate for the reform of services, including the number of doctors and structures of medical schools.

Deputy Sandra McLellan: I welcome the witnesses and thank them for their comprehensive presentations and for sharing their personal experiences. With regard to staffing at the rare diseases office, an administrator on 0.3 of a whole-time equivalent is only working 13 hours per week. That is not even sufficient time to open e-mails or letters. Given that there are between 6,000 and 8,000 rare diseases, it is shocking.

The witnesses clearly outlined what needs to be done. It all sounds very simple and extremely logical. We just need to get to that point. I was very interested and grateful to hear about the life-changing implications for families and how these situations can affect people financially. There is a major lack of understanding as to how rare diseases can impact a family, which has implications for how that impact is assessed.

According to the national rare disease plan, between one in four and one in five rare disease patients wait more than a year for assessment by an appropriate specialist in rare diseases. Respondents, not surprisingly, felt an appointment with a specialist should follow within three months, which is quite logical. For people with rare diseases, the trauma of not knowing what is wrong with them is made much worse by having to wait a year to see a specialist. That is not in any way acceptable.

I have a number of questions regarding cystic fibrosis treatment which I am hoping one of the delegates might be able to answer. How many children in Ireland are being treated under the trials of the ground-breaking drug, Kalydeco? We know this medication can lead to significant improvements for patients with a particular mutation of CF. Can any of the delegates provide an update on the trial participants' progress? Have there, for example, been any side effects? Are there any plans to extend the provision of the drug to other CF sufferers? When are the trials scheduled to continue and does length of time of treatment vary from child to child or from age group to age group? How will it be determined when a person should be taken off Kalydeco? I am not sure whether the delegates can answer those questions, but I would be delighted if any of them can.

Dr. John Devlin: A great number of issues have been raised. I agree completely with the sentiment that rare diseases are, in fact, common. That is very much part of our thinking. I likewise agree with what Senator Crown said regarding clinical focus. It struck us very early on that there was a lack of clinical focus with regard to rare diseases. In the case of cancers, a number of very prominent cancer specialists like Senator Crown were able to take the initiative and from that we got a cancer programme. We had the setting up of things like the clinical research network, iCore, and so on. That type of focus was lacking when it came to rare diseases. This was clear to us very early on, as I said, and we did not really want to wait to publish a plan. We decided one of the first things we had to do was to establish a clinical programme on rare diseases, and that led to the appointment of Professor Eileen Treacy. We talked to the then clinical director of the Health Service Executive, Dr. Barry White, and he agreed to do it. It was very important to achieve that clinical focus which was lacking. It was not that patients were not being treated, but no common themes were emerging and what was being done was happening in isolation in hospitals and so on. We did not consider that a good thing.

In terms of how we moved things on from there, we can talk about structures and all the rest

of it, but our key priority initially was to start making things happen. While we did not have an oversight committee, we did have a number of meetings with the HSE, particularly around the establishment of the office. We considered this a priority to get us going. To do that, we needed to take several actions. First, we needed to secure and tie down the funding from the EU and then match it. That was the basis for the proposal Ms Byrne has outlined today, and it sets out the requirement for further staff and so on.

Deputy Kelleher asked about collaboration. That is very relevant because this is a pan-European issue. Mr. McCormack and I are members of the European Union Committee of Experts on Rare Diseases, EUCERD, where all these issues are debated. They are very practical issues around the development of the European reference networks, how we will tie into them, what the quality criteria for our centres of expertise will be and so on. That work has been a little slower to progress than we expected, but we should be in a position by the end of the year where countries like Ireland will be able to start designating centres. That is reflected in the work Ms Byrne has outlined regarding the three national centres for medical genetics, inherited metabolic diseases and hereditary haemochromatosis. We may be in a position to designate them into Europe.

Regarding training and medical schools, that is dealt with in some detail in our report. It is not that these conditions are not part of the medical curriculum, because they are, but we need to do a bit more. We need more information on rare diseases. Deputy Ó Caoláin referred to registers. Our report lists 40 different sources of information we had to reference to in order to build up a compendium of cases of rare diseases. Within that, there are some ten or 12 registers. Some of them are familiar resources which operate under a particular code of governance and standards, such as the National Cancer Registry and the Cystic Fibrosis Registry of Ireland, while other registers are less developed. We also have a very strong congenital anomaly registry, EUROCAT, in this country. We had a discussion among our group about the best thing to do in terms of bringing all these sources together, whether by establishing a single national register or something else. We concluded that it was probably more important, in the first instance, to develop a network of registers in order that we can apply common standards of governance and ways of working across those registers rather than simply moving straight into a national register for rare diseases without having all those standards agreed.

