Report of the Consultation for a National Rare Disease Plan for Ireland

Synthesis of responses

IPH

An Roinn Sláinte
DEPARTMENT OF HEALTH

HSE

Report of the Consultation
for a National Rare Disease Plan for Ireland

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Report prepared for the Department of Health by the
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1. Introduction

1.1 The Importance of Consultation

Public consultation is an important stage in the policy-making process. Formal consultation processes are now an integral element in the development of policy at a national level to ensure best practice in policy making. While the consultation process is open to any member of the public, it is designed to seek views from those who would be affected by or those who have a particular interest in the new policy. It contributes to good public governance by fostering greater transparency in policy-making; more accountability through direct public scrutiny; better quality decisions based on a wider range of information sources and finally higher levels of implementation and compliance given greater public awareness of policies and participation in their design (OECD, 2001). Consultation is an effective mechanism for alerting policy makers to any concerns and issues not picked up through existing evidence or research. Consultation on rare diseases is then particularly relevant given the noted dearth of evidence on the experiences and needs of people living with rare diseases in Ireland. Large-scale consultation processes can be resource intensive, however new approaches to consultation including online consultation surveys can be powerful tools for enhancing public engagement in policy making as a complement to traditional mechanisms for public consultation (OECD, 2003).

1.2 Policy Context

This report presents findings from the National Consultation on Rare Disease overseen by the Institute of Public Health in Ireland on behalf of the Department of Health to inform the development of Ireland's first National Rare Disease Plan. In 2009, the Council of the European Union recommended that all member countries develop a national plan for rare diseases within the framework of their health and social systems by the end of 2013. The aim is to ensure that all patients with a rare disease in Europe have access to high quality care, including diagnostics, treatments and rehabilitation. Rare diseases were identified as a priority for action at a European level acknowledging the lack of initiatives and health policies at a national level as outlined in European documents such as the '*Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions' on Rare Diseases: Europe's Challenges*

and the *Council Recommendations of 8th June 2009 on an action in the field of rare diseases.* They specifically encouraged the development of plans with a focus on research, establishment of Centres of Expertise at a European level, and empowerment of patient organisations (Council Recommendation 2009/C 151/02). In keeping with this EU directive the Department of Health in Ireland convened a National Steering Group in 2011 to oversee the development of the National Rare Diseases Plan in Ireland and the Institute of Public Health was tasked with supporting the work of the Steering Group to develop a policy framework. In establishing the Steering Group the Department of Health endeavoured to ensure adequate representation of rare disease stakeholders in Ireland. In addition, a national consultation on rare diseases was developed to provide a platform for all rare disease stakeholders to voice their views, engage in the development of the Plan and contribute to policy implications which will ultimately determine rare disease policy and implementation actions in Ireland. While it is difficult to reach all rare disease stakeholders, it is hoped that this three phased approach to stakeholder representation in the development of the Plan adequately captures stakeholder's needs and preferences.

1.3 The Consultation Process

To inform the policy framework a strategic consultation process took place between June and August 2012. On Monday the 11th of June, 2012, at Farmleigh House in the Phoenix Park, the Department of Health hosted a Consultation Day event on the National Rare Diseases Plan with support from the Institute of Public Health in Ireland (IPH) and the Health Service Executive National Advocacy Unit. The consultation event was attended by 120 delegates from a variety of backgrounds. Proceedings from this event were drafted by the HSE and summarised for inclusion in this consultation report.

In order to elicit the views of a wider range of stakeholders and enhance the reach of the consultation process, the Steering Group proposed that an online consultation process also be developed. The Steering Group made this recommendation in order to capture the views and experiences over the broad spectrum of rare disease patients and stakeholders. In recognition of the preponderance of rare disease among children with young parent carers and young adults themselves, an online methodology was felt to be appropriate. Findings from the consultation were critical to informing the content of the National Rare

Disease Plan for Ireland. This is the first time the Department of Health conducted a broad consultation on rare diseases with a view to hearing the views of patients, their carers, health service providers and other stakeholders.

1.3.1 National Rare Diseases Consultation Day

The national rare disease consultation day was organised around presentations from key note speakers and a series of interactive workshops aiming to identify key priorities for a National Rare Disease Plan. These facilitated workshops took place with stakeholders working in small groups to enable greater knowledge and experience sharing. Participants were encouraged to answer a range of predefined questions in relation to five central themes:

1. Research and Data
2. Infrastructure
3. Co-operation and working across sectors
4. Diagnosis
5. Treatment of Rare Diseases including Orphan Drugs & Technology

A suite of documentation was prepared in advance of the consultation workshops to stimulate debate, in addition to key note speaker presentations. Several members of the National Steering group welcomed the consultation initiative on the day.

Mr. John McCormack, Chairman of the Taskforce for Rare Diseases and of the Medical Research Charities Group (MRCG) said

> *"Much work has been done to ensure this plan will meet the needs of people with rare diseases and I welcome the level of engagement which the Department has had with patients' organisations thus far. In particular the launch of the online consultation process offers people with a rare disease and their families the opportunity to contribute to the formation of the plan."*

Left to right: Eileen Treacy (HSE), Caroline Kelly (DoH), Geraldine O'Dea (IMB), John McCormack (MRCG), Eibhlin Mulroe (IPPOSI), Tony Heffernan (Bee for Battens/Saoirse Foundation), Helen McAvoy (IPH), Minister for Health James Reilly, Avril Daly (GRDO), Owen Metcalfe (IPH), John Devlin (DoH), Shaun Flanagan (HSE), Deirdre Mulholland (HIQA) and Catherine Gill (HRB).

Eibhlin Mulroe a member of the Steering Group and CEO of The Irish Platform for Patients' Organisation, Science and Industry (IPPOSI) said

"The move to include a patient's perspective in the process recognises the level of expertise which patients have and their willingness to engage with all the stakeholders. It is good to see that the efforts of the many groups concerned with supporting the individuals and families affected by a rare disease in Ireland and the EU is starting to come to fruition. Establishing and implementing a National Plan for Rare Disease will start to address the marginalisation, frustration and often despair experienced by patients and families of rare diseases."

Avril Daly, Chairperson of the Genetic and Rare Diseases Organisation (GRDO) commented that

"Irish patient groups have engaged with stakeholders nationally and internationally for 25 years to advocate for the development of better systems leading to better health outcomes for patients affected by rare diseases. We welcome the commitment made in the Seanad last March by the Minister to develop a Clinical Care Programme for rare diseases. We are hopeful that by including rare diseases in the National Clinical Care Programme and the establishment of a National Rare Disease Office a clearly defined pathway will be developed to enable patients and medical professionals access the

correct specialists and centres of expertise, new treatments and management options and information regarding ongoing clinical trials."

Ms Daly who was recently elected Vice President of the European Organisation for Rare Diseases (EURORDIS) went on to say

"Ireland will hold the European Presidency in 2013 which coincides with the deadline for member states to have a national rare diseases plan in place. It is therefore important that Ireland provides strong leadership on the rare diseases issue during its presidency."

Tony Heffernan, a Steering Group member and Founding CEO of Bee for Battens, The Saoirse Foundation said

"In the past it has been very difficult for parents to find any information at all about rare diseases. The rarity of some of these disorders means that few doctors have an awareness and parents of course want to know, on a practical level, how they can best help their child. Hopefully this consultation process will give parents an opportunity to input directly into the plan".[*]

1.3.3 Online Consultation Questionnaire

The online consultation questionnaire was developed based on input from a Communications Subgroup of the Steering Group comprising some members of the Steering Group and some members of the Rare Disease Taskforce and with due concern to issues such as accessibility and anonymity. Given the sensitive nature of rare diseases and in keeping with the ethical principles of voluntary participation and anonymity applied in health research, consultation respondents were given the option to submit their views anonymously. Respondents were asked to submit their views on six general themes relating to rare diseases:

- Priorities for people with rare diseases
- Diagnosis and screening
- Treatment and management

[*] see http://www.mrcg.ie/go/news/news/rare-disease-national-plan-consultation

- Orphan Drugs and Technologies
- Involving and empowering rare disease patients
- Data and research.

With the support of the HSE National Advocacy Unit, this questionnaire was made available to the public on the HSE Have Your Say website for four weeks between the 11th of June and the 13th of July. The HSE National Advocacy Unit provided support to respondents requesting assistance with completing the questionnaire.

All potential stakeholder groups were encouraged to respond including rare disease patients, carers, researchers, health service providers, policy makers and industry. The Communications Subgroup of the Steering Group worked to foster engagement with the consultation process by a variety of means including social media, newspaper articles, television interviews and advertisement on websites, organisational mailshots and newsletters. The patient organisations and their representatives were particularly active in this regard. The involvement of a wide array of stakeholders has proved invaluable in the development of other National Rare Disease Plans across Europe including Plans developed in the UK, Belgium and France.

1.4 Structure and Scope of the Report

This report presents a synthesis of findings and a summary account of the consultation responses from both phases of the national consultation i.e. the consultation event and the online consultation questionnaire. *Section 2* of this report presents an overview of the methodological approach to analysing the online consultation. Findings from the online consultation are presented in *Section 3* according to the themes outlined in the questionnaire and other themes arising from the consultation responses. Findings from the consultation day are presented in *Section 4* according to the five workshop themes outlined in Appendix 2.

This report provides an initial overview of the opinions and experiences of many stakeholders with an interest in rare diseases in Ireland. It is important to note that while a wide range of stakeholders submitted their views and provided valuable detailed

responses to the survey, this is a consultation process and therefore not a fully representative survey of all rare disease stakeholders. As such this report does not represent a comprehensive needs assessment for rare disease patients. Capturing the diversity of rare disease patient's needs is a challenge for research as well as for policy and more focussed work may be required. There remains a significant chance that the views of some rare disease patients and carers are less well represented and some perhaps not at all. In particular, minority groups in Ireland such as the Travelling community may be under-represented in this consultation process. Specifically the limitations posed in terms of the accessibility of the consultation process for those with visual, hearing or learning disabilities are also acknowledged. Moreover, some stakeholders may be more or less likely to acknowledge their role in the development of a Rare Disease Plan. Given that we have limited health intelligence on rare disease in Ireland, it is not currently possible to design a fully representative survey of all rare disease stakeholders. Nonetheless, this consultation process engaged a wide representation of relevant stakeholders and provides useful insights into the experiences of rare disease patients, their carers and health care providers as well as some clear direction in terms of priorities for Ireland's first National Rare Disease Plan.

2. Analysis of Online Consultation Responses

2.1 Introduction

A large number of responses were generated from the online consultation reflecting the diverse and significant impact of rare disease on Irish people and the healthcare system. In order to elicit the views of respondents, the survey was designed with a range of closed and open-ended questions, organised according to the main themes outlined in section 1.3.3. Closed-ended questions allowed for a general representation of respondent views while open-ended questions, where respondents were invited to reply to the questions in their own words, provided a more in-depth assessment of respondent's experiences of rare diseases from a variety of perspectives including patients, carers and healthcare providers. In addition, open-ended questions provided a facility for respondents to voice their opinions on diagnosis, care and treatment as well as preferences and priorities for developing a Rare Disease Plan in Ireland. Due to the mixed question design the consultation responses were analysed using both quantitative and qualitative analysis approaches.

A total of 732 survey responses were downloaded from the online survey facility Survey Monkey. All survey responses were saved to an Excel database and reviewed in advance of analysis. Several of the downloaded responses were invalid; either empty, largely incomplete or duplicates of responses. Duplicates and incomplete responses were cleaned and/or removed from the file before analysis. In general, any surveys with a large proportion of unanswered questions were deleted during this phase. In cases where surveys were largely incomplete but respondents had provided qualitative responses, these were included in analysis. In total, there were 481 valid responses to the online consultation survey. All valid open and closed-ended responses were screened and assigned to qualitative or quantitative files and analysed accordingly. In keeping with the Data Protection Acts 1988 and 2003, appropriate data protection standards were adhered to when handling and analysing consultation responses.

2.2 Quantitative Analysis

Survey response data were cleaned in Excel and entered into PASW statistics 18, a statistical package formally known as SPSS. All quantitative analysis was conducted by IPH. Basic descriptive analysis such as frequencies and cross-tabulations were performed. Where appropriate, findings are displayed graphically and data in graphs are based on valid responses to the open consultation process. Due to the sample size, and the limitations evident in terms of full representativeness, it was deemed appropriate to only make comparisons between certain categories of respondents. In some cases statistical analysis were computed including Chi-square tests and ANOVA to ascertain significant differences in opinions between category of respondents, for example between patients and healthcare providers.

2.3 Qualitative Analysis

Open-ended responses were analysed by an independent qualitative researcher in liaison with the research team in the IPH. Qualitative analysis for the purpose of the consultation results relates to seven open-ended questions:

1. *In your opinion, what is currently the most challenging issue for people with a rare disease in Ireland?*
2. *If you have any other comments in respect of new-born screening and rare diseases, please share them with us in the space provided below.*
3. *Do you have any other comments on how the assessment process for Orphan Drugs and technologies in Ireland could be improved?*
4. *In what way should patient organisations be involved in the implementation of the National Rare Disease Plan?*
5. *Do you have any other comments in relation to enhancing rare diseases in Ireland?*
6. *What processes do you think are needed for implementation and monitoring of the National Rare Disease Plan for Ireland?*
7. *If you have any other points you would like to raise in terms of the National Rare Disease Plan, please share them with us in the space provided below.*

An applied qualitative research approach was adopted in the analysis of open-ended data for this report as it can offer the policy maker a theory of social action grounded on the experiences – the world view – of those likely to be affected by a policy decision (Walker

1985), namely rare diseases stakeholders. With this in mind, the methodology deemed to be the most appropriate for collating and analysing the open-ended consultation data was Framework Analysis (Sirvastava and Thomson 2009). A five-step process was applied to the analysis of the consultation data as recommended by Ritchie, Spencer and O'Connor (2003) and included familiarisation with the data, generating a thematic framework representing all themes emerging from the consultation responses, followed by a process of indexing, charting, mapping and interpreting the data according to the identified themes.

A thematic framework was developed, informed by the main themes presented in the online survey. A further set of sub-themes emerged from the survey data. These were sorted and indexed under the main themes. All themes were analysed and cross-referenced between different categories of participants to elicit any differences in opinions between respondent groups. Respondents were characterised into six broad categories:

- Rare disease patient
- Carer of rare disease patient
- Health service provider
- Researcher
- Pharmaceutical/ biotech company
- Other

2.4 Analysis Synthesis

Once initial analyses were complete, both quantitative and qualitative findings were synthesised according to the main themes emerging from the consultation and where comparative analysis is appropriate, responses are attributed to respondent category. However, this is not done in cases where an individual patient with rare diseases may be identifiable. All themes are discussed below and supported by respondent data. Policy implications were developed based on the full consultation findings from the consultation event and the online survey.

3. Findings from the Online Consultation Questionnaire

3.1 Characteristics of Respondents

Given the sensitive nature of rare diseases and in keeping with the ethical principles of voluntary participation and anonymity applied in health research, consultation respondents were given the option to submit their views anonymously with a large proportion of respondents choosing to do so (41.5%). As only a handful of people in Ireland may suffer from a particular rare disease – naming the disease could render a person identifiable, therefore only respondents who elected not to submit anonymously were asked to submit the name of the diseases to which their input relates. Data on specific rare diseases were only available for 58.5% of respondents, given the large proportion of respondents who submitted their views anonymously. This meant that analysis of findings according to rare disease types is not meaningful or appropriate in this report.

It was nevertheless clear from the responses that while diseases and their related needs and experiences vary substantially, there were many common experiences and cross-cutting priorities associated with rare diseases, supporting the need for a National Rare Disease Plan covering this diverse range of rare diseases. While this consultation process is a first attempt to present the views of this uniquely vulnerable and diverse patient group, further detailed research is required to elicit the needs of patients with specific rare diseases which lies beyond the scope of this consultation. There is no way of knowing the characteristics of those who responded anonymously and care should be applied when considering the range of diseases presented in this report as they are unlikely to represent the full range of rare diseases in Ireland. The rare diseases listed by those who did not respond anonymously are summarised in Appendix 4.

The majority of the responses to the online survey were submitted by individuals (88.4%), although a substantial number of responses were also submitted on behalf of organisations (11.4%) including those representing health professionals, patient organisations and pharmaceutical or biotechnology companies. Carers (37%) and patients (29.5%) were the most frequent responders. A significant number of health service

providers and clinicians, and researchers with an interest in rare diseases also responded, representing 13.3% and 6.4% of respondents respectively. Table 1 provides a summary of all categories of respondents by numbers and percentage of valid responses.

Table 1. Online consultation respondent characteristics.

