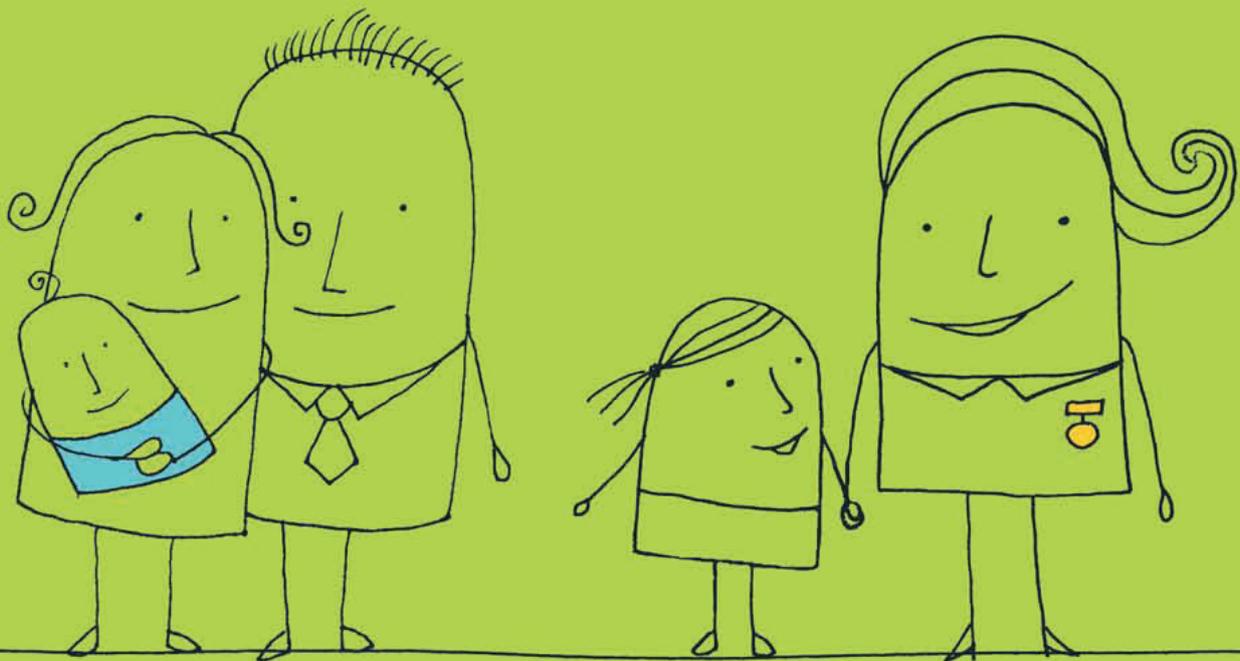


## Patient and Client Council

# Experience of Diagnosis

## Views of patients and carers of diagnosis of rare disease in Northern Ireland

February 2012



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# Foreword

Dear Reader,

People who are affected by a “rare disease” in Northern Ireland tell us that they find their experience to be a very lonely one. They are often misunderstood, frequently misdiagnosed and have little opportunity to be involved in decisions affecting their health care or to have their voice heard and acted upon by decision makers.

This report focuses on the experiences of people with a rare disease, particularly the experience of the time from the onset of the symptoms to the diagnosis of the disease.

The voice of people with rare diseases and their carers can be heard clearly throughout this report. It describes not only their frustrations and difficulties during the process of diagnosis, but also the high value they place on the treatment, care and support that they receive – particularly from the medical profession and from voluntary and community groups.

In welcoming this report, I also welcome the establishment of the Northern Ireland Rare Disease Partnership. This is a new partnership between people with rare diseases, their carers, voluntary groups and the clinical professionals providing treatment and care. The Northern Ireland Rare Disease Partnership is a commitment to work together to improve services for people affected by rare diseases in Northern Ireland.

The Patient and Client Council has been instrumental in the setting up of this partnership over the last two years and will continue to offer its support for a further 12 months to allow the valuable work of this group to be established.

In the recommendations we ask our Health and Social Care colleagues to make a commitment to this important work.

I welcome this report and I endorse its recommendations. I look forward to working with the Northern Ireland Rare Disease Partnership and I wish it success in the future.



Maeve Hully

**Chief Executive of the Patient and Client Council**

## Summary

We know from our work that most people who are affected by a rare disease feel very isolated and are unaware of others similarly affected. They feel that it is difficult to have their voice heard and acted upon in health and social care.

The purpose of this report is to help ensure that the voice of people who suffer from a rare disease and their carers is heard and acted upon by decision makers in health and social care.

The report:

- presents the findings of a survey of 132 people (both patients and carers) who have first hand experience of the diagnosis of a rare disease;
- defines a rare disease;
- highlights the need for the development and implementation of a regional strategy and commissioning plan for rare diseases; and
- outlines the role of the Patient and Client Council in supporting the development of a Northern Ireland Rare Disease Partnership.

The results of the survey clearly demonstrate the difficulties and frustrations faced by people affected by a rare disease. Many people go undiagnosed or misdiagnosed for long periods of time and find that their condition is misunderstood. In some cases this leads to inappropriate treatment and care.

Once diagnosed, patients often experience considerable delays while waiting on the commencement of services, particularly those provided by allied health professions

Many carers provide care for over 20 hours per day and experience little support from health and social care services.

By its very nature the incidence of each rare disease is very low, but collectively the number of people affected by rare disease is significant. The considerable response to this survey and the need to develop a partnership forum indicates a high level of interest and commitment to ongoing involvement among people with rare diseases and their carers.

There is limited information and advice for patients and carers.

This report makes a number of recommendations for the Department of Health, Social Services and Public Safety (the Department), the Health and Social Care Board and the Public Health Agency.

## **1.0. Background and Purpose**

### **1.1 The Patient and Client Council**

The Patient and Client Council was established to provide a powerful independent voice for people.

The Patient and Client Council has four main duties. They are to:

- listen and act on people's views;
- encourage people to get involved;
- help people make a complaint;
- promote advice and information.

In 2010, the Patient and Client Council recognised the need to support the voice of people who are affected by rare diseases in Northern Ireland and set itself an objective to develop a network for people affected by rare diseases in Northern Ireland. From this network, the Northern Ireland Rare Disease Partnership evolved and is expected to be formally established in February 2012.