That is very much part of our thinking and it is one of the functions of the office. Of course, our staff are not the only ones who will contribute to this. Behind the other registers are other personnel and we fully expect they will contribute to the work, including National Cancer Registry staff, EUROCAT staff and so on. It will also require the support of the patient organisations, which have also developed registers. Legislation is important too, and some of it has been enacted or will soon be enacted. The Health Identifiers Act 2014, for instance, will help us out. Even more important will be the health information Bill, which is intended to provide an opportunity for registers to be prescribed. Once that happens, the registers can operate in accordance with common operating procedures and standards, which will make things much easier.

The delegates from the HSE might be better able to answer the question about the national centre for medical genetics. When we are talking about designating national centres, the work that is happening, the mapping of care pathways through the national clinical programme and so on, will bring us to a better understanding of what we need, what that particular centre might look like and how it will operate within a European context. My colleagues from the HSE might wish to say more about that.

I am sorry to tell Deputy McLellan that we do not have the information she requested re-

garding cystic fibrosis medication. We will see if we can secure it for her.

I will conclude by referring to research. A good amount of money is going into rare disease research. Indeed, approximately €6.8 million came from the Health Research Board, HRB, alone from 2010 to 2014. The research covers a variety of different rare diseases. Last November the Minister for Health announced a further €1.9 million in funding for rare disease research through the HRB in co-operation with non-governmental organisations. That is very welcome, but there is more to do in this space. Ireland will not be able to solve the problem of rare disease research on its own. It is very much about being part of those larger European networks like the European Clinical Research Infrastructures Network, ECRIN, the European Research Infrastructure Consortium, ERIC, and collaborating on bio-banking. The very nature of these rare diseases, with some of them being ultra rare, means there is a requirement for pan-European research. Once the centres of expertise are designated and the clinicians can take a lead on this, we would be very much supportive of their coming together, forming a network and making applications as part of a broader European network infrastructure.

Mr. Tony Heffernan: Members asked about people's actual experience of seeking respite and other support and care services. My personal experience was that we were offered respite in an acute hospital full of infection for two children who were prone to infection. For any child with multi-systematic breakdowns of their immune system and other functions, this is not, in my opinion or that of some of the doctors who cared for our children, the best place to put them. However, the practice continues. Access to respite care is exceptionally limited, if there is any. We are self-funding and without State support, but we are transporting children from across Ireland, from Donegal to Kerry, with the LauraLynn children's hospice as one of the principal places. Typically, kids go there for one or two nights' rest after travelling for five hours on a BUMBLEance. We only have one BUMBLEance, with a second on the way.

On the opportunities to access respite care, we should not look at the opportunities but at the benefits. There is a cost saving, on which subject questions have been raised in the Dáil. The amount of money spent by the Health Service Executive on community nursing and similar facilities, including agency nurses, in providing respite and support care is phenomenal. On foot of a question asked more than one year ago, I believe the figure is approximately €78 million. The co-ordination of several areas and the provision of infrastructure such as respite care facilities such as Liam's Lodge will streamline accessibility to respite care for any family to a journey of one and a half to two hours.

Access to respite care will give them head space, which we never had. Speaking personally, we did not receive genetic counselling. One child was already dead and the other one was nearly gone and that was in a five-year period. We received a pre-implantation genetic diagnosis, PGD, and other stuff; frankly, we received information that was not aligned to what it should have been. While the actual personal experiences are strong, we are trying to be proactive. There must be greater emphasis on the patient's needs and those of the parents who are the 24-7 carers. One hears figures of which I seek a correction - one hears there are more home care packages, more home helps and other things. However, it should be quantified in hours and actual time provided to support families, not how many families are receiving home helps and other such services because the inputs and outputs are not the same. This must be improved drastically.

While I agree with all of my colleagues about collaboration, respite care, research and everything else, practical stuff can happen now, as many people are affected. The national office for rare diseases has been identified, which is welcome and a good start to a certain degree.

However, if one considers the whole-time equivalents in the budgets and compares them with the volume of people employed, the investment amounts to approximately €1 per head in year one. Although this affects 6% to 8% of the population during their lifetimes, that is, 275,000 to 330,000 people, the initial budget is €1 per head. We need to prioritise.

