



**An investigation into the  
social support needs of families  
who experience rare disorders  
on the island of Ireland**



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RehabCare  
Investing in People, Changing Perspectives



## Foreword

It is with pleasure that RehabCare presents its report *An Investigation into the Social Support Needs of Families who experience Rare Disorders on the Island of Ireland*.

RehabCare the health and social care division of the Rehab Group provides community-based, person-centred and flexible services which are designed to meet individual need and to support people to achieve their goals. In 2007, we supported over 2,600 individuals with disabilities, the older people and other marginalised groups to maximise their quality of life and to participate in their local communities.

We have been working in the area of rare disorders for many years both in the provision of services and in the identification of the needs of people with rare disorders and their families. In 2003, we developed and opened our first specific rare disorder service for people with Prader Willi Syndrome and their families.

The development of this service indicated that there are clear gaps in the existing information about rare disorders and the support available to people and their families. It was clear that there was little published information about rare disorders.

RehabCare has undertaken to reduce this information deficit by embarking on key pieces of research to look at the experiences of people with rare disorders and their families, both on the island of Ireland and internationally.

Initially, we conducted a scoping exercise about how support was provided to people with rare disorders in other countries and were impressed by the best practice supports available in other jurisdictions. This led to further research and in 2002, RehabCare carried out the first survey of the experience of paediatricians in relation to children with rare disorders in Ireland. This research, carried out in collaboration with key stakeholders, gave an initial insight into the social support needs of families experiencing rare disorders across the Republic of Ireland.

In 2005, we were delighted to be awarded funding by the Health Research Board to complete this current piece of research, which specifically focuses on the social support needs of families in the Republic of Ireland and Northern Ireland considering three key areas: information; current social supports and future social supports.

I would like to sincerely thank all those who have supported this research; people and children with rare disorders, their families, and health professionals. The contribution and commitment of all these people to this research has been immense. RehabCare welcomed the opportunity to work once again with those affected by rare disorders and those working in this area.

This research clearly indicates that there is an urgent need for the development of both information and support services which are tailored specifically to individuals and families affected by rare disorders. International best practice models provide food for thought and included in the reports recommendations is a proposal for the establishment of a centre, which can provide social support, information and outreach for both families affected by rare disorders and the professionals who work with them, to provide a centralised point of expertise to communities throughout the island of Ireland.

The findings of this research are just the beginning. It indicates many areas where change is needed and it is important that all stakeholders work together to implement the recommendations to bring about real change in the lives of people with rare disorders.



Sarah-Jane Dillon,  
Director of Policy and Co-ordination, Rehab Group



## Acknowledgements

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# Executive Summary

## Background

Since 2000, RehabCare has been working with parents and individuals with rare disorders to look at their support needs and how best these needs can be met. During this time RehabCare has been strengthening relationships with key stakeholders, including professionals working in the field of rare disorders, support groups and parent contacts associated with specific rare disorders, and developing links with important national and European networks in the field of rare disorders.

Individual rare disorders by their nature will affect fewer people than more common disabilities. However, when taken together rare disorders affect a significant percentage of the population. According to the European Union, a rare disorder is defined as one that affects five or less people in every 10,000 population. Between 5,000 and 7,000 rare diseases exist and 80 per cent of these have identified genetic origins. The European Organisation for Rare Diseases [EURORDIS] estimates that 75 per cent of rare diseases affect children. These diseases are often life-threatening and/or chronically debilitating, and many rare conditions are so rare that they have not gained funding for the development of drugs and treatments. There are estimated to be approximately 363,000 people affected by a rare disorder in the Republic of Ireland and Northern Ireland combined.

RehabCare was awarded funding from the Health Research Board in December 2005 to carry out research to formally investigate the support needs of individuals affected by rare disorders and their families on the island of Ireland (Republic of Ireland and Northern Ireland). The aims of the research were as follows:

- *To ascertain the information needs of individuals with rare disorders, their families and health professionals in regards to rare disorders.*
- *To ascertain the social support needs of individuals with rare disorders and their families, including siblings.*
- *Where appropriate, to make recommendations for future social support for individuals and families who are affected by rare disorders.*

## Design of study

The research incorporated a mixed-method approach including qualitative and quantitative research. This was structured in two key phases:

**Phase 1** included a comprehensive review of relevant national and international literature, and an examination of European and national policy and practice relating to rare disorders.

**Phase 2** consisted of a consultation phase that included:

- semi-structured interviews with 23 families affected by rare disorders to establish their information and social support needs;
- consultations with five specialist health professionals in the field of rare disorders to determine their views on the social support needs of families and the information needs of professionals and families;
- a postal survey of 1,000 General Practitioners [GPs] to establish the need for information on rare disorders and the GPs' perception of the social support needs of families affected by rare disorders. 232 (23.2 per cent) of the GPs contacted responded.

Data collection took place between December 2007 and March 2008. Responses from the GP questionnaires were analysed using SPSS for Windows v15. All of the interviews with families and specialist health professionals were audio-recorded and transcribed. The transcriptions were thematically analysed and a set of quotations from all interviews was chosen to represent the themes discussed.

## Summary of findings

### Information needs

- Families reported negative experiences of receiving information about the rare disorder, including being presented with inaccurate, inaccessible or worst case scenario information. The requirement to filter or pace the information about the rare disorder and its diagnosis in accordance with the needs of the individual family was emphasised in order to avoid their being overwhelmed by information. The need for ongoing information on a life-span perspective was highlighted.
- Specialist health professionals involved in this research agreed that the information they have on rare disorders can often be in an unsuitable format for parents and that support is needed to help them translate information and compile it for families. This view was supported by GPs surveyed as part of the research, with 75.3 per cent stating that they experience difficulties providing information to families about rare disorders. Of the total sample of GPs 50.4 per cent cited time and workload pressures as the main difficulty in providing information to families about rare disorders. A further 38.4 per cent felt that the information was available but not in an accessible format for families and 27.6 per cent of the total sample of GPs felt that they did not know where to look for information on rare disorders.
- All of the family participants in this research had either used the internet to access information on the relevant rare disorder or had a friend who got them information from the internet; 60.3 per cent of the GPs who cited where they sourced their information on rare disorders stated that the internet was their primary tool. This research has reported on the strengths as well as the risks of internet usage in searching for information on rare disorders.
- The majority of participants in this research, particularly the specialist health professionals, emphasised the importance of appropriate information and support at the time of diagnosis.

### Social support needs

- The majority of the family participants in this study spoke about the stress that having a child with a disability had placed on their parental relationship. Due to the genetic nature of many rare disorders, feelings of guilt can also arise, and this was reported as adding to stress within the immediate and extended family.
- All of the family participants felt that peer support was an extremely important part of coping with rare disorders and plays a major role during difficult times. Lack of understanding about rare disorders was highlighted as a source of frustration by many parents, and therefore support from peers who experience the same rare disorder was deemed essential. Participants also recognised that the uncommon nature of rare disorders meant that peers served as an important source of information.
- Of the 72.2 per cent of GPs who agreed that having a rare disorder gives rise to additional family problems, 28.0 per cent felt that rare disorders can result in feelings of isolation. Two out of the five specialist health professionals in this research felt that peer support was essential in combating feelings of isolation and recognised the role of support groups for rare disorders in the provision of such peer support.
- All of those family participants in the research who came from families with more than one child felt that the brothers and sisters of the child with a rare disorder suffered negative affects. The majority of specialist health professionals in this research agreed that siblings of children with a rare disorder often felt overlooked.
- There is often a loss of income in families where a child has a rare disorder. Rare disorders by their nature can be 'chronically debilitating' and some participants in this research stated that they were forced to cease fulltime employment to care for their child.

## Services and rare disorders

- The majority of family participants spoke about the need for health services to be knowledgeable about the unique and complex needs of people with rare disorders and of the importance of being equipped to provide the right level of support. The participants in this research who had experienced flexibility and person-centred approaches in the provision of health services, which catered for the unique needs of the family experiencing the rare disorder, felt that this resulted in a more positive experience for both themselves and their children.
- Some participants reported that services for rare disorders were too scattered and felt that a one-stop shop approach to social support and therapy provision was needed. Specialist health professionals reported that a centre of expertise with appropriate outreach services is essential to adequately support and inform people and professionals about rare disorders. It was suggested that due to the relatively small numbers of people suffering from each rare disorder, it would be appropriate that this centre would cover the Republic of Ireland and Northern Ireland combined.
- The requirement for advocacy support and information on entitlements and service provision specific to the rare disorder the families were experiencing was emphasised. Some family participants felt frustrated that there was an expectation that they should fit into more generic services which were not sufficiently knowledgeable about their specific needs.

## Summary of recommendations

- ◇ In order to minimise the risk of families who experience rare disorders accessing worst case scenario, inaccurate or out-of-date information it is strongly recommended that a centralised information service and website should be developed for the island of Ireland. This service should be guided by a steering committee made up of specialist health professionals and families. The committee would be ideally placed to oversee and regulate content. It would also guide people on how to access information.
- ◇ There is an urgent need for a support service tailored specifically to individuals and families affected by rare disorders. A centre that can provide social support and information for families and professionals, as well as outreach for professionals, community-based services (including schools) and families is recommended to provide a centralised point of contact and expertise. It is recommended that key issues that were reported by participants in this research would be addressed in this type of service. These services, which would be delivered both in the centre and on an outreach basis where appropriate, would include:
  - facilitation of peer support
  - counselling for parents
  - information programmes tailored specifically to each rare disorder
  - sibling support workshops
  - response to the needs of the extended family
  - respite services
  - access to both established and alternative therapies
  - training for healthcare professionals about rare disorders
  - a centralised point for support groups and parent contacts for specific rare disorders
- ◇ In order to provide the best possible level of care it is essential that current health services be informed and equipped to provide the necessary supports that are specific to the unique needs of people with rare disorders.

- ◇ Families experiencing rare disorders should have access to practical information about their entitlements as well as about the services which are available to them.
- ◇ Greater interaction between rare disorder support groups should be supported. This should include the promotion of links between support groups and umbrella organisations such as Genetic and Rare Disorder Organisation [GRDO] and the Genetic Interest Group [GIG].
- ◇ The successful implementation of the guidelines set out by Harnett et al (2007) as a result of the 'Informing Families of their Child's Disability Project' in the Republic of Ireland would greatly benefit people with rare disorders.
- ◇ Epidemiological data is required to ascertain the exact number of people with rare disorders in the Republic of Ireland and Northern Ireland. Accurate data on the nature and distribution of rare disorders in both jurisdictions is essential in exploring the need for support and the provision of resources.
- ◇ There is a need to consider the additional financial pressures experienced by the families of those with rare disorders. It is important that the families of children with rare disorders are provided with adequate income supports to minimise financial difficulty.
- ◇ Further research is recommended that would explore the level of support each separate family member requires, as well as investigating the needs of families for the long term care of their child through adolescence to adulthood and the specific services required at these times. Further research should also investigate the specific needs of people who have a condition that has not been diagnosed.

# Chapter 1: Introduction and background

## 1.1 What is a rare disorder?

The European Union defines a rare disease<sup>1</sup> as one that affects fewer than five people in every 10,000 of the population<sup>2</sup>. Between 5,000 and 7,000 rare diseases exist and 80 per cent of these have identifiable genetic origins. The European Organisation for Rare Diseases [EURORDIS] estimates that 75 per cent of rare diseases affect children. These diseases are often life-threatening and/or chronically debilitating and many rare conditions are so rare that they have not gained funding for the development of drugs and treatments.

*“Even though the diseases are rare, rare disease patients are many.”* (EURORDIS, 2005a: p20)

Individual rare disorders by their nature will affect fewer people than more common disabilities. However, when taken together, rare disorders affect a significant percentage of the population. Using the European definition, it has been estimated that there are approximately 30 million people affected by rare diseases in 25 EU countries, which is approximately between 6 and 8 per cent of the population. It is estimated that up to four million children and adults are affected by rare disorders in the United Kingdom (Contact a Family, 2007a), which is approximately 6.5 per cent of the population. This includes Northern Ireland, where applying the 6.5 per cent rate would suggest there are approximately 113,000 people living with a rare disorder. In the Republic of Ireland it has been estimated that there are approximately 250,000 people affected by rare disorders (approximately 6 per cent of the population). This would suggest there are about 363,000 people on the island of Ireland living with a rare disorder.

## 1.2 RehabCare and rare disorders

RehabCare provides services to many people with rare disorders and the organisation seeks to support their specific needs. RehabCare’s connection with rare disorders originated in response to a recommendation in A Strategy for Equality, the Report of the Commission on the Status of People with Disabilities (1996: p44) that specified the need for the establishment of a national centre to provide support to people with rare disabilities in Ireland. Since 2000, RehabCare has been working with parents and individuals with rare disorders to look at their support needs and how best these needs can be met. One of the outcomes of this collaborative work was the development of support services for young adults with Prader Willi Syndrome [PWS].

By building relationships with the support groups and parent contacts associated with specific rare disorders and strengthening links with important national and European networks<sup>3</sup> in the field of rare disorders, RehabCare has also been developing a knowledge base which can be used to help families affected by rare disorders. Working closely with Professor Hilary Hoey at the Department of Paediatrics in Trinity College in 2002, RehabCare carried out an exploratory survey with paediatricians to investigate their perceptions of the social support needs of families affected by rare disorders in the Republic of Ireland. The findings highlighted a gap in social support provided to families affected by rare disorders and emphasised a need for further research. The value of partnership and association cannot be overestimated. A notable example of this related to the recent all-Ireland submission by the Irish Platform for Patients Organisations, Science and Industry [IPPOSI] (2008) to the Public Consultation regarding European action in the field of rare diseases.

In December 2005, RehabCare was awarded funding from the Health Research Board to carry out research that would investigate the support needs of both people who have rare disorders and their

1 The terms ‘rare disease’ and ‘rare disorder’ are used interchangeably in a number of publications, and therefore used interchangeably within this research report, depending on how the term is referred to within the literature quoted, or by a respondent. The authors of this report refer to the term as ‘rare disorder’.

2 For the purpose of an official definition we refer to the European Union definition of a rare disease.

3 For a directory of national and international organisations that provide social support and information services to people with rare disorders and their families see Appendix 1.

families. This research was comprised of a comprehensive review of relevant national and international literature and an examination of European and national policy and practice relating to rare disorders. Consultation also took place with three groups of people across the island of Ireland — families experiencing rare disorders, specialist health professionals<sup>4</sup> and General Practitioners. The main aims of the research were as follows:

- 8 |
- To ascertain the information needs of individuals with rare disorders, their families and health professionals.
  - To ascertain the social support needs of individuals with rare disorders and their families, including siblings.
  - Where appropriate, to make recommendations for future social support for individuals and families affected by rare disorders.

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<sup>4</sup> Specialist health professionals, in the context of this research, are defined as professionals with a work remit/professional interest in the field of rare disorders on the island of Ireland.



## Chapter 2: Literature review

This section includes a review of national and international literature related to rare disorders and an examination of national and European policy and practice as it relates to rare disorders. In a number of sections we have used evidence from literature and research into general disabilities in order to ascertain a general picture of the needs of families where a disability is present and to highlight the specific needs of people with rare disorders and their families.

### 2.1 Epidemiological data for rare disorders

An understanding of the nature and distribution of rare disorders on the island of Ireland is essential to any exploration of the need for support and resources. However, data about the numbers of rare disorders is limited, not only within the island of Ireland but across Europe and internationally. The limitations with regard to epidemiological data are well documented. Van Weely and Leufkens (2004) acknowledged the lack of reliable epidemiological data on rare diseases at an international level, stating that people with rare diseases are not generally registered in databases. The authors noted one of the reasons for the lack of data is that the International Classification of Diseases code was not, in practice, convenient for identifying and listing specific rare diseases, where other rare disorders are summed up as “other endocrine and metabolic disorders”. Whilst acknowledging the importance of estimating the total number of people affected by each rare disease, Orphanet (2008) has also stated that there is very limited documented information on the epidemiology of rare diseases, with exact prevalence data being difficult to obtain from available data sources.

Orphanet (2008) carried out a systematic survey of literature to provide an estimate of the prevalence in Europe of ‘most’ rare diseases. The authors noted the “*low level of consistency between studies, poor documentation of methods used, confusion between incidence and prevalence, and/or confusion between incidence at birth and life-long incidence*” (p1). Also, epidemiological data is usually collected in places of high prevalence and this can lead to an overestimation for diseases (Orphanet, 2008). Difficulty in making a diagnosis can contribute to the lack of data, where undiagnosed patients fail to be registered on any database. Grut *et al* (2007) carried out a research project that described the methodological challenges connected to the estimation of incidence and prevalence of a number of rare disorders in Norway. Weaknesses in the tools for coding and insufficient routines for coding were identified as being issues in achieving an outcome of reliable data, with the authors concluding that in order for correct information to be presented on incidence and prevalence there is a need for improvement in public registration of rare disorders.

The Network of Public Health Institutions on Rare Diseases [NEPHIRD] (2007) recommended that immediate action is required in order to promote, support and implement registers for rare diseases. The need for a registry for rare diseases was also highlighted by patients in the EurordisCare3 Survey (2008). This survey of 16 rare diseases was administered to patient organisations across 22 countries. A total of 5,995 patients responded in Europe, including 93 patients in Ireland. Significantly, 59 per cent of responses from Ireland felt that “monitoring the current needs of the patient community of this rare disease through surveys or register of patients” was “essential”. Such a register was also regarded as essential by 42 per cent of the overall European sample. Both the Genetic Interest Group [GIG] (2008), representing the views of patient groups in the United Kingdom [UK] (including Northern Ireland), and the Irish Platform for Patients Organisations Science and Industry [IPPOSI] (2008), representing the views of the rare disorder field in the Republic of Ireland, highlighted the importance of registries for patients with rare diseases when they made submissions to the report ‘Public Consultation on Rare Diseases: Europe’s Challenges’. IPPOSI (2008) stated:

*“In Ireland due to coding issues and lack of patient registries for Rare Diseases obtaining accurate and reliable information is difficult, aggravating planning and the provision of resources to people with Rare Diseases.” (p2)*

## 2.2 Information and rare disorders

Previous research has emphasised the need for the “right kind” of information for individuals with a rare disorder and their families. Allford and Winter (2007) conducted a set of focus groups for patients with one of six rare genetic conditions and their families and carers in the UK. The aim was to gather information, views and respondents’ perceptions of currently available information and services within the UK. The consultations highlighted the importance of good quality and accessible factual information. Van Nispen *et al* (2002) used data from the Patient Panel of the Chronically Ill (PPPZ) in the Netherlands to identify 206 patients who, between 1998 and 2000, suffered from rare chronic illnesses. This group received questionnaires twice a year, from 1998 to 2000. In addition, telephone interviews were conducted with 50 members of this panel for further in-depth consultation. Their results showed that more than one-third of people with rare disorders wanted to get more information on their disease and approximately 40 per cent wanted to get more information on the treatment or on the financial aspects of their illness.

Webb (2005) conducted 15 semi-structured interviews with 23 parents of children with Duchene muscular dystrophy [DMD]. The purpose of the research was to examine strategies parents use to cope with their children who have DMD. The results emphasised the importance of information for parents, stating that vast amounts of information can empower parents to have a more proactive and positive outlook.

Other research has highlighted the deficiencies in information provision to parents and the negative impact this can have on families. For example, Krammer (2003) analysed responses from questionnaires received from 138 parents of children with rare disorders in America and reported that the majority of respondents stated that it was difficult to locate information related to their disorder. Almost three-quarters — 74 per cent — of respondents found it “somewhat difficult” or “very difficult” to get information on voluntary support groups, and 62 per cent stated that they found it difficult to access written information that was easy to understand. RehabCare (2002) carried out a consultation with 14 paediatricians about services and facilities available to people with rare disabilities throughout the Republic of Ireland. Its findings highlighted that 40 per cent of respondents expressed difficulty in accessing up-to-date information for families and children. An overwhelming majority (73 per cent) of respondents admitted to experiencing difficulty in sourcing appropriate facilities in the Republic of Ireland that would provide information and education to the whole family regarding the specific rare disorder.

### 2.2.1 The internet and information on rare disorders

Gallagher *et al* (2007), in their study of internet use and the seeking of health information in Ireland, highlighted that people can and do use the internet to find specific health-related information or to supplement information given by a doctor. A study carried out in the Bath area of England that surveyed parents attending paediatric outpatient clinics found that 32 per cent of parents had used the internet to seek information on their child’s condition (Tuffrey & Finlay, 2002). For rare disorders, there is often a lack of general information as by their very nature they are uncommon. Krammer (2003), looking specifically at the needs of the rare disorder community, found that 81 per cent of patients and families used the internet as a source of information, with the second most frequent source of information being support groups or health charities.

Looking to the general population of disability, searching for peers on the internet as an avenue of information-seeking was emphasised by Powell *et al* (2003) in their research of patients’ use of the internet for healthcare information. The authors also highlighted that peer-to-peer support in virtual communities on the internet can overcome traditional barriers to access. In relation to rare disorders, this type of forum can allow individuals to find peer support and draw on a wealth of health perspectives and experience that they would otherwise not have access to.

A major disadvantage, however, to health-seeking on the internet is that the quality of some health websites is difficult to ensure (Gallagher *et al*, 2007). Powell *et al* (2003) pointed out that although good quality information can be found, information accessed via the internet is often incomplete and inaccurate. Fox (2005) also drew attention to the difficulties with the reliability and quality of health sites.

It is therefore important that information about rare disorders provided on the internet is accessible for families and that it is of a high quality and reviewed by professionals. A main concern is that unreliable/inaccurate information with case studies demonstrating worst case scenarios can frighten families. In the field of rare disorders, there are concerns by health professionals and support groups that the lack of regulation on websites can be more harmful than positive for users. For example, Contact a Family (n.d.), a charity operating in the UK providing support to families experiencing rare disorders, state that they have been contacted by a number of support groups expressing concern over inaccurate and out-of-date information on the internet.

However, steps have been taken to regulate health information and peer support facilities on the internet. For example, Gallagher *et al* (2007) highlight the steps towards regulation of websites providing health information via the development of organisations to oversee and monitor their content. These include Health on the Net (Hon) Foundation and a tool for assessing the quality of websites known as DISCERN. DISCERN is a brief questionnaire that provides users with a valid and reliable way of assessing the quality of written information on treatment choices for a health problem. DISCERN can also be used by authors and publishers of information on treatment choices as a guide to the standard that users are entitled to expect.