This initiative was given further impetus by a letter sent to the Patient and Client Council by the Huntingdon's Disease Association Northern Ireland asking for assistance in ensuring adequate service provision for this rare disease.

The Patient and Client Council Board recognised that people with rare diseases were at high risk of exclusion from involvement in the planning, design and delivery of health and social care services. This was due to the low incidence of the diseases and their impact on the capacity of patients and carers for participation.

### **1.2 Definition of a Rare Disease<sup>1</sup>**

A rare disease is defined by the European Union as one that affects less than 5 in 10,000 of the general population. There are between 6,000 and 8,000 known rare diseases. Around five new rare diseases are described in medical literature each week.

80% of rare diseases have a genetic component. Rare diseases are often chronic and life threatening. 75% of rare diseases affect children. 30% of patients with rare diseases die before their fifth birthday.

In the UK, a single rare disease may affect up to 30,000 people. The vast majority of rare diseases will affect far fewer than this – sometimes a single person in the whole of the UK will be affected.

Taken collectively, rare diseases are not rare. 1 in 17 people will be affected by a rare disease at some point in their lives.

### 1.3 A Strategy for Rare Diseases

The United Kingdom government adopted a European Council recommendation<sup>2</sup> of June 2009 on an action in the field of rare diseases.

Since then work has been ongoing with the UK Government and the Devolved Administrations to develop a plan for rare disease by 2013 in line with this recommendation. A national consultation on a strategy for rare disease is expected during 2012.

**There is a need to ensure that a strategy is developed and implemented in Northern Ireland and that the commissioners of health and social care have a plan for provision of health and social care for people affected by a rare disease in Northern Ireland.**

### 1.4 The Northern Ireland Rare Disease Partnership

The Patient and Client Council is supporting the development of the Northern Ireland Rare Disease Partnership which will:

- Raise awareness of rare diseases in Northern Ireland
- Promote education about rare diseases among clinical professionals
- Provide an independent voice for people affected by rare diseases in Northern Ireland
- Work in partnership with all those involved in the design and delivery of services for people with rare diseases in Northern Ireland

Membership of the Northern Ireland Rare Disease Partnership includes people with rare diseases, carers, representatives of organisations for people with rare diseases and professionals involved with the treatment and care of people with rare diseases.

<sup>1</sup> The data given in these paragraphs is taken from the Rare Disease UK website. ([www.rarediseaseuk.org.uk](http://www.rarediseaseuk.org.uk))

<sup>2</sup> European Council Recommendation (2009/C 151/02) Official Journal of the European Union. 3.7.2009.

## 2.0 Our Approach

In spite of the enormously diverse and complex impact of rare diseases on patients, carers and support groups who were involved in the development of the Northern Ireland Rare Disease Partnership identified six areas of shared concern. One of the key areas of concern identified related to the experience between the onset of symptoms and diagnosis.

To capture the views of patients and their carers a questionnaire was developed. A copy of the questionnaire used is attached in **Appendix 1** of this report. This report summarises the experiences of 132 people around the time of being diagnosed with a rare disease.

The questionnaire was developed with reference to a survey carried out by Rare Disease UK<sup>1</sup> and in consultation with members of the Northern Ireland Rare Disease Partnership and the Patient and Client Council Board sponsor for this project, Dr Sheila Kelly.

The questionnaire was approved by the Research Advisory Group of the Patient and Client Council.

The questionnaire was made available online through Survey Monkey. ([www.surveymonkey.net](http://www.surveymonkey.net)). The survey was then publicised on the Patient and Client Council website and on the websites of member organisations of the Northern Ireland Rare Disease Partnership. Flyers giving information on the survey were distributed to Consultant Neurologists and to a Consultant in Medical Genetics at the Belfast City Hospital in order that the survey could be publicised through outpatient clinics.

Contact names and numbers were given for two Patient and Client Council officers so that respondents could ask questions, get help to complete the questionnaire, or complete the questionnaire over the telephone if necessary.

A communications strategy was drawn up so that potential respondents could be made aware of the survey through the media. An article on this work was published in the Belfast Telegraph.

The survey was available online for completion between November 11<sup>th</sup> 2011 and December 22 2011.

<sup>1</sup>(Limb, L. Nutt, S. Sen, A. (2010). *Experiences of Rare Diseases: An Insight from Patients and Families*. London: Rare Disease UK (Available at: <http://www.raredisease.org.uk/documents/RDUK-Family-Report.pdf>)

### 3.0 Our Findings

132 individuals (both patients and carers) completed this questionnaire. The notes which follow provide some necessary explanation regarding the analysis and presentation of our findings:

- *People with rare disease and carers for people with rare disease responded to this questionnaire. Answers to questions about the rare disease have been given, therefore, both by patients and by carers on behalf of the person with the rare disease.*
- *There were a total of 50 questions on this questionnaire. Not all of the questions have been answered by every respondent. Tables and charts appearing throughout this document relate to the number of respondents to the specific question under discussion. Where a statement of percentage relates to the total number of respondents (132) this is clearly indicated. Appendix 3 of this report details the number of responses to each question.*
- *There has been no editing or exclusion of any response given with the exception of Q.48 where respondent experiences have been summarised and themes highlighted. Quotes from respondents are clearly indicated and verbatim.*

#### 3.1 Respondents

132 people responded to the questionnaire. 49% were people with rare diseases and 51% were carers for people with rare diseases.

57% of the people with rare disease were female and 43% were male. The age distribution of the people with a rare disease is shown in **Table 1** below.

**Table 1 – Age of the Person with a Rare Disease**

Age Range	Number of People	Percentage of Respondents
0-5	18	14%
6-15	21	16%
16-25	7	6%
26-45	29	22%
46-65	42	32%
66-75	10	8%
76+	3	2%
TOTAL	130	100%

The youngest person for whom a response was provided was 7 months old and the oldest person was 91 years old. In this sample, 30% of people with a rare disease were aged 15 and under.

### 3.2 Diagnosis

Respondents were asked a number of questions on aspects of their experience between the onset of symptoms and diagnosis. These responses appear in the following paragraphs.

#### Range of Rare Diseases

66 rare diseases were named by respondents. **Appendix 2** shows a full list of the rare diseases named and the number of incidences.

7 respondents stated that they had not received a diagnosis. 3 of these were awaiting the results of tests. 2 of the remainder stated that there was no test for their condition; another that they were “tired of going to the doctors”.