Question and response	Frequency of response	Percentage %
Organization	55	11.4 %
Individual	425	88.4 %
Total *	480	100 %
Carer	178	37.0 %
Patient	142	29.5 %
Health service provider/clinician	64	13.3 %
Researcher	31	6.4 %
Patient organisations	19	4.0 %
Relative of patient	19	4.0 %
Pharmaceutical or biotechnology company	6	1.2 %
Friend of patient	5	1.0 %
NGO's	5	1.0 %
Health services management/ policy maker	4	0.8 %
Person at risk of rare disease	1	0.2 %
Unclassified	*2*	*0.4 %*
Total †	476	100%

* 1 missing response
† 5 missing responses

Nineteen responses were received from patient organisations; it is not known how many of these are from different organisations as some of the organisations were unnamed. In some cases several responses were received from some organisations. A listing of organisations and individuals responding to the consultation on behalf of an organisation, health service or research group are presented in Appendix 5. Five responders did not identify as a patient organisation, but rather classified themselves as another community or voluntary service. Four individuals responded from a health services management or policy maker perspective. One respondent was a person at risk of a rare disease due to genetic susceptibility and two respondents were unclassified, while five respondents did

not answer this question. It was not possible to compare the numbers or opinions of health service provider or clinicians according to profession as many choose to remain anonymous. Of those who did provide their details, many were employed in acute care settings and there appeared to be an underrepresentation of general practitioners.

Respondent groups were further pooled into six broad categories in order to conduct some more meaningful analysis of respondent views by category of respondent (Table 2).

Table 2. Pooled respondent categories

Pooled respondent category	Number	Percent %
Group 1. Patients	143	30.0%
Group 2. Carer, relative and friends of patient	202	42.4%
Group 3. Health service provider/clinician & health services management/policy maker	68	14.3%
Group 4. Researchers	31	6.5%
Group 5. Patient organisations & NGO's[†]	24	5.0%
Group 6. Pharmaceutical or biotechnology company	6	1.3%
Unclassified	*2*	*0.4 %*

One respondent who identified themselves at risk of a rare disease was categorised with the patient group (30%). Carers, relatives and friends were also grouped together (42.4%). Health service providers, clinicians, managers and policy makers formed a separate category (14.3%). All researcher respondents were classified together (6.5%) and those who respondent on behalf of a patient organisation, or NGO were also pooled into one category (5%). A small number of pharmaceutical or biotechnology companies also submitted responses and were grouped together for comparison purposes (1.3%).

[†] Non-governmental Organisation

Key points - Characteristics of Respondents

- A significant number of responses were received, slightly in excess of the responses to the consultation on the UK Rare Disease Plan.
- The majority of responses were submitted by individual carers and rare disease patients.
- The large number of responses from carers indicates the significant caring burden associated with rare diseases in Ireland and the vulnerability of the rare disease community.
- Responses to the UK Rare Disease Plan were dominated by organisations whereas responses to the Irish Rare Disease Plan were dominated by individuals. This may indicate that for many rare diseases a patient organisation may not exist in Ireland.
- Health service providers and clinicians were significant contributors to the consultation – these were predominantly hospital specialists rather than GPs
- Many respondents elected to remain anonymous, this suggests that anonymity maybe an important prerequisite for patient engagement in rare disease research and policy making processes.
- In and around half of respondents revealed the name of the rare disease(s) to which their input related – over 200 different rare diseases were named.

3.2 Priorities for Service Provision

Respondents were asked to rank the most important recommendations to ensure real benefits for people with rare diseases. These were ranked from 1 to 6 in order of importance, where 1 was most important and 6 was least important. Respondents ranked better access to specialist care as the most important recommendation, followed by improvements in accurate and timely diagnosis, improvements in the quality of healthcare, enhanced research and improved access to orphan drugs and technologies. Accessible and reliable sources of information were not deemed to be as important as other recommendations. However, it should be noted that all benefits were identified as important, and the differences between the perceived importance of each benefit (mean scores) were small.

A one-way ANOVA was performed to test the differences of opinions between respondent categories on the most important benefits for people with rare diseases. Patients and carers were significantly more likely to rank improvements in healthcare as the most important benefit compared to researchers ($P<0.05$). Researchers and patient organisations were significantly more likely to rank enhanced research as the most important benefit compared to patients ($P<0.05$). Researchers were more likely to report improved access to orphan drugs and accessible and reliable information as most important compared to patients and carers ($P<0.05$).

In addition to the priority improvements identified in the closed-ended question, themes emerging from the open-ended responses indicate a range of areas where action is required to improve service provision. Responses referred to a range of challenges facing rare disease patients, centred on critical milestones of the patient's rare disease journey. These challenges included both clinical and broader quality of life issues and concerns. The themes are summarised in the following section of the report and include:

- Publicity and awareness of rare diseases
- Screening
- Diagnosis
- Quality of care
- Waiting times
- Orphan drugs and technologies
- End-of-life support services
- Research
- Equitable care provision

Key points – Priorities for service provision

- Better access to specialist care and improvements in accurate and timely diagnosis were considered the areas which could bring most benefit to rare disease patients in Ireland.
- Awareness of rare diseases was a cross-cutting issue – this lack of awareness was evident at all levels of the health care system as well as in respect of access to public and private services and entitlements both within the clinical setting and beyond it.
- Lack of awareness and understanding of rare disease created significant vulnerability at many stages of the patient journey, most particularly for those seeking a diagnosis and navigating the health care system in the absence of any clear care pathway.

3.2.1 Awareness and understanding – the *patient journey through no-man's land*

The main challenge identified by a large proportion of respondents was rooted in a lack of awareness of rare diseases in clinical practice and in general. This lack of awareness manifested at all stages on the patient journey through the health system. It permeated all areas of living with a rare disease from difficulty in gaining diagnosis to confusion over access and eligibility to appropriate health and social services and entitlements. In addition, respondents voiced issues in relation to accessing health insurance and financial services and supports. One respondent with a rare disease summarised:

'the doctors simply do not see enough cases similar and they do not know what to do!'

Respondents voiced considerable frustration in the limited understanding of rare diseases and the unique care needs associated with specific conditions. They drew attention to the poor awareness of rare diseases and their associated impact on patients both within the clinical setting and beyond it. The need for improved awareness of rare diseases across the health care system was highlighted by respondents as well as the importance of developing tools to foster better understanding pertaining to specific conditions.

Limited awareness and understanding of rare diseases was a commonly cited challenge when dealing with health professionals including general practitioners, consultant specialists and in Emergency Departments. Respondents highlighted the need for a

focused approach to increase the capability of all medical professionals in the field of rare diseases:

> *'I feel we need a much stronger awareness-building programme to educate service providers and GPs. Even currently using a very good system for my child the knowledge base and access to information or desire to be up to date on issues seems to be totally lacking, bar a few keen individuals.'*

> *'…perhaps young medical students could be encouraged to participate in rare disease research…Better training of staff in A&E departments to recognise symptoms of lesser known illnesses.'*

The lack of awareness it was felt, resulted in a lack of understanding and reduced support in health services and in society in general. Respondents highlighted that this lack of support was present at many stages throughout the rare disease illness experience and was most evident at the critical milestones outlined earlier and most particularly in relation to:

- Obtaining services and support without a formal diagnosis
- Obtaining a formal diagnosis after presenting with symptoms
- Navigating between primary care and acute care
- Accessing appropriate medication and treatment
- Absence of care pathways or care plans for long-term treatment
- Accessing patient and family support services such as counselling including genetic counselling and advice on implications for the broader family
- Care at end-of life
- Experiencing bereavement

3.2.2 Screening

Newborn screening

Early detection and treatment of certain rare diseases can prevent illness and disability. Under the national newborn screening programme, all babies born in the State are screened for a number of rare genetic conditions using a heel-prick blood test. These conditions include:

- Phenylketonuria (PKU)
- Homocystinuria

- Maple Syrup Urine Disease
- Classical Galactosaemia
- Cystic Fibrosis
- Congenital Hypothroidism

In addition, all newborn babies are examined at birth to screen for a number of congenital abnormalities such as cleft-lip, talipes, heart murmurs etc. Babies identified as being at risk i.e. born to families with a history of a genetic rare disease, may also be screened for that disease at birth. Respondents were asked to submit their views on the national newborn screening programme.

Development of the newborn screening programme was welcomed by respondents. Nonetheless, a number of respondents queried why it had taken so long to implement the Cystic Fibrosis Screening and several respondents voiced their concern that before the screening programme was introduced cases had been missed:

> *'The newborn screening for CF is so overdue but very much welcome. As a person whose child was diagnosed at 7 months I always wonder how much damage was done in that time…'*

Given the genetic component of many rare diseases, the importance of early screening was highlighted:

> *'…the difference between our newborn diagnosed within 1 week of birth and two children diagnosed after eight very difficult years of illness is incredible…it is very upsetting to know that if they had been diagnosed earlier their quality of life would have improved enormously and our quality of life as parents would have been so much less stressful.'*

Respondents were asked to submit their views on three statements in relation to newborn screening. A large proportion of respondents did not know if newborn screening was operating well at present (30%), while almost half of those who did voice an opinion agreed that it was operating well (50.5%). Approximately 20% of respondents however either disagreed or strongly disagreed that current screening was operating well. Cystic fibrosis patients were well represented in the consultation and the views of other rare disease patients where newborn screening is not available may differ substantially. A large majority of respondents agreed for both the need to introduce legislation to support

newborn screening in line with European best practice and that the listing of conditions screened for should be reviewed in line with international best practice.

Respondents called for a number of further actions relating to screening to be implemented to improve rare disease diagnosis. These included increasing the number of conditions screened for, in line with international best practice, and extending the screening coverage period. Acknowledging that not all illnesses can be recognised in newborns, consideration of antenatal screening was raised as well as repeated screening for young babies. Respondents stressed the need to implement appropriate support structures including care pathways and emotional support following confirmation of a rare disease diagnosis from the newborn screening programme.

Key points – Screening

- Newborn screening was seen as a very positive contributor to the early diagnosis of rare diseases in Ireland.
- The introduction of newborn screening for cystic fibrosis was widely endorsed.
- Responses recognised the need to support the further development of the newborn screening programme through the Rare Disease Plan.
- Appropriate follow-up, referral and support of parents experiencing bad news from newborn screening was emphasised.

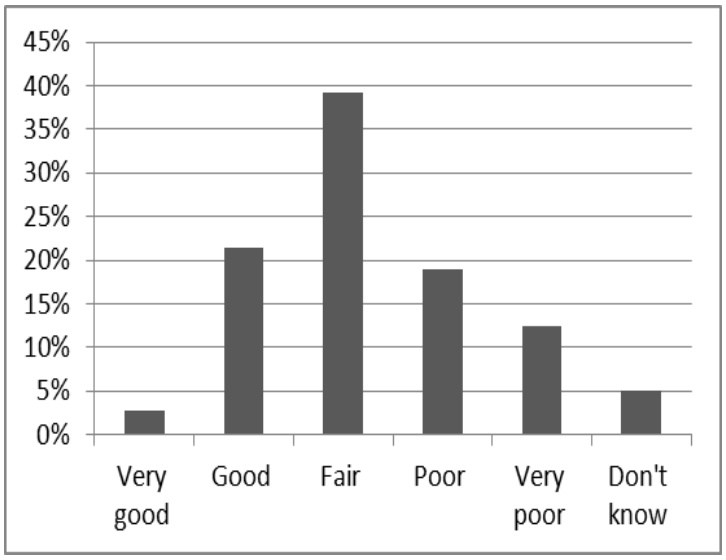
3.2.3 Diagnosis

Obtaining a diagnosis

Due to the rare nature of the diseases under consideration, it can be difficult to obtain a definite diagnosis and it often takes a substantial length of time. While patient and carer respondents acknowledged the challenges involved for clinicians; they felt that the lack of training and knowledge on rare disease diagnosis amongst medical professionals was lacking in general. This was also identified as an issue among clinicians themselves. When all respondents were asked to rate the quality of diagnosis for people with a rare disease, a significant proportion rated it as fair or better (62%) while 32% rated the quality of diagnosis as either poor or very poor. Notably patients and carers were significantly more

likely to rate the quality of diagnosis as poor or very poor compared to health services providers/managers and policy makers ($p<0.05$).

Figure 1. Rating of quality of diagnosis for rare disease patients



Respondents were also asked which aspects of diagnosis were most in need of improvement. The most frequently cited aspect of diagnosis requiring improvement was delays in diagnosis (71.4%), followed by insensitive or ill-informed communication of diagnosis and implications (52.2%). Communication issues were statistically significantly more likely to be cited by those responding on behalf of organisations compared to individuals ($P<0.05$). A further 27% of respondents felt that the frequency of incorrect diagnosis was the aspect of diagnosis requiring most improvement.

While obtaining a diagnosis was problematic in and of itself, respondents also commented on the long drawn out process this entailed for patients. Respondents described a complex journey to diagnosis and felt limited in their options as most were unable to travel abroad for diagnosis:

> *'Getting a timely diagnosis – often involves seeing many specialists, for many years before a diagnosis is made.'*

> *'Getting a diagnosis takes years. Medical insurance does not allow you to travel abroad for a diagnosis, so you have no choice but endure the waiting lists here.'*

Although experiences were mixed, many respondents reported having problems in engaging the interest of health professionals. Patients reported that limited lack of understanding and poor communication from health professionals was distressing:

'I can have an intelligent and productive conversation with my GP but my contact with the specialists have left me phobic and distressed. In particular the neurologist who seem to seek every opportunity to label the symptoms as mental in origin. Their second instinct seems to be to ensure that it is not in their specialist area.'

Another respondent felt that specialists did not take the time to fully examine them and note their symptoms, and highlighted a perceived over reliance on clinical tests. They emphasised the need for a dedicated approach to rare disease diagnosis:

'[The Consultant] took his time questioning me, writing down the key points on my answers and then asked me to return in a week or so, so that he would have time to analyse the results. On my return he repeated some tests and then told me what was wrong and how serious it was. He has no results from the numerous scans and cardiac tests that my earlier consultants got carried out, yet he was right and they were wrong. He spent about two hours with me, the others spent five to ten minutes discussing the tests that were carried out and the next test I would need to have.'

Misdiagnosis

Several respondents described their experiences of misdiagnosis. Respondents stressed that a lot could be learned from previous experiences of misdiagnosis of rare diseases in Ireland. The lack of any system to document, analyse or compensate patients for misdiagnosis was highlighted. Improving information sharing and transparency on misdiagnosis was recommended with a view to learning from mistakes and improving care provision:

'Involve people who are suffering because of misdiagnosis or delays in treatment. I had two serious misdiagnosis from two top professionals in two different areas. Have and maintain a register of all misdiagnosis and value of all specialist treatment.'

Communicating a rare disease diagnosis

Respondents reported on the sense of shock associated with a diagnosis of a rare disease, considered especially difficult when dealing with a diagnosis for newborn babies. They highlighted the need for the diagnosis disclosure to be made in a sensitive way as well as ensuring that patients and families have direct access to support services. This was thought to be especially important for parents of newborn babies:

'With newborn screening comes the sensitive issue of telling new parent, this must be handled by trained personnel, access to other parents with a child with similar condition and reassurance that they are not alone.'

'We received a phone call telling us [diagnosis] nothing else no information, no referrals. I believe no parent should be given diagnosis over phone, if a resource issue GP should be notified.'

'Adequate supports should be on place for parents of newly diagnosed babies. It's hard enough managing a new baby but learning that they have a rare disease is traumatic.'

Important improvements in rare disease diagnosis

Respondents were asked to rate the most important actions required to improve rare disease diagnosis in Ireland. Actions were ranked in order of importance from 1 the most important to 4 the least important. The most important action was identified as up-skilling GP's in the identification and appropriate referral of rare disease patients followed by improvements in access to specialist genetic diagnostic services, training and education of health professionals at all levels. Improvements in laboratory testing was ranked the fourth most important action, however, there was only small differences in the ranking score of all the actions identified. Notably, patient organisations were most unlikely to rate improvements in laboratory testing as the most important action compared to any other group ($P<0.05$).