Ms Eibhlin Mulroe: I am from the Irish Platform for Patients' Organisations, Science and Industry, IPPOSI. I wish to revert to the comment made by Deputy Sandra McLellan on cystic fibrosis and clinical trials. The product ivacaftor was used in clinical trials but is now on the market. Patients in Ireland had access to the clinical trials. This goes back to the comments made by Senator John Crown in the context of registries. We are in a position in Ireland to attract research on cancer and cystic fibrosis because we have a registry of patients. We have information on patients, both clinical and personal information, that is necessary to access trials. Registries are important in the context of research. I revert to the point made by Senator John Crown on specialist care and he is correct. The European Organisation for Rare Diseases, EURORDIS, recently conducted a survey of patients in Europe who were experiencing difficulty. A total of 2,000 patients responded to the survey, of whom 90 were Irish. They had the highest rate of issues in respect of access to specialist care. The issues did not pertain to access to treatment or drugs but access to specialist care. We have an issue in Ireland in that context.

I am thinking of people who have rare conditions, that is, the people I know and with whom I interact, as do Ms Avril Daly and Mr. Tony Heffernan. It is really difficult when one lives in a country in which one does not have health information and one does not have unique identifiers within the hospital system. A person with a rare condition who visits a hospital but then has to vist another the following week because he or she happens to be elsewhere is in trouble. While this is not a rare disease issue, the health information Bill is really important. Its implementation is even more important, that is, the adoption of unique identifiers to ensure all citizens will have their health information collected somewhere. While this is probably a discussion for another day, in the context of rare conditions, it matters.

Another issue that is missing is that of health economics in respect of orphan drugs, of which there are only 72 under the orphan drugs regulations. There are not that many of them, as treatments are not available for every rare condition. There are only a few treatments and where they are available, particularly ultra-orphans where one is talking about perhaps one in 50,000 people, such treatments costs are high. There are fewer people with such conditions, but the cost of developing the drug is the same. In these situations the system of assessment of these drugs is the same as for any other drug. It happens within the National Centre for Pharmacoeconomics and I acknowledge its process is transparent. We can see it and what happened with the health technology assessment, HTA, analysis. The problem is with what happens after this, namely, the decision. What was really good about the cystic fibrosis case was that the people involved communicated with the patient organisation. However, as has been seen within the past six months, a patient who is one of only two or three in the country has nowhere to go to talk to those who make the decisions. The people sitting around this table from both the Department of Health and the HSE have been working with us on how to involve the patient within that process because what then happens is such patients go public and to the media. While they are entitled to do this, we should have a process in place as this issue will come up repeatedly in a country in which budgets matter.

Dr. Avril Kennan: I wish to pick up on Deputy Billy Kelleher's point about research and whether any progress has been made in collaboration. I was a researcher on the genetics of rare diseases for many years. I now work with the patient advocacy organisation DEBRA Ireland.

My worry about research is that it will be perceived as a luxury in the context of the national plan. Within the current constraints, it is easy to focus on the more immediate aspects, which are so important. However, to perceive research as a stand-alone item is to perceive it in the wrong way because it is integral to every single aspect of the plan. Obviously, the developments made in understanding the genetics underlying rare conditions have been an absolutely essential element in making progress towards a diagnosis, but there are still many rare diseases of which the underlying causes are not known. There still are people who never receive a diagnosis. Consequently, there is a great deal of work to be done from that perspective.

It also relates to looking at current supports for patients. The best way to do this is to have an evidence-based way in which one analyses carefully how patients are interacting with health services, as well as the quality-of-life issues for them. Then, of course, there is the one thing about which we all think, namely, a cure and therapies, which are essential. We have made great progress for some, but for others, we still are a very long way off. However, in the case of those conditions where we are a long way off, we cannot underestimate the hope factor. I always think it must be such a dark place for anybody living with a rare disease to know that there is nobody who is interested in his or her condition or in doing something to try to progress treatments. We still have a long way to go in terms of collaboration. Some progress has been made recently and there is a strong centre in UCD, for instance. However, we must take a number of steps to improve matters, one of the main ones being facilitating research within the health service, but we have not been very good at this in Ireland. As Dr. John Devlin mentioned, we need to conduct research within the centres of expertise because, apart from anything else, this has been shown to improve patient care.

One recommendation made in the national plan is to establish a network of rare disease researchers, to which consideration must be given. As everyone has noted, the national office will not have the capability to do this in the short term, yet forming networks would open the door to European Union funding. There are actually reasonable amounts of European Union funding available for research into rare diseases, but we must ensure our researchers are ready to take advantage of it. In addition, we must ensure that under the new strategy for science, technology and innovation, our funding agencies will have the capacity to fund research into rare diseases because in recent times it has been very challenging to obtain funding for research at the more basic end of the spectrum. I would greatly like to see a research representative on the proposed implementation plan oversight committee for the national plan.