#### Age at Diagnosis

Respondents were asked the age of the person when first diagnosed with a rare disease. **Table 2** below illustrates the responses.

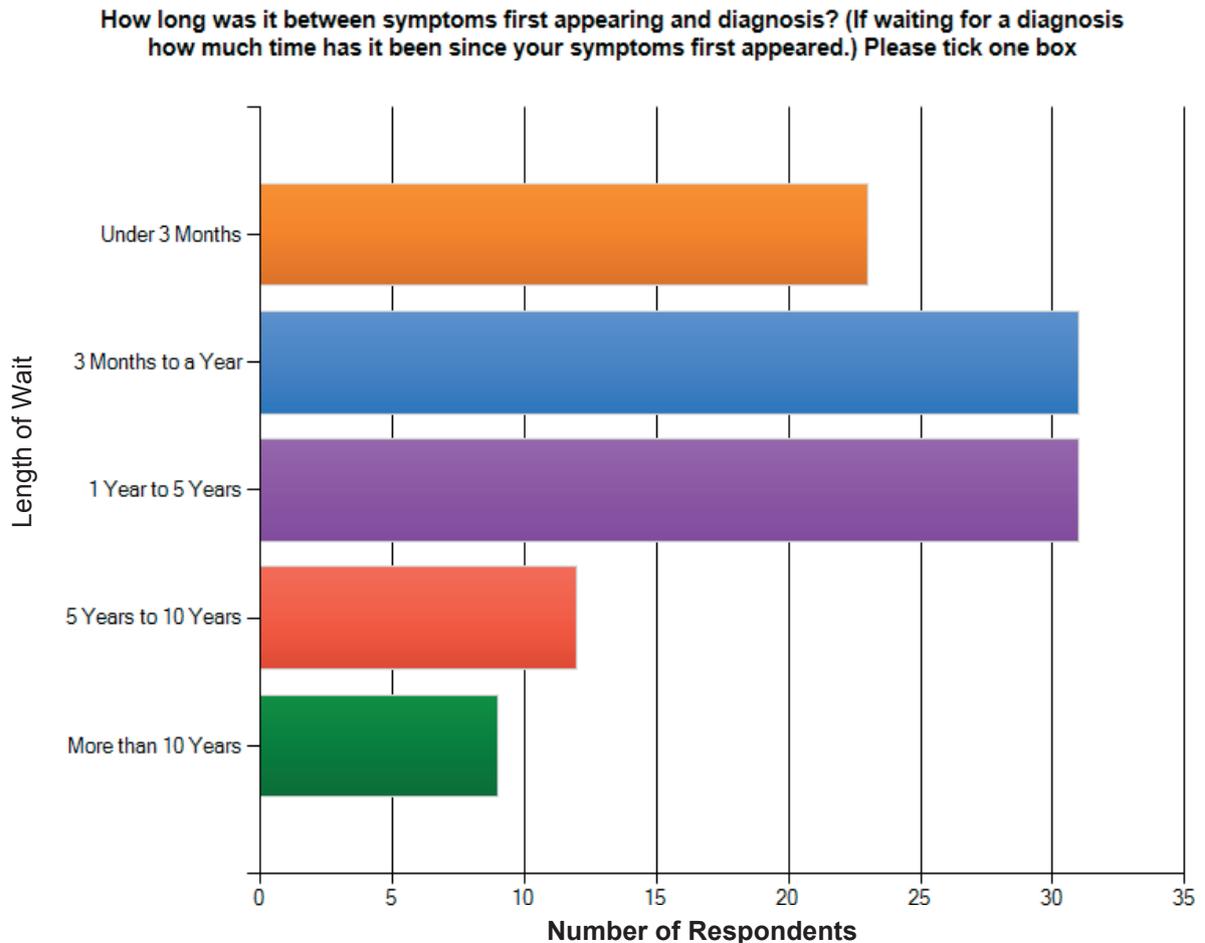
**Table 2 – Age of Patient at the Time of Diagnosis of a Rare Disease**

Age at Diagnosis	Number of Respondents	Percentage of Respondents
Before birth	4	4%
From birth	17	15%
Up to 3 months Old	9	8%
4 to 6 months	1	2%
7 months to 1 year	7	6%
2-5 years	5	5%
6-10 years	2	2%
11-15 years	2	2%
16-20 years	3	3%
21-40 years	21	19%
41-60 years	21	19%
61-80 years	8	7%
81+	2	2%
Not Diagnosed	7	6%
<b>TOTAL</b>	<b>109</b>	<b>100%</b>

## Waiting Times for Diagnosis

**Figure 1** below illustrates the length of time between the onset of symptoms and the diagnosis of a rare disease. Of the 106 respondents, 31 (29%) of respondents had waited between one and five years for a diagnosis.

**Figure 1 – Waiting Time between Onset of Symptoms and Diagnosis**



## Noticing symptoms

Respondents were asked to state who had first noticed the symptoms of their rare disease. The responses were fairly evenly divided across three groups:

1. The person with the rare disease (37%)
2. The carer for the person with the rare disease (33%)
3. Clinical professionals (30%)

## **Emergency Admissions**

Respondents were asked if they had any emergency admissions between the onset of symptoms and diagnosis. 40% stated they had 1 or 2 emergency admissions, 17% had between 3 and 8 emergency admissions. 17% had between 8 and 10 emergency admissions. 7% had more than 10 emergency admissions. 9 people gave a verbal answer; phrases such as “too many to count” characterised 8 of these responses.

## **Travel outside Northern Ireland**

15 respondents stated that they had travelled outside Northern Ireland for treatment and care between the onset of symptoms and diagnosis. 11 people named 9 places to which they had travelled at least once. These were London (Great Ormond Street); Edinburgh; Cambridge (Addenbrookes); Manchester; Leicester; Birmingham; Dublin; Hereford and the USA. 2 people stated they had travelled but did not give the destination. 1 person was offered treatment outside Northern Ireland but declined.

## **Source of referral for treatment and care outside Northern Ireland**

Of those referrals for treatment outside Northern Ireland, the majority (55%) were referred by a Medical Consultant, 25% of respondents had self referred and 15% were referred by a GP.