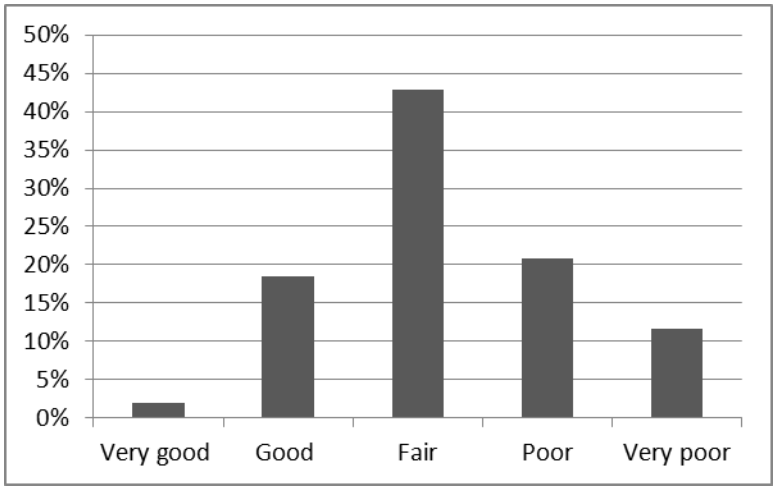
Key points – Diagnosis

- Delays in diagnosis were the predominant concern among respondents. This issue was prioritised by carers, patients and health service providers.
- Around one third of respondents considered the quality of diagnosis to be poor/very poor.
- The challenge of diagnosis of rare disease brought about issues for both patients and their health service providers – some patients perceived a lack of interest in rare disease symptoms and insensitive or ill-informed communication of the diagnosis by clinicians.
- Additional time, effort and clinical skills in history taking and physical examination by the clinician rather than repeated tests were seen as important components of reaching a diagnosis.
- Travel to specialist centres abroad was seen as another opportunity to expedite appropriate diagnosis but this was poorly recognised within the current system.
- The lack of any system to document, analyse or compensate patients for misdiagnosis was highlighted.
- Respondents viewed that improving diagnosis of rare diseases required action across all levels of the health service.
- Supporting GP's skills in appropriate referral of rare disease patients and improving access to specialist genetic diagnostic services represented core actions needed to improve diagnosis.

3.2.4 Quality of Care

Respondents were also asked to rate the overall quality of care for people with rare diseases in Ireland. Almost 5% of respondents did not know how to rate current care provision. Of those who did, few rated it as good or very good (20%). The majority of respondents rated it as fair (42.9%) and a large proportion rated it as either poor (20.8%) or very poor (11.6%).

Figure 2. Rating of care in general for rare diseases in Ireland



High proportions of health service providers/managers and health policy respondents rated the quality of overall care for rare disease patients as good or very good compared to any other respondent category. Conversely, higher proportions of patients, carers, researchers and patient organisations rated the quality of care as poor or very poor.

Patient-doctor/healthcare provider relationship

Experience of the patient-doctor relationship was mixed. Some respondents acknowledged the role of consultants who have shown a keen interest in rare diseases while others perceived a level of disinterest in the context of a rare disease consultation. The lack of knowledge of rare diseases was cited as a challenge by healthcare providers as well as patients in that despite their interest; they struggled with accessing appropriate information and treatment options. Doctors with good intentions did not know what to do or how to proceed, and tended to adopt what one respondent termed a

'…'live with it' approach because of the lack of treatment options available.'

Limited awareness of the diseases under consideration was combined with limited health care options available to the clinician and ultimately the patient. Respondents acknowledged the challenges for interested clinicians within the health services:

'The lack of knowledge on the part of the medical profession. I am lucky to have a consultant who shows great interest but he admits he doesn't have the answers to some of my questions. I rarely come across a medic, be it a doctor, nurse or physio who has actually heard of illness.'

The need for more information about rare diseases and somewhere to turn to for care and support services was a frequently occurring theme cited as a challenge for quality rare disease care provision. One parent expressed concern that they were able to access more information on their child's condition from the internet than from the health care services.

'My child's paediatrician has told me several times that I know more about my child's condition from the internet which is very worrying and in my opinion unacceptable.'

'trying to get help, no one knows anything about the disease…because disease is terminal no one bothers with my son.'

Respondents had accessed both information, and in many cases treatment, abroad, as the health service in Ireland was unable to provide the necessary care for their condition.

From 'revolving door' patients to packages of care

Issues concerning care coordination and management as well as access to support services featured strongly in responses. Respondents referred to challenges with a wide spectrum of care services not just specialist input from clinicians in primary or secondary care settings. The complex care needs of a rare disease patient were evident, as was the requirement for comprehensive packages of multidisciplinary care that encompassed the needs of the carer as well as the patient. Access to rehabilitative services such as occupational therapy, speech and language therapy and physiotherapy were commonly felt to be lacking – either through lack of a service, waiting lists or confusion over eligibility. Many respondents highlighted experiences of limited access to care and inadequate care provision including support services:

'Support…is important and ways of improving the support services should be considered e.g. extra help in the family home which should be available 24/7 if needed, family respite services, suitable transition care from paediatric to adult care and finally appropriate end of life care provided.'

Access to allied health services such as occupational therapy, speech and language therapy and physiotherapy was frequently highlighted. Limited care options as well as the fragmented nature of care provision was also highlighted. Patients felt that their conditions were poorly managed once diagnosed and this added to the treatment burden of their diseases, often resulting in them becoming *'revolving door patients'* in emergency departments or settling for inadequate care:

> *'…there isn't a consistency in the care provided, we often find a different doctor each time we visit a clinic, whereas if we saw the same doctor each time I feel there would be more uniformity in the treatment of the disease.'*

> *'The lack of information for patients, the lack of expertise by the medical professionals, the lack of early diagnosis in primary settings (GP's) lack of funding to send patients to better equipped countries.'*

> *'Multiple trips to A&E is not the answer. When you have an on-going condition, you are not an emergency. I personally just get sent home, and told to wait for my next specialist appointment. Once I was given a two month prescription for paracetamol to help me wait the two months to see a consultant.'*

Respondents consistently emphasised the need for appropriate infection control facilities for relevant rare diseases patients such as cystic fibrosis and called for the provision of suitable units as a matter of urgency.

Barriers to achieving good care outcomes

Respondents were also asked to identify the main barriers to achieving good care outcomes according to a list of options. These are summarised in Table 3 along with the proportion of respondents identifying the main barrier in descending order. The main barrier identified by almost all respondents was the lack of a clear transition from paediatric to adult services. The misunderstanding of the non-clinical needs of patients, such as education, employment and disability challenges was also identified as a primary barrier to good care outcomes for people living with are diseases in Ireland. A large proportion of respondents also cited the financial burden borne by patients as an important barrier. Issues with no defined care pathways for patients as well as waiting lists were identified as significant barriers to good care outcomes, while over half of respondents identified the poor co-ordination of care as an issue. 56.5% of respondents felt there was a lack of knowledge among healthcare providers which was also acting as a barrier to good quality of care for rare disease patients and 41.4% of respondents felt that

late diagnosis of diseases was impeding the standard of care provided to patients in Ireland.

Table 3. Main barriers to achieving good care outcomes for people with rare diseases

Main barriers to achieving good care outcomes for people with rare diseases	Percentage response
No clear transition from paediatric to adult services	97.3%
Misunderstanding of the needs of disease patients outside the health services e.g. education, employment, disability	84.8%
No clear care pathway for patient	76.9%
Waiting lists	76.3%
Lack of respect and support for carers	68.6%
Cost to the patient	58.8%
Lack of knowledge by healthcare provider	58.8%
Poorly co-ordinated care	56.5%
Disease is too advanced at time of diagnosis	41.4%

One respondent emphasised the need for the health service to adopt a proactive approach to identifying rare disease patients rather than assuming a reactionary approach to rare disease care:

> *'Plan should ensure that patients with rare diseases are actually found. Perhaps there should be targets! The doctors should be encouraged to find the patients. Patients whose GPs or other doctors are puzzled should be given a route to diagnosis..'*

It was suggested that an approach to red flagging potential rare disease patients should be adopted in the health services and such patients should be identified for assessment by a rare disease specialist.

> *'If a patient is seeing four or five different consultants it should be red flagged to see a rare disease specialist and save years of misdiagnosis!'*

Key points – Quality of Care

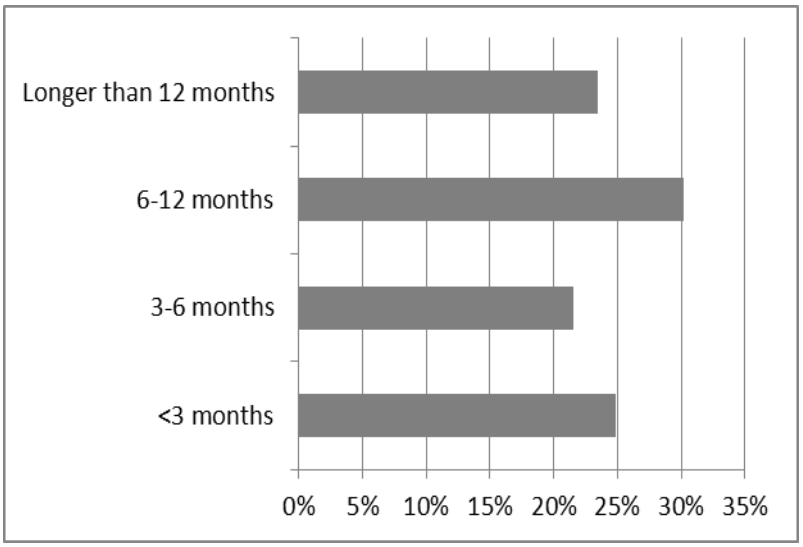
- Quality of care and quality of diagnosis received broadly similar ratings with almost one third of respondents rating the quality of care as poor/very poor.
- Rare diseases brought unique challenges to the doctor- patient relationship– for example addressing limited clinician knowledge on the rare disease, 'patient experts' and the appropriate use of information from many sources including the internet.
- Many responses revealed complex care needs among rare disease patients that required multidisciplinary input.
- Rare disease patients commonly required a high level of co-ordination between health and disability services.
- Issues related to both specialist and general health services.
- Meeting the needs of rare disease patients with chronic, multimorbid and/or disabling rare diseases appeared to be particularly challenging.
- Appropriate support for care provided in the home for significantly disabled rare disease patients was also emphasised including the need for respite care.
- Lack of care pathways was perceived as a barrier to achieving good care outcomes.
- Respondents highlighted the consequences of inadequate care pathways, care co-ordination and continuity of care- multiple trips to emergency departments or primary care, additional carer stress and diminished outcomes for patients.
- Issues relating to access to and eligibility for physiotherapy, occupational therapy and speech/language therapy services were frequently highlighted.
- Appropriate infection-control and age-appropriate hospital facilities for inpatients with cystic fibrosis were seen as a critical component of optimal care for this group.
- The transition from childhood to adult rare disease services was seen as a major barrier to achieving good care outcomes.
- The lack of understanding of rare disease patients outside the health services for example in the domains of education, employment and disability was also highlighted – this would indicate a need to prioritise holistic packages of care with inter-disciplinary input.

3.2.5 Waiting times and access to care

Opinion on the current waiting time for assessment by an appropriate rare disease specialist varied substantially. This may reflect variations in assessment according to the type of rare disease, where some diseases are better catered for than others. In some cases, there may be no designated specialist or specialist service in Ireland for a disorder which would clearly affect the referral period. Several respondents did not know how long

it takes to be assessed by an appropriate rare disease specialist at present (25.6%). Of those who did comment, a slightly higher percentage thought it was six to twelve months (30%) compared to other
categories.

Figure 3. Current waiting time for assessment by an appropriate rare disease specialist



Most respondents expressed an opinion on an appropriate length of time to wait for an assessment after referral from their GP with 70.8% expressing the view that it should be within one month. Almost all felt it should not be any longer than three months (94.8%). When asked about appropriate waiting times for specialist assessment including genetic counselling for family members who may be at risk, the majority of respondents felt that three months was a sufficient waiting time. Less than 1% of respondents felt it was appropriate to wait for 18 months for an assessment of this kind.

Rare disease patients and carers felt they struggled to access treatment and support for their condition. In addition to problems accessing a diagnosis and healthcare treatment, access to support services was also raised as an issue. Some believed this issue further affected the physical health of an already ill person:

> *'Having to fight and use precious energy and oxygen in fighting for home care and a medical card when in need of a double lung transplant.'*

Moreover, they highlighted the disabling nature of rare diseases and the impact of multimorbid conditions on their health and physical functioning. There was limited recognition of the need for and access to auxiliary services. The dichotomy of health and social services was seen as problematic, especially for patients with no formal diagnosis or those with lesser recognised conditions.

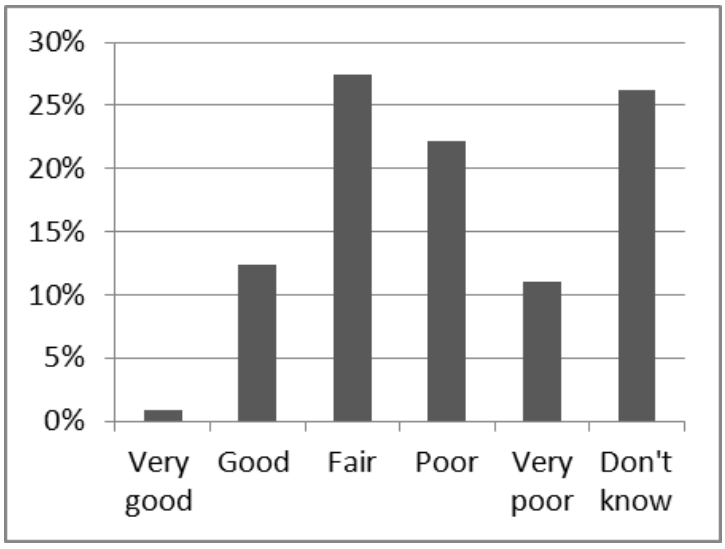
Key points – Waiting times

- Opinions on the current waiting times for assessment by an appropriate rare disease specialist varied substantially
- Responses suggest that between 1 in 4 and 1 in 5 of rare disease patients in this consultation wait over a year for assessment by an appropriate rare disease specialist
- Respondents felt that an appointment with a specialist should follow within 3 months of referral by a GP
- Respondents considered that assessment including genetic counselling for potentially affected family members in families where a genetic disease has been detected, should occur within 3 months also.

3.2.6 Access to Orphan Drugs and Technologies in Ireland

Respondents were asked to rate current access to orphan drugs and/or technologies for people with rare diseases in Ireland. 26.2 % of survey respondents did not know how to rate this, suggesting that it was not relevant to all rare disease patients. However, of those who did a substantial proportion rated access as fair (37%), almost half (45%) rate it as poor or very poor and only 18% rated it as good or very good.

Figure 4.Rating of current access to orphan drugs and/or technology.



When it came to accessing orphan drugs, there was a strong aspiration that more could be done to speed up the process and make drugs more readily available as warranted by the specific needs of a person with a rare disease. Respondents felt that the process should be made more transparent and fair, and that the current process is too slow and constrained:

'make it less bureaucratic and complicated to process the assessment..'

The expense of orphan drugs was widely discussed, and a number of respondents expressed a view that certain drugs were beyond reach due to cost. Respondents expressed strong concerns that the difficulties accessing appropriate medications experienced by rare disease patients should be acknowledged and addressed. Furthermore, a number of respondents highlighted a perception that medications to treat certain conditions were available in others countries but not in Ireland.

Respondents felt strongly that treatment for rare diseases should not be constrained by financial considerations. Decisions on drug treatment were viewed as medical decisions solely that should be, left to the doctor, the patient and the medical team.

'At present the availability of medicine is based on a case by case basis and the opinion of the person making the decision. Often a person in Ireland will have to involve media or go on the Joe Duffy Show in order to get access for the medicine. These decisions should be fair and equitable to everyone and based on policy.'

Respondents stressed that there should be a fair process in place for accessing medications and treatments for all patients, regardless of how rare a condition may be. In addition, some respondents believed there should be a special reprieve applied to

treatment of rare disease patients, and that orphan drugs treatment should be funded from a ring-fenced budget, acknowledging the high cost of some rare disease medications. Pharmaceutical companies responding to the consultation were supportive of new ways of assessing the needs for orphan drugs, and emphasised that the process should promote fairness and equity.

A large proportion of respondents (35.1%) did not comment on the need for a second assessment pathway for rare diseases, distinct from the Main Gateway for new drugs and technologies in Ireland. However of those who did comment, the vast majority (89.5%) believed a second assessment pathway was a requirement. Likewise, a large proportion (28.5%) of respondents did not know if the commissioning of medicines for rare disease patients should be the responsibility of an Advisory Committee mandated and reported by the Minister for Health. Of those who did voice an opinion, 82.2% believed that the commissioning of medicines for rare diseases should be the responsibility of such an Advisory Committee.

Issues relating to the availability of orphan drugs through clinical trials were also raised. One respondent depicted the diverse experiences of being on the text treatment or standard treatment arms of a clinical trial for her family members with the same condition:

> *'My family members…[signed up for a drug trial] one…received the new drug to try with great results, the other was given a drug which she had already been on many times before with no improvement – she stayed with the trial and ended up [in hospital for condition][…..] If both…would have been given the new drug on the trial instead of it being a lottery, they would both be benefitting.'*

Key points – Orphan Drugs and Technologies

- Many respondents did not respond to this section suggesting that the issue of orphan drugs, while important, was either poorly understood or not as relevant to some rare disease patients and carers as others.
- Of those who responded, nearly half considered that access to orphan drugs and technologies in Ireland was poor.
- Concerns were raised in terms of the transparency and fairness of decisions made on the assessment of orphan drugs.
- Over one third of respondents were unable to comment on issues relating to assessment pathways for rare diseases in Ireland but among respondents the majority favoured a new system.