Ms Helen Byrne: Most members mentioned the resources available in the national office. Deputy Caoimhghín Ó Caoláin remarked on how it was not mentioned in the service plan. In developing the service plan for 2015 the HSE was working with a limited budget and lots of requests for resources, as the committee can appreciate. We were not in a position to make money available for the office, but subsequent to publishing the service plan, we were able to allocate money. It was not mentioned in the service plan because there was no funding specifically available at the time. The HSE, however, recognised the need and sourced the funding which it made available in 2015 on a half-year basis, with a view to providing funding for the office for a full year in 2016. I acknowledge that it probably seems to be a small number of staff. We were working from a low base where there was nothing in place - there was no central registry or information available centrally. For the HSE to plan and deliver services on an ongoing basis we need information. Having the office is not what it is all about; we need to be able to provide the specialist services required, but in order to ensure this we need to have the registry in place and the information needed. It is a start, although it is not from where we want to start.

On the national centre for genetics, I know that the issue has been brought up by Senator John Crown and Deputy Billy Kelleher. There are going to be genetic services available in the children's hospital. There have been numerous international benchmarking activities. We have considered whether the full service should be located in the children's hospital. The committee has to appreciate that the ratio in genetic services between adults and children is 60%:40%. There has to be consideration given to whether the service should be placed on the site of the new children's hospital.

Chairman: We were briefed by the patient groups involved in the rare diseases task force and they stated there had been virtually no effort to inform or consult patient groups. Is that the case and can it be changed? Can they be accommodated and included?

Ms Helen Byrne: I will come back to the Chairman on that issue. I will receive confirmation on whether there was consultation.

On having it part of the service in Crumlin versus locating it in the national centre, a review has been undertaken of the services being delivered by the genetics service in Crumlin. Senator John Crown made a very good point that it was considered as part of the funding for Crumlin hospital and when budgets were cut, that budget was cut also. A steering committee has been set up with some international expertise to advise on what the genetics service should look like and it is to report to the HSE within the next six months.

Ms Avril Daly: There is funding for rare diseases research. There is a matching funding process through the medical research charities group and the Health Research Board. It is important to mention that we have not received that funding this year. We are I hope going to receive it next year, but that means that €1 million has not been put the research into rare diseases this year.

In the context of the relationship between common diseases and rare diseases, a lot of common diseases, particularly in the case of cancer and heart disease, are being broken down into rare conditions because we are beginning to understand and characterise them better. We need to understand better that what we are talking about is how we are going to provide care to meet future needs. What we now consider to be rare conditions are going to have an impact on how we provide care in treating more common diseases.

Deputy Billy Kelleher: Am I to take it that we will have a national centre for medical genetics, regardless of the location? The purpose is to conduct genetic testing in both paediatrics and for adults. Is it going to be specifically designated a national centre? Why does where it is located make a difference as long as it does what it is meant to do?

Ms Helen Byrne: We will report back to the HSE in the coming months on that issue. We will have more information at that time.

Deputy Caoimhghín Ó Caoláin: On the same issue and the response to Senator Jillian van Turnhout that it was intended to incorporate such a facility within the new national paediatrics facility, presumably the St. James's Hospital site, that is the way it is to proceed, but it is not expected to come on stream until 2019 at the earliest. What is to happen in the interim? Can the delegates anticipate that we will at least see an interim facility up and running pending the commissioning of the new national paediatrics centre?

Dr. Devlin glossed over my point a little about oversight and an oversight committee. There were 48 recommendations made in the context of the national plan. Will there be an oversight

committee? Will the steering committee carry out this function in the interim? How inclusive will the oversight committee be? I happen to believe it needs to be more representative of many of the groupings involved. There is a need for more direct involvement and the oversight requires to be very focused and an intimate relationship where the people most directly affected will have an opportunity to ensure what is proposed will actually be delivered at the earliest opportunity.

Dr. John Devlin: I did not mean to gloss over the Deputy's point. It was a very relevant question. Our focus initially was on engaging with the HSE on a number of key issues around the programme, the office, access to orphan drugs and so on. The national steering committee is due to meet in the next couple of weeks to discuss this issue and is representative, as the Deputy knows. Mr. Heffernan has expressed a view in that regard. We are open to considering what works best. We did not actually have a meeting of the oversight committee as such because a lot of changes were taking place at the time, but we wanted to continue to do business with the HSE. The question of whether it should be the steering committee or something different that takes on this role is less significant, as long as it does the right things and we monitor the plan and everything else. Let us not forget that we will be reporting on a pile of key indicators in relation to the rare diseases plan. There are about 20 or 30 indicators on which we are going to report and which will very much form part of the information we will be making available to everybody.