With regard to rare disorders specifically, EURORDIS (2004a), in response to concerns raised by a number of agencies, released a list of organisational principles for those providing information to individuals with rare disorders and their families. It highlighted the importance of recognising patients' expertise, the value of inclusiveness, accessibility, sensitivity, human resource obligations and the use of an advisory expert committee. It also advised within the principles that information must be validated and revisited regularly and provided in a clear and accessible way in terms of content, format and appearance.

### 2.2.1.1 Rare disorders and internet-based support groups

Many support groups for people with rare disorders use the internet as a forum for information and peer support. A study was carried out in the Netherlands where 44 patient organisations and 167 patients and parents responded to a survey on the supply of information by healthcare professionals and the availability of information to patients suffering with rare diseases. The survey showed that most information received by patients and their families in the Netherlands was from patient organisations (Huizer, 2002). Support group websites in the rare disorders field are popular sources of health information but these can be unregulated. As with health information on the internet, the literature has also pointed out the need for regulation of communities on the internet that facilitate peer support. For example, Powell *et al* (2003) pointed out that there are some concerns that the lack of a system of moderation in virtual communities could lead to the dissemination of inaccurate messages. Together, Contact a Family and the Information Management Research Institute [IMRI] (2003) recognised that the role of support groups and the information and support they provide needs to be widely advertised. However, they pointed out that it is essential that these websites contain good quality information and are well designed. The group issued guidelines entitled the Judge Project<sup>5</sup> for both consumers and support groups for judging the quality of health information on the internet.

One example of good practice in the provision of information on the internet is the online Directory of Specific Conditions and Rare Disorders created by Contact a Family (2008) that is accessible to individuals, families and professionals. This directory was developed in response to individuals, parents and professional workers frequently needing to access reliable, up-to-date information about specific conditions and rare disorders and the related support organisations. The directory is written in co-operation with relevant specialist representatives from the medical profession.

<sup>5</sup> [www.judge.org.uk](http://www.judge.org.uk)

## 2.2.2 Knowledge of rare disorders among healthcare professionals

McInerney (2008), in an overview of genetics education for non-genetics health professionals, pointed out that comprehensive knowledge is required to provide patient-centred care and that a recognition is needed that effective genetic patient care requires the expertise of a diverse range of healthcare professionals.

In an exploration of genetics in medical practice in the US, Korf (2005) highlighted that, in general, medical courses suffered from persistent and pervasive deficiencies in genetics education.

Specifically in relation to primary care, Van Nispen *et al* (2002) found from their research with people who have rare diseases in the Netherlands that 93 per cent of their sample contact their General Practitioners [GPs] and that on average they contact their GP five times a year. Gutmacher *et al* (2007) in their study on educating healthcare professionals about genetics in the United States, pointed out that a crowded curriculum, poor teaching of genetics and a lack of confidence and knowledge about genetics among GPs resulted in poor knowledge of genetics among non-expert health professionals. Hsia *et al* (1979), in their research on genetic services and education, felt that GPs must have adequate knowledge to recognise a problem as genetic and that they should have enough familiarity with genetic principles to use the literature wisely or consult in-depth with a geneticist.

Knight and Senior (2006) found in their overview of problems arising with rare diseases in general practice that guidelines were needed for GPs on their role in dealing with rare disorders as many patients with rare diseases often present their symptoms first to their GP and then continue to use their GP for general preventative healthcare between their specialist visits. The authors therefore argued that a systematic generic primary-care approach to rare disease could reduce problems such as lack of co-ordinated care, lack of information, delayed diagnosis and other difficulties encountered by people with rare disorders and their carers. Drury *et al* (2007), in their research on genetics support to primary care practitioners, argue that GPs need to be supported by specialist genetics services to enable them to cope effectively with their patients. The authors also evaluated an outreach service in the UK in which a genetic counsellor supported 10 general practices in the areas of genetic information, education and service delivery. Though the clinics proved expensive, they were found to be extremely effective in increasing both patient-centred care and education for the GPs involved.

Health professionals also access the internet to retrieve information. Powell *et al* (2003) considered the use of the internet as an information source for professionals looking for information on general health, pointing out that clinicians benefit from increased access to evidence, policy and guidelines and training and professional development. The rarity of the disorder health professionals are dealing with can cause them to search for information on the internet. This behaviour has been acknowledged by the French Ministry for Health, Institut National de la Santé et de la Recherche Médicale [INSERM] and the EU, who have collaborated to fund an initiative called Orphanet<sup>6</sup>. Orphanet acts as a free portal for rare diseases and orphan drugs allowing health professionals across Europe to access information on specific rare disorders via an online database. The portal also has a section containing information for patients. The information for patients includes an online facility to contact other patients. In 2008, this facility was only available in French. The other information provided is mainly medical, including details about the disease, clinics, reference centres, diagnostic tests, research projects and registries, but also includes information and links to patients' organisations.

## 2.2.3 Diagnosis

### 2.2.3.1 The absence of diagnosis

*"The absence of a diagnosis or a late diagnosis may lead to an unnecessary deterioration of the patient's condition."* (Van Weely & Leufkens, 2004: p8)

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6 [www.orpha.net](http://www.orpha.net)

Krammer (2003) reported on diagnosis time for 138 rare disorder respondents in the United States. In this study, 17 per cent of patients went undiagnosed for four years or longer; 64 per cent of respondents cited “doctor confused by symptoms”, and 58 per cent reported “doctor unable to diagnose” as reasons for delays in diagnosis.

EurordisCare2 (EURORDIS, 2004b) is a survey of diagnostic delays for eight diseases (all defined as rare) which was carried out across Europe. The study received 5,980 completed questionnaires from a co-operating 69 European rare disease organisations. It was found that 25 per cent of patients had to wait between five and 30 years from early symptoms to confirmatory diagnosis of their disease, leading to medical interventions that were not based on a correct diagnosis. These issues highlighted very specific dilemmas of having no diagnosis due to the rarity of a disease, including the loss of confidence of patients in the healthcare system and the medical profession.

Hansen and Ege (n.d.) conducted a survey on the living conditions of people with rare disabilities in Denmark. A key conclusion highlighted the difficulties experienced by parents and the family during the period of no diagnosis. Contact a Family is one of the support groups that provide information to families who have a child with no diagnosis (other organisations include Syndromes Without a Name [SWAN] and Unique). Contact a Family (2005) states that feelings of frustration, confusion and worry can arise in a family when there is no diagnosis. One worry is that patients may be deprived of appropriate services because there is no “label” on the disorder. As described by Early Support (n.d.):

*“Without a clear diagnosis, it can feel like you’re in limbo, not knowing what’s happening to your child or what the future holds. It can also feel frightening if you don’t know how a condition might progress.” (p1)*

### 2.2.3.2 Support at diagnosis

The literature reports that when a patient receives a diagnosis of any disability there are important considerations in relation to information provision and method of delivery of the diagnosis. Woolfe and Bartlett (1996) for example, found that, at diagnosis, parents wanted clear unambiguous information and continued contact and support. Kerr and McIntosh (1998) in their exploration of disclosing a disability diagnosis found that the most important factors for parents were that they were told straightaway and in private. They also concluded that it was essential for healthcare professionals to realise the impact of diagnosis for parents so that parents can be guided and supported through the emotionally difficult early days.

Specifically looking to rare disorders, EurordisCare2 (2004b) found that 45 per cent of respondents reported poor communication at the time of their diagnosis. Starke *et al* (2002) indicated that a parent’s experience of receiving diagnosis of Turner Syndrome was affected by the doctor’s ability to provide relevant information and to correctly manage the psychosocial aspects. They also found that in order to effectively counsel the parents at the diagnosis stage, doctors should have a better understanding of what the parent experiences when hearing the diagnosis.

Harnett *et al* (2007) completed a comprehensive consultation and research report in the Republic of Ireland that aimed to develop evidence-based national best practice guidelines, as well as education and training on appropriate procedures to inform families of their child’s disability when communicating a diagnosis or concern. The report entitled ‘Informing Families of their Child’s Disability’ recognised that every disclosure event is unique and outlined seven guiding principles that the author felt should be applied in every case. Particularly notable in the area of rare disorders is the inclusion of “appropriate, accurate information” within these seven guiding principles.

The seven principles were:

- Family-centred disclosure
- Respect for child and family
- Sensitive and empathetic communication

- Appropriate, accurate information
- Positive, realistic messages and hope
- Team approach and planning
- Focused and supported implementation of best practice

(Harnett *et al*, 2007: p8-9)

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## 2.3 Rare disorders and the social support needs of the family

The Swedish Association of Rare Disorders (2002) analysed responses to questionnaires from more than 2,000 of their members that aimed to establish the extent of the problems which people with rare disorders experience in daily life. The results highlighted the unique and complex problems experienced by a person and their family due to the rarity of their disability. The review of the literature below acknowledges that there are common social support needs experienced by all families who experience any disability, with each section focusing also on the relevant studies that relate directly to families experiencing the very specific and unique needs associated with rare disorders.

### 2.3.1 Emotional support

Davies and Hall (2005) in their UK-based evaluation of parents and professionals in partnership suggested that parents often feel isolated, lonely and unsupported when caring for a disabled child. Family relationships suffer as parents are too tired to have time for each other and for their other children. Isolation and a feeling of stigmatisation are commonly experienced by parents of disabled children as their lives revolve around the child.

Redmond *et al* (2000) reported in their study of fragile babies and children with developmental delay that mothers experienced social isolation due to giving up work and caring for their baby full-time. The authors found that disability had a negative impact on how families functioned in terms of relationships with the other children and also between parents. The lack of an advocate who could help families to obtain information and access support services was reported as a further source of stress.

Contact a Family and One Plus One (2003) carried out a survey of more than 2,000 parents of children with disabilities in the UK. Findings about the effect of having a child with a disability on parent relationships concluded that 23 per cent thought that it had brought them closer together and 19 per cent thought it had little effect, with more than half the sample reporting negative effects of having a child with a disability — 31 per cent felt that it had caused some problems, 13 per cent felt that it had caused major problems and 9 per cent felt that having a disabled child had led to separation.

In relation specifically to rare disorders, Mayo (2003), using interview material from 51 parents of children with rare disorders in the UK, found that parents in particular will feel isolated, have anxiety about their child's future and feel concern for their child's siblings. Delve *et al* (2006) in their report on stress and wellbeing among parents of children with rare disease pointed out that mothers of children with a rare disability reported high parental stress and high physical and emotional strain. The authors found that mothers experienced higher levels of stress compared to fathers and that parental stress was related to incompetence, role restriction, social isolation, spouse relationship problems and health problems. In the Republic of Ireland, 13 out of the 14 paediatricians surveyed by RehabCare (2002) felt that having a child with a rare disorder gave rise to more family problems than the normal issues experienced in a family with a child with a disability, including marital tension and stress.

## 2.3.2 Peer support<sup>7</sup>

### 2.3.2.1 Peer support: theoretical perspectives

Previous research has demonstrated the importance of social support for families of children with special needs (Hartman *et al*, 1992; Ainbinder *et al*, 1998). The research shows that peers can play a crucial role in supporting people with rare disorders and in combating feelings of isolation, as often the idea of real understanding can only come from a peer with similar experiences. Kerr and McIntosh (2000) for example, in their qualitative study of 63 families with limb deficiencies, found that social support can be an important buffer against the stress and isolation faced by parents. They also found that when a child has a disability, parents experience the typical stressors associated with parenthood plus a host of additional stressors unique to their child's disability.

Mead *et al* (2001) described peer support as being about understanding another person's situation empathically through the shared experience of emotional and psychological pain and pointed out that people find affiliation with others they feel are like them. Dass and Gorman (1985) described peer support as being about normalising what has been named as abnormal because of other people's discomfort. Curtis (1999) pointed out that positive peer support can offer a culture of health and ability as opposed to a culture of illness and disability.

Kerr and McIntosh (2000) described the following as being the benefits of peer support among families:

- The realisation you are not alone — this marked a crucial turning point for many families.
- Someone understands — partners felt that only others with the same situation could truly understand.
- A glimpse into the future — fear for their child's development was assuaged by some positive examples from peers.
- Coming full circle — many parents discussed the fact that after a number of years receiving peer support they themselves felt ready to offer peer support to others.
- Lessening need — generally as the children got older parents felt less of a need for peer support but often maintained contact with peers. (p317-318)

Peer support provides parents with the benefits of learning from experience — something that could not be derived from other sources (Kerr & McIntosh, 2000). This also supports previous research findings that parents of children with special healthcare needs are uniquely qualified to help each other (Ainbinder *et al*, 1998).

Despite the positive affects of peer support, there can be some negative implications and Kerr and McIntosh (2000) maintain that care must be taken not to introduce parents to veteran parents with unresolved problems or negative attitudes toward their child's disability as this could have a potentially damaging effect.

### 2.3.2.2 Peer support: groups and organisations

Van Nispen *et al* (2002) in their research of patients living with rare disorders in the Netherlands found that peer support and patient organisations are of vital importance for people with rare disorders. More than a third of participants in their study had joined support groups and they listed information supply and contact with fellow sufferers as being the most positive supports. The authors pointed out that support groups did not exist for every rare disorder and suggested that umbrella patient organisations could play a part in supporting such individuals. A number of support organisations do acknowledge the importance of peer support specifically for rare disorders by facilitating contact between parents and/or individuals with rare disorders. Two examples of support groups responding to the need for peer support include the Making Contact website<sup>8</sup> based in the UK and the National Organisation for Rare Disorders

<sup>7</sup> Peer support in the context of this report refers to people meeting in order to exchange information and emotional support with others facing similar experiences and challenges.

<sup>8</sup> [www.makingcontact.org](http://www.makingcontact.org)

[NORD] Networking Program<sup>9</sup> for members of this US-based organisation. Even though access to these facilities is available, there is no forum for families on the island of Ireland to make contact with one another.

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Within the island of Ireland support groups do exist that are specific to the rare disorder being experienced by the families. Many of these groups are run by parent contacts who recognise the importance of peer support. However, most of these groups are not funded in their efforts to facilitate families meeting one another. Due to the uncommon nature of the rare disorder, many families have to travel a considerable distance to meet with others.

### 2.3.3 Siblings

Perrin (1999), in a study carried out in the Republic of Ireland, described the feelings of children who have a sibling with a physical and sensory disability. The children were involved in age-appropriate 'sibshops' where they could meet other children who have siblings with a disability. They were encouraged to share experiences, ask questions and learn coping strategies. The author concluded that the children involved in the groups were extremely well-adjusted, they exhibited feelings of happiness, appreciation, consideration and love for their family and brother or sister. Negative feelings included unfairness at the uneven balance of parental attention. The group process was described as a stimulating and challenging way to link with children and a learning experience for the facilitators.

Cate & Loots (2000) investigated the experiences of 43 siblings of children with physical disabilities, including the sibling relationship, the relationship with parents, and the relationship with others. The children took part in one-to-one sibling relationship interviews with the researcher. The study found that stressful situations do occur more often for the children interviewed than for children without a disabled sibling. However, there were no serious problems in psychological adjustment. Everyday problems the siblings experienced included difficulty with managing to do activities together and communication difficulties due to speech impairments. Relationships with friends were not affected. They did report encountering difficulties with strangers who often directed stares and unpleasant remarks at their disabled sibling. The children had a positive view of their relationship with parents, reporting that the main characteristics of the relationship were open communication and trust (Cate & Loots, 2000).

A number of rare disorder support groups have made reference to the importance of caring for the needs of siblings (e.g. Contact a Family, 2007b; Unique, 2005). Unique (2005) point out that the whole family has to adjust to the changes a child with a disability brings to the family. The organisation highlights that parents' attitudes and mechanisms for dealing with their child's disability are important in enabling siblings to accept and value their disabled brother or sister. Common difficulties which siblings have experienced include a lack of understanding and knowledge about their brother or sister's disorder, also the feeling of being left out and potential embarrassment at their brother or sister looking and behaving differently. Barretstown in the Republic of Ireland provides activity-based programmes for children affected by serious illnesses and their families. The organisation supports the view of caring for the needs of all the family in providing summer camps for both the whole family and for siblings.

### 2.3.4 Financial issues

Financial issues have been reported in the literature regarding families affected by rare diseases. Van Nispen *et al* (2002), for example, in their study with rare disease patients in the Netherlands, found that approximately 40 per cent of their sample of people with rare diseases wanted to get more information on the treatment or on the financial aspects of their illness. The authors stated that there was an indication that people with rare diseases were far more likely to face difficulties with housing and finance:

*"...the need for information on the treatment or on financial aspects is stronger among people with rare diseases than among people with more prevalent diseases...this higher rate could be due to the*

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9 [www.rarediseases.org/programs/networking](http://www.rarediseases.org/programs/networking)



*fact that people with rare diseases more often indicated to face financial and housing problems.”*  
(Van Nispen et al, 2002: p7)

Redmond *et al* (2000) interviewed 17 mothers who were the primary carers of vulnerable and fragile babies under four years of age, with a significant and sometimes life-threatening level of developmental disability. Fifteen out of the 17 cases reported a financial pressure on families, due to the loss or diminution of the mother’s income.

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Krammer (2003) stated that 77 per cent of respondents reported that their rare disorder had caused the patient or their family a financial burden, with 32 per cent stating that the burden was “extreme”.

## 2.4 Services for rare disorders

The literature reports that people with rare disorders generally use generic disability services and there are few services tailored to people with rare disorders. An example of this is EURORDIS (2005) which recognised the need for dedicated services for people with rare disorders both nationally and across Europe. These include day care services, respite centres, emergency units, socialisation and rehabilitation centres, summer camps, education services and professional training.

The Genetic Interest Group (1999) consulted with clinicians and support workers for families and presented 20 case histories with the aim of making recommendations for the achievement of integrated services and support for families with rare genetic disorders in the UK. The report concluded that *“the experience of many families is of delayed or absent diagnosis, non-referral to sources of expert help and inadequate social support”* (p25).

The importance of a holistic, family-centred view of service provision is highlighted in the literature. Hennepe (1999) in his study in the Netherlands states that parents of children with rare diseases experience problems such as lack of knowledge, late diagnosis, lack of continuity in care, stress within the family and lack of knowledge about where to get support. The author stated that a multi-disciplinary approach to the provision of supports is needed. The Genetic Interest Group (1999) also stated the importance of multi-disciplinary medical care having a holistic view of the affected person and his or her family. Hernandez *et al* (2006) reported on a study carried out in America that examined perceptions of state health and human service programs on the family-centred nature of services for children with genetic conditions and their families. Findings indicated that there was a need to enhance family input into service design, evaluation and policy development processes of existing services. Van Nispen *et al* (2002) stated that people in the Netherlands with rare diseases need more care co-ordination than people with more prevalent diseases.

## 2.5 Policy context

### 2.5.1 National disability policy activity

#### 2.5.1.1 Disability policy in the Republic of Ireland

The recommendations of the 1996 Report of the Commission on the Status of People with Disabilities in Ireland and the subsequent debate about provision of services to people with disabilities has led to many legislative and policy developments in the area of disability in the last 10 years.

In 2004, this culminated in the launch of the National Disability Strategy and the ongoing implementation of this Strategy has had considerable impact on the delivery of services to people with disabilities in the Republic of Ireland.

The National Disability Strategy comprises:

- The Disability Act 2005 which included the introduction of the Independent Assessment of Need process; provisions for increasing the public service employment of people with disabilities to 3

per cent; the establishment of a Centre for Excellence in Universal Design and access to buildings and services for people with disabilities

- The creation of Sectoral Plans by the six government departments seen as having the most impact on the lives of people with disabilities, outlining their plans to improve the services they provide to people with disabilities
- Commitments by government to multi-annual funding
- The Citizens Information Act 2007 which provides for personal advocacy services for people with disabilities
- The Education for Persons with Special Educational Needs Act 2004 which sets out a new process for the assessment of the educational needs of children and adults with disabilities

It is predicted that the Independent Assessment of Need process under the Disability Act 2005 will have wide-reaching consequences for the provision of services to all people with disabilities. Under this system people with disabilities are entitled to apply for an independent assessment of their health and educational needs, resulting in a statement of the supports they have a statutory right to receive. The process commenced for children between 0 and five years in 2007 and will be extended to all adults with a disability by 2011.

These legislative developments have been supported by the creation of two key bodies in the disability sector. The National Disability Authority [NDA] was established in 1999 and is the lead state agency on disability in Ireland, providing independent, expert advice to government. In 2007, the Health Information Quality Authority, an independent authority designed to ensure that the health services in Ireland are of the highest quality, was established and is currently developing national standards in a number of areas of health and social care service provision to people with disabilities.

Information about the number and type of disabilities represented in the Republic of Ireland has been collected by the Health Research Board's National Intellectual Disability Database since 1995 and the National Physical and Sensory Disability Database since 2002. The databases are designed to provide comprehensive and accurate data to support future service planning.

Internationally, the Irish Government has shown its commitment to recognising the rights of people with disabilities and in 2007 was one of the first countries to sign the UN Convention on the Rights of People with Disabilities.

#### **2.5.1.2 Disability policy in Northern Ireland**

On May 8, 2007, the Northern Ireland Assembly regained devolved powers and is currently the prime source of authority for all devolved responsibilities and has full legislative and executive authority. The Department of Health, Social Services and Public Safety has responsibility for policy and legislation for hospitals, family practitioner services and community health and personal social services, as well responsibility for policy, legislative and administrative actions to promote and protect the health and well-being of the population. Disability issues are a key responsibility of this Department.

In order to provide protection for disabled persons against discrimination on the grounds of disability, the Disability Discrimination Act came into force in 1995. This was the UK's first anti-discrimination legislation specifically for disabled people and it enshrines in law disabled people's rights to participate in civil society. Since then it has been amended, for the Northern Ireland case, by a number of Orders and Regulations. Legislation relevant to the care, welfare and support of children and adults with a disability in Northern Ireland including the Children (Northern Ireland) Order 1995; the Chronically Sick and Disabled Persons Act 1978; and the Disabled Persons (Northern Ireland) Act 1989.

Many initiatives developed by the UK government and its agencies have an effect on the services and funding available to disabled people in Northern Ireland. The UK Department of Health's Children's National Service Framework (2004) set standards for services for disabled children and families against which services will be inspected. One other important government initiative in recent years was Aiming

High for Disabled Children: Better Support for Families, which was announced in May 2007. This initiative is UK-based and includes families living with a disability in Northern Ireland.

In Northern Ireland, the report *Equal Lives: Review of Policy and Services for People with a Learning Disability in Northern Ireland* (2005) emerged from the Review of Mental Health and Learning Disability. It set out a vision for developing services for adults and children with a learning disability for the next 15 to 20 years in Northern Ireland and made recommendations to the Government about the integration and participation for people with disabilities in their communities and accessibility to the full range of opportunities that are open to all other groups.