3.2.7 End-of-life

Palliative care

The need for appropriate palliative care provision was raised as a priority issue in the consultation. Respondents expressed a clear need for appropriate end of life support for rare disease patients of all ages and respondents felt this should be expressly stated in the Plan:

> *'A section on palliative care should be huge in relation for the plan as for most this is the result. A plan for children's hospices countrywide with respite care and end of life care for children with rare diseases is a must.'*

The familial aspect of rare diseases was seen as particularly problematic in that children may be familiar with the end-of-life care needs of parents who had the same condition. Respondents further stressed the importance of dignity and respect in relation to end-of-life care needs at all ages and the importance of consulting with patients and families in relation to advance directives and other end-of-life care decisions.

Bereavement support

Bereavement counselling was also considered as an essential support service for families struggling to cope with the loss of a loved one, often at a young age. One bereaved relative described their experience:

> *'Nothing has, or perhaps could have, prepared you for the shock, the finality, the hopelessness, the isolation, the depression, the despair, and ultimately the challenge of how and where to find the resolve and fortitude that will be necessary in order to die, or hold the hand with dignity and peace of a loved one who has been told that they are now going to die sooner rather than later. Or how to survive the sorrow and loneliness when they are gone'.*

Key points – End-of-life

- Respondents emphasised the need to develop appropriate provision for the end-of-life phase of rare disease patient journeys.
- A significant proportion of responses focussed on end-of-life issues for children with terminal rare diseases including hospice and respite services.
- Bereavement issues also featured in many responses. For some families with rare genetic disease, the bereavement process was complicated by affected children observing their parent dying from their disease or families with more than one child affected.

3.2.8 Research

Respondents stressed the need for generating an evidence base on rare disease diagnosis and care in Ireland. Improved research into rare diseases and the need for in-depth knowledge of specific diseases as well as the care needs of patients was emphasised throughout the consultation responses. Respondents highlighted the gaps in research for particular conditions but also for rare diseases in general. Funding for rare disease research and ethical concerns emerged as two major themes in the consultation.

The importance of having a national registry of rare diseases was highlighted by a number of service providers, researchers and patient organisations. One respondent also pointed out the need for a record of all patient groups or organisations associated with rare diseases. The need for a registry to guide the development of a National Plan was stressed by one respondent:

> *'Start with registry development with reviews built in. It will give [a] basis to develop [the] strategy based on [the] info provided.'*

Another respondent stressed the need to develop existing registers of major congenital malformations. It was not specified if this should be part of an overall rare disease registry.

> *'A national registry of major congenital malformations is essential and is present in other EU countries. This is mandatory to allow audit and assessment of patient needs and diagnoses....'*

There were also calls for strengthening of prevalence data and more comprehensive epidemiological research into rare diseases in Ireland in order to guide care provision:

'Epidemiological data is required to ascertain the exact number of people with rare disorders on the island of Ireland. This information is essential in exploring the need for and the provision of resources.'

'Research and registries to collate epidemiology data on rare diseases is important. We need this information to be able to plan appropriately for rare diseases present in the Irish population. Health research into the effects on patients regarding their experience of living with a rare disease and experience of health related services they have received to help evaluate the current state of play and what is needed for the future.'

Improvements in research were widely considered as an essential first step to increasing awareness and knowledge. Conversely, it was also pointed out that the lack of awareness of rare diseases limited the potential for researching rare condition. Funding for rare diseases is limited and there are limited resources for interested professionals to focus on research interests. Gaining funding support for research was seen as a major challenge. One researcher described the current research situation:

'There is almost no funding for basic research at present – graduate and post-graduate students have no career progression and are being lost to science.'

A healthcare professional completing doctoral research summarised her inability to secure funding for a rare disease project:

'…I have applied to many organisations for support […] however I have been unsuccessful so far for any form of support…I am a full-time employee and a part-time researcher, all my annual leave and my time I have dedicated for this study.'

Conventional funding application assessments are seen as problematic and respondents highlighted that funding for rare diseases should not be assessed in the same way as funding for research into more common illness:

'Applications for funding to national finding bodies are judged in part according to their scale of impact. This will count against applications in the field of rare disease research which may benefit only a small number of patients within Ireland compared to come prevalent medical diseases.'

In addition, some respondents perceived an over reliance on international research and while research collaboration with European partners was seen as positive:

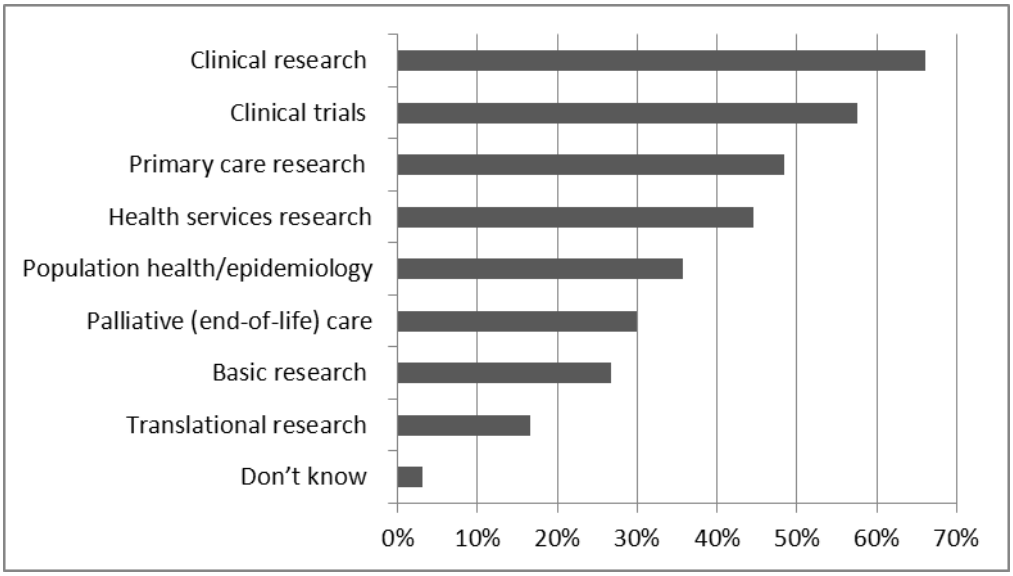
'We currently have designated centres of excellence. One such centre have [sic] advised me that they are not interested in doing research. Ireland is more likely to be involved in clinical trials which are organised from Europe as happens in cancer treatment.'

Respondents were also concerned about establishing an appropriate ethical framework for rare disease research in Ireland. Researchers highlighted the need to provide clear guidance in the hope that research priorities would be protected within an ethical

framework. The importance of retaining the genetic information of screening samples for future research was reiterated throughout the consultation.

Respondents also indicated which areas of research they felt should be prioritised in the rare diseases plan (see Figure 5). Clinical research (66.1%) as well as clinical trials (57.6%) appeared most often in respondent's top four listing of research priorities. These were followed by a focus on health services research (44.5%), population health or epidemiology (35.8%) and to a lesser extent palliative care (29.9%), basic (26.8%) and translational (16.6%) research. These findings should be interpreted with caution as the distribution of research priorities may be in part reflecting understanding and familiarity of the terminology of different research disciplines. In addition, qualitative findings support the need for prioritising a wide range of research areas to develop an evidence base in Ireland.

Figure 5. Research priority areas



Respondents were asked to rate the importance of a range of items in enhancing rare disease research in Ireland. These were ranked from 1 to 5 in order of importance, where 1 was most important and 5 was least important. Mean response scores were calculated and responses were ranked in order of importance from the highest to the lowest response sore. Establishing Centres of Expertise was widely considered the most important action for enhancing rare disease research, followed by sustainable funding mechanisms, international collaboration and networks to support and develop rare diseases in Ireland. Developing leaders in research was considered the least important action compared to all

others however, again this should be interpreted with caution as all actions were considered important, with very little differences in mean response scores.

A large proportion of respondents did not know if Ireland should develop its participation in the main European approved portal Orphanet (23%). However the majority of those who did have an opinion agreed with this development (76.3%).

Respondents were asked to comment on important issues to ensure that maximum benefit can be achieved from rare disease registries. Support for analysis and sharing of findings from registry data and clear funding mechanisms and integration with clinical outcomes were seen to be the most important benefits. Other important issues to ensure maximum benefit can be achieved from rare disease registries include ability to provide data to international registries where no Irish data is available, to provide infrastructure to support a single rare disease registry and support a federation of rare disease registries. Very little differences were detected between responses on these benefits, although the importance of registries in providing lists of patients for clinical trials was considered as important. 9% of respondents did not know if this was an important benefit from registries, while 8% did not agree that it was important.

Key points – Research

- Better basic health intelligence on the number of people with rare diseases was seen as critical to awareness and advocating for appropriate service and policy development.
- Clear policy, data protection and ethical frameworks were seen as critical for rare disease research.
- Many types of research were recognised as important with clinical research and clinical trials particularly emphasised.
- Health service and primary care research were also prioritised.
- Development of basic epidemiology of rare diseases in Ireland through rare disease registries was seen as a priority.
- The development of designated centres of expertise and sustainable funding mechanisms were the proposed priority actions for enhancing rare disease research.
- Irish involvement in Orphanet was seen as positive but underdeveloped.
- Rare disease registries were considered a critical tool to support many types of rare disease research. Respondents were supportive of either a single rare disease registry or a federation of rare disease registries.

3.2.9 Geographic and Social inequalities in care provision

Respondents highlighted differences in access to and quality of care between geographic regions pointing to the concentration of expertise and speciality services in Dublin. The lack of suitable local facilities forces many patients to travel to services in other areas, namely Dublin. This geographic disparity in services was seen as unfair and an added burden to patients and families in other urban or rural regions in Ireland. Some respondents felt the disparity in service provision resulted in poor diagnosis and care outside of Dublin:

> *'We live in X and if there are any issues with our son it's a long way to Dublin also if we lived in Dublin he wouldn't have spent 2 weeks in the local hospital with no one knowing what was wrong and as a result of that would not have brain damage from his disease.'*

While some services are provided at a local level for children with rare diseases, once they reach adulthood they no longer have these services available locally and must travel to regional hospitals for treatment:

'[hospital] has a consultant in the area but does not have the support services. When children reach the age, they must then go to Cork/Limerick/Dublin for treatment. This is unacceptable…'

Travel was a major issue both in terms of the burden of travel in and of itself and also the additional cost it incurs:

'The lack of local facilities – there is very little understanding or consideration given to the fact that we live so far outside Dublin…the difficulties involved in travelling so far, overnight stays, expense, time away from work etc are completely underestimated…with three children with [illness], two of whom are also autistic, travel is incredibly stressful and it is hard to understand why more services cannot be provided locally…'

In response to the geographic disparities in services, respondents called for Centres of Expertise to be dispersed throughout the country rather than concentrated in Dublin. They emphasised the importance of easily accessible locations for Centres of Expertise, with appropriate transport links including motorway and rail routes.

In addition, the paucity of adequate services, the often serious financial implication of accessing services, and the need to fight for services and supports, may reinforce the social gradient in health, where some families with greater resources are in a better position to access necessary care compared to others:

'At the moment families and organisations have to do so much. Families with higher incomes and better education do much better in managing their child's rare disorder as they have the education and financial resources to research. This is not fair on the disadvantaged/lower socio-economic families.'

Key points – Inequalities in service provision

- The concentration of expertise and speciality services in Dublin created particular challenges for rare disease patients nationally.
- However the need for efficient referral from local hospitals to specialised tertiary referral hospitals for rare disease patients was recognised.
- Inequalities in outcomes for rare disease patients were also related to issues of family income, access to transport and level of education.

3.3 Emerging themes from qualitative responses

3.3.1 Coping with a Rare Disease – isolation and loss of independence

The unique position of rare disease patients, their families and their carers was expressed throughout the consultation. Feelings of isolation, loss of independence and social exclusion due to illness were widely described by those with a rare disease. It was perceived that a lack of information about the particular illness and access to informed health professionals often exacerbated the situation. There was a resounding call for a more holistic approach to caring for people with rare diseases including a focus on counselling and psychological support for patients:

> *'Isolation. There is no one really to talk to about coping with the disease. It is very lonely.'*
>
> *'Isolation, no other with similar condition to share with/compare unknown future..'*
>
> *'Isolation – difficult to meet others who share the same issues..'*
>
> *'I went from being a very independent 25-year old to someone who needs the support of others in almost all areas of my life…My GP while helpful has not the time to try and understand all the complications that are part and parcel of my disease and so often I feel desperately alone because nobody understands'.*
>
> *'I will have this disease for the rest of my life…life for me is like dominoes, one complication triggers another and another and there is no let up. My life has been turned upside down by this, I worked full time, I was always happy and lived life to the full…I shouldn't be this sick or unhappy, I should be able to go about my daily routine as normal…Lazy was never a word that would have been associated with me, I almost died out of not wanting to take time off work to attend a doctor…now I can barely walk down the street without pain and it's only a matter of time before people see this healthy looking girl as a lazy layabout…Counselling is a massive issue, I have yet to see anyone…So all I would ask is that this National Rare Disease Plan take in a holistic approach to supporting patients who suffer from these rare diseases and really try to understand what a devastating disease this is.'*
>
> *'…dealing with the social exclusion involved as well as loss of income and independence at age 45…'*

Patients stressed the burden this experience brought on in what they perceived as a largely unsupportive environment.

Key points – Coping with a Rare Disease – isolation and loss of independence

- Social isolation emerged as a common theme – this included an element of isolation from other rare disease patients with the same disorder.
- Adapting to disability, loss of employment and loss of independence at a young age were common experiences.
- Social exclusion was also featured in responses.
- Counselling and psychological support were recognised as important services on the rare disease journey.

3.3.2 Financial implications and access to entitlements

The critical need for financial support for rare disease patients, carers and families was strongly expressed throughout the consultation responses. Rare diseases directly impact on a patient's and family's financial situation, often exacerbated by limited access to financial support. The main threats to their financial stability were cited as:

- limited employment options;
- out-of-pocket treatment and medical expenses, especially when required to travel for treatment abroad;
- limited access to entitlements including the long-term illness card and medical card;
- limited access to health insurance and life assurance.

For example a person may have to stop working due to their illness, or if they continue to work they may not be entitled to a medical card and will not be in a position to pay out-of-pocket for expensive medications and treatment. A parent may have to give up work to care for their child and sometimes several children with a rare disease. Financial issues come from many directions, creating more stress in an already difficult situation. It can be especially difficult in families with one or more members affected by a genetic condition or where parents and children have a rare disease. The lack of care and treatment options provided by the State for rare diseases may force a patient or family to pay for their own care, including especially allied health services such as physiotherapy and counselling. Limited service provision often contributed to the general financial difficulties described by many respondents:

'It's a major struggle for patients and carers trying to cope as best they can when the government keep cutting the vital services and financial help they need.'

One person with a rare disease described the constant worry about her financial situation:

'At present, I do not have a permanent position…This has left me struggling to pay rent and food bills, without even considering the cost of medication, consultant appointments and annual hospital visits. I understand that the government is under huge pressure with it comes to finance and budgets, but my life would be greatly enhanced if I did not have to worry so much each month about covering my medical cost.'

Entitlements and financial burden

Another respondent described how the medical card issue made accessing medication difficult for her, as she was struggling to retain her day-to-day life despite her illness:

'…it is impossible to receive a medical card while working. I work with my condition to keep my self-esteem but struggle with medication costs and require private health insurance.'

It was pointed out that early intervention and support for patients and families would be cost effective for the State:

'…I need the HSE to see the huge gap in services for children at the severe level, my husband…[is] self-employed working 14 hours every day to pay our mortgage we are using all the carers money and more to fund the [specific treatment] …I am trying desperately to save the state from [son's name] needing a full care package when we are no longer able to manage his behaviours when he is older, I am trying to save the state €100000 a year by helping my child when he is young enough to learn…'

In other instances, the financial pressures come from other parts of the system. This lengthy quote describes the difficulties of getting a pension:

'As I was a homemaker and the mother of X children never having the time or opportunity to be employed outside over the years and unable to work because of my disease I then had no stamps to entitle me to a state pension which is a disgrace for any government…I am deemed not worthy of ANY pension from the state and more serious still having reared X children and funded their education on my husband's single salary we had to borrow exorbitant amounts of money over these years to pay doctors and consultants in this country and in London for the care and help necessary with my condition.'