## **Private Treatment and Care**

25 people (19%) of respondents had arranged private treatment and care between the onset of symptoms and diagnosis. Private treatment was most commonly (11 of 25 people) used for diagnostic tests and scans. A further 9 people used a term like “consultation” or “specialist advice”.

## **Misdiagnosis**

45 people (34%) of respondents stated that their rare disease had been misdiagnosed before a correct diagnosis had been reached. In total, 30 people stated a number of misdiagnoses. 9 had been misdiagnosed once, 13 people twice or three times and 6 people more than three times.

## **Inappropriate Treatment**

26 people (20%) of respondents stated that as a result of misdiagnosis they had received treatment and care that was not appropriate for their rare disease. The most common treatment people stated they had received inappropriately was prescribed medication. 15 people stated that they had been prescribed medication unsuited to the disease. This included 5 people who were prescribed anti-depressants.

## Number of Doctors Seen Between Onset of Symptoms and Diagnosis

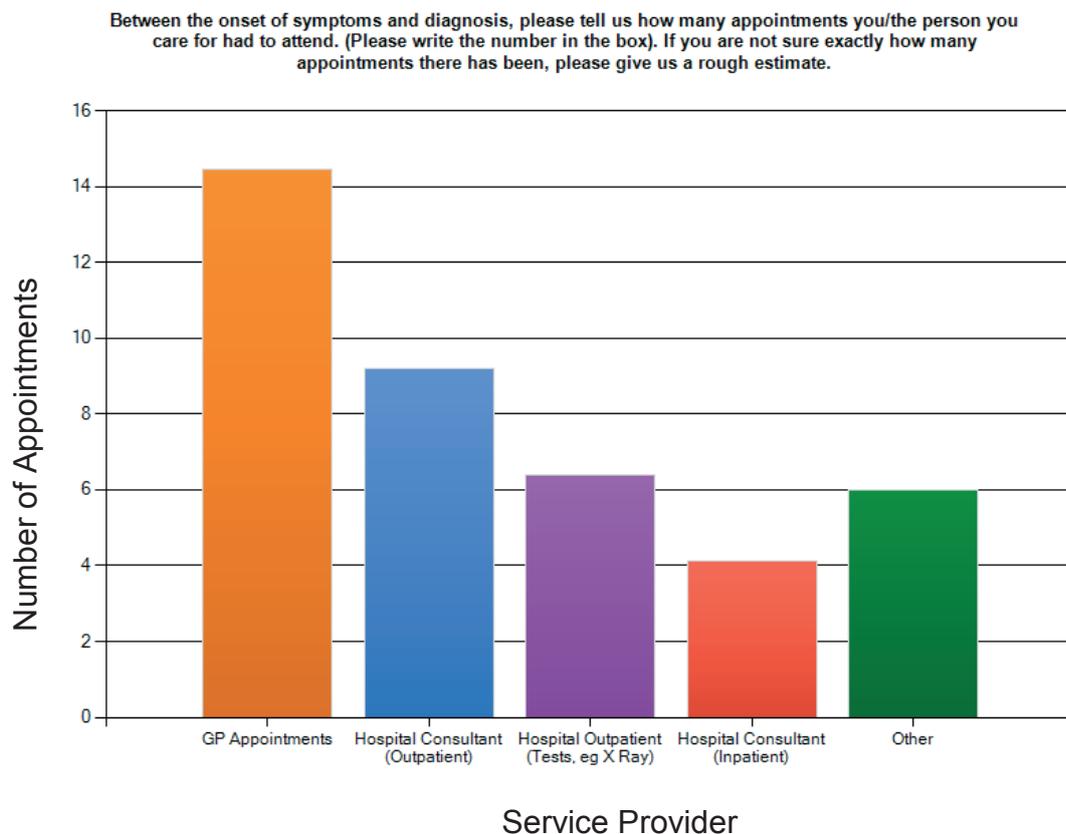
Respondents were asked if they saw more than one Doctor between the onset of symptoms and diagnosis. 63% of respondents stated that they had seen Doctors from different specialties.

Where respondents had seen more than one Doctor, they were asked to state how many they had seen. 43% stated that they had seen between 2 and 5 different Doctors. 15% stated that they saw between 5 and 10 Doctors and 7% stated that they had seen more than 10 Doctors.

## Number of Appointments between Onset of Symptoms and Diagnosis

Respondents were asked to indicate how many appointments they had attended between the onset of symptoms and diagnosis. **Figure 2** below illustrates the average number of appointments by respondents in different areas of service delivery.

**Figure 2 – Average Number of Appointments Attended between Onset of Symptoms and Diagnosis**



## Sources of Information and Support between Onset of Symptoms and Diagnosis

Respondents were asked to indicate all sources from which they had received information and support between onset of symptoms and diagnosis. **Table 3** below illustrates the responses to this question.

**Table 3 – Sources of Information and Support between Onset of Symptoms and Diagnosis**

**NOTE:**

- Respondents were asked to tick all that applied from a range of options

Source of Support	Number of Responses
Hospital Consultant	58
General Practitioner	49
Internet	41
Voluntary Group/Charity	33
Specialist Nurse	11
Social Worker	11
Health Visitor	9
Counsellor	6
District Nurse	4
Practice Nurse	4

17 further responses under “Other” were received. These included 4 which stated that no support had been received and 3 who named “family” as a source of support.

### Information and Advice

Respondents were asked to indicate the most important source of information and advice to them between the onset of symptoms and diagnosis. **Table 4** illustrates the responses received.

**Table 4 – Most Important Sources of Information and Advice between the Onset of Symptoms and Diagnosis**

Source of Information and Advice	Number of Respondents
Internet	21
Hospital Consultant	19
Charity	16
None Received	11
Support Group	6
Family; Having a Diagnosis; Hospital; Other	3
Counsellor; Medical Literature; Specialist Nurse	2
Care Worker	1