## 2.5.2 Policy activity and rare disorders

### 2.5.2.1 Rare disorder policy activity in Europe

The European Union [EU] member states who have developed specific public policies on rare diseases at a national level are Denmark, France, Italy, Sweden, Spain and the UK. France is the only country in the EU with an established and comprehensive National Plan on Rare Diseases.

The first EU effort in the area of health protection in relation to rare diseases in Europe was the Community Action Programme on Rare Diseases 1999-2003. The aim of the programme was to ensure a high level of health protection in relation to rare diseases, with specific attention given to improving knowledge and facilitating access to information about rare diseases. This was followed by the EU Public Health Programme 2003-2008 Community Action Programme for Public Health. This Programme also focused on the exchange of information as well as encouraging co-operation between nations within Europe. The Sixth Framework Programme 2002-2006 included, amongst many other actions: *“the fight against ... rare diseases: to improve prevention and care, to share means dedicated to research on these diseases”*. (Orphanxchange, 2007).

The European Project for Rare Diseases National Plans Development [EUROPLAN] is funded by the European Commission and commenced in the first semester of 2008 for three years. EUROPLAN aims to identify and promote best practices in order to share information on effective strategies to address rare diseases and to instigate the creation of national plans on rare disease by all participating European countries.

Many elements of EU rare disease policy and programmes are related to clinical research, orphan drugs and scientific advances. One innovative project that relates to the social needs of people affected by rare disorders is funded by the Public Health Programme of the European Commission, DG SANCO, called The Rare Disease Solidarity Project [RAPSDODY]<sup>10</sup>. This project is led by EURORDIS and aims to respond to the needs of European Rare Disease patients by improving the quality of care, information and social services for them. The project concludes in 2008 and to date has established European networks of help lines, respite care services and therapeutic recreation programmes.

The Public Health Programme of the European Commission, DG SANCO, launched the Public consultation regarding European action in the field of rare diseases at the fourth European Conference on Rare Diseases, 2007. The consultation included discussion around orphan drugs, coding and classification of rare diseases, the need for registries, clinical screening and testing, national action plans and partnerships between charity and industry. In relation to the social and information needs of families, the consultation asked for public responses on the kinds of specialised social and educational services for rare disease patients and their families that should be recommended at both EU level and national level. This consultation yielded more than 600 contributions, which the European Commission announced was a record response to a public consultation on health. The responses were being collated in early 2008.

A number of groups from the Republic of Ireland and Northern Ireland made submissions to the consultation. IPOSSI (2008) made a submission representing the views of science, industry and patients in Ireland. It stated that national action plans were vital for the provision of adequate services and information to people with rare diseases and their families in Ireland and advised that European

<sup>10</sup> [www.rapsodyonline.eu](http://www.rapsodyonline.eu)

recommendations and guidelines on this would assist the acceleration of the process in Ireland. This view was echoed by GIG (2008) in the UK who stated that they strongly support developing action plans to impact directly on services for patients and families with rare diseases, and on research and therapeutic development. The adoption of such national plans was recommended by the EU and within the UK, devolution of health services means action plans need to be developed at regional level (England, Wales, Scotland, Northern Ireland).

On February 29, 2008, the first Rare Disease Day was held across Europe. Organised by EURORDIS and national alliances for rare diseases, this was the first time that patient groups from different countries and representing a variety of rare diseases collaborated on a co-ordinated large-scale communication campaign to make rare diseases a public health priority. The day ended with the first Public Hearing on Rare Diseases at the European Parliament in Brussels. The day was also marked in the Republic of Ireland and the UK. On the February 29, 2008, The Genetic and Rare Disorders Organisation [GRDO], the Medical Charities Research Group [MCRG], IPOSSI and RehabCare collaborated to take part in the Irish section of the event. The occasion aimed to raise awareness amongst Irish policymakers of the medical and social needs of rare disease patients and their families. The UK event was organised by GIG and 200 patients and representatives from across the UK travelled to Westminster to meet parliamentarians to raise the awareness of the needs of patients with rare diseases and to discuss policy developments at a UK and European level.

### **2.5.2.2 Rare disorder activity in the Republic of Ireland and the UK (including Northern Ireland)**

Two organisations which are engaged in lobbying activities specifically for people with rare disorders operate in the UK and the Republic of Ireland. In Ireland, the Genetic and Rare Disorders Organisation [GRDO] is a non-governmental organisation that aims to act as an alliance for voluntary groups representing the views and concerns of people affected by or at risk of developing genetic or other rare disorders. GRDO also aims to act as a watchdog in relation to legislation concerning disability to ensure the rights of people with genetic or other rare conditions are protected. The Genetic Interest Group [GIG] is a national alliance in the UK and Northern Ireland for patient groups which aims to increase the public/government profile of genetic conditions including rare disorders.

During the Irish EU Presidency in 2004, University College Cork [UCC] hosted a European Awareness Conference 'Empowering the Rare Disease Community'. The one-day conference was organised by EURORDIS jointly with the European Centre for Clinical Trials in Rare Diseases, based in UCC, and highlighted the needs of the rare disorder community at a national and European level.

## **2.6 Practice in Europe: three models of best practice**

The literature has mentioned a number of support groups providing practical advice to families (e.g. Contact a Family, Unique) and patient-driven alliances (e.g. NORD, EURORDIS) within Europe and beyond<sup>11</sup>. The authors have noted particular examples of good practice that provide services to families with rare disorders.

### **2.6.1 Contact a Family, UK**

Contact a Family is a UK-wide charity providing support, advice and information for families with disabled children. The charity provides information and support specifically to families experiencing rare disorders. Services include:

- A bi-monthly e-newsletter specifically aimed at parent support groups, professional workers and anybody interested in policy issues concerning rare disorders.
- A helpline for rare disorders, as well as information officers across the UK including Northern Ireland.

<sup>11</sup> For a directory of national and international organisations that provide social support and information services to people with rare disorders and their families see Appendix 1.

- Services across the UK organise a wide range of training workshops, meetings and information events.
- A web-based linking service whereby families can register free to be contacted by other families across the world affected by a particular condition by e-mail.
- Families in the UK who do not have access to the internet can register to a manual linking service, whereby the charity looks for a match from others who are looking to make contact either via the Internet service or by telephone.
- The Contact a Family Directory contains a short medical description of a variety of conditions, including a comprehensive list of rare disorders, together with details of inheritance patterns and prenatal diagnosis. This information is followed by details of the related support networks, their activities, publications, and what they offer to families.

### 2.6.2 Agrenska, Sweden

The Agrenska Centre in Gothenburg, Sweden provides services to children with rare disabilities and their families. This includes a family programme where families who have children with the same rare disorder meet each other and health professionals to exchange information. Other services include respite and summer camps. A research and information function exists and conferences on the subject of rare disorders are held. In order to study the effects of interventions by the Agrenska family programme specifically, the Nordic School of Public Health and the University of Gothenburg set up a research programme in 2002 (Olauson, 2002). Feedback from participants on the Agrenska programme indicated the following positive outcomes of programmes specifically tailored to families affected by rare disorder:

- Parents feel “normal” for the first time due to peer support.
- The family feels empowered as a result of meeting others in the same situation.
- Parents get the knowledge they need to take better care of their own lives through sharing experiences with others who understand.
- Children with the disorder meet others in the same situation.
- Siblings meet other siblings.
- Professionals meet other professionals and thereby develop their own network.

(Olauson, 2002)

A similar centre based on the Agrenska model has recently been developed in Estonia. The Eesti Agrenska Rehabilitation Centre aims to be a nationally-based family programme offering individual multidisciplinary assessments, training and rehabilitation planning as well as lectures and discussions for parents on medical, psychosocial and educational aspects.

### 2.6.3 Frambu, Norway

Frambu is a national centre for rare disorders and disabilities in Norway, funded by the national Ministry of Health and Care Services. It is one of 17 centres of competence for rare diseases in Norway, which is equivalent to one rare disease centre per 300,000 people in the country. Frambu’s mission is to improve the quality of life for those with rare disorders and their families at home, at school, at work and in the community so that they can better cope with their daily lives. Frambu has a multidisciplinary staff, covering medical, social and educational aspects of life for families with a member who is diagnosed with one of the listed disorders. Patients, family members and professionals can contact Frambu for information, advice and guidance regarding designated rare disorders. As with Agrenska, family programmes take place at Frambu where families and children with rare disorders meet each other along with relevant health professionals to share experiences and exchange information. The needs

of siblings are catered for in summer camps and the extended family (grandparents) are included in specially designed family programmes. Frambu aims to:

- Enhance knowledge and competence regarding the rare diagnosis amongst the users, their families and the local professionals.
- Focus upon a life-span-perspective.
- The knowledge base is based upon the competence obtained through the direct work with people having the rare disorders, their families, and upon collaboration with other professionals with similar or other useful competence.
- Knowledge, information and guidance is systematised and disseminated to users, families and professionals at a local level including schools.
- Individual services are offered — but only as a supplementary service to what is available in the community, county or region.

(Frambu, n.d.)

Frambu has received positive feedback both from family, clients and professionals. Family clients have reported on the significance of having Frambu as a centre where families in the same situation can meet each other along with professionals. Feedback from professionals has highlighted the value of Frambu catering for needs beyond the medical — including the importance of peer support (Frambu, 2005).

## 2.7 Conclusion

Many of the themes in this literature review have been researched extensively when investigating the experience of individuals who have more common disabilities and their families. We have found that research and information specific to the psychosocial experience of individuals and their families with rare disorders in Ireland is limited. Much of the research and information gathered for the literature review that does refer to the needs of families experiencing rare disorders comes from support groups in the UK or national alliances within Europe and beyond.

The research on rare disorders often fails to make clear distinctions between the experiences that result due to the rarity of the disorder as opposed to more common disabilities. However, of the research relating to rare disorders that has been carried out in Europe and further afield, it is positive that it is based on the direct experiences of individuals and families who have been consulted. Models of best practice do exist with regard to meeting the needs of families affected by rare disorders, where evaluation and research has concluded in stating the positive effects of these types of services for all members of the family. This literature review has demonstrated the significant degree of lobbying activity relating to rare disorders policy in Ireland, the UK and European member countries.

The lack of research relating to the unique social support and information needs of people with a rare disorder and their families in the Republic of Ireland and Northern Ireland has provided the impetus for this current study being undertaken. Using both quantitative and qualitative methods, including a direct consultative approach, this research aims to provide some insight into the social support needs of families affected by rare disorders, the information needs of families and health professionals on rare disorders and, where appropriate, to make recommendations for future support.

## Chapter 3: Design of the research study

### 3.1 Introduction

The aims of the study were as follows:

- To ascertain the information needs of individuals with rare disorders, their families and health professionals with regard to rare disorders.
- To ascertain the social support needs of individuals with rare disorders and their families, including siblings.
- Where appropriate, to make recommendations for future social support for individuals and families who are affected by rare disorders.

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### 3.2 Scope of the study

#### 3.2.1 Geographic area

Due to the relatively low numbers of people experiencing rare disorders it was decided to take an all-island approach to the research. Therefore sampling took place in both the Republic of Ireland and Northern Ireland.

#### 3.2.2 Inclusion criteria

- Individuals with one of the particular rare disorders selected for the research (see below)
- parents (mother and/or father) of individuals with one of the rare disorders selected for the research
- general practitioners [GPs]
- specialist health professionals with a professional remit/interest in rare disorders
- the geographical area for inclusion was the Republic of Ireland and Northern Ireland.

Families who experienced the following rare disorders were selected to be included in the research<sup>12</sup>:

- Cornelia de Lange
- Cri du Chat
- Fragile X
- Neurofibromatosis
- Prader Willi Syndrome
- Rett Syndrome
- Sotos Syndrome
- Williams Syndrome
- Other very rare congenital disorders (less than 5 cases in Ireland) at time of study

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<sup>12</sup> Details on the criteria and selection process for the chosen rare disorders are detailed in Appendix 2

### 3.3 Data collection/methodology

#### 3.3.1 Overview

The research incorporated a mixed-method approach, using both qualitative and quantitative methods. Individuals with rare disorders, their families, specialist health professionals and general practitioners were consulted about information and social support needs relating to rare disorders.

Data collection took place between December 2007 and March 2008, with the exception of one interview with a specialist health professional which took place in early 2007. This interview was of a more informal structure, taking place after a meeting with RehabCare representatives about this research. The views of this respondent are included in the analysis.

Prior to data collection, during late 2006 and during 2007, contacts were made with the relevant support groups and parent contacts for rare disorders in order to inform them about the research. Other organisations with a remit for disability in the Republic of Ireland and Northern Ireland were informed about the research during this time as well as key health professionals in the field of rare disorders.

#### 3.3.2 Consultation with families

##### 3.3.2.1 Rationale

The format for interview with families was open, with one or both parents attending the interview. The child or adult with a rare disorder was also invited. The researchers were keen to hear the views of all family members hence an informal approach was taken with regard to those present at the interview. Initially the researchers sought to hold focus groups with siblings of people with rare disorders as part of the research. Following initial access difficulties due to the sensitive nature of the topic the researcher sought instead to elicit data about the experiences of siblings of people with rare disorders from parents in this study.

A qualitative method was chosen in order to ensure an in-depth exploration of the issues. The interview schedule was designed based on the themes on social support for families experiencing rare disorders that arose in the literature review:

- information needs: type of information and information from health professionals
- practical social support needs: financial and access to services/support
- social and emotional needs: including family relationships and peer support

For the full interview schedule see Appendix 7. This interview schedule was adapted if the individual with the rare disorder was being interviewed alone. If the individual had a learning disability similar themes were covered in a broader scope.

##### 3.3.2.2 Sampling

The sample of families was selected from volunteer participants recruited by advertising the project within support organisations representing people with specific rare disorders within the island of Ireland. The gatekeeper in cases where no specific support organisation existed was a disability organisation with a general disability remit or a third party professional who works in the field of rare disorders. This assisted in reaching the more isolated cases. A press release (see Appendix 3) was issued in the Republic of Ireland and Northern Ireland that outlined the nature and aims of the research, including contact details for the researcher. This yielded further participants who were interested in taking part in the research.

##### 3.3.2.3 Planning

After recruitment and selection, an initial phone call was made to either the individual with a rare disorder or the parent in each participating family in order to provide further details of the potential nature of the involvement of their family.



A pack containing a letter was sent to all families who volunteered to take part. The pack contained detailed information about the purpose of the research (see Appendix 4), an information leaflet about the research (see Appendix 11) and an outline of the research process. For people with a learning disability, a separate information leaflet that was in a user-friendly, accessible format was enclosed (see Appendix 12).

#### **3.3.2.4 Participants**

Semi-structured interviews took place with 23 families.

- In nine cases a mother was consulted alone.
- In three cases a father was consulted alone.
- In four cases a mother and father were consulted together.
- In two cases an individual with a rare disorder was consulted alone.
- In five cases a mother and individual with a rare disorder were consulted together.

### **3.3.3 Consultation with specialist health professionals**

#### **3.3.3.1 Rationale**

Specialist health professionals were selected on the basis of their professional interest and/or work in the rare disorders field in the Republic of Ireland and Northern Ireland. A survey with paediatricians about the information needs of professionals and families and the social support needs of families experiencing rare disorders was carried out previously in the Republic of Ireland (RehabCare, 2002). This survey rendered comprehensive findings outlining the views of this profession about rare disorders. It was therefore decided that although paediatricians would not be excluded from the sample, the emphasis would be on other specialist health professionals for this piece of research. Geneticists were the main profession targeted, because of their role in diagnosing rare disorders and follow-up medical support. Due to the fact that rare disorders are, by definition, uncommon it was noted that only a small number of specialist health professionals would be equipped to comment on the topic. A qualitative method was chosen in order to ensure an in-depth exploration of the issues. The interview schedule (see Appendix 8) was designed based on the information arising in the literature review and included the following themes:

- information needs of families and health professionals
- practical support needs of families
- social and emotional support needs of families

#### **3.3.3.2 Sampling and planning**

The researchers identified a number of specialist health professionals who had a professional interest in the field of rare disorders, and who would have been in contact with families at diagnosis and/or follow up meetings. The selected specialist health professionals were contacted directly by the researcher to request involvement in one-to-one semi-structured interviews. Those who agreed to take part received a letter outlining further details of the research. This was adapted from the family letter, an information leaflet about the research and a consent form adapted from the family consent form. The letter also included an outline of the nature of their involvement.

#### **3.3.3.3 Participants**

Five specialist health professionals took part in semi-structured one-to-one interviews.

### 3.3.4 Survey of GPs

#### 3.3.4.1 Rationale

General practitioners have a role in the primary care of a patient. Because of the nature of their work they often interact frequently with rare disorder patients (Van Nispen *et al*, 2002) and traditionally have contact with the whole family. It was decided that a quantitative survey approach would be incorporated for this phase of the research. This method was chosen in order to obtain opinions from a large sample of GPs within a specified time period and in a cost-effective way. Questionnaires were designed based on the information arising in the literature review. The questionnaire included questions about the information needs of health professionals, and GPs' perception of the information and social support needs of families in regards to rare disorders.

#### 3.3.4.2 Sampling

It was decided that 1,000 of the total combined number of GPs in the Republic of Ireland and Northern Ireland combined would be selected to receive a postal questionnaire (see Appendix 9 for the cover letter and Appendix 10 for the questionnaire). Of these, 300 were selected for the Northern Ireland sample and 700 were selected for the Republic of Ireland sample. A systematic probability sampling method was applied using two alphabetical lists of GPs. An alphabetical list of GPs practicing in the Republic of Ireland was sourced from the Irish Medical Directory (2007-2008) where every fourth name was selected out of a total sampling frame of approximately 2,400 GPs. An alphabetical list of GPs practicing in Northern Ireland was sourced from the Central Services Agency (2007), where every fourth name was selected out of a total sampling frame of 1,111 GPs.

#### 3.3.4.3 Pilot

Prior to the full postal survey, a pilot study took place with six GPs. The questionnaire and cover letter were consequently revised as a result of the feedback gained from this pilot. The main issue was the time pressures experienced by GPs in regards to completing a questionnaire. The questionnaire was therefore shortened and the cover letter was made more concise while still containing detailed information about the research and its purpose. Contact details for the researcher were on the letter, enabling participating GPs to raise any queries or concerns.

The revised and final 'survey packet' included a stamped addressed envelope addressed to RehabCare, an information leaflet about the research (see Appendix 11), a cover letter (see Appendix 9) and the questionnaire (see Appendix 10).

#### 3.3.4.4 Planning

A press release was issued by Rehab Group Communications & Public Affairs Team to various medical and health-related newsletters across the Republic of Ireland and Northern Ireland one week prior to the administration of the survey. The reason for this was to publicise this phase of the research within the health sector, with the aim of gaining a higher response rate. This included the *Irish Times* Health Supplement, *Irish Examiner* Feelgood, *Irish Medical Times*, *Irish Independent* Health and Living, *Irish Medical News* and *Medicine Weekly*. See Appendix 3 for press release.

A two-phase approach was applied to the administration of the questionnaires. Initially 600 questionnaires were posted (400 in the Republic of Ireland and 200 in Northern Ireland) in January 2008. Due to the positive response rate the second phase began in February 2008, when the remaining sample of 400 questionnaires (300 in the Republic of Ireland and 100 in Northern Ireland) was sent out. The total of the two phases matched the initial sampling target of 1,000 GPs.

#### 3.3.4.5 Respondents

232 GPs (23.2 per cent response rate) responded to the postal survey. Of these 177 were from the Republic of Ireland (25.3 per cent response rate) and 55 were from Northern Ireland (18.3 per cent response rate).

Table 1. Number of GP respondents by catchment area of practice in the Republic of Ireland and Northern Ireland.

	Republic of Ireland		Northern Ireland		Total	
	Number of respondents	%	Number of respondents	%	Number of respondents	%
Urban	60	33.9	12	21.8	72	31
Rural	36	20.3	18	32.8	54	23.3
Mixed urban rural	80	45.2	24	43.6	104	44.8
Unknown	1	0.6	1	1.8	2	0.9
Total	177	100	55	100	232	100

### 3.4 Data analysis

#### 3.4.1 Data analysis and family/specialist health professional consultations

Each interview with families and specialist health professionals was audio-recorded and transcribed. The transcriptions were coded and thematically analysed. A set of quotations from all interviews was chosen to represent the themes discussed.

The views of families experiencing the different rare disorders and the views of all family members who were consulted (i.e. the individual with a rare disorder, father and mother) were combined to be thematically analysed. The distinction was made in the findings between an individual with a rare disorder being quoted or a parent (mother and/or father).

#### 3.4.2 Data analysis and GP survey

Responses from the GP questionnaire were coded, entered into a dataset and analysed using SPSS for Windows v15.

### 3.5 Ethical considerations<sup>13</sup>

Ethical considerations were of the utmost importance due to the sensitive nature of the topic, particularly for families involved in the research.

#### 3.5.1 Informed consent

All potential participants were notified by letter about the purpose of the research and that the results of the research would be published as a report. Potential participants were made aware of the contact details for the researcher and were encouraged to make contact if they had any queries or concerns

<sup>13</sup> Ethical approval was received from the Department of Paediatrics, Faculty of Health Sciences, Trinity College Dublin.

prior to the consultation. Participants were told of the nature of their involvement, including that their participation was voluntary and they had the right to withdraw at any time with no negative consequences. Participants were informed that no names would be attached to anything quoted in the report and the final report will be disseminated in an accessible format for all participants. Participants were told that they were consenting to the interview and to the publication of the research which would be used as an evidence base of the social support needs of families experiencing rare disorders. Participants were also informed that their interview data would be used for purposes of this current research only.

Considerable effort was made to ensure that material provided about the research was as accessible as possible. For example, both a text-based and pictorial version of the consent form was made available. In addition, all ethical considerations, including a verbal explanation of consent and the right to withdraw from the study at any time, were explained again at the commencement of one-to-one interviews. Permission for electronic recording of interviews was sought prior to consultation and participants were also informed of the steps that would be taken to protect the confidentiality of the tapes.

Consent was obtained prior to the interview taking place (see Appendix 5). For adults with learning difficulties the researcher strongly advised prior to the research that if the person requested it, an advocate could be present at the interview (an advocate chosen by the participant). A pictorial consent form was devised for prospective participants with learning difficulties (see Appendix 6).