One particular issue was the non-inclusion of many rare diseases on the Long Term Illness (LTI) list - a very high proportion of respondents asked that the list be updated. Some of the more common rare diseases are not on the LTI list, and an even lesser number of ultra-rare diseases are present. Also, the procedure of having to re-apply for a medical card

every two years was frustrating and seen as an unnecessary burden imposed on patents and carers:

'I would ask that the National Disease Plan update/extend the list of long term serious illnesses which are automatically granted Medical Card statuses…This enables the person to access OT support, Speech & Language etc which they will badly need without an exhausting battle from their carer. Also, for all these conditions added to the list of long term illness please make it easier to renew the Medical Card without the traumatic form each two years where the carer has to almost prove again that their loved one has not 'recovered' from their illness.'

'My rare disease is not covered by the long term illness scheme and I do not 'qualify' for a medical card, yet I spend the full amount on the drug payment scheme every single month without fail, have to fork out for health insurance as well as paying €100 every three months to see my consultant, or even more frequently if I am not in remission.'

The issues faced by patients who do not have access to Long Term Illness protection or medical cards was exacerbated when other expenses were added in, and many patients expressed their experience of poverty borne as a result of the high costs associated with their illness. Patients are forced to cut-back on spending often at the expense of medical treatment:

'…without insurance or medical card I still have to pay €132 for meds, €75 every eight weeks for hospital treatment and €50 every time I need to see my GP…My mortgage is over €1450 a month before I even buy food or pay a bill so guess where I have to cut. Yep the meds and GP visits if I miss work I need to provide a doctor's note which I cannot obtain without spending €50!'

Health insurance and life assurance

A number of patients raised the issue of inability to access insurance and problems with receiving insurance pay-outs and the difficulty this can present and they highlighted the need for research to raise publicity in this area and specifically training for health insurance companies:

'I have a critical illness with a life expectancy of less than 5 years, I have a critical illness policy I took out many years ago, with [insurance broker], yet they say that my illness is not recognised as a critical illness and refuse to pay up.'

'Getting adequate insurances such as mortgage protection and health insurance. Medical underwriters don't have the knowledge or understanding of rare medical conditions.'

'We need to make people aware, we need a voice, we need to be heard. A rare disease normally means a lifetime disease, which I have fought with insurance companies etc. on recognising it. I mean nothing to them, and because [illness] is genetic, I have to fight for my children, so they will have a path paved for them..'

Key points – Financial implications and access to entitlements

- A reduced capacity to engage in paid employment on the part of rare disease patient and carer was a feature of the financial burden of rare disease.
- Out-of-pocket expenses were an issue, including medication, travel and disability needs.
- Paying for private care or care abroad where a service not available or accessible in Ireland – including private physiotherapy and counselling – also featured.
- Medical card eligibility for rare disease patients was often unclear and was commonly a cause of frustration.
- Many respondents emphasised that the long term illness scheme does not adequately encompass rare diseases.
- Rare disease patients were left in financially precarious situations due to issues relating to health insurance, life insurance payouts, critical illness policies, mortgage protection.

3.3.3 The Role of Carers

A substantial number of consultation respondents were carers, emphasising the disabling impact of many rare disease conditions and the high level of dependency of many rare disease patients. Several of those who responded had a rare disease and were also caring for their child or children with a rare disease. These respondents emphasised the burden felt by the families and carers of patients with rare diseases exacerbated by poor support services. They argued the need and benefits of supporting carers in their support role of caring for rare disease patients such as homecare support:

> *'Please do not underestimate the toll it takes on a family to care for a person with a rare disease. It is of primary importance to support the family with as much as possible. It is much, much easier for a family to function effectively with support provided from home. It is also more cost effective for the family and the state providing the care…one other massive advantage is that all family members become very medically skilled in taking care of their love done. Hospitals are way less overcrowded, doctors have more time to troubleshoot, the State is less burdened in so many ways..'*

The pivotal role of family carers was highlighted throughout the consultation. Their feelings of responsibility and heartbreak associated with delayed diagnosis and missed opportunities for treatment, and the struggle to access treatment was highlighted many times throughout the responses:

'I hate that my son has brain damage and will never walk or talk due to the lack of [type of screening] and it kills me every day to know I knew something was wrong before the damage set in but no one would listen to me and then it was too late…that is very hard to deal with and another issue is that I knew the next child to be diagnosed with [son's] disorder is also going to have brain damage and that rests on my shoulders as I feel I should be fighting to stop that happening.'

'My experience while caring for my wife was I received no help at all as I was self-employed. Now I care for my daughter I have been able to access much more assistance, but because she is only 44 some services are not available, even though her condition is terminal. Why???'

'Access to public health services was extremely difficult to organise and there was a huge amount of paper work involved. Thankfully I was able to access information for my mother as she would not have had he strength or the ability to look up information via computer.'

The fact that many of those with rare diseases are children put even more pressure on their parents as carers to provide for them. In one case, an entire family had to emigrate from Ireland to access care abroad:

'…from my own experience with a son who is incredibly complex medically the support system to enable us to care for him at home is terribly lacking…with the right support we could have taken care of him at home. As a result we had to emigrate…'

Difficulties in accessing information about drugs and treatments contributed to the carer burden, again even more so for those who were fighting for their children. Given the significant burden borne by carers of persons with a rare disease, the carer's allowance was widely seen as insufficient, especially considering the high level of care support that many rare disease patients require.

Key points – Role of Carers

- Carers were the most common respondent to this consultation, representing 37% of responses.
- Many carers were parents of children with a rare disease, carers of spouses and parents were also commonly represented in responses.
- Caring for more than 1 effected family member was commonly described.
- Carers of rare disease patients commonly provided a high level of care in the home as well as acting as advocates.
- Support services for home care including allied social welfare allowances and benefits were perceived as inadequate.

3.4 Implementation of the Rare Disease Plan

3.4.1 Commitment to implementation

The development of a National Rare Disease Plan was widely welcomed as an important basis to improve services for rare disease patients and greatly encouraged. However, the need to follow through on the implementation of the Plan was emphasised to ensure that necessary improvements in service provision are realised.

> *'The political will to actually act upon any findings in the Plan. It shouldn't just be a paper exercise'*

There was a call for independent monitoring to ensure effective implementation of the Plan:

> *'There needs to be real time frames to bring about changes and improvements, it's not good making plans if they are not going to actually [be] pursued vigorously. It needs to be monitored by an independent group which will not bow to any sources, be that government or HSE. And guaranteed funding is needed so as not to be dependent on however the current financial climate is at the time.'*

There existed a strong sense that accountability was the only way the Plan had a chance of being successfully implemented. One carer envisaged the accountability process thus:

> *'...a named person needs to be responsible for implementing the Plan. A dedicated unit and budget must be ring fenced for implementation of the Plan…'*

Also, patient feedback on the efficacy of the Plan was mentioned as a way to ensure effective implementation.

> *'Let their voices be heard. Health providers don't know people with rare diseases exist unless they speak up.. If they speak up then people will recognise that they exist.'*

Some participants recommended that preparatory work needed to be under-taken in advance of developing an Implementation Pan. An emphasis was placed on development of infrastructure to support rare disease research and care in Ireland. One respondent called for improved recording and monitoring of rare diseases in Ireland as priority to provide an adequate evidence base to inform implementation.

3.4.2 Centres of Expertise

Almost all respondents (92%) held the view that a newly developed clinical care programme should be developed within the Health Service Executive (HSE). While 6.5% of respondents did not know if this was the best approach, and 1.5% of respondents disagreed with this approach. Similarly large numbers of respondents agreed that Centres of Expertise (CoE's) should be designated by the clinical care programme (90.8%) while 3% disagreed with this and 6.1% of respondents were unsure. Respondents also put forward their views on the core functions of the CoE's. The vast majority of respondents agreed with the pre-specified functions listed in the questionnaire. Treatment and delivery of multidisciplinary care was identified as the most important core function of CoE's followed by support for the development of care plans in partnership with specialists and GP's. The role of CoE's in support for research into rare diseases and education and training of health services was also widely endorsed. While monitoring the quality of diagnostic and treatment services and informing service planning and development yielded lower numbers in agreement, a large majority of respondents believed these to be important functions for CoE's.

Table 4. Core Functions of Centres of Expertise

Suggested core functions	Percentage in agreement	Percentage not in agreement	Don't know
Treatment and delivery of multidisciplinary care	95.2%	1.7%	3.0%
Support the development of care plans in partnership with specialists/GP's	93.5%	2.8%	3.7%
Support research into rare diseases	92.4%	4.1%	3.5%
Education and training of health service professional	91.9%	4.8%	3.3%
Monitor the quality of diagnostic and treatment services	88.7%	4.6%	6.6%
Inform service planning and development	82.9%	4.1%	13.1%

Respondents were asked if they agreed that protocols for treatment and monitoring very rare diseases and if these should be linked to Centres of Expertise. Of the 81.3% who submitted a view, the majority (81.7%) were in agreement with this statement.

Respondents overwhelmingly agreed that in cases where there is no suitable specialist service at home, Irish patients should be provided with access to European reference networks (95.7%). A small percentage of respondents were not sure (4.3%), however no respondent felt that this access should not be provided. Opinion was more divided on the role of the State in reimbursing travel expenses for consultations, treatment and clinical trial involvement outside of the State for ultra-rare conditions when there is no licensed treatment or appropriate specialist available. In total, 81.4% of respondents agreed that reimbursement should be provided for consultation and participation in appropriate clinical trials, however substantially more researchers (30%) and health service providers (22%) and company's (20%) disagreed compared to patients or carers and friends of patients (<5%). 59.4% of respondents agreed that reimbursement should be provided for treatment only. Fewer researchers and health service providers agreed with this compared to any other respondent group. Over 90% of respondents agreed that the State should reimburse travel expenses for treatment and follow-up for ultra-rare disease patients. Again, there was a clear distinction in levels of agreement between health service providers and researchers who were more inclined to disagree compared to other groups. Almost 70% of respondents agreed that reimbursement should be provided for participation in clinical trials of new therapies, although health service providers and researchers again were less inclined to agree (42.6% and 48.3% respectively).

For ultra-rare conditions where there is no licensed treatment or appropriate specialist available in Ireland, respondents were asked if they agreed that the State should reimburse travel expenses for a range of healthcare expenses. Of those who expressed an opinion, a large majority of respondents agreed that expenses should be reimbursed for all treatment procedures and follow-up (97.4%). Between 9% and 17% of respondents did not know whether patients should be reimbursed for consultation and participation in appropriate clinical trials and participation in clinical trials for new drug therapies. Of those who did express an opinion, 81% and 70% respectively, agreed with reimbursement in such cases.

Table 5 State reimbursement of travel expenses for ultra-rare conditions.

State reimbursement of travel expenses for ultra-rare conditions.	Percentage yes	Percentage no	Don't know
Consultations and participation in appropriate clinical trials	81.4%	9.4%	9.2%
Participation in clinical trials of new therapies	69.9%	13.6%	16.8%

3.4.3 Funding and resources

One healthcare group representing clinicians argued that for the Plan to be successful additional resources are required, mirroring the concerns of patients and carers:

> *'…existing health, social care and educational resources would not be sufficient to meet the challenges posed by the plan, if persons with rare diseases, and their families are to receive the most appropriate, earliest and best possible are when, and there, they need it.'*

In order to fund the Plan, 12.8% of respondents believed that additional resources should be sourced from taxation, while the majority believed it should come from elsewhere within the health system (44.5%) and a large proportion of respondents did not know how to answer (27.4%). 15.3% of respondents cited other sources as alternative means of funding additional rare disease services. Fundraising and public donations were suggested as a way of increasing funding as well as finance from drug companies, the National Lottery, encouraging philanthropy through tax based schemes and international bodies. Several respondents commented on the need to increase efficiency in the HSE and in public services to fund important health care for people with rare diseases. One respondent queried if current resources were being utilised properly and others suggested that more efficient functioning of the health services overall would mean a reduced need for extra funding in rare disease services, for example improved IT infrastructure and integrated care pathways. Another respondent advocated for universal access to health services where rare disease patients would have the same level of access to diagnosis and treatment as other patients. In addition one respondent called for VAT exemptions on government funding of research projects.

3.4.4 International collaboration

There was a strong emphasis placed on the need for Ireland to look to international best practice and access to care and research outside of the State. Establishing links to European Reference Networks was seen as important step in developing international links. The vast majority of respondents called for a greater collaboration with international research and for Ireland to follow international best practice in the approach to rare diseases. In particular, international collaboration was emphasised for those with very rare conditions who are not represented by rare disease patient organisations in Ireland:

> *'Many of the severe rare disorders in the paediatric population do not have a national rare disease patient group to represent them and so increased links with international groups is vital.'*

The call for international collaboration was particularly strong when it came to clinical trials. Due to the nature of rare diseases it is essential with full international as well as domestic collaboration, as this carer states:

> *'Need to combine and network the efforts of researchers across the country, and link to international research groups (particularly for clinical trials) in order to achieve critical mass and benefits to patients.'*

Also, as some people with rare diseases go abroad for treatment, collaborating internationally on research may bring benefits to patients based in Ireland who cannot access treatment here.

Domestic collaboration between hospitals and cross-border initiatives was encouraged; however the vast majority of respondents advocated broad international collaboration. This is necessary for research and for broadening expertise, but can also be of benefit for patients who have to travel to receive a diagnosis or to access care. One respondent had gone 12 years without a diagnosis until she was diagnosed abroad, and writes:

> *'There is a big need to fund a patient to get the urgent necessary investigation and treatment wherever and in whatever country that is available in when a patient needs it urgently. In my case I had to go to [abroad] to find a neurologist who was able to provide this and again…when my condition had deteriorated seriously. Because of the urgency in the need for proper help…we had to borrow from the credit union to fund this…'*

Key points - Implementation

- The development of healthcare infrastructure was seen as a core component of a Rare Disease Plan. In particular, participants called for the development of appropriate rare disease registries to inform a rare disease evidence-base in Ireland, a new Clinical Care Programme in the HSE dedicated to rare diseases and establishment of Centre's of Expertise.
- Respondents highlighted the core functions of the CoE's as treatment and delivery of appropriate care and support for rare disease patients as well as the development of care plans in partnership with relevant providers.
- Establishing links to European Reference Networks was seen as important as well as support for treatment and follow-up abroad when appropriate care is not available in Ireland.

3.4.5 Involving and empowering rare disease patients

There was strong support for the involvement of patient organisation involvement at all levels of the Plan development including during the implementation phase. Respondents expressed a preference for a more holistic approach to rare disease care, including a focus on access to care and support services for rare disease patients as well as clinical care. They felt that appropriate involvement of patient organisations would ensure a balanced Plan with a focus on patient's quality of life as opposed to an over emphasis on biomedical care. Important roles played by rare disease patient groups or organisations were clearly identified by respondents, listing the most important roles as: support (85.2%); information provision (78.8%) and advice (72.8%). Other roles identified as very important included advocacy (66.8%) and to a lesser extent fundraising (51.4%) and input on research or clinical trials (50.1%).

The diversity in patient organisation was highlighted and it was noted that that not all rare diseases have a patient advocacy group, and that there may only exist a handful or indeed only one person with a particular rare disease diagnosis in Ireland. People with no established diagnosis; those with an illness that has not yet been recognised; or those with an illness without a patient advocacy group tended to have even less of a voice than

others with a rare disease. This in turn created a specific type of burden as the patient and often their carers felt voiceless.

> *'There is huge variability regarding the size, resources, time and effectiveness of patient organisations.'*

> *'…there is so few patients with that particular disease it can be very difficult for that disease to have a voice and can be easily overlooked for a louder disease affecting more patients…just because you are in the minority doesn't mean yours is any less of an issue and that it still needs the same participation as the other diseases.'*

> *'Enable existing patient representative organisations to cater for patients which fall between two stools. For example, my husband's (RIP) [illness] was not catered for by either [patient organisation X] or [patient organisation Y], even though it has features of both. Perhaps there should be a wider group, e.g. Neurological diseases, overarching these..'*

Key points – Involving and empowering rare disease patients

- There was strong support for the involvement of patients organisations at all levels of the plan
- The most important roles of patient organisations were viewed as support, information provision and advice
- Considerable diversity in patient organisation roles, resources and effectiveness were evident
- Many rare disease patients do not have a relevant patient organisation in Ireland, including those not yet diagnosed
- For diseases not easily categorised, patient alliance type organisations were seen as offering benefit

4. Findings from the Consultation Day at Farmleigh

A series of workshops were held during the consultation day at Farmleigh to guide discussion on key topics of relevance to the development of the National Rare Disease Plan. These included:

Workshop 1 - Data and Research
Workshop 2 - Infrastructure
Workshop 3 - Cooperation and Working Across Sectors
Workshop 4 - Diagnosis
Workshop 5 - Treatment of Rare Diseases including Orphan Drugs and Technologies

The aims of the workshops were to:

- Provide a forum for discussion on the development so far in terms of a National Rare Disease Plan;
- Gather feedback on the comprehensiveness and appropriateness of the subgroups deliberations to date;
- Discuss opportunities and threats in respect of the implementation of preliminary subgroup recommendations;
- Foster further engagement by a range of stakeholders with the online consultation process through the involvement of workshop participants and their contacts.