Mr. Tony Heffernan: We are talking about the provision of services in dealing with rare diseases and symptom management. The symptoms of many of those affected by rare diseases are common and similar to those affected by common conditions; therefore, they can be managed in the same way. We must consider what is happening now. The State relies on families and carers to look after all those affected and most of them receive no financial benefits. The least the State could do is give them a break, by means of respite care and additional support. This would them give them a better future in terms of their mental health and also support the families directly. We have to take a proactive approach to supporting them while all of the other work is continuing.

Ms Helen Byrne: I can confirm for Deputy Caoimghín Ó Caoláin that there will be genetic services in the interim in advance of the opening of the new children's hospital.

Senator John Crown: Will there be national funding for the existing local unit? Will there be an acknowledgement that it is providing a national service in order that it will not have to fight with management of Crumlin hosital for a share of the local pie?

Ms Helen Byrne: That is the report we are going to receive in the next couple of months from the steering group on how genetics services should be developed and run separately.

Senator John Crown: I will make a little bet with Ms Byrne that it will tell her that she needs to have a service there right now and should just provide for a modest increase in funding to allow it to perform a national function without there having to be nickels and dimes from local hospital management pending the development of the national service, but we can wait for the report. My opinion is, however, free.

Senator Colm Burke: I raised the issue of the cross-border health care directive and the treatment abroad office. No one responded to that and it is a big issue.

Dr. Colm Henry: The treatment abroad scheme currently operates within the health and

well-being division of the HSE. This scheme deals with much more than rare diseases.

Senator Colm Burke: I am aware of that.

Dr. Colm Henry: Much of its work is to engage with protocols. The treatments and care pathways for many of the diseases are well defined. It is currently run largely by public health consultants. The director of acute hospitals, Liam Woods, recently met the director of the well-being division, because there is recognition that the public health consultants need more specialist support in arbitrating on treatments, especially for rarer conditions. Our division is working with the well-being division to decide how we can give them access to timely, specialist support for conditions that are not protocol-driven, for which answers are required in a much more timely way.

Senator Colm Burke: Why not set up a more streamlined process for dealing with it? This is where the problem is arising. People are left in mid-air for a quite a long time, not knowing whether they will be able to get the required support for that treatment abroad. I have come across a number of cases where it went around in circles for three or four months, which is unfair to the families and to the patient concerned.

Dr. Colm Henry: I agree that it is not fair. That arises where for some reason the treatment abroad scheme refuses or does not allow the application to proceed. In the majority of cases the treatment is approved, and in most cases there is a protocol that allows people to sign off on whatever treatment is required. The system must be streamlined. Quicker access to specialist opinion is required for those rarer conditions that are not protocol-driven.

Senator Colm Burke: What about the transposition of the directive and the setting up of structures for dealing with the cross-border care directive?

Dr. Colm Henry: I am not clear on what the question is.

Senator Colm Burke: Has the EU cross-border health care directive been transposed and have we set up a procedure for its implementation?

Dr. Colm Henry: I cannot answer that question. We will come back to the Senator.

Ms Eibhlin Mulroe: The question regarding the transposing of the cross-border directive is probably best directed to the Department of Health. It is completely different from the treatment abroad scheme. I know Senator Burke is familiar with this, because we had an event dealing with this last year when the representatives of the Commission were over to try to get Ireland to transpose the cross-border directive. It makes a great deal of sense, from what we have talked about this morning. We have to work outside Ireland to get treatment for conditions that only one person in the country has. That person must be able to travel. We are seeing, more and more, that patients with rare conditions who need to travel, even to London, are finding it very difficult. They are waiting longer than three months. They are waiting years to get access. We would urge that rare diseases be treated in a slightly different way. The political theorist Rawls used to say that in democracies, where people are discriminated against, we need to positively discriminate in their favour. I am stealing that from Professor David Smith, who quoted it at a meeting of ours this week. We need to look at that within the context of rare diseases, within the treatment abroad scheme, and within access to treatment and care.

Chairman: That is a good point to end on. We might ask that the patient groups involved in the rare disease task force might update the briefing to include the fact that the HSE has made

funding available, as Ms Byrne commented, although it is not mentioned in the service plan - just to be fair in terms of outlining both the positives and the negatives. I thank the group for its excellent presentation and research. I thank everyone for coming. We all need to advocate the implementation of the plan and to understand, as Ms Daly rightly said, that "rare" does not mean "few". As a committee, we are committed to working with all of the witnesses to ensure the implementation of the plan is strong and successful.

The joint committee adjourned at 11.45 a.m. until 9.30 a.m. on Thursday, 5 March 2015.