### **3.5.2 Confidentiality**

For those participants taking part in one-to-one interviews, only designated members of the study team (researcher and line manager) were aware of the names and details of participants. The researcher clarified verbally, at the commencement of interviews, the steps taken to protect confidentiality and what responsibilities family members had in relation to the protection of each other's confidentiality. The tapes from all interviews were titled by the date and time. No participants had their full names recorded on the tapes and no identifying information was included with transcripts of one-to-one interviews.

The questionnaire for GPs was anonymous. The only personal information requested was the county of practice and whether its catchment area could best be described as rural or urban. The GP was given the option of including their name if they were interested in receiving updates on the project.

All electronic data (transcripts, and files containing personal information about participants) was, and is, subject to appropriate strict controls in the form of password-locked files. Hardcopy files holding the recorded information are held securely in fire-proof, locked cabinets kept at the Head Office of RehabCare which has limited access.

### **3.5.3 Location of fieldwork**

Arrangements for the interview were made at the convenience of the participant. In many cases the interviews with family members were carried out in the participant's homes or in a convenient café or restaurant in their locality. For their convenience the interviews with specialist health professionals were carried out in their offices. Privacy and confidentiality were major considerations when interviews took place in public venues — these were in a location of the participant's choice and in a private area away from other members of the public.

The researcher often carried out the fieldwork alone and appropriate control measures were put in place in order to ensure their safety as a lone worker.

## Chapter 4: Research findings

In this chapter, the findings from all consultations, including families who experience rare disorders, specialist health professionals and GPs, are presented. The findings are presented under the following headings:

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### Support needs

- ◇ peer support
- ◇ sibling support
- ◇ emotional support

### Information needs

- ◇ accessing appropriate and sufficient information
- ◇ diagnosis
- ◇ shocking imagery and worst case scenarios
- ◇ understanding information and medical terminology
- ◇ lack of information
- ◇ parents informing healthcare professionals

### Financial issues

- ◇ loss of income due to disability
- ◇ access/eligibility to benefits and supports

### Services

- ◇ location of services
- ◇ social worker support
- ◇ alternative therapies/services
- ◇ services that meet the needs of people with rare disorders

## 4.1 Support needs

### 4.1.1 Peer support: views of family participants

All of the participants in the study who used peer support (i.e. contact or interaction with other individuals with a rare disorder and/or their families experiencing a rare disorder) found it to be an extremely important part of coping with rare disorders. Those participants who had experienced peer support felt that it played a major role during difficult times and also recognised peers as having an important information function.

One participant spoke about his first experience with the support group for his disorder.

*"I went along initially because I wanted to meet other people and I'm really, really glad I did. I have made a couple of good friends. We are constantly in touch with one another...I had so many fears... but when I went I cried so much...when I saw the rest of them, what they were able to do."*

One participant spoke about her main reasons for attending the annual conference on her daughter's rare disorder.

*"Our little one really adores it, you know... the conference is great for the information but it is more meeting all the parents and seeing how the kids are doing and everything. You learn so much."*

One participant felt that the peer support provided them with knowledge of what to expect from the future.

*"We went when our son was only a few months old...we met other parents then and from there we started to get a kind of life experience, ideas of what it was about and we saw other children. It was eye-opening. It was tough, but it was necessary."*

Some parents felt that they learned practical information and coping mechanisms from their peers who had gone through similar situations with their children.

*"They may say something daft like 'we lay him on his side' and you think, 'we haven't tried that'. It is just simple things that you'd never think of. They will tell you something and you try it and you know, 'we don't feed them this, we feed them that'. 'Right well we'll try that'."*

One participant mentioned the importance of peer support for her son who has a rare disorder. She describes the first time her son met another girl with the same disorder as him and how much he enjoyed visiting her.

*"I mean the two of them knew they were the same so when they met each other they both knew they were similar. We would go there three times a year. I mean the minute you drove up he'd clap his hands in the back of the car because he knew where he was going. Like the two of them would give each other a hug."*

One participant spoke about her frustration at not having peer support due to the extremely rare nature of her child's disorder.

*"...if there had been a network or something, we could have possibly phoned up and said: 'Look, what the [expletive] can I expect here, because I really do not know, or where can I get help, or where can I get support?'"*

One participant, who has two children with a rare disorder, felt that the only people who fully understood her were the parents of other children with the same rare disorder. She recalled how in the early years of her children's disorders she would get angry at what she felt were the futile complaints of other parents she socialised with.

*"...their conversation is entirely different because they're talking about, 'Oh God he drove me mad because I had to bring him to the hurling fields like five times last week' and you're there saying: 'Jesus give me that, like what I wouldn't do...why are you giving out like'....and you're sitting there*

*and in three minutes you're just angry and you just want to go home because they're so feckin' selfish and they can't see it like."*

The participant felt that her main source of comfort and the only people who completely understand her are parents of children with the same rare disorder. She felt these people had similar experiences. She describes the level of understanding between peers.

*"It's just a level of understanding. It's innate. You don't have to explain...When you just meet for lunch that, you know, Coronation Street and anything else, you know we're not going to talk about the boys, we're not going to talk about how wrecked we are...and we sort of nearly give each other a look at the start and it's like you know do you need to go there or no?"*

One of the participants spoke about the support she receives from parents of children with more common disabilities and recognises that they can have common support needs and therefore are able to support one another.

*"There is a group of parents who've started in the special needs mother and toddler group together. Now most of the kids are Down syndrome but we are still all in contact ...we go out for dinner, we go away on holiday together some of us you know and that's very important even though they are different conditions. It's really... it is important because we are all in the same boat."*

Another participant recognised the common support needs she feels parents of children with rare disorders have and felt that there are practical issues which people from different rare disorder backgrounds could inform each other about. This participant recognised that this exchanging of information could prove beneficial to those with extremely rare disorders — especially those without support groups.

*"You're going to hit the same obstacles...You would have a lot in common with them... even to talk to parents and say, 'Well look, we went through this like with the long-term illness card, we had this problem, we couldn't get a medical card', this kind of thing. That helps because you know that you're not sitting at home frustrated on the phone every day looking for this, that and the other and there's other people doing it as well, so it doesn't feel so bad."*

#### **4.1.1.1 Risks associated with meeting peers**

Two of the participants recognised that there can be negative implications in some instances from peer meetings. These participants pointed out that there can be a tendency for some people to use peer support environments to voice their frustrations and complain about services.

*"Some of the families tend to load all their problems on to you and you think, 'I don't really want to know all your problems to be honest'."*

Another participant came across this kind of negativity at a peer meeting. However, he was happy that the majority of people at the meeting did not tolerate the behaviour and were positive in their approach to life.

*"There were some people there who thought that they were going to meet like-minded people who were down-at-heel and all browned off about it but in fact the people who were trying to play that particular card and just go for the, 'Oh poor me' lark, didn't really get a look in. We don't stay in touch on an 'Oh well how's your creaking bones this weather?' you know...there may be the occasional reference but you are much more positive."*

Another participant recognised that attending peer conferences and coming across worst case scenarios can deeply impact on some families. In cases where conditions can become progressively debilitating over the years, peer support environments can highlight this.

*"You were seeing girls with a condition in their teens and as adults and that was devastating. So I think a lot of people realise that they don't want to see where their children are going to be at in*

*the future because it is too much. When our children are small they look very normal...so to see what it is going to be like in 20 years time. It's devastating."*

#### **4.1.2 Peer support: views of specialist health professionals**

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One specialist health professional highlighted the commonalities between different groups regardless of rare disorder being experienced. Although she felt that people tend to look to their peers with the same rare disorder for emotional support or details of the future, she stated that for practical information on such things as services and financial assistance, there is a lot to be gained from rare disorder support groups joining forces.

*"...it seems a huge source of frustration to my families that they kind of campaign to get classroom assistants or what happens when the wheelchair breaks down. There's a lot of that goes on where families feel they are battering their heads against brick walls. They have been turned down for financial aid for the hundredth time when it's obvious their child needs it. In terms of making stuff happen, I think there is quite a big role that groups could actually work together for the sort of advocacy."*

Another specialist health professional felt that the support of peers is very important in combating the feelings of isolation in families affected by rare disorders.

*"They don't feel so alone. You know that there is somebody going through the same thing that they are going through."*

One of the specialist health professionals felt that support groups and peer support play a vital role in helping families cope with their child's disorder.

*"Essentially you are talking about a human need for people to communicate about their tragedy with somebody else who hopefully has a humane and kind approach, having been through it themselves. And I don't think that we should object to any format that helps people to process what is essentially a grieving situation. After all, they are being told in my clinic that their child isn't normal... We must facilitate them to process their grief in the way that they find greatest comfort from and some of them will find it from speaking very directly to me and others will find it from talking to the mother of a child in Nevada who has a website."*

This specialist health professional also felt that an umbrella organisation representing numerous support groups for rare disorders could provide an even stronger base to support people with rare disorders and allow groups to advocate more powerfully as a joint force.

*"I think it would not be a bad idea if support networks are being established in Ireland for them to be linked to a wider organisation such as Contact a Family because there's a certain quality control of the data and information. One feels that the information being given out is accurate and sure and nothing too untoward or too unorthodox is going on. Orthodoxy has a certain reassuring value in these situations."*

#### **4.1.3 Sibling support: views of family participants**

##### **4.1.3.1 Lack of attention**

The majority of parents felt that the reality of having a child with greater needs results in an uneven balance of time and attention, with the lesser of both going to the children who do not have a rare disorder. One couple went through constant sleep deprivation during the early years of the life of their child with a rare disorder. They struggled to cope and felt that they were almost always too tired to spend adequate time with their other daughter.

*"She got a bit lost, especially the first three years... Most of our sleep would be with [daughter with rare disorder]... you would lie down beside her and she would sleep for maybe 2½ hours max... we*



*were going around like zombies and our other daughter lost out, I know she lost out. She gets on with and loves her sister but she would have felt very left out for a few years."*

Another participant recognised that the siblings of a child with a disability inevitably feel left out as they receive less attention.

*"From a child's point of view, the equilibrium of the family will be disrupted. If it's another child who needs a little bit more care and attention... a child is by nature self-centred and self-focused anyway so anybody needing more care and attention would probably put them out of kilter anyway."*

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One participant felt that having a child with a disability means that the family unit cannot perform normally and that there is no way of avoiding the lack of attention that other siblings receive.

*"Automatically you're dealing with a dysfunctional family. It's a simple hard fact of life, once there's disability in the family, the person with the disability gets the attention, and rightly so. But the other children do suffer and they always will... I'm very conscious of that and we try to give them the best we can and then you end up overcompensating for it and that's not right either."*

Another participant felt that the presence of a significant physical disability can be even more time-consuming for parents. This leaves parents with little family time for the other siblings.

*"...in a lot of cases, families are housebound, they miss out on a lot of opportunities and a lot of normal outings and there is a lot of focus, a lot of time and attention that goes into the physical management of the child with the disability, and generally speaking you find that children don't complain about it but it does impinge on their quality of life so siblings do definitely suffer considerably in those circumstances."*

#### **4.1.3.2 Feelings of resentment**

Some parents recalled feelings of resentment among siblings due to the extra time and attention required by the child with the rare disorder. One participant recalled the relationship between two of her sons when they were young:

*"He resented his brother when he was young when my husband was alive. He used to think that we were giving [brother] more attention than him."*

Another participant felt that even though her children were very understanding of their brother's needs and demands, they were still affected by the presence of a rare disorder in the family.

*"You know, you do hear, 'You always give him more time than me'."*

#### **4.1.3.3 Altered behaviour**

Some of the participants spoke about the increased speed with which some siblings are forced to mature. One participant felt that this could often become apparent in the way some siblings become more caring and patient individuals as a result of their circumstances.

*"It's a false maturity and it's a false well-being. In my case I think they are genuinely stronger and it's a good thing. They have learnt to be more responsible and they have learnt to cope with things and they have learnt to be more sensitive and things like that."*

One participant spoke about the overprotective nature some siblings adopt and highlighted that as they recognise the difficulties their parents experience with their sibling children often modify their own behaviour.

*"A lot of siblings are very overprotective...you get this kind of pseudo maturity thing with quite young kids, where they are almost overly responsible and they take on a lot of things that kids their age don't normally take on. They are better behaved than one would expect for a child of that age or I suppose there is a kind of a mentality with some of these kids where you see them as almost being too good because they are aware of the burden on the family as a unit."*

#### 4.1.3.4 Facilitation of sibling support: needs and benefits

Many of the participants expressed a desire for a sibling support network or workshop stating that they often did not have the time to broach the concerns and issues they needed to, due to the high demands of their child with a rare disorder.

*"I mean, parents won't, I mean, you don't exclude any of your kids, but if one kid needs more, you automatically give it to them and by doing that, whether you like it or not, you're giving less to the other...if there was some sort of club that maybe two or three times a year...a workshop like that would be really special for them...it would be something totally different and they'd feel special. I think they desperately need that."*

One participant spoke of the concerns her 16-year-old son had about the future care of his brother with a rare disorder and how he worried about the burden of care if his widowed mother passed away. She felt that a workshop or counselling support could help him deal with any issues he has.

*"Like he worries and he won't talk to me so...I don't know if there was something there...maybe a workshop for him to address this....anything to help him cope you know."*

Parents of children who have received some peer support or workshop support spoke of the important benefits of such support for their siblings. One participant had brought her whole family to a peer group meeting in the UK and she found that not only was it beneficial to her and her child in terms of information but that the siblings really gained from it.

*"They found it hugely beneficial...because that was their first opportunity to meet with the siblings... The families just really met first of all informally, where you just sit around and have a cup of tea and have lunch together. There were bouncy castles, there was football for the kids...the older kids then could do face painting or making jewellery. You know there were activities for every age group. My daughter is still in email contact with one of the girls she met there."*

Another participant agreed that siblings can benefit hugely from meeting peers from a similar background. He brings his whole family along to the annual conferences for the rare disorder his son has and finds that it helps them greatly.

*"My kids come along to all the conferences or parties or whatever, so they see the other people and I think that's important at this stage. When he goes now, I see him going off with the other guys around his age and the other girls and I know they're talking about it and it's great. 'Off you go guys and have your chat because they're dealing with things in their own way'... I think that's crucial for them to be honest. I think it's vital."*

#### 4.1.4 Sibling support: views of specialist health professionals

A specialist health professional felt that siblings are often overlooked and inevitably affected in their everyday lives as the child with the rare disorder must take priority as they generally have high needs.

*"They have a really high rate of bereavement because there's a higher rate of illness, death and early death in some of the rare disorders. I think it's probably quite hard — especially when you're going through your teenage thing where you think the whole world should revolve around you and it actually never ever does — not for one second are you first, you're always second to the illness which happens to attach to your brother and sister. I don't think siblings hate their brothers and sisters but it's a different way of being brought up."*

This specialist health professional also felt that medical staff can often neglect to speak to the siblings who inevitably are often present in consulting rooms when parents come in to discuss the rare disorder.

*"Even when it comes to the clinic room you know you have to make sure you actually do speak to them because I think quite often nobody speaks to them. 'Hi Mum, Hi Dad' and there's this little child or whatever and then there's somebody else sat there who has to be there because Mum and Dad couldn't leave them at home. You know, they're like the appendage."*

Another specialist health professional felt that teachers in schools have a role to play as siblings need support that they cannot always get at home due to the extra demands on their parents.

*“I really think siblings of handicapped kids get a rough deal. I think very little is done for them, I think schools are not helpful and I do think they [the siblings] have a role as sort of semi-unpaid carers because they sort of have to be because they’re what props the system up. They wouldn’t be able to do other stuff that kids can do like swimming lessons or football or whatever due to the time constraints on their parents.”*

The specialist health professional referred to a support organisation for a rare disorder which is very inclusive of siblings in their conferences and seminars, feeling that this inclusive style helps siblings immensely.

*“They get some of the siblings to speak, sometimes about the disease and sometimes about being the sibling of somebody with the disease and...I think it’s quite nice. It gives the sibling a certain amount of recognition in the rare disease world because whether you like it or not I think as a sibling you’re part of that world, that’s it. It’s a club you probably didn’t have choice to belong to but you’re there. This wasn’t ever how you thought you might spend your life as a 13-year-old...It’s quite nice that they do get their views on things.”*

#### 4.1.5 Emotional support: views of family participants

Many of the participants spoke about the strain that having a child with a rare disorder can place on your relationship. One participant said she felt her husband initially refused to accept that there was anything wrong with their son and described how this impacted on their relationship.

*“It was actually difficult on the two of us, because I was kind of saying, ‘No there’s something wrong’, and I used to go to all the appointments and I’d be coming home and I’d be telling him and he’d be kind of looking at me as if I was mad... We were both kind of, not arguing but trying to find...I was kind of hoping [he] would take more, not interest, but get more information into his head, so he knew what was happening because I hadn’t a clue.”*

Another participant described how she and her husband would end up blaming each other and even their families due to the frustration.

*“You know, and I’d be saying, ‘oh your mother doesn’t understand’ and he’d say and ‘your mother doesn’t’, you know ...but it was hard that because we didn’t know what to do for [their son] and how to help him. So I think that’s the thing, if you know and have support there to let you know what you’ve to do, but when you’ve to try and find it out the hard way it is tough.”*

One participant who also runs a support group recognises the importance and need for counselling for the entire family in situations where rare disorders are present.

*“In any disorder I would believe that patients require counselling. It’s like being struck by lightning; it destroys your whole world. Generally people become emotional. I get it here all the time — particularly from women ringing up here, they’re crying, they’re upset, they can’t handle the situation. They say ‘I could take this for myself but I can’t take it for my child’.”*

One couple (participants A and B) who have two children with rare disorders, felt that there is a widespread tendency for parents to forget about their own needs and that of their partner because the needs of the child are so great. They felt that a partner can fall by the wayside very easily and that there is a danger that the parents of children with rare disorders can become completely enveloped and absorbed in the lives of their children.

Participant A felt that such parents can easily lose their partner to a large degree as they cannot spend as much time with them or on their relationship.

*“Definitely, your partner is going to fall by the wayside in an awful lot of ways and you know you really have to kind of make yourself remember to spend some time with the other people in your*

*life because you just get swallowed...you lose yourself completely. You have no time for 'what do I want from my life?' You know, that's just a complete luxury really...there are so many daily challenges...you can't really switch off with it and go out and have a meal or anything because you will end up talking about them."*

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Participant B felt that counselling and peer support would be a great help for everyone and is disappointed that because of the rarity of the disorder from which his child suffers there was no support service for him to avail of.

*"It would be great for males to be able to talk to fathers who are in the same situation or whatever because I would say a lot of the parents must do a runner. I would say a lot of fathers can't cope and just leg it...it would be really helpful to have somebody who can give you suggestions...another human being that has been through it."*

#### **4.1.6 Emotional support: views of specialist health professionals**

One specialist health professional who participated in the consultation recognised that the absence of a counselling service is a huge drawback for families and places pressure on relationships. Due to the genetic nature of many rare disorders he felt that the whole family can be affected whether as carriers of the defective gene, through having the disorder themselves or having a family member with the disorder.

*"So at diagnosis time the first thing I would say is the entire family would need to be seen and secondly, I think even the extended family because it may go back to the grandparents and there may be other members of the family affected."*

The specialist health professional sees that there is often a blame game attached to genetic disorders which can create a strain within families. Parents often struggle to accept that the disorder their child has been diagnosed with is as a result of their own genes and this specialist health professional finds that often parents blame themselves and become consumed by guilt.

*"One thing I think is an added strain for genetics is the blame game. We, one of us, both of us, us together made this happen and no matter how much support or if you try and say look you couldn't choose this, you didn't make this happen, you inherited your genes, you pass them on."*

The specialist health professional also often finds that fathers cannot accept a diagnosis as easily as a mother can. This tendency can manifest itself in fathers not attending appointments with their child and also refusing to accept a diagnosis altogether.

*"...sometimes couples come to the clinic and I'm suddenly realising actually Dad has no idea quite how handicapped their child is and they're going 'oh so he's never going to get better?' I'm thinking: 'How do you not know this? Of course he's not going to get better...'"*

Another specialist health professional felt that involvement of older generations in the blame game often places more stress on families.

*"We can tell the parents it's not your fault but behind the scenes the grandparents are the ones saying 'well it's not on my side'...you know that the grannies are doing the chattering and that, I think, places tension in the wider family circle — the jolly christening with both sides blaming the other side. It's very stressful for them."*

This specialist health professional felt that counselling is essential for families and that, although health services do their best to follow up with families, a lack of resources can hinder them from helping everyone. She felt that the genetics team are the best people to support families with rare disorders as other healthcare professionals often do not know enough about genetics to be of practical value.

*“We would try and phone their GP. I think we probably would like to do a little more with them... it is not as much as we would like but unless I can learn how to clone our counsellors it’s what we are stuck with but I think they do need a huge amount of support and I think, other healthcare professionals actually are not as good as they should be and that is because they have absolutely no knowledge of genetics.”*

One specialist health professional felt that there is a large strain put on a relationship after diagnosis and finds that fathers often take a step back in order to cope.

*“Whatever strains existed between the parents previously are now becoming absolutely irreconcilable in light of a diagnosis of a child who is never going to be normal. Usually, though not always, this takes the form of a mother becoming very fixated upon the child’s needs; the father feeling that he is no longer part of the mother’s time and less important in her life. It is not always the mother. I have seen fathers keep the child. But it is more commonly the other way around. I think these are important issues and I actually externalise them and verbalise them quite frequently in my clinic myself.”*

Another specialist health professional also felt that there is a chronic lack of counselling services for families with rare disorders and that there is a pressing need for counselling in most cases post-diagnosis. This specialist health professional offers some guidance where he can while realising this is not entirely his role.

*“I don’t see my clinic as a psychotherapeutic environment but I will say to families: ‘Look, what you have heard today does change your lives forever. It is very important that you rationalise this and process this yourselves within your own relationship. The balance of the life you thought you were embarking upon has changed’...What I am hoping I am doing there is taking away their fear about saying to one another afterwards: ‘Will we be alright...? If I throw the ball in for them to have the dialogue that needs to be had between them and sometimes maybe no dialogue is necessary and in other times I’m afraid an enormous amount of dialogue isn’t even enough to save the day.”*

#### **4.1.7 Emotional support: views of GPs**

GPs were asked if they felt having a rare disorder gave rise to additional family problems. More than two-thirds of those who responded to this question — 164 (72.2 per cent) — felt that having a rare disorder did give rise to additional family problems with 60 (26.4 per cent) feeling that having a rare disorder did not. Three (1.3 per cent) of the GPs stated they did not know. There were five GPs who did not respond to this question. When asked what problems GPs felt that having a child with a rare disorder gives rise to, it is interesting to note from Table 2 below that 46 (28.0 per cent) of responses referred to isolation and lack of peer support and 33 (20.1 per cent) of responses referred to frustration due to lack of services and information.