### 3.3 Treatment and Care

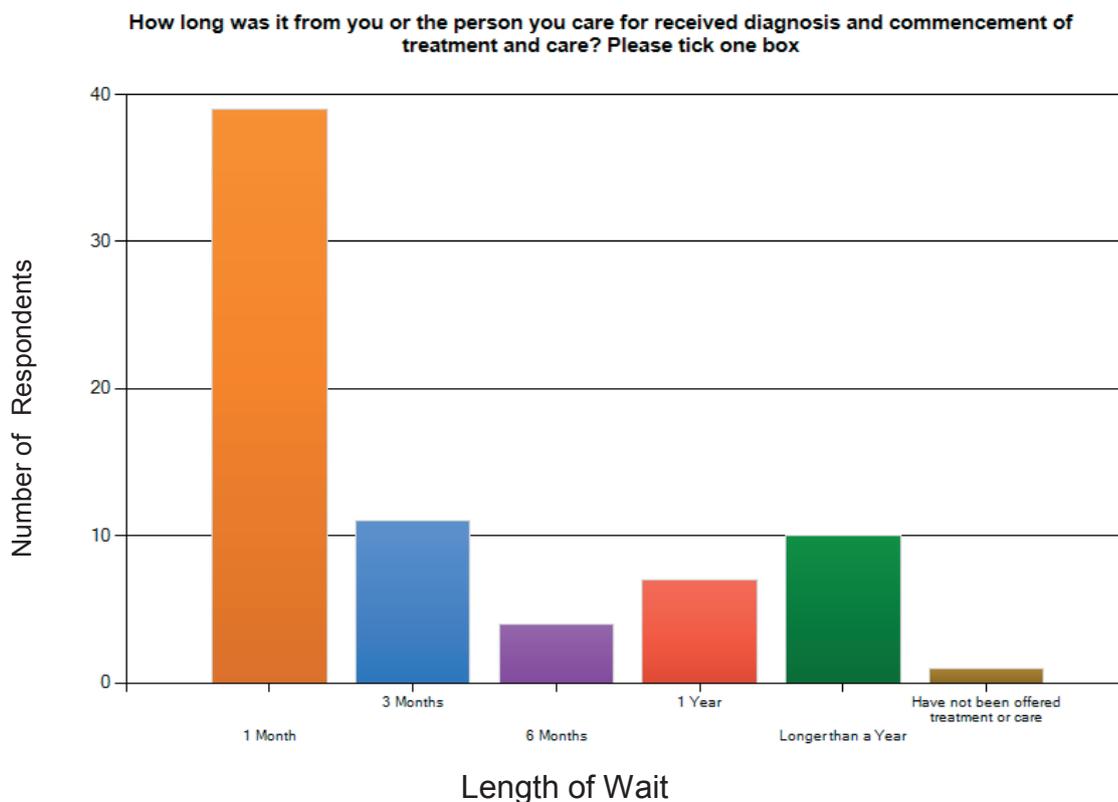
Respondents were asked a number of questions about the treatment and care they received following diagnosis. The answers to these questions appear in the paragraphs following.

#### Waiting Times for Treatment Following Diagnosis

Respondents were asked to state how long they had waited for treatment and care to commence following their diagnosis. **Figure 3** shows the responses to this question. Of the 72 responses, 39 (54%) of respondents stated they had waited one month for treatment and care to commence. 17 (24%) of respondents stated they had waited a year or more for treatment and care to commence.

3 people stated that they had not been offered treatment or care. One person said that this had been the case for 10 years. Another respondent stated that their treatment had started immediately and without a wait and another person stated that they had transferred from private to HSC treatment and care.

**Figure 3 – Waiting Time for Treatment and Care Following Diagnosis**



## Clinical Professionals Involved in Care

Respondents were asked a series of questions about the clinical professionals involved in their treatment and care. 61% of respondents said that they were receiving treatment from more than one medical professional. Respondents were asked how many professionals they were seeing and from which specialties. **Table 5** details the responses received.

**Table 5 – Clinical Professionals Involved in Care of Respondents**

**NOTE:**

- Respondents were asked to tick all that applied

Clinical Professional	Number of Times Named
Medical Consultant	131
Allied Health Professional	52
Nursing and Health Visiting	20
General Practitioner	17
Other*	16

The term “Other” refers to respondents who gave a number only (e.g. “5”) or where the service could not be confidently assigned to a speciality or profession – e.g. “continence clinic”.

### Specialism of Medical Consultant

25 different clinical specialties were named by respondents as involved in their treatment and care. The specialties named most frequently were:

1. Neurology (23 respondents)
2. General Practitioner (17 respondents)
3. Respiratory (12 respondents)

### Non Medical Clinical Professionals

A range of nursing and allied health professionals were named by respondents as involved in their treatment and care. Those most frequently named were:

1. Occupational Therapist (13 respondents)
2. Specialist Nurse (11 respondents)
3. Physiotherapist (11 respondents)

### Number of Contacts with Clinical Professionals by respondent

Respondents were asked for the total number of clinical professionals involved with their care. **Table 6** illustrates the responses to this question. 31% of respondents had between 3 and 5 clinical professionals involved with their treatment and care. 29% of respondents had between 6 and 10 clinical professionals involved with their care.

**Table 6 – Number of Contacts (All Professionals by Respondent)**

Number of Professionals Involved in Care	Number of Respondents	Percentage of Respondents
1	7	11%
2	17	27%
3-5	19	31%
6-10	18	29%
11-20	1	<1%
20+	1	<1%
<b>TOTAL</b>	<b>62</b>	

### Management of Treatment and Care

58% of respondents stated that there was one doctor in overall charge of their treatment and care. 25% said there was not and the remainder of respondents were unsure.

### Services Being Received

Respondents were asked to state what treatment and care they were receiving from a list of services on the questionnaire. They were also asked to indicate if they were waiting for any service. **Table 7** illustrates the responses to these questions.

**Table 7 – Services Being Received/Waited for by Respondents**

<b>Service</b>	<b>Receiving a Service (No. Of Responses)</b>	<b>Waiting on a Service (No. Of Responses)</b>	<b>Total Responses</b>
Outpatient	55	7	62
GP	46	1	47
Support Group	32	1	33
Occupational Therapy	30	5	35
Social Services	24	4	28
Physiotherapy	23	6	29
Speech/Language Therapy	21	4	25
Nurse Clinic	20	1	21
Benefits Advice	13	5	18
Inpatient	12	4	16
Counselling	11	4	15
Home Help	7	4	11
Helpline	7	2	9
Respite Care	5	3	8

### **Waiting Times for Services**

Respondents who said they were waiting for a service were asked to indicate which service they were waiting for and how long they had been waiting. 18 people responded to this question, with 12 named services and times waiting.

People waited most commonly on the Allied Health Professional services with most stating they had waited between 4 and 6 months for these services. Consultant waits were mentioned twice with 6 and 9 month waits described.