For each workshop a set of key points were developed in advance to guide discussion, and participants were encouraged to discuss and elaborate on these key points or raise additional points of relevance. Workshops were facilitated by appointed delegates and key points were noted by workshop rapporteurs. A list of workshop leads, facilitators and note takers is provided in Appendix 2. The key points presented for discussion in each interactive workshop are presented below along with key findings generated by the discussion, summarised below under each workshop theme.

4.1. Theme 1 - Data and Research

4.1.1 Key points for discussion

- *If you are involved in rare disease research – tell us about your experience.*
- *What do you believe are the key outcomes we should expect for information and research in the national rare disease plan?*
- *Data/Information/Research: strengths and weaknesses of the current system?*
- *Recommendations in terms of improving the data, information and research we have on rare diseases in Ireland.*

4.1.2 Key findings

- A strategic approach to the development of research into rare, very rare and ultra-rare diseases is required. Research capacity should be developed by supporting researchers in collaborating and applying for funding in Ireland and abroad.
- Significant strengths in rare disease research were identified in Ireland – the common denominator is strong clinical leadership and integration with active patient organisations. Other characteristics include close links with specialised treatment centres and integration with wider European/international networks.
- An increased focus on a broader range of research is required in addition to the current basic biomedical research focus. For example, health service professionals should be facilitated to conduct health services research for rare diseases. Protected time to enable professionals to engage in and conduct research was believed to be an important factor in supporting health services research.
- There was general support for the concept of a single core dataset to underpin a rare disease registry to ensure high quality, standardised data collection. Careful planning around the details, such as purpose, scope and implementation of a single registry would be required. Respondents also felt that appropriate investment would be required to establish and maintain a rare disease registry. The appropriate ethical and legal/policy frameworks need to be in place to support recommendations and actions to enhance information systems and research in rare disease – notably the Health Information Bill.
- Ireland's involvement with Orphanet was believed to be useful but in need of development.

4.2.1 Key points for discussion

- *Towards a national rare disease office – when, where, how?*
- *Centres of Expertise for rare disease in Ireland – aspirations and concerns.*

4.2.2 Key findings

- The development of infrastructure to support a strategic approach to rare disease care in Ireland was seen as an essential component of a National Rare Disease Plan. The development of a National Rare Disease Office (NRDO) and Centres of Expertise (CoE's) were seen as core to the development and co-ordination of rare disease care in Ireland.
- There was widespread agreement that there was a clear and immediate need for the establishment of a NRDO as a priority.
- It was believed that establishment of a NRDO would improve patient lives, remove unnecessary burden from health services and prove cost effective because of synergies in service provision through:
    - generating awareness,
    - closing information gaps and speeding up diagnosis,
    - avoiding misdiagnosis and multiple referrals, unnecessary appointments and incorrect and/or duplication of treatment.
- To ensure the provision of a strategic and sustainable service the majority of respondents agreed that the NRDO should be State funded, provided through the HSE or Department of Health and that it should not be dependent on charitable donations. Others believed that it should be an independent entity. Regardless of where it resides appropriate governance of the NRDO was considered very important. Delegates envisaged that it would be managed by a voluntary, independent board involving a range of relevant stakeholders. It was also considered important that there would be clear demarcation between the role of patient groups and the NRDO.
- Delegates agreed that it should be a virtual central information hub to signpost both patients and health professionals to existing local and international information/knowledge about rare diseases. It would provide information on 'whole life' services for paediatric and adult onset rare diseases. It would have its own

identity with an independent website. It could also house the Irish Orphanet database.

- As the core function of an NRDO would be a supportive role, predominantly IT based, it was envisaged that it would only require a minimum number of staff, for example a health professional (nurse, medical background) to provide information and a data manager to manage access to safe and reliable information. Physical location of the office would not need to be a central concern. It was proposed that the new National Children's Hospital would be an ideal location for such a service, given the large numbers of children affected by rare diseases. It could be housed in the short term in the National Centre for Metabolic Disorders (Temple Street) or the National Centre for Medical Genetics (Crumlin Hospital). It was considered that the NRDO should be bound to clinical care pathways as part of the Clinical Care Programme. Establishment of care pathway guidelines was seen as essential to offer care pathway information for both patients and health professionals.
- It was also considered important that the NRDO would link to patient registries but not house them.
- Participants felt that the NRDO should provide a platform for rare disease patients and organisations to address common needs. Although patient organisation representatives voiced concern over the potential loss of autonomy for individual patient groups.
- It was proposed that a Rare Disease National Clinical Programme should be established within the National Clinical Programmes (Programme Manager in place, Clinical lead TBC) under the auspices of the Directorate of Quality and Clinical Care.
- Participants widely acknowledged that *transitional care* is a significant problem that needs to be addressed. CoE's were regarded as core infrastructure to enable the provision of co-ordinated care pathways for rare disease patients and guide integrated care provision. Although it was acknowledged that a scoping of existing services and expertise would be required in advance of the establishment of CoE's to identify and evaluate current care practices and the gaps in expertise e.g. medical genetics.
- Participants felt strongly that a network of CoE's should be established rather than one centre and delegates recommended a *Hub and Spoke* model with CoE's as

central elements connected to a range of satellite clinics, designed to enable much broader geographic coverage. Delegates discussed reference models such as the:

- *Haematology Group* model - located in St James Hospital, Dublin 8
- *Motor Neuron Disease* model - located in Beaumont Hospital, Dublin 9.

- A lot of discussion centred on the need for a definition of CoE's and the quality standards involved. Some participants felt that CoE's for rare disease should be organ specific, based on the systems affected e.g. metabolic diseases or liver diseases etc.
- It was acknowledged that patient groups should be facilitated to contribute to the establishment of co-ordinated care pathways.
- Respondents felt strongly that CoE's must be integrated into national funding plans with provision for clinical investigation/research time for health service professionals.
- Collaboration both nationally and internationally was considered essential to avoid competition and ensure streamlining of services.
- The costs of patient travel warrants consideration in the planning for infrastructural developments such as CoE's.
- Respondents stressed the need to lobby for cross-border shared services e.g. access to a child's hospice in Belfast for Republic of Ireland patients.
- It was considered that better facilitation of the homecare package for rare disease patients would be a cost effective measure.
- Finally, delegates discussed the need to direct patients for treatment within designated approved CoE's within the EU rather than travelling to the US for the same treatment at a greater expense to the exchequer.

4.3. Theme 3- Cooperation and Working across Sectors

4.3.1 Key points for discussion

- *What role for patient organisations in the plan?*
- *Getting the most out of cross-border working on rare diseases – clinical and research networks.*

- *Beyond health services - working across sectors (social, financial, educational and disability services) to deliver fair outcomes for rare disease patients.*

4.3.2 Key findings

The discussion focused on the three key discussion points above. The need to empower patients through the involvement of patient organisations was widely recognised as well as the need for multi-sectoral working within Ireland and international collaboration. Participants strongly felt that voluntary organisations should not have to raise funds for the treatment of rare disease patients, however they identified a wide range of supportive roles for patient organisations including:

- dissemination of information,
- general awareness raising,
- acting as a source of information for patients, carers and healthcare professionals and as an educational resource for service users and families,
- representing rare disease patients in the development of clinical care pathways,
- provision of emotional support at time of diagnosis and while service user and family are 'finding their way through the services,
- to fill gaps in existing services and build on existing capacity and
- advocacy and support throughout the rare disease life-cycle, fund-raising, assisting in supervising the implementation of the Plan.

It was widely considered that working across sectors (social, financial, educational and disability services) would lead to improved communication between departments and would provide cost savings to the health service. Participants also felt it would ensure improved provision of services for adults, and that cross-sectoral collaboration was essential to develop appropriate services across the life-cycle. They felt that current funding for services was not adequate and that services could be strengthened by:

- better organisation of services,
- more focused testing,
- a review of care packages and
- more focused referrals though the system.

Participants also called for a review of the Medical Card and Long Term Illness Scheme for people with rare diseases.

Commitment to cross-border working on rare diseases was considered an important aspect of the Plan. Participants identified several benefits and areas for improvement with a specific focus on clinical and research networks. These included:

- pooling resources and sharing services with implications for cost-effectiveness,
- information sharing and networking, participants highlighted however that research from abroad should be adapted to ensure it is appropriate for an Irish context,
- participation of Irish patients in clinical trials abroad,
- linkages to European registries,
- comparative assessment of international service provision and evaluation to identify what we do well and areas for improvement.

It was considered that greater international collaboration would lead to greater opportunities for rare disease patients. Participants believed that international collaboration would lead to increased opportunity for securing research funds, for example through EU grants. Participants also commented on a need to clarify and improve the structure and governance of the Treatment Abroad Fund and to extend this fund beyond Europe.

4.4. Theme 4 – Diagnosis

4.4.1 Key points for discussion

The work of the diagnosis sub-group to inform the National Steering Group is divided into seven key considerations or recommendations. Participants were asked to comment on the 7 preliminary considerations (outlined in table 9) in terms of comprehensiveness and appropriateness.

4.4.2 Key findings

In general, participants agreed that the considerations outlined by the subgroup were appropriate for the development of the Plan. However opinion was mixed on whether they were comprehensive enough.

Table 6. Key considerations of the diagnosis sub-group to inform the development of a National Rare Disease Plan

Seven key considerations
1. National Office for Rare Disease
2. Map out existing services
3. Grouping of Centers of Expertise
4. Funding
5. Quality Assurance of Centers of Expertise
6. Education
7. Newborn screening strategy and implementation

Participants discussed the recommendations of the subgroup according to the seven key recommendations put forward above.

- Participants felt it was important to base CoE's on existing models of care and pointed to The Cancer Control Strategy as a good example of a care model. They also highlight the example of HSE designated centres for cystic fibrosis patients.
- Some participants suggested broadening and amending the terminology of CoE's to reflect the different role and competencies of CoE's and their significant role for providing care at local level.
- They called for ring fenced-funding for CoE's to be recognised in National Plan and funding to follow the patient. In addition, they identified the need for funding infrastructure to support clinical research in CoE's and to promote basic research.
- Participants favoured separate funding mechanisms for drug reimbursement and service delivery to CoE's.

- Participants highlighted that the Cross-border Directive provides opportunities to designate CoE's and they felt it was important to look at appropriate standards of care for designated centres.
- They emphasised strongly the need to aspire to current European Guidelines for CoE's and the need for continuous external review, quality control and accreditation of clinical centres as well as laboratories.
- Several actions were identified in relation to the subgroup consideration of education on rare diseases including:
    - improvements in access to information for GPs and the community in general,
    - the need to address undergraduate, postgraduate and on-going education needs and the need for specialist training to address succession planning.
    - provision of a concise introduction to rare diseases for physicians, perhaps in undergraduate modules.
- The group were undecided on the sub-group recommendation for a Newborn Screening Strategy and Implementation Plan and felt that they needed more information. In particular, both groups were unaware of the recent EU EAHC Newborn Screening report (July 2011) and its implications for newborn screening in Ireland. In addition, the group indicated that they were not sufficiently informed to comment on a need for legislation review in regard to the newborn bloodspot screening programme.
- Several participants emphasised the need to urgently introduce a national governance model for population screening in general (beyond new born screening) to include screening for specific high risk groups and develop guidelines on consent.
- All participants considered it important to introduce on-going governance and review of screening requirements.
- Participants also felt it was important to acknowledge the importance of and improve holistic care provision to rare disease patients including psychosocial support. Improved care pathways were viewed as integral to holistic care provision.
- Participants stated that 3 months would be the optimal waiting time to be seen by a rare disease specialist for diagnosis following referral from a GP.

4.5. Theme 5 - Treatment of Rare Diseases including Orphan Drugs and Technology

4.5.1 Key points for discussion

- *Define where there are gaps in the system for patients*
- *Recommend a pathway for new orphan drugs which might fall outside the current system.*

4.5.2 Key findings

- Participants identified the need for increased research to inform and guide the treatment of Rare Diseases in Ireland. There was an identified need to raise awareness and educate the Irish public about Rare Diseases in general.
- Participants in this workshop also identified the need for a national office (NRDO) to co-ordinate and develop the management of rare diseases in Ireland.
- It was felt that Ireland needs to collaborate with experts across the EU and share treatment protocols and monitor patient outcomes as the wide range of expertise required to treat rare diseases in Ireland is limited. It was considered that adopting such international practice will drive improvements in clinical practice in Ireland and improve outcomes for rare disease patients in general.
- The important role of patient advocacy/support groups and organisations in decision-making processes was highlighted. Participants believe they have built up a valuable resource of expertise and feel they have a lot to offer clinicians in treating and monitoring rare diseases however, as there are no established communication mechanism; their input into rare disease care is currently limited.
- A commitment to the development of national infrastructure was called for, including: high quality health information, independently governed Disease Registers to monitor how Orphan drugs are performing and CoE's with international links and clinical protocols for all rare diseases.
- Participants identified the need to maintain current budgetary access for rare diseases whilst the National Plan for Rare Diseases and structures such as the National Advisory Committee for commissioning medicines is being established and set-up.
- There was widespread support for making training on rare disease compulsory for health professionals.

- Levels of knowledge & understanding of the current system for orphan drugs and technologies varied across participants**.** Patient support representatives were unclear of the current processes, specifically as to how orphan drugs are made available for use in Ireland.
- It was widely acknowledged that the decision-making criteria for cost-benefit assessment of orphan drugs are unclear and inaccessible at present. Orphan drugs and expensive medicines are assessed using Health Technology Assessment (HTA). Decision-making processes and criteria need to be more transparent and inclusive of patients' experiences and should include other cost-benefit measures. Some further considerations should include for example, the person's ability to continue working/remain in employment or the potential for drug therapy to reduce the need for costly acute hospital treatment such as dialysis and transplantation. A clear pathway for application for drugs, with appropriate and transparent process for assessing eligibility for orphan drugs was called for.
- At present there is no specific budget for treating rare diseases in Ireland. Drug treatments are currently paid for by individual hospitals or local community care services and as a result drug treatment decisions vary across the country. It was highlighted then that patients' access to drug treatment can be disadvantaged because of their geographical location. Irish people with rare diseases in general are disadvantaged because of low population numbers and the relative cost of treatment for such small numbers.
- Participants highlighted the need to review the development, pricing and funding for orphan drugs and other expensive medicines and suggested that a scheme akin to the '*High Tech Scheme'* is required.
- A review of medical care for patients with rare diseases was also called for to include a new alternative assessment pathway with clear, transparent, accountable processes and the right to appeal.
- Attention was drawn to compliance with the *Transparency Directive* specifically in relation to delays in the release of new drugs approved for market; legal timeframe for releasing new drugs is currently 180 days but this is due to be reduced to 120 days.
- Rapid access to orphan drugs requires timescale to be put in place and establishment of appropriate clinical protocols.

- An audit of the medicines currently available or due to become available in the future is required and participants felt this information should be made publically available.
- Evidence-based studies to review how we develop and fund orphan drugs and other medicines were also strongly advocated for.

Summary of the Consultation Findings

5.1. Consultation Response

This consultation process was a first step towards engaging with rare disease stakeholders in the development of Ireland's first National Rare Disease Plan. The three phased approach including: representation of patient organisations on the Steering Group; the consultation day at Farmleigh and the online consultation survey was designed to capture a wide range of rare disease experiences. The diverse range of conditions and large numbers of not yet diagnosed patients present a challenging task for meaningful consultation with those affected by rare diseases. Nonetheless, there was a large attendance at the consultation day and the online consultation yielded a substantial number of responses, representing a broad spectrum of stakeholder views, most especially patients and carers. Almost 40% of respondents were carers reflecting the vulnerability of the rare disease patient by virtue of age and/or disability and the importance of carers in giving a voice to the rare disease experience. The responses to the Irish consultation far outnumbered that of the UK consultation. The substantially higher proportion who submitted personal responses to the consultation, compared to the UK, perhaps indicates a lower level of representation of patients by patient organisations in Ireland compared to the UK.

It is acknowledged that some rare disease groups may remain under represented; in particular those not yet diagnosed, the Travelling community, and rare disease patients with visual and hearing impairments and learning difficulties. Nonetheless, the significant volume and diversity of responses indicate the clear need for addressing rare disease services in Ireland. Respondents identified a wide range of issues affecting rare disease patients, families, carers and healthcare providers. Patients are faced with a range of issues including the general lack of awareness of rare diseases in Ireland, the lack of awareness of specific rare disease conditions and the issues associated with diagnosis, treatment and management of relatively unrecognised, under researched and poorly understood conditions. Findings from this consultation process provide insight into the needs of persons living with rare diseases, their families and carers as well as those of service providers, thereby offering clear direction in terms of priorities for Ireland's first National Rare Disease Plan. The lack of infrastructure specific to rare disease patients,

namely information systems and healthcare structures were seen as major priorities in the development of a National Rare Disease Plan.