Table 2. If you agree that having a rare disorder gives rise to additional family problems what problems do you feel it gives rise to?

	Number of Responses	Percentage of responses
Isolation/lack of peer support	46	28.0
Frustration due to the lack of services/ information	33	20.1
Fear/anxiety	23	14.0
Stress	17	10.4
Conflict	14	8.5
Lack of understanding among friends/family	12	7.3
Self-esteem issues	6	3.6
Relationship problems	4	2.4
Drug/alcohol problems	2	1.2
Problems with school/work	1	0.6

## 4.2 Information needs

### 4.2.1 Information needs: views of family participants

#### 4.2.1.1 Accessing appropriate and sufficient information

All of the participants spoke about their difficulties accessing information about rare disorders. Many found that the information was inaccessible, inappropriate or in a format they could not understand. All of the family participants had either used the internet to access information on the relevant rare disorder or had a friend get them information from the internet.

#### 4.2.1.2 Diagnosis

Some participants felt that there was a lack of understanding shown by medical professionals when providing a diagnosis and that the lack of support and information at this stage proved especially traumatic.

*"...it would be so much more helpful to be handed some kind of a booklet or something a little bit more human or, you know, just for her to have more kind of consideration of the impact it would have. As I say, she didn't have the research or the training."*

One participant recalled how after receiving the diagnosis of her child's condition there was no support or anyone to talk to.

*"...so when they went and they wrote up their little handwritten diagnosis, when they called us back in, I would imagine we were only in there 15 minutes max. So we had so many questions... afterwards, but we had nobody to ask them to because as far as they were concerned, they had finished with us at that stage."*

Many of the participants referred to the information that they received at diagnosis stage as being insufficient and reported on how they found it difficult to cope with this lack of knowledge.

*"You are totally thrown in at the deep end when you get a diagnosis... There was nothing for us. We were handed a leaflet that the paediatrician had downloaded from the internet herself which was one page printed out from a site... it was unbelievable. And you're in shock and you're walking around with this one piece of paper that is your child's future and you are just completely at a loss."*

One participant stated that there is an inherent need for sufficient information at the point of diagnosis in order for the parents to cope.

*"Information is a huge, huge thing, not to be left completely stranded with a child that's got this really rare disorder that you know nothing about."*

Many of the participants referred to being given worst case scenario information at the point of diagnosis. One participant talked about how she was told the worst case scenario for her child and that this prediction for her child's future was false.

*"...she had a very severe scoliosis. They called her a banana girl because she was literally like that... we were told she would never walk...She would have feeding problems, she would have behavioural problems, that she wouldn't be able to stop herself falling over, that she would need to be in a full body splint and basically it was horrific and untrue."*

#### 4.2.1.3 Shocking imagery and worst case scenarios

One couple recalled how they were horrified by the imagery on some information they received on their child's disorder.

*"She gave us a pamphlet...I still have it upstairs ...the only words I could choose quite frankly is [expletive] awful picture of a child on the front. The [information provider], God love them, at the time, had done it but it was this awful document."*

One participant recalled how he attempted to hide the shocking imagery from his wife.

*"The doctor ...produced a medical textbook that day and there were things jumping off the page, like the mobility, the obesity...jumping out...dreadful. I hadn't got internet access at the time. My sister-in-law downloaded pages and they were worst case scenarios and I literally censored what I was going to let her see."*

One participant recalled the 'awful' imagery of her child's disorder that she sourced on a support group website and how it affected her.

*"Sure even when we went onto the [support group] website when [name of daughter] was born it was oh my God, I mean I said to [name of husband]: 'Please don't show me any of those pictures I don't want to see them.' They were so out of date and really for a first-time mother it's awful. You know some of the pictures of the children were just horrendous."*

#### 4.2.1.4 Understanding information and medical terminology

Many of the participants felt that medical information they received about the rare disorder tended to concern itself with genetics and that this was often presented in an unsuitable format for families.

One participant felt that her science background lent itself to her understanding of the medical information she received on her child's disorder but she had great difficulty relaying this information to other family members who had no science training.

*"I found it very, very difficult to try to translate that into layman's English to explain to my mother and father-in-law. I understood it was a life sentence that was going to be with us until the day we die. Straight away I understood it meant no grandchildren, it meant the consequence of a future pregnancy all of that but no one else could."*

Another participant recalled how she and her husband could not understand the genetic information they received on their child's disorder. This information was found on the internet.

*"It was all the chromosomes and x and y's and it just went over our heads because we didn't understand ...full mutation, half mutation...so we thought he was going to turn into an alien or something. I would advise people not to go on the internet after being diagnosed because it's very scary... you would actually look at your child and wait for two heads to pop, that's how bad it is. It's all doctors' term as well. And there's very long words which have no meaning... I don't know how the next person would take it but it had no meaning to us whatsoever."*

#### 4.2.1.5 Lack of information

The majority of participants used the internet as a source of information immediately after diagnosis because of the lack of information provided to them at this stage.

One participant recalls how after eventually receiving a diagnosis she was then shocked by the lack of information available.

*"To have a diagnosis initially it was a relief and then, they didn't have much information. She printed everything she had and gave it to us, but it was very medical I had to get a nurse to decipher it."*

One participant's child had an extremely rare disorder and she found that the information was simply not available on the disorder on the internet.

*"There is very limited information. You can type in [the disorder name] on the internet and you can get some information, it's very limited. Everything is based on the medical model. There is nothing around, you know, quality of life for the child."*

Many of the participants referred to the difference in the level of support you receive with a rare disorder when compared with more common disabilities.

One participant felt that if her daughter had suffered from Down Syndrome she would have been supported much more.

*"For instance, people with Down Syndrome, when your baby's born in hospital you've immediately got somebody in from Down Syndrome Ireland, straight away and immediately you get like bumper packs of, you know, whatever and whereas with us there was nothing nobody."*

#### 4.2.1.6 Parents informing healthcare professionals

The majority of the participants had on at least one occasion found themselves having to inform a health professional about their child's disorder as they would have had little, if any, information about it. Some parent support groups had compiled information packs on each disorder for parents to take with them on health visits. One participant who runs a support group has gone to great efforts to ensure that health professionals have information on the rare disorder he is concerned with.

*"If you were to visit any doctor anywhere and ask them for a leaflet on [the name of the disorder] they would not have as much as what you would write on the back of a stamp. The only information that has been provided about the condition comes from the [support group]. We supply it, we get it printed. We get doctors to write up information on our behalf and we circulate it. And we have*



*circulated information to every doctor and public health nurse in Ireland but unfortunately, I would say 90 per cent of it saw the bin because they get thousands of leaflets."*

Another participant felt that one health professional was taking the wrong approach in dealing with his son. He offered advice and information he had received from a support group to the health professional so that he could adequately treat his son.

*"I just kind of went, 'listen, I'm just going to have to tell you a few things where you're going to have to change your approach here.' Very simple things, so obvious and you know, I brought him in and he was very sceptical as well of me telling him this. So I had a load of leaflets at home from the [English support group] and I was able to highlight stuff from them and drop them into him the next week and just hints on how to engage with our son without being kind of confrontational."*

Another participant echoed this idea of educating professionals about their child's disorder.

*"We were the ones trying to educate the medics and the nurses and the teachers and everybody else because nobody knew anything about it."*

Every time one of the participants goes into hospital she takes an information pack on her child's disorder with her. She feels that a basic website with information for medical professionals would be very useful.

*"We spent a lot of time educating staff when she is in hospital and educating GPs when you see them and most of them are open to that. I don't think they have time to attend training and they don't have the time to read much information...If there was a basic pack that they could get ... 'this is [name of disorder], this is what it means, these are contacts for information, this is the basic website' whatever."*

Two participants felt that there needs to be a filtering process at which parents are given information, as it can be a lot to take in at one time.

*"During the process of first accepting the diagnosis there is so much that you don't need to know ... There almost should be a chronology to it, how the information is presented to you, so that you can kind of grow with it...because if you are just bombarded it's very overwhelming and it takes you a long time to get your head around it."*

#### **4.2.2 Information needs: views of specialist health professionals**

All the specialist health professionals who participated in the research felt that there is a very large need among their patients for information on rare disorders.

One of the specialist health professionals felt that people will do anything to get information on the disorder they are concerned with. This participant felt that without direction on where to look for information people can end up accessing inappropriate information.

*"I think people are desperate for information, absolutely desperate for it. In most cases... if you don't give it to them they will go and hunt it out or try to hunt it out and sometimes what they get is not suitable so I think there is a huge need for patient-friendly information. I think there are some websites where somebody has brought their own personal burden of the disease to the website and has decided to share it with the entire world. Those websites I think aren't helpful but a lot of them are very good. I think what people need is a gateway into it."*

One specialist health professional felt that GPs and other healthcare professionals need to be aware of how to access information in order to support families affected by rare disorders.

*"Parents say that they saw my GP and he said 'oh I don't know anything about that etc' but I know people who are dustbin men who manage to Google their kid's disease and find out stuff about it and I think we're in so much of an information-easy society not only do we need to have information for families but I think we need to educate health professionals about where they can*

get information. It's just not good enough any more to say 'oh I have never heard of it, no, no, no idea' and we hear that a lot that what's happened."

This specialist health professional felt that the information available to people can often be biased and inappropriate.

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One specialist health professional stated that genetic information can be very complicated and that support is needed to translate this into an appropriate format for families.

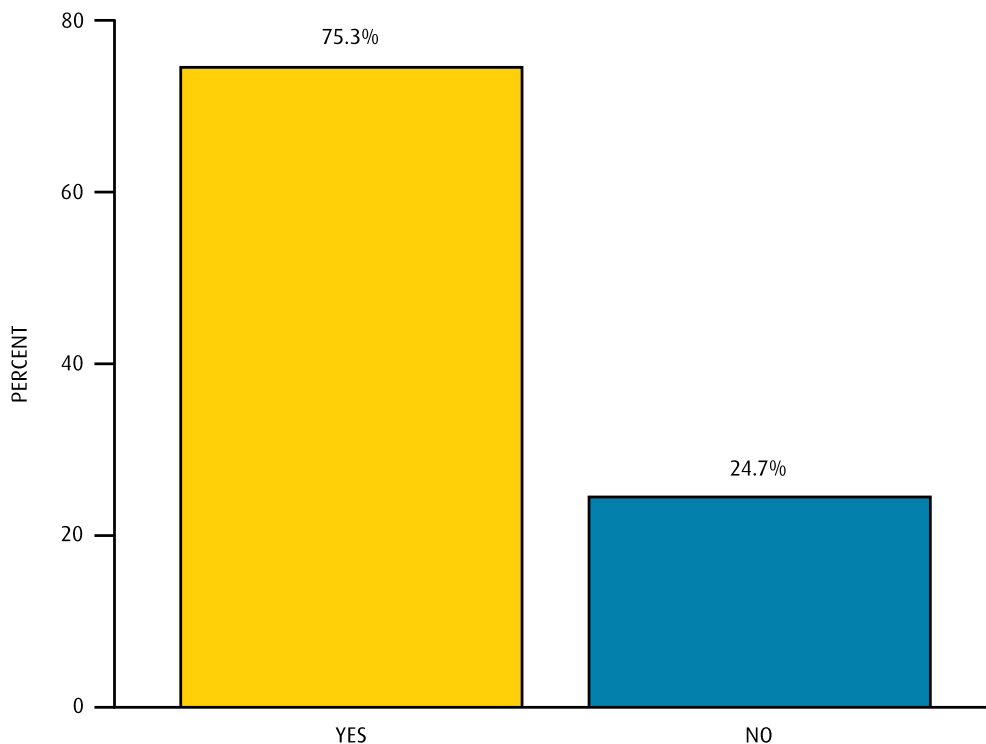
*"Obviously the genetics of these situations can be very complicated and difficult for ordinary people, even educated people to understand. It's a bit more easy for me because I am thinking about these things every day. People to whom the concepts are entirely new can struggle with them, very understandably. Now less educated people, sometimes we have to accept that a sophisticated understanding of the genetic situation is perhaps asking too much and in those situations we try and simplify things. Explain it in a way which they might be able to understand it and try and ensure that they have access to support systems such as exists."*

### 4.2.3 Information needs: views of GPs

#### 4.2.3.1 The need for information

The findings of the GP survey highlighted that primary care professionals also acknowledge a need for information to assist professionals in supporting families with rare disorders. As can be seen from Figure 1 below, 174 (75.3 per cent) of GPs who responded felt that they currently experienced difficulties in providing information on rare disorders and 57 (24.7 per cent) of those surveyed did not experience difficulty providing information on rare disorders to families. One GP did not respond to this question.

Figure 1. Do you experience difficulty providing information on rare disorders?



Analysis was carried out in order to ascertain if there was any difference between the Republic of Ireland and Northern Ireland in the responses of those GPs who reported they experienced difficulties providing information to families about rare disorders. This is outlined in Table 3 below, and taking into account the differences in sample sizes when looking at per cent figures, it can be seen that there were small differences between the Republic of Ireland and Northern Ireland and whether the GP was from urban, rural or mixed urban/rural catchment area.

Table 3. GPs who reported that they do experience difficulties providing information to their patients per region and per catchment area.

	Republic of Ireland		Northern Ireland		Total	
	Number of respondents	%	Number of respondents	%	Number of respondents	%
Urban	46	33.8	7	18.4	53	30.4
Rural	26	19.1	15	39.5	41	23.6
Mixed urban rural	64	47.1	15	39.5	79	45.4
Missing	0	0	1	2.6	1	0.6
Total	136	100	38	100	174	100

GPs were asked why they had difficulties providing information to families about rare disorders. As can be seen from Table 4 below, the two most frequently reported reasons that GPs gave for having difficulties in supplying information to patients were workload pressures (50.4 per cent) and the information not being in an accessible format suitable to families (38.4 per cent).

Table 4. Why did you have difficulties providing information on rare disorders?

	Number of responses	Percentage of total sample
Due to workload pressures I don't have time to look	117	50.4
The information is available but not in an accessible format that is appropriate for families	89	38.4
I don't know where to look	64	27.6
The information is available but not in an Irish context	63	27.2
The information is available but not up to date	22	9.5
The information is not available at all	15	6.5
Other	14	6

When GPs were asked what they feel could help them better support people and families experiencing rare disorders the majority of respondents referred to information services, including websites, as being appropriate. This included 101 (51.3 per cent) of GPs who responded to this question feeling that a specialised rare disorders website alone could help them to better support people with rare disorders. While 50 (25.4 per cent) felt that both a website and an information service would help them, 24 (12.2 per cent) felt an information service alone would help them better support patients. Just 12 (6.1 per cent) felt that postgraduate training would help them while eight (4.1 per cent) felt that other resources could help them. The 'Other' category included Irish College of General Practitioners [ICPG] guidelines and electronic contact with Genetics teams. Two (0.9 per cent) GPs stated that they did not know what could help them. There were 35 GPs who did not answer this question.

When GPs were asked where they normally source information on rare disorders, 135 (60.3 per cent) of those who answered said they used the internet as their primary tool with 54 (24.1 per cent) using both the internet and textbooks. There were eight GPs who did not respond to this question.

#### 4.2.3.2 Training and rare disorders

A total of 166 (74.1 per cent) of GPs who responded to the question on training in rare disorders felt that they did not receive sufficient training to help them inform and adequately support people with rare disorders. Just over a quarter — 58 (25.9 per cent) — felt that they had received sufficient training. There were eight GPs who did not answer this question. Of the 58 (25.9 per cent) who felt that they did receive sufficient training to help them support people with rare disorders, the key areas of training that were listed as being beneficial were communication skills, stated by 14 GPs, and work experience during college, cited by eight GPs.

## 4.3 Financial issues

### 4.3.1 Financial issues: views of family participants

#### 4.3.1.1 Loss of income due to disability

More than half of the participants stated that they experienced financial difficulties due to having a child with a disability. Owing to the high level of support needed by many children with rare disorders, in many cases one or more parents had to give up work to take care of their children. One participant explained the difficulty his family were going through due to his wife having to care for their son on a fulltime basis.

*"...financially we're under immense strain...we were structured to be a double income family. We're a single income family now...Last night I said to my wife: 'You're going to have to get a job' and she says: 'Well how can I? I work long hours as it is and she literally can't.' She has to give the time to [child's name]. So there is a financial penalty having a child with a disability."*

Another participant recalls the reality of having to give up full-time work. She had always intended to return to work fulltime but this has proved impossible.

*"I had intended to go back part-time after but I couldn't. There was no way I could work. And I've only just now, when she was eight, gone back to work and it's only possible to do it part-time."*

For an individual with a rare disorder, financial loss was reported as almost always a reality. One participant was forced to reduce his working hours due to having a rare physical disorder which caused him to suffer arm and bone pain which became worse after working. He was advised that, in order to qualify for financial aid, he would have to sell a property he had bought as an investment. After selling the house he was still ineligible for any benefit due to the equity he had received from its sale.

*"I mean I didn't ask for this, she didn't ask for it, my dad didn't ask for it, nor did my ... sisters ask for it but the bottom line is I'm feeling physical less and less able to work. I'm still not entitled to anything, I have to spend all I had managed to accrue, which is for my child, not for me."*

One participant who is self-employed felt that the amount of time she needs to take off work due to having a child with a disability is not conducive to a normal working environment. While being self-employed means she can take off the required time for hospital appointments, she is beginning to feel the impact financially and is worried about the future of her company.

*"I wouldn't be able to hold the job down I don't think. He's got so many consultants' appointments. When you have your own company you can take the time off, but time off equals no income, you know, so we've really, really, really taking a walloping and you know only time will tell whether the company can sort of sustain that type of neglect."*

#### 4.3.1.2 Access/eligibility to benefits and supports

Due to the lack of information and support for people with rare disorders many of the participants initially had great difficulty getting information on their entitlements, and in some cases found out very late that they were actually eligible for a range of allowances. One participant was not aware that she was entitled to financial support until a speech therapist dealing with their child informed them about the availability of domiciliary care allowance. However, they were not entitled to it as their child was less than three years of age.

*"But at that stage a child couldn't claim it until they were three. Now that has been reassessed since but we were on the poverty line back then, unable to work and no support."*

Another participant described how she was forced to give up work due to the care needs of her daughter. She then failed to qualify for carer's allowance due to her husband's income exceeding the threshold accepted. They suffered financially as they spent everything they had on their daughter's therapies.

*"My wage was gone, we were surviving on [partner's wage]. There was nothing there. Absolutely nothing there... not entitled to the carer's allowance because you are over the limit... I mean the last two years we've pumped out thousands just because she needs it...We'll do whatever it takes."*

The son of one participant who started a parent support group for a rare disorder referred to the issues that people can have in accessing financial basics such as insurance.

*"...if they get it as I say they have to pay a higher premium, being refused a mortgage, they have a house bought and they go for their medical check-up, the building society will give them the loan but the insurance will not insure it, dreadful situation."*

One participant felt that she could not return to work as her child had high medical needs due to the nature of the rare disorder. He therefore had a lot of hospital visits and she felt that she could not trust a third party such as a nanny to take her child to these appointments while she went out to work.

*"Realistically, I was the only one completely tuned in to his needs so I just knew that if I tried to give that information to a third party, they wouldn't be interested in your child enough to do something. They might do it haphazardly and, as far as I was concerned, it was my responsibility — I had to do it. So yeah, I didn't go back to work."*

One parent was angry at the lack of support aids she was given and felt that she had to put up a fight to get a free incontinence sheet for her son's bed even though she was supposedly entitled to one.

*"It was absolutely disgraceful. The only thing I ever sought was an incontinence sheet for the bed. He used to wet the bed up to quite an advanced stage...It took six months for them to produce that, during which time I actually sourced it and bought two myself."*

Another participant who was entitled to a special style of bed for her daughter also felt that she had to put up a fight to get the bed she required for her daughter.

*"I had to fight, fight, fight ... in the end I had to pay half and they paid half. Oh I had my TDs and everybody because they wouldn't, they kept saying: 'This is not on contract. They're not one of our contractors.' And I was saying: 'Well the bed you're offering me is not suitable for my child, it's not as if it's a luxury and I'm looking for something really flash and nice.' It really, really is a fight."*

## 4.4 Services for people with rare disorders

### 4.4.1 Services: views of family participants

#### 4.4.1.1 Location of services

Some of the participants felt that they had to constantly chase up services and experienced frustration at the fragmentation of services for people with rare disorders. The large number of people involved in the process of acquiring a service was also frustrating. One participant felt that the structure was poor since applicants had to deal with several different people to get one piece of information.

*"It was really just waiting indefinitely and ringing people and ringing people and just being told to call one after the other... a lot of people just, they just don't even ring back. They're obviously just so used to parents ringing that they don't even return your call any more. You know just before he started speech therapy I rang five different people several times in one week. Not one of them rang back."*

Another participant spoke about the lack of services for his son and how the delay has been detrimental to his progress. Once the participant's son moved from a special needs pre-school to a mainstream school he lost his speech therapy, physiotherapy and occupational therapy services which had been onsite weekly in the pre-school. Now to receive any services the family have to travel and this involves a lot of stress and planning.

*"We live outside the city, so for half an hour physio it takes us five hours to organise... I take time off work...we do it because it's vital...but he still has no OT [Occupational Therapist]. I've campaigned,*

*I've gone to TDs and everything about it and six months later I haven't heard anything back... His education is like a tower, they're pushing the blocks in, but there is no foundation... They're trying to teach my son in school and yet he can't hold a pen, so he's going to fall way behind the other kids. He's really being let down."*

One participant highlighted the difficulties involved with travelling for services and the impact that geographical location can have on the supply of certain services.

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*"They wouldn't do the operation in [local city]. I am very disappointed about that because my support structure is here. Like, if I go to Dublin, if I bring him on my own, the nurse might come up with me but she is not going to be up the day before and like there's medication the day before, medication during the operation and three days after. So you have got a five-day stay in hospital."*

#### **4.4.1.2 Social worker support**

Only two of the participants had dealings with social workers after they received diagnoses. These participants found that the social worker played a crucial role in providing them with information on their entitlements, supports and advocating for them with service providers. This was particularly helpful when dealing with rare disorders because information was so difficult to access.