Other responses to this question included one person who stated they had waited “years” for home help and respite services. 3 respondents stated that there was no treatment and care for their condition – only the management of symptoms.

### **3.4 Carers**

65 people who responded to the survey were carers. This is just over half of all respondents. Of these, 76% were carers for one person, 18% were carers for two people and the remainder - 6% - were carers for three or more people.

## Carer and the Relationship to the Person Cared For

**Table 8** below shows the relationship between the carer and the person for whom they are providing care:

**Note:**

- *This question required respondents to write their response. It did not give tick box options. The table gives the responses actually given by respondents. The word “Mother” where it appears was the response written by the respondent in each case.*

**Table 8 – Relationship of Carer to Person with Rare Disease**

Carer Relationship	Number
Mother	28
Parent	13
Spouse/Partner	10
Daughter	9
Other*	4
Father	2
Sister	2
<b>TOTAL</b>	<b>68</b>

\* Other = Daughter in Law; Grandmother; “Child” and Cousin

Parental relationships account for 60% of all carer relationships in the responses given.

### ***Hours of Care Provided by Carers***

Carers were asked for how many hours a day they provided care. **Table 9** below shows the answers given.

**Table 9 – Hours per Day Spent Providing Care**

Hours providing Care	Number of Respondents	Percentage
0-4	10	16%
5-9	9	14%
10-14	3	5%
15-19	4	7%
20+	27	44%
Other*	9	14%
<b>TOTAL</b>	<b>62</b>	<b>100 %</b>

44% of carers stated that they provided care for 20 hours a day or more from the options given on the form. Of nine verbal responses, eight used a phrase like “all day” or “constant”. Therefore, 57% of responses indicated a caring commitment in excess of 20 hours daily.

## Sources of Support to Carers

51% of carers stated that they received additional support in providing care. Asked what sort of support this was, a wide variety of responses were received which fell into three categories:

1. Family only (39%)
2. Paid carers only (36%)
3. Family and paid carers combined (21%)

The remainder (4%) named other support including “childminder” and “taxi”.

### 3.5 Patient/Carer Experience

Respondents were asked a series of questions which addressed their feelings about the services they had received and their experiences around raising concerns about these services. 70 people (53% of all respondents) provided additional comments and information in addition to the responses to the specific questions included in the questionnaire. The responses to these questions are included in the paragraphs following.

#### Rating of Service Response

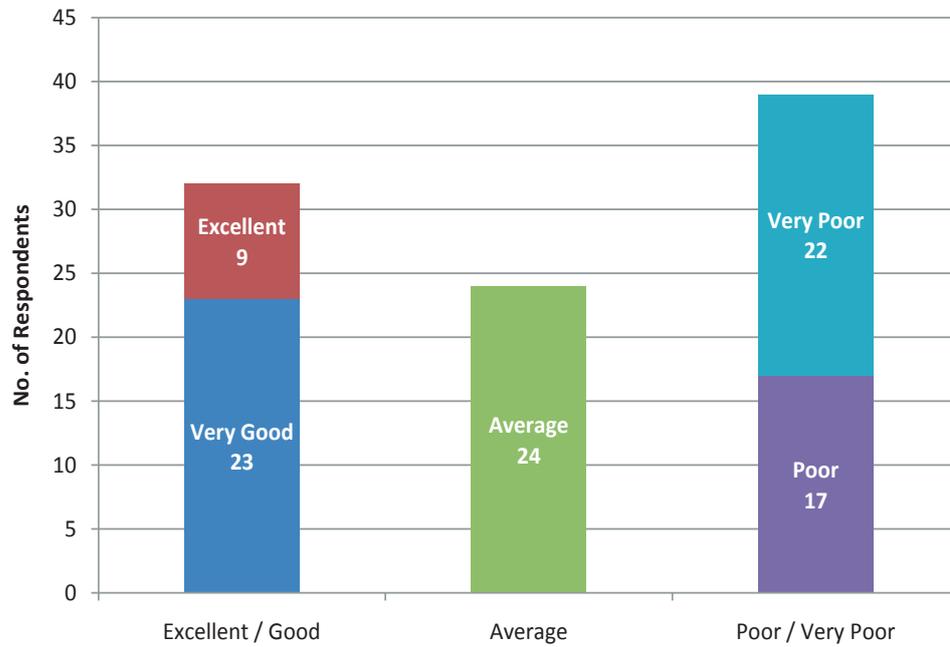
Respondents were asked to rate from options given on the questionnaire the way that Health and Social Care Services had responded to their needs.

**Figure 4** illustrates the responses to this question.

95 people (72% of all respondents) answered this question. 32 (33.7%) rated the response of the service as “Very Good” or “Excellent”. 41.1% of respondents rated the service as “Poor” or “Very Poor”. The remainder described the service as “average”.

**Figure 4 – Rate the Way that Health and Social Care Services Responded to Your Needs between the Onset of Symptoms and Diagnosis**

Between the time symptoms started and diagnosis, how would you rate the way the health and social care services have responded to your needs?