5.1.2 Access to and Quality of Screening and Diagnosis

Recent developments in rare disease services were welcomed including extended screening for newborn babies. Emphasis was placed on the need to introduce a national governance model for population screening and extension of the current newborn screening programme. Several respondents pointed to the benefits of screening for additional conditions in newborns. A large majority of respondents called for the alignment of Irish screening in line with European best practice and extending screening services to include repeated screening for babies at six months. The vast majority of respondents also agreed with the need to introduce legislation on screening in line with European best practice. The practice of screening for rare conditions during pregnancy also emerged in the qualitative responses. Respondents explained that they would prefer to be informed of a diagnosis during pregnancy, enabling them as parents to prepare for the condition in their newborn child.

Individuals presenting to the health services with rare disease symptoms described arduous journeys to diagnosis and poor understanding of their condition among healthcare professionals. While the quality of diagnosis was generally rated as fair, almost a third of respondents rated diagnosis quality as poor or very poor (32%). Respondents cited the need to address delays in diagnosis as the most important factor to improve diagnosis quality. Inaccurate diagnoses, as well as ill-informed or insensitive communication of a diagnosis were highlighted as problematic. Often respondents reported a lack of awareness of symptoms of rare conditions among clinicians, a perceived lack of interest, an over reliance on medical diagnostic tests and/or an eagerness to move the patient on through the services if a specialist was unable to diagnose. Misdiagnosis and late diagnosis of rare diseases were seen as substantial barriers to good quality care. The need for increased awareness of rare diseases and training for health services staff in general as well as for general practitioners and specialist consultants was emphasised throughout the consultation.

Improvements in diagnosis was considered the most important area for bringing real benefit to patients with rare diseases, specifically timeliness and accuracy of diagnosis as well as sensitive communication of a diagnosis and the availability of services once a diagnosis has been made. The most important action to improve diagnosis of rare diseases in Ireland was up-skilling GP's in the identification and appropriate referral of rare disease patients as well as improving access to specialist diagnostic services.

5.1.3 Access to and Quality of Care

The need for improved care pathways for patients at every stage of their illness trajectory was identified as a priority as well as support for holistic care including psychological support.

Respondents highlighted a lack of support throughout the rare disease illness experience and it was most evident in relation to particular stages:

- Obtaining services and support without a formal diagnosis
- Obtaining a formal diagnosis after presenting with symptoms
- Navigating between primary care and acute care
- Accessing appropriate medication and treatment
- Absence of care pathways or care plans for long-term treatment
- Accessing patient and family support services such as counselling including genetic counselling and advice on implications for the broader family
- Care at end-of life
- Experiencing bereavement

The majority of respondents rated quality of care as fair (42.9%) while just over 30% rated it as either poor or very poor. The main barriers to achieving good care outcomes included no clear transition from paediatric to adult services and misunderstanding the needs of rare disease patients outside the healthcare services, for example, education, employment and disability. No clear care pathways and waiting lists were also cited as main barriers to quality care provision. Several respondents cited poor coordination of care, no care plans and multiple visits to Emergency Departments for unmanaged symptoms resulting in what was referred to as 'revolving door' patients.

While health care providers were more likely to report care provision as good compared to patients, they too report feeling limited by a lack of awareness, training and poor availability of care and treatment options within Ireland. The need for appropriate treatment facilities was also highlighted, specifically in relation to suitable infection control units for patients with cystic fibrosis. Respondents described inequitable provision of care with specialist care concentrated in large urban centres. Some respondents described poor experiences of care in local hospitals where there was limited expertise for their condition.

5.1.4 Centres of Expertise

There was widespread support for developing Centres of Expertise on rare diseases in Ireland, State funded to ensure equity and sustainability and operated by the HSE. The core functions of such centres, in order of importance, were seen as: multidisciplinary care; support for the development of care plans; supporting research; education and training of health service professionals, to monitor the quality of diagnostic and treatment services and inform planning and development. Overwhelmingly respondents supported enabling Irish patients to access European reference networks when necessary. While most agreed that travel and costs of treatment should be reimbursed, opinion was divided with fewer health service providers and researchers in support of financial reimbursement for patients.

5.1.5 Patient-doctor Relationship

The patient-doctor relationship with regards to both general practitioners and consultants emerged as a central theme throughout the consultation. Patients viewed this relationship as pivotal in obtaining a diagnosis and appropriate care. Patients reported experiences of poor communication from some doctors and felt their symptoms had been dismissed as they did not fit neatly into any one category or specialism. In some cases patients or carers reported insensitive communication of a diagnosis, this was found to be particularly difficult for parents of a newborn child. Often this was accompanied by a lack of support or direction towards services. However, several respondents also reported positive experiences of care providers and pointed to the lack of infrastructure and

medication as limiting care options, highlighting the difficult position of healthcare providers when dealing with a rare disease.

5.1.6 Allied Health and Auxiliary Services

As well as issues with clinical care there was an identified gap in the provision of allied and auxiliary health services such as physiotherapy and counselling services. Counselling services were thought to be especially important when receiving a diagnosis, particularly for new born babies and in terms of end-of-life and bereavement support. Patients and carers reported paying for such services out-of-pocket and were limited by their own precarious financial position. Many rare disease patients are young children and it was highlighted that palliative care services for this group are underdeveloped. The vital need to extend palliative care services to end-of-life care for patients of all ages with rare diseases was stressed.

5.1.7 Orphan Drugs and Technologies

Many respondents expressed concern that the current process for accessing orphan drugs and technologies lacks transparency and is too slow and constrained. Initiatives to improve access and increase transparency to these would be welcome. Although it was acknowledged that treatments for rare diseases are often expensive, respondents felt strongly that their treatment should not be curtailed by financial considerations. They stressed the need for an equitable approach to decisions on medication and a review on the development, pricing and funding for orphan drugs in Ireland.

5.1.8 International Collaboration and Research

In general respondents felt limited by diagnostic and treatment options in Ireland and many wanted the option of accessing these abroad. The role of international collaboration was emphasised, in particular for research into all rare disease conditions but its value was noted especially for very rare diseases where the capacity to diagnose and treat is currently limited in Ireland. Respondents identified the need for high quality data and research, central to good quality care. They supported the establishment of rare disease registries in Ireland and an increased emphasis on a broad spectrum of research initiatives

into rare diseases. Increasing capacity for rare disease research in Ireland was considered as an essential first step in promoting awareness and improving healthcare provision. Providing protected time for health professionals to engage in and conduct research was seen as a priority. Respondents felt that strong clinical leadership was required for successful development of rare disease research in Ireland. The importance of developing a legislative framework for research was also emphasised and a large majority of respondents agreed that Ireland should develop its participation in the European approved Orphanet.

5.1.9 General Challenges for Rare Disease Patients in Ireland

The burden brought on by living with a rare disease is substantial and under supported both in terms of clinical management and wider provisions for ensuring people with rare diseases have equal opportunity to enjoy a good quality of life, compared to other patients and the population in general. Distinct from many other chronic illnesses, rare diseases are often genetic with a disproportionate prevalence occurring in children and susceptible families. A rare disease can be experienced by several members of the same family, increasing the familial burden with considerable consequences on care capacity.

Respondents identified a general lack of awareness and expertise on rare diseases in Ireland in general and in the Irish healthcare system. This was generally associated with the low profile of rare disease conditions. Patients often felt their symptoms were poorly understood and reported a perceived lack of interest on behalf of some healthcare providers. Due to the unique nature and low prevalence of many rare diseases, the impacts of such conditions, often profound and disabling, were also poorly understood, resulting in limited support for rare diseases in the health and social services. Both carers and patients felt that they were struggling to access services and entitlements due to limited awareness and understanding of rare disease symptoms. This was especially true for allied health and auxiliary services such as counselling and home care support. Respondents reported the need for improved infrastructure to support appropriate rare disease diagnosis and care in Irish health and social care services, supported by some healthcare providers.

In addition to challenges in accessing and maintaining entitlements such as the medical card and the long-term illness card, respondents also reported challenges in obtaining health insurance and life assurance. Persons with rare diseases are often forced to choose between continuing to work and giving up work in order to qualify for the aforementioned entitlements; often opting for the latter as the burden of rare disease treatment proves too costly for the individual. Many patients were no longer in a position to work and found themselves in an increasingly precarious financial position, sometimes faced with excessive out-of-pocket costs for treatment and medications. Loss of employment then also leads to increased isolation and financial dependence. The pivotal role of carers for rare disease patients was stressed in the consultation response.

5.1.10 Role and Involvement of Rare Disease Organisations

In general the importance of patient organisations was emphasised, particularly for their role in information dissemination, advocacy and support for those with a rare disease. However several respondents also highlighted their lack of representation by patient organisations that largely represent a single rare disease condition. This may be reflected in the large number of individual respondents who responded to the online questionnaire. These patients expressed feelings of isolation and felt voiceless and would welcome a broader focus of rare disease patients groups to encompass their needs.

5.1.11 Priorities for Service Provision and Commitment to Implementation

The development of a National Rare Disease Plan for Ireland was widely welcomed and supported by all stakeholders. Respondents expressed the vital need for a well developed and implemented rare disease Plan for Ireland. There was a call for independent monitoring to ensure effective implementation of the Plan and involvement of stakeholders in the evaluation of its implementation. Patient organisations are seen as an essential voice within the development and implementation of the Plan. Several priorities were identified by respondents in the consultation. These include:

- Access to appropriate specialist care.
- Improvements in timely and accurate diagnosis.

- Improvements in the quality of healthcare supported by improved infrastructure, including the establishment of a National Rare Diseases Office, a Rare Diseases Clinical Care Programme and a network of Centres of Expertise.
- Improved health information infrastructure and research supports specific to rare diseases in Ireland.
- Enhanced research across a wide range of domains including clinical, epidemiological and health services research to develop the evidence-base on rare diseases in Ireland and support for development of rare disease registries.
- A focus on improved access to allied and axillary services including palliative care and counselling.
- Dedicated approach to increase rare disease awareness and relevant training among health and social care professions.
- A focus on co-ordinated care pathways to ensure appropriate care for rare disease patients throughout their illness.
- Increased cross-sectoral working essential to develop appropriate services and supports across the patient's lifecycle.
- Increased focus on international collaboration.

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Appendices

Appendix 1 Consultation Event Programme

The Consultation Event Programme included the following:

2.1 Welcome from Dr. James Reilly, TD, Minister of Health,
Chair: Dr John Devlin, Deputy Chief Medical Officer, Department of Health

2.2 Introductions from the key note speakers

2.3 Facilitated interactive sessions were held before and after lunch in relation to each of the subgroup reports, enabling participants to participant in two workshops of their choice.

2.4 Feedback from the workshops on recommendations and priority areas to be addressed in the plan were summarised and presented to the audience by each of the workshop facilitators.

2.5 Closing remarks were made by Dr. John Devlin, Deputy Chief Medical Officer, Department of Health and Mr. Owen Metcalfe, Institute of Public Health.

Appendix 2 Facilitated Workshop Sessions

Theme 1 – Data and Research

Workshop Lead:	**Helen McAvoy, Institute of Public Health in Ireland**
Facilitator:	**Caoimhe Gleeson, National Advocacy Unit, HSE**
Note Taker:	**Catherine Gill, Health Research Board.**

Theme 2 – Infrastructure

Workshop Lead:	**Avril Daly, GRDO**
Facilitators:	**June Boulger, National Advocacy Unit, HSE**
Note Taker:	**Oonagh Ward, Health Research Board.**

Theme 3 – Cooperation and working across sectors

Workshop joint Leads:	**John McCormack, Taskforce for Rare Diseases and MRCG.** **Tony Heffernan, Bee for Battens and The Saoirse Foundation.**
Facilitators:	**Winifred Ryan, National Advocacy Unit, HSE**
Note Taker:	**Shane Shannon, National Advocacy Unit, HSE.**

Theme 4 – Diagnosis

Workshop Lead:	**Eileen Treacy, Children's University Hospital**
Facilitator:	**Juanita Guidera, National Advocacy Unit, HSE**
Note Taker:	**Larissa Silva, Fighting Blindness Association.**

Theme 5 – Treatment including Orphan Drugs and Technologies

Workshop Lead:	**Eibhlin Mulroe, IPOSSI**
Facilitator:	**Geraldine Quinn, National Advocacy Unit, HSE**
Note Taker:	**Lisa McCormack, National Advocacy Unit, HSE.**

Appendix 3 Organisations represented at the Consultation Day in Farmleigh

Organisation
Ataxia Ireland
22q11 Ireland support group
Adelaide and Meath Hospital, Tallaght
Alexion
Alpha One Foundation
Bee for Battens and The Saoirse Foundation
BioMarin Europe Ltd
CanTeen Ireland
Celgene
Core Counselling and Psychotherapy
CUH, OLCHC, NCH
Cystic Fibrosis Association of Ireland
Cystic Fibrosis Registry of Ireland
Cystinosis Foundation Ireland
DEBRA Ireland
Department of Health
Disability Federation of Ireland
Drury Communications
Dublin EUROCAT Registry, Health Service Executive
Duchenne Ireland
Dystonia Ireland
EUROCAT - East - HSE
Fabry Ireland
Fine Gael
Genzyme
Genzyme Therapeutics
George Hunter Entertainment
GlaxoSmithKline Pharmaceuticals
GRDO (Genetic & Rare Disorders Organisation)
Health Research Board

Health Service Executive
Huntington's Disease Association
Institute of Public Health in Ireland
IPPOSI
Irish Cancer Society
Irish Clinical Oncology Research Group (ICORG)
Irish College of General Practitioners
Irish Fragile X Society
Irish Haemophilia Society
Irish Heart Foundation
Irish Lung Fibrosis Association
Irish Medicines Board
Irish Patients' Association
Irish Pharmaceutical Healthcare Association
Irish Stem Cell Foundation
Marfan Syndrome Support Group
MDI
Molecular Medicine Ireland
MRCG
MRCG
MUH & UCD
Muscular Dystrophy Ireland
Multiple Sclerosis Ireland
National Cancer registry
National Centre for Medical Genetics
National Children's hospital Tallaght
National Emergency Medicine Programme
National Paediatric Mortality Register
NCMG
NCSRN Ireland at the Irish Heart Foundation
Niemann Pick
NMH, OLCH, RCSI &UCD
Northern Ireland Rare Disease Partnership
Novartis

Our Lady's Children's Hospital Crumlin
parent of a child with a very rare condition
Patient - IPPOSI; MDI
Pfizer
Pfizer Healthcare Ireland
Polychondritis
Prader-Willi Sybdrome Association Ireland
Rare Disease Taskforce
RCPI / HSE
Rehab Group
Royal College of Physicians
Shire Pharmaceuticals Ireland Ltd
Society for Mucopolysaccharide Diseases
St James's Hospital
St James's Hospital and HSE
St Vincent's University Hospital
Tallaght Hospital
Tarlovcyst Disease Support Group
TCD
TCGI
The Alzheimer Society of Ireland
The Jack and Jill Foundation
Trinity College Dublin
University College Dublin
Individual Participants

Appendix 4 Rare Disease Listing from Online Consultation

Please note that this list only refers rare diseases named by respondents who elected not to submit their views anonymously (representing 58.5% of all respondents) and is therefore not fully representative of the spectrum of rare disease respondents in the consultation. For some of the rare disorders listed several responses were received. The disorders are presented in alphabetical order.