All of the Northern Irish participants had access to a disability liaison nurse who provided them with basic details on entitlements and where to go to seek advice and information on financial issues. Only two of the Republic of Ireland participants had received similar information at diagnosis and in both cases this was provided by a social worker. One participant who had a social worker advocating for them felt that their family had no problems getting any services or supports from the outset.

*"The key is the social worker. Because the social worker intermits between you and the services and finds out about services...you know, makes that contact on your behalf to get your child a service. I dunno what we'd have done without her."*

#### **4.4.1.3 Alternative therapies/services**

Four of the participant families had used alternative therapies for their children when they failed to get much progress from standard services and therapies.

In all these cases the participants felt that they had excellent results from alternative therapies. However, none of the alternative services that parents sought are provided through public healthcare and therefore costs had to be borne by the parents themselves.

One couple who used an alternative therapy to help treat their daughter's severe scoliosis were so happy with the success of the therapy for their daughter that they have dedicated their lives to helping other people access the therapy and spent extra money on the therapy for their daughter. They shunned any restrictive aids such as splints and braces in favour of this therapy.

*"I mean they said she'd never walk...or roll...and when we took her home after the therapy...and she started climbing. And using her left arm and we were both crying...the results...it was like a miracle. I mean we've spent thousands but we'll do anything. We can't get any help because you know...ooh it's different it's not OT [Occupational Therapy] or physio."*

Another couple visited an osteopath because they could not get any help for their son's diarrhoea. He had been suffering with it for over a year, awake all night and screaming in pain and they felt that being told it was part of being a toddler by their GP was not good enough. After visiting an osteopath they were advised to put their son on an exclusion diet. They found eventually that he seemed to be intolerant of a few different foods. After removing these foods from his diet the diarrhoea stopped and his behaviour improved.

*"He was thin and weak and miserable and totally withdrawn... about a week and a half later he took his first steps and the difference was just unbelievable. You know, after the professional telling you get over it, it's just toddler diarrhoea... We couldn't get a referral for anything,... the idea of a gluten*

*or dairy intolerance is kind of looked upon by professionals as being almost like you're fussy. This has changed the course of his life...he was mute, he was banging his head off things, he was completely withdrawn into himself and going away from us further by the minute and he changed completely once we changed his diet and he was just not sick any more."*

#### **4.4.1.4 Services that meet the needs of people with rare disorders**

Many participants felt that there were often unsuitable service placements for people with rare disorders. Some felt that the expectation that they would fit into more generic services that did not recognise the unique needs of the individual with the rare disorder often did more harm than good in terms of their progress.

One participant's son had to use a number of different services before finding a suitable place for his education.

*"He went to special school which was too severe for him really, it didn't suit him. There was nobody in his class talking, so he ended up going home with tantrums and just going backwards rather than going forwards and then I took him out of there and brought him to the local school for a year and then I realised after the end of the first year that this is not working."*

Her son, who has a rare disorder, echoed these difficulties and spoke about being in a school where he found it difficult to interact with other people. He found it hard to read out loud and felt that he had no friends there.

*"Mmm, it was hard, it was just so hard reading in the class with other people and people are not talking to me. Well I found it so hard. There was nobody to talk to."*

Another participant who has a rare disorder felt that a service he had been using was not appropriate. He had used services abroad which he felt catered to his needs much better than they did in Ireland. After a more person-centred approach was taken with him his goal for independent living was realised and he is now much happier in his placement.

*"...I felt it was not the place for me...we didn't have much of a social life in the community, now I feel I do...the staff are great and I am really happy...they listened to me."*

Another couple who have a child with a rare disorder were extremely frustrated due to constantly searching for a suitable service for their son. They eventually got referred to a local school which, although mainstream, had a special needs section which they found to be very person-centred. They moved their son to the school and have never looked back.

*"When we called in just to look at the place, they spent an hour and half showing us around and giving us their time. Four different people, vice principal, the principal, the resource teacher and the home liaison officer all came to meet us...I had to fight to stop myself from crying...It is just such a relief to find somebody who is actually interested and wants to help your child reach his best possible potential."*

One participant who is very frustrated with the fragmentation of services echoed the view that service providers need to have a flexible approach to service provision. He finds the bureaucracy a major barrier in actually getting services and entitlements and often feels like he is forced to go around in circles to apply for services and entitlements which he may not even receive in the end. He also feels that a one-stop shop with advice and advocacy is essential.

*"Go drive to one centre, go and drive to the dole office, get this stamped, go to the hospital then and get that signed... They should be able to walk in and say that's my PPS number, I'll come back next week. Would you have it all sorted for me? Instead of some [expletive] pen pusher from the Civil Service...it's cruel. Either that it's all under one roof or there is a person whose job it is to go and get your stuff for you."*



Another participant felt that having a rare disorder with a more severe intellectual disability means that you come last in the queue for service delivery and supports when compared with other more common disabilities.

*“There has been a lot of input into what I call the able-bodied disabled and people who are mildly or moderately disabled, who can be assisted by advocates and as a result I think that severely and multiply disabled, especially with rare disorders, tend to basically come at the bottom of the pecking order when it comes to service delivery. There is no doubt about it.”*

#### 4.4.2 Services: views of specialist health professionals

A specialist health professional felt that the health services are under-funded in every area and that this impacts very negatively on patient care.

*“...it is very dependent upon the good will of individual consultants...Often times, consultants don't even have proper secretarial service to file files, to reliably file letters, to do very basic things that we should all be able to take for granted ... the hospital administration frequently does not put in place an adequate clerical network to facilitate the doctor's work. So oftentimes, the doctor is working really very hard but the patient doesn't see it because the essential clerical link hasn't been put in place and I speak from personal experience.”*

Another specialist health professional stated that specialist clinics for rare disorders at a local level are essential to inform families in order to provide best practice in care and service provision. This specialist health professional described how these clinics take place throughout the year for different disorders where experts meet families. She feels that the clinics are extremely beneficial.

*“Specialist clinics are quite good...we do these two massive clinics and that is really good because it is a way of them getting specialist advice on their own doorstep. You know, it's a big hassle as well if you've got a handicapped child trying to take them on a plane.”*

All of the specialist health professionals felt that one centre of expertise is essential to help people with rare disorders.

*“A multidisciplinary one-stop shop....sometimes it's the eye clinic, the heart clinic, the kidney clinic, the genetics and they're up and down like yo-yos whereas if you could get funding when they can come in and everybody is in the one place, a bit like a child development clinic but for rare diseases, that would be excellent. Plus it's great for the families and I think it is also good for the professionals themselves because they can talk to each other and get ideas from where their expertise is and what would you do in this situation and I think that is great.”*

Another specialist health professional stated that managing and referring people with rare disorders can prove problematic as there are no services specific to their disorder.

*“And often even when you make the rare diagnosis, your problem is trying to manage them then because the services do not embrace them...Often they need specialist therapies that are very unique ...I would be hopeful over the next five years with the new national children's hospital when they bring under one roof a very large number of people, some specialist things may improve as one can be seen by large numbers of people in the same room very rapidly because most of these disorders are multidisciplinary.”*



## **Chapter 5: Discussion**

In this chapter the research findings are discussed under the following headings:

### **Information and rare disorders**

- ◇ information needs of families
- ◇ the internet and information on rare disorders
- ◇ healthcare professionals and knowledge of rare disorders
- ◇ diagnosis

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### **Supports and rare disorders**

- ◇ peer support and support groups
- ◇ sibling support
- ◇ financial support
- ◇ emotional support

### **Services and rare disorders**

- ◇ rare disorders and flexible person centred-service provision
- ◇ alternative therapies
- ◇ social workers and advocacy
- ◇ location and structure of services

### **Limitations and reflexivity of the research**

## 5.1 Information and rare disorders

### 5.1.1 Information needs of families

Previous research has emphasised the information needs of both individuals with a rare disorder and their families. The Genetic Interest Group (1999) and Webb (2005) highlighted the importance of information for both categories. All of the family participants in this research stated that clear and accessible information on the specific disorder they were experiencing was essential to aid their understanding. Some participants expressed a need for non-medical explanations about the actual disorders as they could not make sense of the medical and genetic terminology. Other participants referred to the need for a filtered information process as too much information can be frustrating and overwhelming. It can be seen that this need for information is ongoing, not only about the disorder, but about the supports available to the family on an age-appropriate, life-span perspective.

Specialist health professionals involved in this research agreed that the information they have about rare disorders can often be in an unsuitable format for parents and that support is needed to help them translate information and compile it for families. This was supported by 75.3 per cent of GPs surveyed as part of the research stating that they experience difficulties providing information to families about rare disorders with 50.4 per cent of the total sample of GPs citing workload pressures as the main difficulty. Further, 38.4 per cent of GPs felt that the information was available but not in a format accessible to families.

This would suggest that there is a great need for information and education for both health professionals and families affected by rare disorders. There appears to be a lack of awareness as to how to access the information and also considerable time constraints and pressures on professionals that often inhibits them from dedicating the necessary time to search for suitable information for the families. Harnett *et al* (2007), as part of the 'Informing Families of their Child's Disability' project in the Republic of Ireland, listed meeting the family's information needs as a key priority in order to relieve levels of parental stress. The author pointed out that it is essential to pace the information in accordance with the needs of the individual family in order to avoid information overload. It can be seen from this research that this finding is particularly relevant to those families who experience rare disorders.

With such an obvious requirement for clear and sufficient information for families reported in this research, and a need for information on the part of professionals as highlighted by both GPs and specialist health professionals, it is apparent that an information service on rare disorders is required on the island of Ireland.

### 5.1.2 The internet and information on rare disorders

Research has shown that searching for health information on the internet is an extremely popular choice for both professionals and families (e.g. Gallagher *et al*, 2007, Powell *et al*, 2003).

Although the internet has obvious benefits for professionals and families alike, both Fox (2005) and Powell *et al* (2003) drew attention to the lack of regulation of internet information and the risk of people coming across inappropriate and inaccurate information. All of the family participants in this research had either used the internet to access information on the relevant rare disorder or had a friend who sourced them information on the internet. Of the GPs surveyed about where they source their information, 60.3 per cent stated that the internet is their primary source for information on rare disorders. Some of the specialist health professionals who took part in the research rate the internet as a highly valuable information resource but recognised that GPs and other health professionals need to be educated about where to access the information on rare disorders. Also, 51.3 per cent of GPs who responded to the question on what would assist them to support families experiencing rare disorders stated that a rare disorders website alone would help them. In addition, 25.4 per cent of GPs who responded to the question on what would assist them to support families experiencing rare disorders felt that both an information service and website would help them support people.

Given the increasing internet use and the popular use of the internet to source information on rare disorders by both families and professionals, it is clear that some regulation is needed to ensure information is appropriate. EURORDIS (2004a) issued guidelines for those providing information to individuals with rare disorders and their families and outlined that information must be validated and revisited regularly and provided in a clear and accessible way in terms of content, format and appearance. Gallagher *et al* (2007), in the context of the Republic of Ireland, highlighted the steps towards regulation of websites providing health information through the development of organisations to monitor the content. Contact a Family and the Information Management Research Institute [IMRI] (2003) compiled the Judge Project which issued guidelines for both consumers and support groups for judging the quality of health information.

Two specialist health professionals felt that if people with rare disorders and their families are not given sufficient information they will go to extreme lengths to access it and can therefore end up with inappropriate information.

Orphanet is a portal for information with rare diseases and orphan drugs aimed primarily at health professionals across Europe. This is a free service that also has a section for patients with rare diseases. It is important that Orphanet receives a high profile within the island of Ireland amongst primary care professionals in particular. The UK-based organisation Contact a Family (2008) has developed an online Directory of Specific Conditions and Rare Disorders. This information source is a model of good practice for information provision that could be extended to the island of Ireland.

Although it is clear that there are guidelines surrounding health information in a European and UK context, this research suggests that specific guidelines are needed for support groups and websites on the island of Ireland. With the large number of GPs and families accessing the internet for information on rare disorders this research would suggest that a dedicated rare disorders website for localised information, with sections for both professionals and families, could, if properly regulated, be hugely beneficial. Links with initiatives such as Orphanet and Contact a Family could greatly assist in this process.

### 5.1.3 Healthcare professionals and knowledge of rare disorders

The majority of the family participants had on at least one occasion found themselves having to inform a non-specialist health professional about their child's rare disorder and found that, in general, knowledge on rare disorders was lacking. Knight and Senior (2006) felt that guidelines are needed as many patients with rare disorders often present their symptoms initially to GPs and continue to use their GPs for general preventative healthcare between specialist visits. The authors also argued that a systematic generic primary-care approach to rare disease could reduce problems such as lack of co-ordinated care, lack of information, delayed diagnosis and other difficulties encountered by people with rare disorders and their carers.

Some participants involved in this research who were part of support groups have compiled information packs on each disorder for parents to take with them on health visits. One support group compiled information on the particular rare disorder that was circulated to all GP surgeries across Ireland. Some participants who experienced a lack of knowledge in healthcare settings felt that this could have been detrimental to the treatment of their child's condition had they not been there to explain the differing manifestations of the disorder.

Many of the participants felt that a basic website with information on the different disorders could be extremely useful while some of the participants emphasised the need for a filtering process from health professionals — i.e. to be given the information in phases in order to avoid information overload.

In regards to primary care, 27.6 per cent of the total number of GPs surveyed felt that they did not know where to look for information on rare disorders and 74.1 per cent of GPs who responded to the question on training in rare disorders felt that they did not receive sufficient training to help them inform and adequately support people with rare disorders. It is reasonable that GP training cannot cover every aspect of rare disorders. However, it can be suggested that training can include how GPs

should access relevant information to be presented to individuals and families. A centralised source of information on rare disorders is therefore key. A specialist health professional involved in this research felt that healthcare professionals and GPs need to be informed as to how they can access rare disorders information in order to adequately support their patients.

54 | This again would suggest that there is an obvious need for a regulated information service and website for both healthcare professionals and people affected by rare disorders. As suggested previously, links with Orphanet would assist in this process — particularly for a website section for health professionals. A dedicated role to the development and management of the service, along with a steering committee of specialist health professionals and families to oversee and regulate content and guide people on how to access information would be required.

#### **5.1.4 Diagnosis**

Kerr and McIntosh (1998) felt that it is crucial for healthcare professionals to realise the impact of diagnosis for parents so that parents can be guided and supported through the emotionally difficult early days. Woolfe and Bartlett (1996) found that parents at diagnosis wanted clear unambiguous information and continued contact and support. The majority of family participants involved in this research felt that, due to the rarity of the disorder, they had received minimal or inappropriate information at diagnosis and that this had impacted negatively on them. Many of the participants had been forced to access unregulated websites to try and comprehend their child's disorder. Some also perceived a distinct lack of understanding by healthcare professionals at the diagnosis.

A result of the 'Informing Families of their Child's Disability' project (Harnett *et al*, 2007) was the creation of National Best Practice Guidelines in the Republic of Ireland on informing families of their child's disability. The report recognises that every disclosure event is unique but outlined seven guiding principles that the author felt should be applied in every case. These included: family-centred disclosure, respect for child and family, sensitive and empathetic communication, appropriate, accurate information and positive, realistic messages and hope. This current research has suggested that parents of children with rare disorders can be very vulnerable at the diagnosis stage and therefore require both sufficient information and emotional support. The successful implementation of these National Best Practice Guidelines throughout the island of Ireland would benefit people with rare disorders hugely.

## **5.2 Supports and rare disorders**

### **5.2.1 Peer support and support groups**

All of the participants in this study felt that peer support was an extremely important part of coping with rare disorders and plays a major role during difficult times. Participants also recognised peers as having an important information function. Due to the rarity and lack of understanding of the disorders experienced by families in this research, peers were particularly appreciated.

Of those GPs who agreed that having a rare disorder gives rise to additional family problems, 28.0 per cent felt that rare disorders can result in feelings of isolation in families. Two of the specialist health professionals in this research felt that peer support was crucial in combating these feelings of isolation and recognised the role of support groups in the provision of such peer support.

Previous research has demonstrated the importance of social support for families of children with special needs (Hartman *et al*, 1992; Ainbinder *et al*, 1998). Mead *et al* (2001) described peer support as being about understanding another person's situation empathically through the shared experience of emotional and psychological pain and he pointed out that people find affiliation with others they feel are like them through feeling a connection.

All of the family participants who experienced peer support felt that the only people they could really relate to are people who have had similar experiences to themselves. All of the participants found peer support and support groups to be positive in their approach to rare disorders. This finding is in line with

Curtis (1999) who pointed out that peer support offers a culture of health and ability as opposed to a culture of illness and disability.

Some participants in this research referred to the frustrations of accessing peer support due to the rarity of the disorder they were experiencing. Many family participants felt that they had felt isolated and removed from society due to the lack of understanding of their child's rare disorder. They felt that they only began to realise that they were not alone when they met other families who experienced the same issues. This is in line with Mead (2001) who described peer support as being about normalising what has been named as abnormal because of other people's discomfort. Some families found that they had experienced positive peer support from families of children with more common disabilities as they had similar needs. Others found that people with more common disabilities were supported much more by their representative organisations and therefore they had little in common with them in terms of experiences.

All of the support groups for people with rare disorders on the island of Ireland primarily exist for peer support and information. Almost all of these groups are operated by parent contacts in their free time and receive no funding. One specialist health professional felt that it is important for rare disorder support groups to remain in the social realm in terms of information provision. A number of specialist health professionals involved in the research felt that an umbrella organisation representing all support groups could provide a stronger voice to lobby for much needed supports and benefit people with rare disorders across the board.

Some family participants stated the negative side to peer support, including the 'devastation' of seeing an older version of your child with a disorder that is progressive. This highlights an important point about the age-appropriateness of peer support, where families can meet others of similar ages.

This research has shown that support groups and peer support of an age-appropriate manner is essential for the participants in terms of combating isolation, providing information and increasing understanding. The lack of funding of support groups and the absence of support groups for extremely rare disorders would suggest that a central support group or umbrella group representing families experiencing rare disorders could have huge benefits and could lobby more effectively for information services and funding.

### **5.2.2 Sibling support**

The research literature on siblings of individuals with a rare disorder was found to be lacking. Support groups for rare disorders such as Unique and Contact a Family in the UK provide information to parents on how to support the brothers and sisters of their child with a rare disorder. The information provided to families does, however, relate also to more common disabilities. Perrin (1999), for example, discussed the benefits of 'sibshops' where siblings of children with a physical and sensory disability took part in structured workshops allowing them to talk and learn about the disability. The author concluded that the children involved in 'sibshops' were extremely well adjusted and had overcome negative feelings such as unfairness at the uneven balance of parental attention. In the Republic of Ireland Barretstown recognised the need for sibling support and provides activities for the whole family, including the siblings of children with serious illnesses.

All of the participants in the research who had more than one child felt that having a brother or sister with a rare disorder had a negative affect on their other children. Many parents in this research felt that they inevitably gave more attention to their child who had a rare disorder as they had greater needs. Although many parents made concerted efforts to redress the balance among their children they felt that support such as sibling workshops or summer camps would help siblings come to terms with their situation. A small number of participants had brought their children to conferences and other events with the sole purpose of enabling their children to mix with other siblings of children with rare disorders. In all these cases the sibling support had proved extremely beneficial and educational.

The specialist health professionals in this research agreed that siblings often felt overlooked, with one health professional suggesting that schools could play an important role with siblings in terms of

education. Another specialist health professional made reference to a support group for a rare disorder that is very inclusive of siblings in their conferences and seminars. This specialist health professional feels 'whole family' inclusive efforts need to be adopted across the board as they are hugely beneficial for families.

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This research suggests that there can be an uneven balance of time and attention afforded to siblings of children with rare disorders by their parents. Siblings who have experienced peer support have benefited hugely from it. However, rare disorder support groups often don't have funding and therefore they cannot provide such specific supports. There is a suggestion that funding for sibling supports such as workshops would be hugely beneficial to siblings and parents alike, taking the stress off parents while their children come to terms with the rare disorder in a supportive setting. However, it is important to note, as previously stated, that there is lack of research on siblings of people with rare disorders, as compared to the needs of siblings in families experiencing more common disabilities. None of the participants (both family and specialist health professionals) in the research who commented on the needs of siblings distinguished between rare disorders and more common disabilities. Further research, following on from this current research, that would focus in-depth on siblings within families experiencing rare disorders would tease out the differences, if any, in experiences due to the rarity of the disorder.

### 5.2.3 Financial support

Financial pressures are seen amongst families experiencing all disabilities. Redmond *et al* (2000) in their study of parents of children with disabilities, found that 15 out of the 17 cases reported a financial pressure on families due to the loss or diminution of the mother's income.

Parents in this study reported difficulties accessing information about their entitlements, made more difficult by the rare nature of their child's disorder. Rare disorders are defined as often being 'chronically debilitating' meaning that high support needs are present, resulting in the need for more round-the-clock attention. Consequently in this research it was found that in some cases one parent was forced to cease fulltime employment in order to care for their child. Some of the family participants therefore stated that they had suffered a loss of income due to having a child with a rare disorder. Some of the participants felt that the carer's allowance was insufficient to replace the loss of a fulltime income and that they were struggling to survive. Some participants failed to qualify for allowances due to either their assets or their partner's income exceeding the threshold required. Those participants who were entitled to allowances recall struggling financially before finding out about these entitlements through chance meetings with other families in similar situations. Two family participants experienced great difficulties getting basic aids such as incontinence sheets or specialised beds and felt that they had to battle to get these goods for free.

This research suggests that people need practical support on financial issues and that the provision of help and advice on financial issues is essential. The relevant government citizens' advice agency in both the Republic of Ireland and Northern Ireland are important sources of information on this and this research would recommend links would be created with a centralised information service for rare disorders, in order to exchange and enhance knowledge bases.

### 5.2.4 Emotional support

Davies & Hall (2005) suggested that parents often feel isolated, lonely and unsupported when caring for a disabled child and pointed out that family relationships suffer as parents are too tired to have time for each other and for their other children. Redmond *et al* (2000) found that disability had a negative impact on family functioning in terms of relationships between partners. These findings were substantiated in this current research, where 72.2 per cent of GPs who responded on emotional issues felt that rare disorders can give rise to additional family problems. Among the problems listed by GPs were isolation, stress, conflict, lack of understanding from friends/family, anxiety and relationship problems.