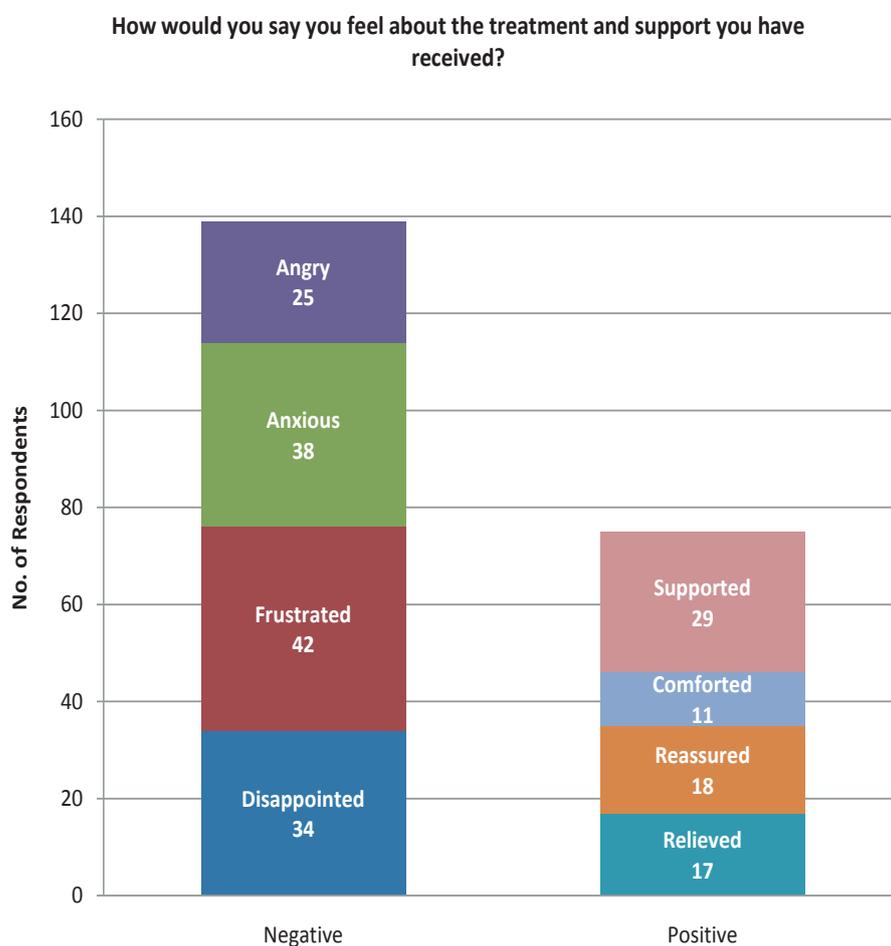


## Feelings about the Services Received

Respondents were asked to state how they felt about the services that they received by choosing from a range of options given on the questionnaire. **Figure 5** below illustrates the responses that were received.

Of a total of 214 responses, 139 (65%) expressed negative feelings about their experience between the onset of symptoms and diagnosis and 75 (35%) had positive feelings about their experience at this time.

**Figure 5 – How would you say you feel about the treatment and support you have received?**



## Raising Concerns

45 people (34% of all respondents) stated that they had raised concerns about their treatment and care between the onset of symptoms and diagnosis. 30 people (23% of all respondents) stated that they had felt concerned about their treatment and care but had not raised these with the people providing their treatment and care.

People who had felt, but had not raised concerns were asked why they had not raised their concerns. 30 people (23% of all respondents) responded to this question. Their responses were:

- 10 people responded by describing their concern but did not state why they had not raised it
- 4 people said that they were concerned to maintain good relationships with service providers
- 4 people stated that the service could not be expected to deal adequately with a condition of which it had little knowledge and that they understood that there were limited resources.
- 3 people stated that they felt dismissed and ignored and did not think their concerns would be listened to as a result
- 2 people stated that staff were always too busy to talk through any issues that patients or carers might have.
- 1 person stated that too much time had passed for them to raise their concern.
- 1 person stated simply that they did not feel comfortable raising their concerns.
- 1 person said that they were young at the time and felt that the doctor knew best.

### **Explanation of Diagnosis**

Respondents were asked if, when they received their diagnosis, it was explained to them by a medical professional. 96 people answered this question of which 79% stated that a medical professional had explained their diagnosis to them. Just over 20%, therefore, stated that their diagnosis had not been explained to them by a medical professional.

### **Personal Experiences**

70 people chose to provide additional comments on their experience. These comments are summarised in the following paragraphs.

The additional comments covered a wide range of subjects and experiences. However, four key themes emerge from these responses where, in each case, the issue was raised by respondents on 10 or more occasions. The key themes are:

1. Patient/Carer ignored/misdiagnosed
2. Supportive doctors
3. Lack of advice/information
4. Importance of self advocacy

### **Patient/Carer Ignored/misdiagnosed**

This was the concern most frequently raised by respondents.

Comments made by respondents included:

*“Since diagnosis, we have had excellent support, but I feel very strongly that in some cases, parents are dismissed as overly concerned...even as first time parents we knew things were not right...”*

*“GP insisted depression... reason for no follow up? ‘We only get one of these a year and we’ve already had one’”*

*“Because my disease is very rare, the GP hadn’t heard of it...putting everything down to post natal depression...”*

*“Prior to diagnosis, I was basically told the illness was in my head and to get on with it. I took this advice and ended up at death’s door”*

### **Supportive Doctors**

This was the second most common theme raised by respondents. Many praised the service that they had received. Often, one doctor is singled out for praise in contrast to the response received up to that point by the respondent. Comments made by respondents include:

*“my Mum was lucky as our GP had experience of one other person with sarcoidosis and so suspected it quickly...”*

*“if it hadn’t been for a chest consultant listening to me... I think I wouldn’t be here today”*

*“I feel forgotten apart from my GP...without his care and support I would not be here today...”*

*“The only reason a diagnosis was made was because she was seen by an excellent paediatrician who took the time to fully examine the child and listen to our concerns...”*

## Lack of Advice and Information

This was the third most common theme raised by respondents. Comments made by respondents include:

*“When I was first diagnosed, I was not given the correct advice. This ultimately caused me to try to ‘push through’ the symptoms causing a major relapse...”*

*“The treatment and care we received was excellent for the vast majority of the time, however, some support when we were not at hospital...someone we could contact with any concerns/worries would have helped...no real idea what to expect or how to manage at times...”*

*“When we were first told of a suspected genetic disorder for our son we were told completely out of the blue and then offered absolutely no support or help...or what to do or where to go next...hugely disappointing and frustrating...”*

*“Generally with a rare condition it hits you hard...where do you go after that? Internet and use of information can be frightening, outdated or wrong...”*

## The Importance of Self Advocacy

This was the fourth most common theme raised by respondents. The examples given were of patients, parents and carers who were persistent and assertive in seeking diagnosis, treatment and care from a service that initially failed to recognise the rare condition. Comments made by respondents include:

*“I was told my requests for referral to other services were only being done to please me as a difficult mother. I am glad I had the determination to continue”*

*“The support we have had has been exceptional...however, we have had to do most of the contacting and in some cases I have had to complain...this saddens me as it goes against the grain...I often think of those sufferers who do not have a voice...”*

*“...it took lots of visits to convince my GP something was seriously wrong...once I was referred, diagnosis and treatment was excellent.”*

*“While I now have a diagnosis, care path is a fight and I can’t get answers...only my persistence and education background meant I got any help or support...”*

## 4.0 Conclusions

### 4.1 Respondents

In total 132 people took part in this survey with 50 questions, but not all respondents answered every question. The responses reflected a diversity of age, sex and condition. 39 of the people for whom a response was provided were children (ages 0 – 15). 70 people provided detailed comments on their experiences and feelings about services. 65 people gave their contact details so that they could remain involved.