Acromegaly
Adult growth hormone deficiency
1p36 micro deletion syndrone
22q 11.2 deletion syndrome
ADEM
Acanthamoeba sclerokeratitis
Addison's disease
All genetic disorders
Alper's syndrome /disease
Alpha-1 antitrypsin deficiency
Anaphylaxia
Angelman syndrome
Ankolosing spondylitis
Arterio venous malformations
Arthrogryposis
As yet unknown genetic condition
Avm
Balanced reciprocal translocation between short arm no. 6 and long arm no.
Battens disease
Beamer Langer syndrome type 4 (short rib syndrome)
Behcets syndrome
Bilateral schizencephaly with hydrocephalus and global developmental
Bleeding disorder of unknown aetiology
Born without adrenal glands
Broad spectrum genetic disorders due to chromosomal abnormalities
Bronchiolitis obliterans organizing pneumonia (BOOP)
C1 inhibitor deficiency (hereditary angioedema)
Carcinoid cancer - neuroendocrine tumours midgut with metastases to the
Cardiofaciocutaneous syndrome CFC
Cavernoma angioma. (with the CCM3 gene).
CDKL5
Chri Du Chat syndrome (5P- syndrome)
Chromosomal abnormality 2q24.3 deletion
Chronic demyelinating peripheral neuropathy
Churgg Strauss syndrome
CINCA or Nomid syndrome
Coats disease
Cohen syndrome

Common Variable Immunodeficiency (CVID)
Complex 1 Respiratory Chain Deficiency (Mitochondrial Disease)
Congenital myasthenia Gravis-Slow Channel Syndrome
Congenital myasthenia syndrome
Congenital sucrase isomaltase deficiency
Craniopharyngioma
Crohns disease
Cushings disease
CVID primary immune deficiency PANDAS syndrome Crps
Cystic fibrosis
Cystinosis
Dandy Walker malformation
Dermatomyositis lymphangioleiomyomatosis
Diabetes insipidus
Duchenne muscular dystrophy
Ectodermal dysplasia bladder extrophy episadias
Ehler Danlos syndrome
Emma
Enzyme disorders such as Pompe disease, Gaucher disease, Fabry disease, Epidermolysis bullosa
Extremely rare neurological immune related condition, currently linked to Fabry disease
Familial C3 glomerulopathy
Fragile X
Friedreich's ataxia
GLUT1 DS
Glutaric aciduria Type 1
Glycogen storage disease
Guillain Barré syndrome
Gut failure of unknown origin TPN dependant
Haemophilia.
Hemi hypertrophy
Hereditary angioedema
Hidradenitis suppurativa
High ANCA associated vasculitis
HLA B27 positive
Hodgkin's lymphoma
Hunter syndrome/disease (Mucopolysaccharidosis II)
Huntington's disease
Hyperinsulism
Hypopituitarism
Rare neurological diseases, such as Krabbes, Metachromatic leucodystrophy
Idiopathic pulmonary fibrosis
,Mowat Wilson ,Ohdo syndrome,Wolf Horshorn syndrome,Chromasome 9
Inborn errors of metabolism, e.g. Maple Syrup Urine Disease, urea cycle
Inflammatory bowel disease
Inherited cardiac conditions - Long QT syndrome, hereditary DCM, ARVC,
Inherited cardiac diseases - ion channelopathies and cardiomyopathies
Inherited metabolic disorders including Phenylketonuria, Maple Syrup Urine
Intracranial hypertension Arnold Chiari malformation

Kearns Sayre Syndrome
Kennedys syndrome which is a motor neuron disease
Keratoconus
Krabbes leukodystrophy
Langerhans cell histiocytosis
Automatism - sleep automatism, epileptic automatism, drug induced
Leucoencephalopathies and no diagnosis
Lisincephly
Lissencephaly, + CVA
Loeys-Dietz Syndrome
Long QT syndrome Rare cardiomyopathies
Lung carcinoid cancer
Lungs- and breathing
Lupus including lupus nephritis, CFS, fibromyalgia, vasculitis, endometriosis,
Lysosomal disorders e.g. Hurlers disease, Morquio, San Fillipo, Hunters
Male breast cancer
Marfan Syndrome
Mercury and heavy metal poisoning (cadmium, lead, arsenic)
Mesothelioma
Metachromatic luekodystrophy
Metastatic neurendocrine tumour NET Tumor, Metastatic Carcinoid Tumor.
Miller-Dieker Syndrome
Mitochondrial disorder - complexes I,III & IV.
Mixed connective tissue disease. Systematic sclerosis,with psoriatic arthritis.
Motor neuron disease Prior polio
Motor neurone disease
Moya Moya
MPGN Familial
MPS 1VA
Mucopolysaccharide disease type IIIB San Filippo syndrome
Mucopolysaccharidosis(MPS)
Multiple system atrophy
Muscular dystrophy Facioscapulohumeral MD
3p deletion syndrome
Aicardi syndrome
Mixed connective tissue disease and secondary Raynauds
Myasthenia gravis
Myelodysplastic syndromes
Neurofibromatosis
Neurological sarcoidosis
No specific disease as our members are involved in investigation, diagnosis
No specific disease but my PhD study concerns fathers' experiences of
Non-functioning tumours
Nutrition related conditions
Ohdo
Organic acidemias, Fat oxidation defects, amino acidopathies, purine and
Orofacial granulomatosis
Osteogenesis imperfecta
Rehab provides a wide range of health and social care services, training and

P.O.T.S. Syndrome..(Postural Orthostatic Tachycardia Syndrome)
Pan-hypo-pituitarism
Paroxysmal nocturnal haemoglobinuria Gaucher's disease
Penile cancer Testes cancer
Periodic fever (PFAPA), auto inflammatory disease, Familial Mediterranean
Periodic paralysis
Peroxisomal Biogenesis Disorder.
Phelan McDermid Syndrome
Phenylketonuria, Galactosaemia
Pierre Robin syndrome
Pituitary tumour
Pleural mesothelioma
Polychondritis
Pompe Disease
Postpolio myelitic syndrome
Postural Orthastatic Tachycardia Syndrome
Prader Willi Syndrome
Primary Immune Deficiency (Hypogammaglobulinaemia)
Primary immunodeficiencies (PID)
Progressive multifocal leukoencephelopathy
Progressive muscle atrophy
Progressive supranuclear palsy
Prolactinoma
Pseudomyxoma peritonei
Pulmonary fibrosis
PVNS - Pigmented villonodular synovitis
Rare forms of dementia
Rare Kidney Diseases: Hereditary kidney diseases: FSGS, Familial MPGN,
Rare neurological/ metabolic disorders
Raynaud's & scleroderma
Retinitis pigmentosa
Rett syndrome
Retts Angleman Conradi Hunerman Miller Dicker Zellweger Battens
Rubenstein Taybi syndrome
Sarcoidosis
Ewings sarcoma
Scleroderma
Several forms of vasculitis and connective tissue diseases
Sotos syndrome
Spinal muscular atrophy Type 3 / variant.
Stargardts Disease

Sturge Weber Klippel Trenauney
Syingomyelia Arnold Chiari Malformation
Syndromes and conditions related to global developmental delay/
Systemic Lupus Erythematosis including lupus nephritis, cerebral vasculitis,
Antiphospholipid Antibody Syndrome
Systemic vasculitides and systemic rheumatic diseases (e.g. Lupus, Sjogren's,
Systemic vasculitis
Rare and severe diseases present in the neonatal period: congenital

Those rare diseases that are life limiting in nature; affecting children; adults -
Trisomy 14 mosaicism
Trisomy 17p
Ulcerative colitis
Rare musculoskeletal diseases - Systemic sclerosis vasculitis juvenile
Undiagnosed genetic syndrome presenting with global development delay,
Undiagnosed global development delay
Ushers syndrome
Vascular malformations
Vasculitides (~ 20 disorders) Connective tissue diseases Juvenile idiopathic
Vasculitides (~20 individual syndromes) Connective tissue diseases Auto
Vasculitis Lupus Von Hippel Lindau
Wilson's disease
Cystic fibrosis -Epidermolysis bullosa -Friedreich's ataxia -Huntington's
X Linked ichthyosis

Appendix 5 Online Consultation Respondent Type

Patient Organisations

Genetic & Rare Disorders Organisation (GRDO)	Bee for Battens	Irish Primary Immunodeficiency Association (IPIA)	Advocacy for Neuroacanthocytosis Patients
European Haemophilia Consortium	Irish Motor Neurone Disease Association	Raynaud's & Scleroderma Ireland	The Alzheimer Society of Ireland (ASI)
The Irish Heart Foundation	Cystic Fibrosis Association of Ireland	Huntington's Disease Association of Ireland	DEBRA Ireland
Rare Disease UK	Post-Polio Support Group	Neurofibromatosis Association of Ireland	22q 11 Ireland Support Group.
Disability Federation of Ireland (DFI)	Syringomyelia Self Help Group (Irish ANTS Co. Ltd)	Irish Hospice Foundation	The Warm Shoulder Club
The Jack & Jill Children's Foundation	Rehab Group		

Service Provider/clinician

Dr Eamonn Molloy - St Vincents University Hospital.	Dr Joanne Balfe - Consultant Paediatrician, Tallaght Hospital and LAura Lynn House at The Children's Sunhine Home	Dr Deirdre Ward - Consultant Cardiologist Centre for Cardiac Risk in Younger Persons Tallaght Hospital	Dr Eamonn Molloy Department of Rheumatology St Vincent's University Hospital Elm Park Dublin 4
Catherine Mc Donnell - Clinical Nurse Manager 3 National Center for Inherited Metabolic Disorders, Temple St. Dublin 1	Eimear Daly -CNS Paediatric Intellectual Disability, Mercy University Hospital Cork.	Eleanor Molloy - Neonatologist and Paediatrician , National maternity hospital & Our Lady's Children's Hospital, Crumlin Associate Prof of Paediatrics, RCSI	Yvonne Rogers-National Centre for Inherited Metabolic Disorders Children's University Hospital Temple Street
Ms Rosie O'Shea - Genetic Counsellor The National Centre for Medical Genetics Our Lady's Children's Hospital Crumlin	Peter Lavin - Consultant Nephrologist Tallaght Hospital Dublin 24	Dr Helen Enright Consultant Haematologist Department of Haematology Adelaide and Meath Hospital Tallaght Dublin 24	Naomi O'Malley - Clinical nurse manager Enzyme replacement co-ordinator Metabolic outpatients Childrens University Hospital Temple St
Denis Kerin Paramedic HSE Kerry	Children's University Hospital, Dublin	Catherine Wall. Nephrologist. Tallaght Hospital.	Philip Murphy - Tallaght Hospital
Dr. Ina Knerr - Children's University Temple Street, NCIMD.	The Academy of Medical Laboratory Science	Dr G canny - Formerly OLCH, Crumlin	Clare Curtin - RCN Mercy University Hospital Cork
Dr Rosemarie Watson	Dr Mark Ryan	SA Lynch - NCMG	Dr Kate Russo

Dr Joseph Galvin
Policy/Health Service Mgt

Central Mental Hospital - Prof Harry Kennedy *Executive Clinical Director National Forensic Mental Health Service Dundrum Dublin 14 and Clinical Professor of Forensic Psychiatry Department of Psychiatry TCD*	National Centre for Medical Genetics - David Betts	Irish Association of Speech and Language Therapists	HSE - Fenton Howell

Pharmaceutical/biotechnology company

CTI Science Limited	Genzyme Therapeutics	Shire Pharmaceuticals Ireland Ltd.

Researchers/Research Institutes

Dr Honor Nicholl School of Nursing & Midwifery TCD	Prof Mark Little on behalf of the Vasculitis Rare Disease Working Group	Valerie Urbach National Children Research Center Crumlin	Marion Rowland MB PhD Lecturer in Clinical Research School of Medicine and Medial Science University College Dublin
Dr Tony McElligott, Trinity College Dublin	Catherine Greene, Royal College of Surgeons in Ireland	Douglas Veale, Director of Translational Research, Dublin Academic Medical Centre	Ms.Suja Somanadhan, Doctoral Student, University College Dublin, Ireland.
Steven G. Gray, PhD Trinity College Dublin Dept of Clinical Medicine	Dr Warren Thomas Royal College of Surgeons in Ireland	Irish Clinical Research Infrastrcuture Network (ICRIN) www.icrin.ie	Catherine Norton,PhD researcher School of Psychology, University College Dublin .
NUI Galway	RCSI	Mai Harris	Deirdre Kelleher, lecturer School of Nursing Midwifery and Health Systems, University College Dublin.

Appendix 6 Glossary of Terms

Basic biomedical research:

Research conducted to increase understanding of the physical, chemical and functional mechanisms of life processes and disease. It is often called fundamental or „pure" research and is not directed at solving any particular biomedical problem in humans or animals. It provides the building blocks upon which other types of biomedical research are based.

Centres of Expertise:

Centres of Expertise (Centers of Expertise) are care centres that bring together a group of multidisciplinary, specialised competencies, from offering consultations, medical examinations, genetic testing and counselling and social services to facilitating inclusion in research protocols and clinical trials. The core aim of Centers of Expertise is to deliver a patient-centred service ensuring timely diagnosis and appropriate follow up care.

Clinical research:

Research with the goal of improving the diagnosis and treatment (including rehabilitation and palliation) of disease and injury; improving the health and quality of life of individuals as they pass through normal life stages. It involves research on, or for, the treatment of patients.

Clinical trials:

These are trials which use human volunteers to evaluate prospectively the effectiveness and safety, and optimum dosage schedule (where appropriate) of medications or medical devices by monitoring their effects on large groups of people.

European Reference Networks:

It is not possible to diagnose and treat each of the 5 to 8,000 rare diseases in a specific Centre of Expertise. The use of European Reference Networks may be cited as the 'networking of knowledge and expertise' through either physical or virtual expertise and/or reference centres and teams of experts – these are fundamental to address the issue of rare diseases at both European and national levels

The goal of an ERN is the improvement in the overall quality and management of care of a single rare disease or a group of RDs by complementing, supporting and providing added value to the existing services and expertise at national level. Such networking activity between national Centers of Expertise promotes the sharing and mobility of expertise.

ERNs can improve knowledge by sharing and creating mutual databases and registries of information and resources.

Epidemiology:

Epidemiology is the study of the distribution and patterns of health-events, health-characteristics and their causes or influences in well-defined populations. It is the cornerstone method of public health research, and helps inform policy decisions and evidence-based medicine by identifying risk factors for disease and targets for preventive medicine. Major areas of epidemiological study include outbreak investigation, disease surveillance and screening (medicine), biomonitoring, and comparisons of treatment effects such as in clinical trials.

Genetic counselling:

An educational counselling process for individuals and families who have a genetic disease or may be at risk for a disease to facilitate informed decision-making.

Health services research:

Research with the goal of improving the efficiency and effectiveness of health professionals and the healthcare system, through changes to practice and policy. Health services research is a multidisciplinary field of scientific investigation that studies how social factors, financing systems, organisational structures and processes, health technologies, and personal behaviours affect access to healthcare, the quality and cost of healthcare and, ultimately, health and well-being.

HSE Clinical care programme:

Clinical Strategy and Programmes has been established by the HSE to improve and standardise patient care by bringing together clinical disciplines and enabling them to share innovative solutions to deliver greater benefits to every user of HSE services.

The directorate has established a number of National Clinical Programmes. The Programmes are based on three main objectives

To improve the quality of care we deliver to all users of HSE services

To improve access to all services

To improve cost effectiveness

http://www.hse.ie/eng/about/programmes/

Orphanet:

Orphanet is the reference portal for information on rare diseases and orphan drugs, for all audiences. Orphanet"s aim is to help improve the diagnosis, care and treatment of patients with rare diseases. (www.orpha.net) Orphanet is led by a European consortium of around 40 countries, coordinated by the French team. National teams are responsible for the collection of information on specialised clinics, medical laboratories, ongoing research and patient organisations in their country.

Orphanet offers a range of freely accessible services:

- An inventory of rare diseases and a classification of diseases elaborated using existing published expert classifications.
- An encyclopaedia of rare diseases in English and French, progressively translated into the other languages of the website.
- An inventory of orphan drugs at all stages of development, from EMA (European Medicines Agency) orphan designation to European market authorisation.
- A directory of specialised services, providing information on specialised clinics, medical laboratories, ongoing research projects, clinical trials, registries, networks, technological platforms and patient organisations, in the field of rare diseases, in each of the countries in Orphanet"s network.
- An assistance-to-diagnosis tool allowing users to search by signs and symptoms.
- An encyclopaedia of recommendations and guidelines for emergency medical care and anaesthesia.
- A bimonthly newsletter, OrphaNews, which gives an overview of scientific and political current affairs in the field of rare diseases and orphan drugs, in English and French.
- A collection of thematic reports, the Orphanet Reports Series, focusing on overarching themes, directly downloadable from the website.

Orphan Drugs and Technologies:

An orphan drug is a pharmaceutical agent that has been developed specifically to treat a rare medical condition. The European Union (EU) has enacted legislation, Regulation(EC) No 141/2000, in which pharmaceuticals developed to treat rare diseases are referred to as 'orphan medicinal products.

Population health research:

Research with the goal of improving the health of the population, or of defined sub-populations, through a better understanding of the ways in which social, cultural, environmental, occupational and economic factors determine health status or through the identification of effective interventions for improving health status and reducing health inequalities.

Rare disease:

The European definition considers a disease to be rare if it occurs with a prevalence of ≤ 5 per 10,000 of the European population (EU legislation regulation (EC) No. 14/2000 on orphan medicinal products). This definition is recognised in Ireland. This means that some diseases with a slightly higher prevalence in the Irish population may still be considered rare diseases for the purposes of the Irish plan.

There are approximately 6000 diseases considered rare and the list is available on the Orphanet database (www.orpha.net).

In the case of rare congenital anomaly prevalence at birth is generally used.

The definition of a rare cancer in Ireland differs from the overall definition used for rare disease. Cancers with an annual incidence of less than 6 per 100,000 European population are considered rare (RARECARE)

Registry/ Patient Registry:

Disease or patient registries are collections of data related to patients with a specific diagnosis, condition, or procedure.

Translational research:

Research that is specifically concerned with the application of basic research findings into innovative strategies, devices, products or services for the diagnosis, treatment or prevention of human disease.

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