Many of the participants in this study spoke about the stress that having a child with a disability had placed on their relationship. Particularly with rare disorders, this came from genetic guilt as well as refusal to accept the child's disability. Many participants experienced difficulties with their spouse. In some cases the mother ended up taking the role of primary carer until the father was able to deal with the situation at hand. The specialist health professionals involved in this research all recognised that feelings of guilt and stress pervade parents of children with rare disorders. One health professional recognised that relationships can suffer because the 'blame game', often originating from older generations in the family, can add pressure to parent relationships. The emotional impact on the extended family was also mentioned by family participants in this research. The Frambu model, based in Norway, has in recent years recognised the role of grandparents in families experiencing rare disorders and has responded to these needs by providing specialised programs for grandparents of people with rare disorders.

One specialist health professional felt that fathers often find it difficult to accept that they may have created an imperfect child and that support is essential for fathers in these cases. All the specialist health professionals recognised counselling as essential, with the absence of this type of emotional support as being possibly detrimental to parent and family relationships.

It is suggested from this research that parents of children with rare disorders often suffer additional problems such as stress and guilt. These added stressors can often affect parent relationships and counselling is essential to help alleviate this stress and combat potential family problems. Further research into the specific needs of different members of the family including fathers and grandparents is also required in order to set a context for planning of specialist provision of support.

## 5.3 Services and rare disorders

### 5.3.1 Rare disorders and flexible person-centred service provision

Previous research (e.g. Hennepe, 1999, Hernandez *et al*, 2006) referred to the importance of a holistic approach to service provision for people with rare disorders.

Hennepe (1999) stated that parents of children with rare diseases experience problems such as a lack of knowledge, late diagnosis and lack of continuity in care. The author stated that a multi-disciplinary approach is needed for this population. Many family participants in this research felt that people with rare disorders are often unsuitably placed within generic services. Some participants felt that there was an expectation for them to fit into these more generic services and that this often did more harm than good in terms of their progress. One participant for example stated that with their child being part of a generic service this held him/her back developmentally. The majority of the family participants in this research felt that there was a huge need for all services to respond to the unique and complex needs of rare disorder patients.

The Genetic Interest Group (1999) consulted with clinicians and support workers who worked with families affected by genetic disorders and presented 20 case histories. Their report stated the importance of medical care having a holistic view of the affected person and his and her family. The participants in this research who had experienced flexibility in services and person-centred approaches within services felt this resulted in a more positive experience for both themselves and their children. Most family participants who experienced person centred, individualised services commented on their appreciation of understanding staff who operated flexibility in their approach in order to meet the needs of the person with a rare disorder.

One specialist health professional referred to lack of funding in healthcare being an issue and that this can impact very negatively on patient care.

This research suggests that flexibility and person-centred approaches which fit the service to the individual's specific needs should be adopted by all service providers, as the very nature of rare disorders means that often they will not blend effectively with generic disability services. Current services that are

informed about rare disorders and equipped to be able to provide the necessary support that is specific to the unique needs of people with rare disorders are required in order to provide the best possible level of care.

One specialist health professional referred to the need for people involved in education to be appropriately informed about rare disorders and the specific needs of the family. This was not explored further in this research but highlights a requirement for all services within the local community that impact on the life of the family to be informed and educated regarding the rare disorder experienced by the family. The Frambu model in Norway provides this type of information service within their outreach work. There is evidence to suggest this type of outreach service should be considered within the island of Ireland.

### 5.3.2 Alternative therapies

Some of the participants in this research had used alternative therapies for their children when they failed to get much progress from standard services and therapies. In all these cases the participants felt that they had achieved extremely positive and successful results from the chosen alternative therapies.

However, none of the alternative services that parents sought are provided through public healthcare and costs therefore had to be borne by the parents themselves.

This research suggests that alternative therapies play an important role for some families with children with rare disorders and that they need to be given consideration in a tailored rare disorders service.

### 5.3.3 Social workers and advocacy

Redmond *et al* (2000) pointed out that having no advocate who could help families to obtain information and access to support services could be a further stressor. Only two of the family participants in this research had dealings with social workers after they received diagnoses. These participants found that the social worker played an essential role in providing them with information on their entitlements, supports and advocating for them with service providers. All of the participants in Northern Ireland had access to a disability liaison nurse who provided them with basic details on entitlements and where to go to seek advice and information on financial issues. All these participants reported that this was a positive assistance in their search for information. One participant had an especially positive experience with a social worker, reporting that they had no problems getting any services or supports from the outset, as the social worker acting as their advocate, carried out all the enquiring on their behalf.

It is important to consider the benefits of practical social support services that advocate for and inform families with rare disorders of the services available to them.

### 5.3.4 Location and structure of services

EURORDIS (2005b) recognised the need for dedicated services for people with rare disorders. They maintained that the social aspects and implications of rare diseases have to be kept in mind while the community and financial services to support families and patients have to be organised and developed on a local basis (EURORDIS, 2005b: p10).

Some family participants in this research referred to the location of services as being a barrier to access. Some felt that there were better services in urban areas and that they were forced to travel for these services. Some participants reported that services were too scattered and felt that a one-stop shop approach to social support and therapy provision was needed. Two specialist health professionals supported this position by suggesting that one centre of expertise is essential to adequately support people with rare disorders.

One specialist health professional recognised that in order to achieve best practice in care and service provision, localised specialist clinics are essential to inform families. This health professional made reference to outreach clinics for different disorders where experts are brought over to meet families and

stated that these are extremely beneficial because they serve all localities and therefore people are not required to travel to access services. This research suggests that the location and fragmentation of therapy services must be considered as they can often put excessive travel pressures on families affected by rare disorders. Outreach specialist clinics would help bring the services to the people and could support the provision of best practice for people with rare disorders.

Looking to models of best practice identified in the literature review (Frambu, Norway and Agrenska, Sweden) it can be suggested that the island of Ireland would benefit from a centre that has expertise in rare disorders, acting as a portal for information for professionals, families and the wider community, including schools. Evidence gathered from this research suggests also that social support is required in one centre, and on an outreach basis, to include counselling for parents, facilitation of peer support (tailored specifically to each rare disorder), sibling support workshops, response to the needs of the extended family, respite services and access to both established and alternative therapies.

With regards to the geographical area studied in this research, the analysis of GP responses in this investigation did not render any significant difference between the Republic of Ireland and Northern Ireland in regards to information needs of primary care professionals or their perception of the social and information needs of families. Family participants and specialist health professionals came across similar issues when dealing with rare disorders in the Republic of Ireland and Northern Ireland. Considering the population size of the Republic of Ireland and Northern Ireland combined, and the low numbers of specific rare disorders, it can be suggested that if a centre of expertise were to be developed this would cover both jurisdictions. There are added complexities due to differences in policy and service provision in the two jurisdictions of the Republic of Ireland and Northern Ireland which would need to be taken into consideration.

#### **5.4 Limitations and reflexivity of the research**

A longitudinal design would have worked well in this research as it would have provided a better picture of long term support needs of families on a life-span perspective. This would also have generated an important evidence base reporting on the needs of families for the long term care of the child with a rare disorder through adolescence to adulthood, and the specific services required at these times. A longitudinal design would have sufficiently increased researcher rapport with the participants. Due to the time constraints of this study it was not possible to incorporate such a design.

In retrospect, the interview schedules for both families and professionals were not strictly adhered to. It is felt, however, that the flexible approach to interviewing coupled with some probe style questions systematically helped the researcher gather the broad range of data required.

One of the questions on relationship difficulties for parents was recognised as being a sensitive issue and where a participant parent and their child with a rare disorder were interviewed together the researcher chose to omit this question.

The researcher had initially sought to hold focus groups with siblings of people with rare disorders as part of the research. Following initial access difficulties due to the sensitive nature of the topic the researcher sought to elicit data about experiences of siblings from parents in this study. Although the researcher recognised that the gathering of data via a third party can be unrepresentative it was felt in this case that the data received from parents was very valuable to the research and is included in the analysis and discussion. Recommendations for further research with siblings are made in the final section of this report.

All of the families who participated in this research had received a clear diagnosis of the rare disorder they were experiencing. As reported in the literature review, there are also specific needs of people who have no diagnosis. This study did not extend to an investigation of these needs, and would recommend that further research be carried out with families on the island of Ireland who have not received a diagnosis, in order to appropriately plan for the provision of support.



## Chapter 6: Conclusions and recommendations

This report has presented a study that was completed in two phases. Phase 1 was a comprehensive review of the literature, policy and practice context with regard to the social support needs of families experiencing rare disorders. Phase 2 was a consultation with families, GPs and specialist health professionals on the social support needs of families who experience rare disorders. The findings of the study have been presented and discussed and arising from this there are very clear conclusions and recommendations that can be put forward with regard to social and information support for families who experience rare disorders. This chapter begins by making conclusions within three themes carried over from the discussion, including information and rare disorders; social support and rare disorders and services and rare disorders. Finally, recommendations on how to support both families experiencing rare disorders and health professionals in the field of rare disorders are presented as a response to the research.

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### 6.1 Conclusions

#### 6.1.1 Information and rare disorders

- This research has highlighted the need for appropriate, accessible, clear and up-to-date information about the disorder experienced by the family. Families reported negative experiences of receiving information, particularly at diagnosis.
- The requirement for an on-going, life-span approach to information provision was highlighted. This approach needs to be age-appropriate and filtered or paced in accordance with the needs of the individual family in order to avoid an overwhelming information vacuum.
- GPs and other non-specialist health professionals often experience difficulty accessing appropriate information on rare disorders. Accessing this information can be time-consuming for GPs. This information vacuum can be distressing for families and health professionals, with parents often informing professionals about their child's disorder.
- There is no website in the Republic of Ireland offering information on rare disorders that specifically focuses on the social support needs of families, yet the internet seems to be the main point of information for both health professionals and families. Families in Northern Ireland can find some localised information from UK websites and services such as Contact a Family.
- Some support groups for rare disorders have their own websites and provide information to families. However, these are not always funded or regulated. Due to their very nature some rare disorders have no support groups and therefore parents are accessing content from the internet that is unregulated and not in a localised context.
- Although this research has highlighted the benefits of the internet as a source of information on rare disorders it also highlights the dangers of unregulated websites providing inappropriately presented and sometimes inaccurate information.

#### 6.1.2 Support and rare disorders

- This research has shown that peer support is crucial for families with rare disorders as they often experience isolation due to the uncommon nature of the disorder. It was highlighted that peer support should be age-appropriate.
- The need for peer support to be specific to the disorder experienced by the family was highlighted by participants.
- Support groups play an essential role in terms of information provision and peer support. However, they are not funded and are often run by family contacts. Where disorders are very rare and

where there may only be one or two known cases in the country, these families are often without support groups or any type of peer support and can experience isolation.

- It was highlighted that a centralised point for rare disorder support groups to work together would provide a stronger base for advocacy and efficiency.
- It is suggested from this research that parents of children with rare disorders often suffer additional problems such as stress and guilt. These added stressors can often affect parent relationships.
- The research presented arguments for the need to support specific family members, including fathers and the extended family, particularly grandparents. This is due to the genetic nature of many rare disorders adding to feelings of guilt from some family members.
- This research highlighted the support needs of siblings in families with rare disorders, with parent participants in this research often feeling that they cannot afford to give sufficient attention to both their child with a rare disorder and their other children. Some participants stated that workshops for siblings or an inclusive approach by the support group to whole family involvement had been hugely beneficial.
- There is often a loss of income in families where a child has a rare disorder. Rare disorders by their nature can be 'chronically debilitating' and some participants in this research stated that they were forced to cease fulltime employment to care for their child. The need for guidance on practical information such as entitlements was highlighted as essential for families.

### 6.1.3 Services and rare disorders

- This research suggests that flexibility and person-centred approaches which fit the service around the individual's specific needs should be adopted by all service providers because the very nature of rare disorders means that often they will not blend effectively with generic disability services. This, therefore, suggests a requirement for training and information about rare disorders for those working in the health services.
- Both family participants and specialist health professionals reported that a one-stop shop style of service for rare disorders would be beneficial in order to act as portal for information and service provision.
- The research has highlighted the importance of one centralised area of expertise for rare disorders that could act as an information function and provide social support services to families. Having taken an all-island approach to the research the authors noted the appropriateness of one centre for the Republic of Ireland and Northern Ireland combined.
- Some families found that alternative therapies proved extremely beneficial in helping their children with rare disorders. However, since these treatments are not part of mainstream healthcare, parents have to pay for them privately.
- This research suggests that the location and fragmentation of therapy services must be considered as they can often put excessive travel pressures on families affected by rare disorders. Outreach specialist clinics offering both therapy and information can bring these services to families experiencing rare disorders.
- It was suggested that other services (for example, education) that impact on the lives of families experiencing rare disorders should be informed about the specific rare disorder. It was highlighted this could be addressed via outreach services.
- Epidemiological data on rare disorders is limited in Europe and on the island of Ireland. This impacts negatively on the information available to assist in planning for service provision.

- There is a suggestion that further research, perhaps incorporating a longitudinal design, would report on the needs of families for the long term care of their child through adolescence to adulthood, and the specific services required at these times.
- Where the literature review provided some insight into the needs of families where no diagnosis has been made, this current research did not explore this any further. The needs of these families should be researched further in order to plan for appropriate provision of support and information.

## 6.4 Recommendations

### **There is an essential need for an accessible website and a centralised information service on rare disorders on the island of Ireland.**

In order to minimise the risk of families who experience rare disorders accessing worst case scenario, inaccurate or out-of-date information it is strongly recommended that a centralised information service should be developed. This service should be guided by a steering committee made up of specialist health professionals and families. The committee would be ideally placed to oversee and regulate content. It would also guide people on how to access information. Information for both health professionals and families that is specific to the island of Ireland should be contained in a website, in separate sections where relevant. It is essential that an on-going, life-span approach to information provision is adopted. This needs to be age-appropriate and filtered in accordance with the needs of the individual family in order to avoid an overwhelming information vacuum. Links to recognised models of good practice such as Orphanet and Contact a Family would be extremely beneficial in the development and presentation of information about rare disorders on the island of Ireland.

### **There is an urgent need for a support service tailored specifically to individuals and families affected by rare disorders. A centre that can provide social support, information and outreach for both professionals working with rare disorders and families is recommended to provide a centralised point of contact and expertise.**

The bringing together of expertise in rare disorders in one centre would ultimately benefit both health professionals and families. Due to the population size and uncommon nature of rare disorders, it would be appropriate geographically to establish one centre to cover both the Republic of Ireland and Northern Ireland combined. A centre where the knowledge base of rare disorders is built up as a result of direct work with individuals with rare disorders and their families and where the facilities are available for social support, as well as periodic therapeutic interventions, is also recommended. In order to respond to the unique needs of families experiencing rare disorders it is essential that any such support should be holistic and person-centred in its approach. It should be multi-disciplinary in its approach. It is recommended that key issues that were reported by participants in this research would be addressed in this type of service. These services, which would be delivered both in the centre and on an outreach basis where appropriate, would include:

- facilitation of peer support
- counselling for parents
- information programmes tailored specifically to each rare disorder
- sibling support workshops
- response to the needs of the extended family
- respite services
- access to both established and alternative therapies
- training for healthcare professionals about rare disorders
- a centralised point for support groups and parent contacts for specific rare disorders

It is strongly recommended that this centre would provide outreach services relating to support and information for families in order to minimise the need for travel and the difficulties this can create. There is a need also for an outreach facility to provide information about rare disorders to community-based services that impact on the life of a family, including schools.

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**In order to provide the best possible level of care current health services should be informed and equipped to provide the necessary supports that are specific to the unique needs of people with rare disorders.**

In order to reduce the difficulties faced by individuals and families who experience rare disorders when accessing current health services, including primary care, it is emphasised that staff within these health services would significantly benefit from specialised training on rare disorders, with access to a centralised source of information on rare disorders. Health professionals should also be supported to meet with other professionals of the same or another discipline.

**Families experiencing rare disorders should have access to practical information about their entitlements as well as about the services which are available to them.**

The relevant government citizens' advice function in both the Republic of Ireland and Northern Ireland are important sources of information in relation to practical areas such as entitlements. This research recommends that links be created with a centralised information service for rare disorders, in order to exchange knowledge.

**Greater interaction between rare disorder support groups should be supported. This should include the promotion of links between support groups and umbrella organisations such as the Genetic and Rare Disorder Organisation [GRDO] and the Genetic Interest Group [GIG].**

Umbrella organisations represent the voices of rare disorder support groups and those individuals without a support group within their respective national legislative contexts. They provide a valuable voice to promote the views of people with rare disorders and their families in a lobbying forum, and therefore all rare disorder support groups should be informed of their activities.

**The successful implementation of the guidelines set out by Harnett *et al* (2007) as a result of the 'Informing Families of their Child's Disability Project' in the Republic of Ireland would greatly benefit people with rare disorders.**

This research has suggested that parents of children with rare disorders can be very vulnerable when first hearing about their child's diagnosis and therefore, require appropriate information and emotional support. It is important that these guidelines are rolled out throughout the island of Ireland to bring maximum benefit to the families of those with rare disorders.

**Epidemiological data is required to ascertain the exact number of people with rare disorders in the Republic of Ireland and Northern Ireland.**

Accurate data on the nature and distribution of rare disorders in the Republic of Ireland and Northern Ireland is essential in exploring the need for support and the provision of resources. The development of coding procedures and patient registries in both jurisdictions is essential in obtaining reliable and accurate information.

**There is a need to consider the additional financial pressures experienced by families of those with rare disorders. It is important that the families of children with rare disorders are provided with adequate income supports to minimise financial difficulty.**

Rare disorders are defined as often being 'chronically debilitating' which results in more round-the-clock attention for the individual. In this research it was found that many parents are forced to give up fulltime employment, thus resulting in a loss of income in families where a child has a rare disorder. The need for adequate income supports is therefore highlighted as essential for families experiencing rare disorders.



**Further research is recommended that would explore the level of support each separate family member requires, as well as investigating the needs of families for the long term care of their child through adolescence to adulthood and the specific services required at these times. Further research would also investigate the specific needs of people who have a condition that has not been diagnosed.**

The very specific needs of an individual with a rare disorder, their siblings, their mother and their father arose from the current research that explored the family support needs as a whole. The need to investigate the requirements of family members for the long term care of the individual with a rare disorder through adolescence to adulthood and the specific services required at these times, was also highlighted. Further in-depth research is recommended in order to study the experiences of each family member on a life-span perspective, and to make recommendations for future specialised social support.

The needs of both people who have a condition that has not been diagnosed and their families were not explored in this research and it is recommended that an exploratory investigation would be carried out in the context of the island of Ireland in order to appropriately plan for the provision of support.



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## Appendix 1: Organisations that provide social support and information services to people with a rare disorder and their families

The list below may not be exhaustive. The authors and RehabCare came across the organisations and services it contains during the process of our research. We have included those that have made reference to having specialised information or support with regard to rare disorders and direct support to families. The list does not include information about support groups specific to each rare disorder.

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### Ireland and the United Kingdom

#### Barretstown

[www.barretstown.org](http://www.barretstown.org)

Barretstown is a specially-designed camp, providing a programme of adventure, activities and fun — backed by the medical world — which helps children with serious illnesses regain their confidence and self-esteem. Barretstown also runs programmes for siblings and parents.

#### British Paediatric Surveillance Unit [BPSU]

<http://bpsu.inopsu.com>

The British Paediatric Surveillance Unit is a joint project of the Royal College of Paediatrics and Child Health, the Health Protection Agency, the Institute of Child Health, Health Protection Scotland and the Faculty of Paediatrics of the Royal College of Physicians of Ireland. The BPSU provides information to doctors and researchers about how many children in the UK and Ireland are affected by specific rare disorders. It also provides information collected about the disorders to aid the treatment of patients.

#### Climb

[www.climb.org.uk](http://www.climb.org.uk)

This is a UK-based charity for families affected by a metabolic disease. The organisation funds research and facilitates medical treatment, provides information and support for families and professionals and financially supports families to help meet the cost of equipment and other expenses. It also aims to educate professionals and others about metabolic diseases.

#### Contact a Family

[www.cafamily.org.uk](http://www.cafamily.org.uk)

Contact a Family provides advice, information and support to parents of disabled children. The organisation has a specific interest and expertise in rare disorders. Makingcontact.org puts families with disabled children in touch with each other. The Contact a Family Directory is a database of information about specific rare disorders. It includes entries for disorders which are life-limiting to, or life-threatening in, childhood or adolescence, chronic disorders affecting children and adults, disorders with paediatric and adult forms and adult onset disorders.

#### Genetic and Rare Disorders Organisation [GRDO]

[www.grdo.ie](http://www.grdo.ie)

GRDO acts as a national alliance for voluntary groups in Ireland representing the views and concerns of people affected by or at risk of developing genetic or rare disorders.

## **Genetic Interest Group [GIG]**

*www.gig.org.uk*

The Genetic Interest Group is a national alliance of patient organisations with a membership of more than 130 charities in the United Kingdom which support children, families and individuals affected by genetic disorders. It aims to promote awareness and understanding of genetic disorders so that high quality services for people affected by genetic conditions are developed and made available to all who need them.

## **National Centre for Medical Genetics**

*www.genetics.ie*

The National Centre for Medical Genetics provides a comprehensive service for all patients and families affected by or at risk of a genetic disorder. The website provides information about the centre, as well as information leaflets for affected families aimed at helping them to understand genetic testing. It also offers lists of support groups and links to other sources of information about genetic testing and rare disorders.

## **Syndromes without a Name [SWAN]**

*www.undiagnosed.org.uk*

S.W.A.N. aims to preserve and protect the health and promote the welfare of children who suffer from undiagnosed conditions. The organisation provides support and information to families where their child has no diagnosis. It also offers facilities to make links with other families.

## **The Jack and Jill Foundation**

*www.jackandjill.ie*

An Irish charity that provides early intervention home respite care to severely developmentally delayed children from birth to four years of age throughout the Republic of Ireland.

## **Unique**

*www.rarechromo.org/html/home.asp*

A UK charity, Unique is a source of information and support to families and individuals affected by any rare chromosome disorder and to the professionals who work with them. Unique aims to act as an international family support group, to promote awareness of rare chromosome disorders, to assist research and centralise information.

## **WellChild**

*www.raredisorder.org.uk*

A site developed by WellChild was set up in response "to the need for rare disorders in the UK to be given a higher profile". It includes an online community for professionals, a FAQ on rare disorders section, research and policy sections. The organisation is also carrying out a mapping project to establish a comprehensive qualitative and quantitative dataset on rare disorders in the UK.

*NB: RehabCare provides a dedicated residential service in the Republic of Ireland to young adults with Prader Willi Syndrome. For more information see [www.rehabcare.ie](http://www.rehabcare.ie)*

## Europe

### Agrenska

[www.agrenska.se](http://www.agrenska.se)

The Agrenska centre is a Swedish national competence centre for rare disorders. It provides programmes for children and young people with disabilities, their families and for professionals. Agrenska's family program is directed towards families who have children with rare disabilities and arranges about 20 family stays each year for people with a variety of diagnoses.

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### European Surveillance of Congenital Anomalies [EUROCAT]

[www.eurocat.ulster.ac.uk](http://www.eurocat.ulster.ac.uk)

EUROCAT is a European network of population-based registries for the epidemiologic surveillance of congenital anomalies.

### European Organisation for Rare Diseases [EURORDIS]

[www.eurordis.org](http://www.eurordis.org)

EURORDIS is a non-governmental patient-driven alliance of patient organisations and individuals active in the field of rare diseases, dedicated to improving the quality of life of all people living with rare diseases in Europe. It was founded in 1997 and is supported by its members and by the French Muscular Dystrophy Association (AFM), the European Commission, corporate foundations and the health industry.

### Frambu

[www.frambu.no](http://www.frambu.no)

Frambu is a national centre for rare disorders and disabilities in Norway. Its aim is to improve the quality of life for persons and families affected by one of approximately 40 rare, designated disorders. These include most progressive neurological disorders that have been diagnosed in Norway. Frambu's mission is to improve the quality of life for those with rare disorders and their families at home, at school at work and in the community so that they can better cope with their daily lives.

### Information Centre for Rare Diseases and Orphan Drugs [ICRDOD]

[www.raredis.org](http://www.raredis.org)

Information Centre for Rare Diseases and Orphan Drugs is a free educational and information service in Bulgarian and English languages, providing personalised replies to requests from patients, families and medical professionals.

### Orphanet

[www.orpha.net](http://www.orpha.net)

Orphanet is a database providing information on rare diseases and orphan drugs. This is accessible to both professional and families. It includes information specific to Ireland.

## **The Rare Disease Solidarity Project [Rapsody]**

*[www.rapsodyonline.eu](http://www.rapsodyonline.eu)*

The Rapsody project is led by EURORDIS and its objectives are to improve access to, and quality of, fundamental services primarily for patients, families and patients' organisations, but also for health professionals. The project was conceived in order to create new European networks for services. The Rapsody online project provides information about respite centres, therapeutic programmes and helplines for rare diseases across Europe.

## **Rarelink**

*[www.rarelink.org](http://www.rarelink.org)*

This site is a useful source for qualified information aimed at both patients and experts. It also includes information for families in Norway, Sweden, Finland and Denmark to make contact. The site is available in English.

## **International**

### **Canadian Organization for Rare Disorders [CORD]**

*[www.cord.ca](http://www.cord.ca)*

CORD is Canada's national network for organisations representing all those with rare disorders. CORD provides information on more than 6,000 rare disorders. The information is available in easy-to-comprehend language. Through a network system, CORD links individuals/families together with the same rare disorder.

### **National Organization for Rare Disorders [NORD]**

*[www.rarediseases.org](http://www.rarediseases.org)*

NORD is a non-profit, voluntary health agency serving rare disease patients and their families in America. NORD provides information about diseases, referrals to patient organizations, research grants and fellowships, advocacy for the rare-disease community. It also offers Medication Assistance Programs that help patients obtain certain drugs they could not otherwise afford. The site contains access to support sites, general information, developmental drugs and a database of medical reports on more than 1,100 rare diseases.

### **New Zealand Organisation for Rare Diseases**

*[www.nzord.org.nz](http://www.nzord.org.nz)*

A charitable organisation set up in 2000. Its website gives details of information portals for rare diseases, information on support groups in New Zealand, disability resources and research. The organisation also monitors rare disease issues and policy matters and builds partnerships between patients and their families, support groups, clinicians, researchers, policy-makers and industry.

### **NIH Genetic and Rare Diseases Information Center [GARD]**

*<http://rarediseases.info.nih.gov>*

United States-based portal to rare disease information and research including information on Patient Advocacy Groups, Genetics information and services, research and clinical trials, patients travel and accommodation, reports and publications, and rare disease news.

## **Office of Rare Diseases [ORD]**

*<http://rarediseases.info.nih.gov>*

The Office of Rare Diseases is an American government office within the National Institutes of Health. The site has information on more than 6,000 rare diseases, including links to current research, publications from scientific and medical journals, completed research, ongoing studies, and patient support groups. The site provides free access and is user-friendly. The ORD home page provides easy-to-follow links and more detailed descriptions.

## Appendix 2: Selection process for choosing 10 rare disorders to study in research report

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*The research team consulted with two health professionals in the field of rare disorders in Ireland to devise a criteria and selection process for the rare disorders to study in this report. The criteria was finalised as follows:*

The criteria listed below were based on the list of disorders supported by Frambu, a recognised model of best practice for the support of individuals and families affected by rare disorders. The rare disorders supported by Frambu were selected in consultation with the Ministry of Health in Norway and identified as rare disorders requiring social support for the individual and the family.

### **1. All rare disorders chosen for the research were congenital.**

NB: Congenital disorders are present at birth but symptoms may not present until later in life. Therefore the criterion below (No. 2) was taken into consideration.

### **2. Symptoms were present from birth or early childhood.**

### **3. The study included a cross-section of people affected by rare disorders with regard to the levels of support they currently receive in Ireland:**

- Six rare disorders of varied prevalence, all with established support groups (of varying sizes) in Ireland.
- Two rare disorders, less common, with a lower prevalence and with parent contact names in Ireland as opposed to an established support group.
- Two rare disorders of very low prevalence (5 cases or less in Ireland) at time of study — no support group or parent contact in Ireland.

The final list of rare disorders where families that were approached to take part in the research consisted of:

- Cri du Chat
- Cornelia de Lange
- Fragile X
- Neurofibromatosis (Type 1 and 2)
- Prader Willi Syndrome
- Rett Syndrome
- Sotos Syndrome
- Williams Syndrome
- Two other rare disorders of very low prevalence (disorder names cannot be published in order to ensure confidentiality of the participants in the research)

## Appendix 3: Press release about the research

### GPs to Assist in New Research on Rare Disorders in Ireland

A research project by RehabCare, the health and social care division of Rehab Group, is set to examine the support needs of people affected by rare disorders, across the island of Ireland including Prader Willi Syndrome, Cornelia de Lange, Rett Syndrome and Fragile X. In 2003 RehabCare opened Ireland's only residential service for young adults with Prader Willi Syndrome.

Funded by the Health Research Board, the research will initially include consultation with a randomly-selected sample of approximately 700 GPs in the Republic and 300 GPs in Northern Ireland. In the coming months, GPs will receive a short questionnaire seeking their views on the provision of information on rare disorders and on the specific support services for patients and their families.

Other rare disorders to be examined as part of the research, although not an exhaustive list, are: Williams Syndrome; Cri du Chat; Neurofibromatosis; and Sotos Syndrome.

In addition to consulting with GPs on both sides of the border, RehabCare researchers will also speak individually with other professionals, including geneticists. A number of focus group sessions with individuals with rare disorders and their families will also take place early in 2008.

According to Caroline Hart, Research Officer, RehabCare, the study will benchmark the current provision of information and services for individuals with rare disorders, their families, and professionals working in the field and so help inform future service development. The information compiled will form the basis of an information resource on rare disorders.

The research follows a 2002 consultation between RehabCare and the Royal College of Physicians into the support needs of all stakeholders affected by rare disorders. This revealed that information on rare disorders was difficult to access for both professionals and families alike. Issues around mental health, general health and stress, were also cited as being particularly prevalent in families where a child has a rare disorder.

Welcoming the research, Professor Hoey, Head of the Department of Paediatrics, Trinity College, Dublin and Consultant Paediatric Endocrinologist with the National Children's Hospital, Tallaght, said,

"Previous studies have shown that patients with rare disorders, their families, and indeed medical professionals, have faced huge difficulties in the past getting relevant and timely information. This research by RehabCare is to be welcomed as an important step in filling this vacuum."

Medical professionals or anyone with experiences of rare disorders who are interested in contributing to the research should contact [name and contact details]

Ends:

For further information [name and contact details]

## Appendix 4: Letter to parents

RE: Rare Disorders on the Island of Ireland:  
An investigation of the support needs of families



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Dear

Thank you for expressing an interest in taking part in the research about rare disorders in Ireland. Your views and opinions are extremely important and RehabCare are very grateful you are taking the time to talk to us about your experiences.

The research we are carrying out is funded by the Health Research Board and is explained in detail in the enclosed leaflet. RehabCare have recognised a need for support for people with rare disorders and their families from as far back as the mid-1990s. The organisation have been working with individuals with rare disorders and their parents to look at their support needs and how best these needs can be met. We have also carried out a survey with Paediatricians on the topic and have continued to lobby for funding to progress the work on rare disorders. We are hoping, through this current research, to provide recommendations for the development of support on the island of Ireland for people with rare disorders and their families.

RehabCare look forward to the potential of becoming a resource for information for both families and professionals with regard to specific rare disorders. We recognise that families affected by different rare disorders have very common support needs, and as stated recently at a European conference for rare disorders "even though the diseases are rare, rare disease patients are many".

If you decide to take part in the research, to enable you to put your views across we will arrange for you to take part in an interview. We will discuss your support needs, the support needs of your child with a rare disorder and the support needs of any siblings. You will have the opportunity to discuss the challenges you face as a parent of a child or adult with a rare disorder. The discussion will not be longer than an hour to an hour and a half.

[Include if their child with a rare disorder under 18 is taking part]. If your child who has a rare disorder decides to take part in the research, I will also be discussing his/her support needs and what he/she feels would be important for him/her and the whole family with regard to support.

Your participation in this consultation is voluntary and you have the right to withdraw at any time during the research process. Please do note that anything you or your children say at the one-to-one will be kept confidential; no names will appear on any publication or information seen by anyone other than me, the researcher. The results of the research will be published as a report to be presented to funders (Health Research Board) and used as an evidence base for future support.

We will also give you and your children an opportunity to discuss any queries or concerns about the research process on the day. If you have any queries before that about any of the research and/or your involvement please don't hesitate to contact me, Researcher at RehabCare [Researcher name and contact details]

If you would like to take part, please fill out the enclosed consent and assent forms. There is one for yourself, and if your children are under 18 you are requested to also sign theirs. Please fill out a separate form for each family member taking part. As soon as we receive the forms we will be contacting you by telephone to discuss convenient times and places. Please note, we will arrange a location and time that is convenient for you and you will be reimbursed for any expenses incurred in participation, including travel expenses. If I have not heard from you within two weeks of you receiving this letter I will assume you do not want to take part in the study.

Yours sincerely

[Researcher]



## Appendix 5: Family consent form

Project Title: Rare Disorders on the Island of Ireland:  
Investigation into the support needs of families



[Researcher Name]:

### Consent Form

I \_\_\_\_\_ (name of participant) have read and understand the information about the research on rare disorders, and give my consent to participate in the consultation. The consultation will involve me talking to someone from RehabCare about what it is like to be affected by a rare disorder and what supports I feel are needed for families like ourselves. I understand that taking part is totally voluntary, that I can withdraw at any time, and this will have no negative consequences. I also understand that all the information I provide will be kept strictly confidential within the research team, and, when it is written up as a report, all identifying information will have been removed. I know that if I have any questions I can call the researcher [Researcher name and contact details]

Name (please print): \_\_\_\_\_

Signature: \_\_\_\_\_

Date: \_\_\_\_\_

Contact telephone number: \_\_\_\_\_

# Appendix 6: Pictorial consent form

## Pictorial Consent Form



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Project Title: Rare Disorders on the Island of Ireland:  
Investigation into the support needs of families  
Researcher: [researcher and organisation name]

This research involves talking to someone from RehabCare about what it is like living with a rare disorder and what kind of help I feel me my family and I would like. This would take about an hour and the researcher would meet me where and when is easy for me.

Name of Participant: \_\_\_\_\_

Signature of Participant (if appropriate): \_\_\_\_\_

Name of person explaining consent: \_\_\_\_\_

Signature of person explaining consent: \_\_\_\_\_

I have looked at the info about the research on rare disorders

I know that taking part is my choice

I know that I can leave the project if I want to

I know that all my information will be kept private and confidential by RehabCare staff.

I know that name my will not be used in the report.

If I have any questions I can call:  
[researcher name and contact details]

I agree to take part

I do not agree to take part

## Appendix 7: Interview schedule for parents

### Theme 1: Information Needs

#### Type of Information

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Do you feel your information needs are different or similar to the information needs of parents of children with more common disabilities?

What kind of information do you feel it is important for you as parents to receive? (see below prompts)

- Symptoms of your child's condition
- Development and future of your child
- Results of recent genetic research including clinical trials
- Local support groups
- Entitlements
- Information about your rights
- Other

#### Searching for and Accessing Information

If you have searched for information yourself, where did you search?

- Internet
- Local Library
- Local GP clinic
- Asked family and friends
- Other

If you search on the internet what sites do you access, do you find them useful?

In your search, what kind of information did you find? Was this useful?

#### Information from health professionals

What is your experience of accessing/receiving information about your child's disorder from health professionals?

Did you receive information on diagnosis about support groups from your GP, Paediatrician, anyone else?

### Theme 2: Practical Support Needs

#### Financial Considerations

##### Employment

Are you aware of where to access up-to-date information financial advice and/or help?

- Access to health professionals and other help
- What is your experience of accessing the services of relevant health professionals e.g. physiotherapy, speech therapy etc

Who do you go to if you need any advice or if you have any issues with wheelchairs / transport / child care / adapted devices and other equipment?

How satisfied are you with the response you get in requesting advice on these practical matters? If not, why not?

84 | What do you feel are the similarities or differences in accessing information and advice on these practical matters for rare disorder as opposed to more common disorders?

What do you feel would help when you are attempting to access health professionals or requesting advice on practical matters?

### **Theme 3: Social and Emotional Support Needs**

#### **You as a Parent**

Who do you talk to when you need it? / Why do you choose that person? / How does that person support you emotionally?

Are you in contact with anyone else whose child has a disability? Does that child have the same disorder as your son / daughter? What kind of things do you talk about with that parent?

#### **Family Relations**

What, in your view, is the experience of your other children in having a brother or sister with a rare disorder? Do you know who do they talk to?

What kind of support do you think your other children would like / need

Do you think your other children have support needs that are different or similar to the support needs of siblings of people with more common disabilities?

Can you think how marital relationships could be affected by having a child with a rare disorder, as opposed to more common disabilities – positively / negatively?

#### **Positive aspects of having a child with a rare disorder**

What are the positive things for you about having a child with a rare disorder?

What do you think are the positives for your family as a whole?

### **Theme 4: Common Support Needs**

As a group with children of differing disorders, what do you feel are your common support needs?

What are the differences in your support needs as specific to each disorder?

## Appendix 8: Interview schedule — specialist health professionals

### Theme 1: Information needs

- Searching for and accessing information
- Type of Information
- Information from other health professionals
- Information needs at diagnosis

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### Theme 2: Practical support needs

- Financial Considerations
- Access to health professional services for families
- Proposed services

### Theme 3: Social and emotional support needs

- Individuals with rare disorders
- Parents
- Siblings
- Family Relations
- Information needs of families at diagnosis
- Positive aspects of having a child with a rare disorder

### Theme 4: Common support needs

- Differing rare disorders – common support needs?
- Needs of families affected by rare disorders as opposed to more common disabilities.

## Appendix 9: Cover letter to general practitioners



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Dear General Practitioner,

Funded by the Health Research Board, RehabCare are currently conducting research into rare disorders in Ireland. The project involves an exploration of existing need for specific support services for individuals with rare disorders and their families. The aim is to establish an accessible working store of information and to make valuable recommendations for support to meet the needs of individuals with rare disorders and their families.

RehabCare are currently in the initial stages of the research; reviewing the literature on the common support needs of individuals with rare disorders and their families, collating information on support groups in Ireland available to those affected by rare disorders and exploring existing information sources in Ireland for rare disorders. The next stage will be to consult directly with individuals with rare disorders, their families and professionals.

As part of the first stage of this project we are requesting the views of a sample of General Practitioners in Ireland by means of a confidential survey. The survey explores GP's information needs in relation to rare disorders, information and support needs for families, and the questionnaire also requests opinions on proposed types of support for families affected by rare disorders.

RehabCare would be extremely grateful if you could take five minutes to complete the attached questionnaire and return to RehabCare by Friday February 1st using the enclosed stamped-addressed envelope. Your answers are anonymous and will only be used for the purposes of this piece of research.

If you have any queries regarding the questionnaire or would like more details about the project please don't hesitate to contact [Researcher name and contact details]

Yours Sincerely

[Researcher]

# Appendix 10: Questionnaire for general practitioners

## Questionnaire for GPs



### Background Details

County of Practice: \_\_\_\_\_

Nature of Practice (please tick):                      Group                       Individual

Other (please specify): \_\_\_\_\_

### 1. Please state below if your patients are mainly from an urban catchment, a rural catchment or mixed urban and rural (please tick box)

Urban Catchment Population

Rural Catchment Population

Mixed Urban and Rural Catchment Population

### 2. Information about rare disorders

Do you experience difficulty providing relevant information to individuals with a rare disorder and their families about their specific rare disorder e.g. Medical or service information or info about support groups?

Yes                       No

### 2a If yes, why do you experience difficulty? Please tick the relevant box(es) below:

The information is available but it is not up-to-date

The information is available but it is not in an accessible format that is appropriate for families

The information is available but is not in an Irish context

Due to workload pressures I don't have time to look

I don't know where to look

The information is not available at all

Other (please specify):

-----  
-----  
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### 3. In your experience, do you consider that having a rare disorder rather than a more common disability may give rise to any additional, unique problems within a family e.g. family conflict, self-esteem issues, substance abuse etc?

Yes                       No

**3a. If Yes what problems do you feel rare disorders give rise to?**

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**4. Where would you usually source your information on rare disorders?**

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**5. Do you feel that you had sufficient professional training to help you inform on and adequately support people with rare disorders?**

Yes  No

**5a. If yes can you please specify key areas of training:**

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**6. Finally, is there anything you feel would help general practitioners to deal with patients with rare disorders if it was made available to them e.g. Post graduate training, a rare disorders information service, a website etc.**

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If you would like to receive further information on rare disorders from RehabCare please leave your contact details below and if you would like to take part in a more in-depth interview or focus group as part of the research then please do not hesitate to contact us.

Thank you for taking part in this survey. If you have any queries please contact  
[Researcher name and contact details]



# Appendix 11 – Information leaflet about the research



**RehabCare's Rare Disorders Project**

RehabCare is the health and social care division of the Rehab Group. Established in 1996, the organisation provides a wide range of community support services to marginalised groups and people with a disability, their families, carers and service providers, assisting them in developing to their full potential and obtaining the appropriate supports to allow them to live as independently as possible and interact in their community. Individuals with rare disorders come into contact with RehabCare on a day-to-day basis, and RehabCare seeks to support their specific needs. An example of this is the support service for people with Prader Willi Syndrome (PWS) developed by RehabCare.

In 2005 RehabCare was delighted to receive funding from the Health Research Board to carry out research that will examine rare disorders on the island of Ireland. This research will investigate their prevalence and incidence, and explore existing need for specific support services for individuals with rare disorders and their families.

**What is a Rare Disorder?**

The European Union defines a rare disease as 'a disease affecting 5 or less per 10,000 population'. It is estimated there are currently between 5,000 and 8,000 distinct rare disorders, and they affect between 6% and 8% of the population in the European Union. About 30 million people have a rare disease in 25 EU countries. *"Even though the diseases are rare, rare diseases patients are many"* (ERCD, 2005).

*"There are not just medical needs in living with a child with a rare disorder, there are practical and emotional needs..."<sup>1</sup>*

*"We desperately needed more information, which neither the GP nor our health visitor could offer, both grieving and confused we did not know who to ask..."<sup>2</sup>*

**Background to the Research**

RehabCare's interest in the topic of rare disorders originated in response to a recommendation in *A Strategy for Equality, the Report of the Commission on the Status of People with Disabilities* (1996) specifying the need for the establishment of a national centre for rare disabilities in Ireland.

Since 2000 RehabCare has been working with parents and individuals with rare disorders to look at their support needs and how best these needs can be met. One of the outcomes of this collaborative work was the development of support services for young adults with PWS.

In 2002, RehabCare, in partnership with the Faculty of Paediatricians in the Royal College of Physicians of Ireland, carried out a survey among paediatricians practicing in Ireland to obtain professional views of those who work with individuals with rare disorders on existing support services. This preliminary work found that access to information on rare disorders was seen as challenging for both professionals and families alike. Mental health problems, general health problems and stress were cited as being prevalent in families where a child has a rare disorder. The current research, as outlined in this leaflet, is building upon these findings.

**Aims of the Project**

- To establish a picture of the prevalence and incidence of rare disorders on the island of Ireland
- To become an information point for individuals with specific rare disorders, their families and professionals working in the field
- To develop links with other European and international service providers and to review international models of best practice in relation to support services for individuals with rare disorders
- To make valuable recommendations for the development of a holistic all-island support service to meet the needs of individuals with rare disorders and their families

*"I think it would be wonderful to have a source of information and support to families, professionals."<sup>3</sup>*

1 & 2 Contact a Family (NI), Family Information Group, Parent Contributors, 2002. Parents' Stories, Derry: Guildhall Press  
3 RehabCare & Royal College of Physicians of Ireland, 2002. Results of Research into the Support Needs of all Stakeholders Affected by Rare Disorders Dublin: RehabCare

## Appendix 12: Information about the research for an individual with a learning disability

### Rare Disorders



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#### What is a Rare Disorder?

A rare disorder is a disability that not many people have. If there are 10,000 people only 5 of those people will have a rare disorder.

#### What are RehabCare doing to help people with rare disorders?

RehabCare are asking people with rare disorders and their families what kind of everyday help they want or need.

#### What you can do

If you decide you would like to talk to someone about what it is like to have a rare disorder, someone from RehabCare will come to your house and talk to you.

We will make a time and place that is easy for you.

#### Your opinion is very important to RehabCare!

RehabCare will put together everything you and other people say about living with a rare disorder into a report. We will use the report to help make services better for people with rare disorders and their families.

Some other things to know before deciding if you want to take part:

The researcher is the only person who will know your name— Your name won't be put on the report or anything that anyone other than the researcher will see.

If you decide at any time (before, during or after the research) that you don't want to be part of this research you don't have to be anymore.

Any questions call [Researcher name and contact details]

## Notes

# Notes



*For more information, please contact us.*

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