**There is a high level of interest and commitment to ongoing involvement among people with rare diseases and their carers.**

### 4.2 Diagnosis

30% of respondents had waited between one and five years for a diagnosis. 81% had emergency admissions between the onset of symptoms and diagnosis. 34% had at least one misdiagnosis before the correct diagnosis was reached. 20% had begun treatment that was not appropriate for the rare disease.

**There are clear issues around the diagnosis of rare disease in Northern Ireland and a need to support initiatives that raise awareness of rare disease and improve access to timely diagnoses.**

### 4.3 Treatment

The key sources of information and support for patients and carers are Medical Consultants, General Practitioners, the Internet and voluntary groups active in rare disease. 54% of respondents had commenced treatment and care within one month of diagnosis. 24% stated they waited a year or more.

**While the responses make clear that there is evidence of timely responses to a majority of patients once they are diagnosed, there is evidence also of a significant minority waiting for services. There is a need for additional work focussed on the experience of treatment and care by people with rare diseases beyond the initial response of the service to diagnosis. Future work should involve the four key sources of information and support identified by respondents.**

### 4.4 Carers

60% of the carers who responded were parents of a person with a rare disease. 57% of carers stated they provided care for more than 20 hours per day. 40% of the people with rare diseases for whom a response was submitted were children.

**The needs of carers should continue to be considered alongside those of people with rare diseases. Work should be considered to identify any particular needs of parents and families in providing care for a child with a rare disease.**

#### **4.5 Patient/Carer Experience**

38% of people rated the response of the service to their needs as “Good” or “Excellent”, while 41% rated the service response “poor” or “very poor”. Almost twice as many responses expressed negative feelings about the experience of Health and Social Care as expressed positive feelings. 34% of respondents had raised concerns about their treatment and care. 23% of respondents had felt concerns but not raised them.

**There is a need to support patients and carers in their self advocacy and to ensure that Health and Social Care services are able to listen effectively to patients and carers.**

## 5.0 Recommendations

In making these recommendations, the Patient and Client Council is mindful that, from February 2012, the Northern Ireland Rare Disease Partnership will be established as an independent organisation and will set its own agenda and programme of work. It is therefore recommended that:

- This report should be presented by the Patient and Client Council and the Northern Ireland Rare Disease Partnership to the Department of Health and Social Services, the Health and Social Care Board and the Public Health Agency. These organisations should be asked by the Patient and Client Council to make a written response to the report. In particular it is important they develop and implement both a regional strategy and commissioning plan for care of people with a rare disease.
- The Patient and Client Council should include the Northern Ireland Rare Disease Partnership with all other stakeholders in a major initiative it is undertaking on advice and information for patients and the public on health and social care in Northern Ireland.
- The Patient and Client Council strongly recommends that the Health and Social Care Board and the Public Health Agency participate in further survey work with the Northern Ireland Rare Disease Partnership on an agreed area of service. Priority should be given to exploring the experience of ongoing treatment and care.
- Links should be established between the Northern Ireland Rare Disease Partnership and professional educational groups to raise awareness of rare disease and to enhance early diagnosis and management of patients who present with symptoms of these conditions.
- The Patient and Client Council will continue to facilitate administrative support to the Northern Ireland Rare Disease Partnership for one year while the new organisation establishes itself. It will also support the partnership to work with the Health and Social Care Board and the Public Health Agency to ensure sustainability for the partnership, in order that patients and carers affected by a rare disease have a voice in the commissioning and delivery of appropriate health and social care services.

**QUESTIONNAIRE**  
**PATIENT AND CLIENT COUNCIL**  
**RARE DISEASE PARTNERSHIP**

**YOUR EXPERIENCE OF DIAGNOSIS**

The Patient and Client Council and the Northern Ireland Rare Disease Partnership want to hear from you. If you have a rare disease, or if you care for someone who does, and you live in Northern Ireland, please take a few minutes to fill in this questionnaire.

The Northern Ireland Rare Disease Partnership is a new initiative in Northern Ireland supported by the Patient and Client Council. It aims to make sure that the voices of people affected by rare disease are heard by decision makers in health and social care.

This questionnaire focuses on your experience of diagnosis. You can give us information anonymously or, if you want to remain in contact with us, please give your name and address at the end of the questionnaire. All information we use will be anonymised.

We will use what you tell us to produce a report for decision makers that is based on your experience. We will make recommendations for changes and improvements based on what you tell us.

Please answer the questions as fully as you can and return your completed questionnaire by post or e-mail to:

Richard Dixon  
Belfast Area Manager  
Patient and Client Council  
1<sup>st</sup> Floor  
Lesley House  
25-27, Wellington Place,  
BELFAST  
BT1 6GD

e-mail to: [richard.dixon@hscni.net](mailto:richard.dixon@hscni.net)

If you have any questions before filling in the questionnaire, please contact either Richard Dixon or Sarah McCandless at 02890321230.

Thank you for taking the time to complete this questionnaire. Your views are important.

**SECTION ONE: ABOUT YOU OR THE PERSON YOU CARE FOR.**

1. Are you/have you been:  
Diagnosed with a rare disease?

Caring for someone who has a rare disease?

2. Please state age of person diagnosed with a rare disease

3. Please state gender of person diagnosed with a rare disease  
Male  Female

4. If you are a carer, how many people are you a carer for?

5. How many are affected by rare disease?

6. What is your relationship to the person/people you provide care for?  
(e.g. parent; brother/sister; child; partner)

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7. If you are a carer approximately how many hours per day do you spend providing care?

8. If you are a carer do you require additional support?  
Yes   
No

9. If you answered yes to Question 8 please give details of who provides this support. For example family members; friends; voluntary helpers; others

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**SECTION TWO: DIAGNOSIS.**

**All of the questions in this section are about the time between symptoms first appearing and a diagnosis of a rare disease being made. If you are still waiting for a diagnosis, but a rare disease is suspected, please make sure you answer Question 11 so that we know this.**

10. Please print clearly below the name of the rare disease which has been diagnosed

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11. If a rare disease has been suggested however there are reasons a final diagnosis cannot be confirmed please provide details.

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12. Those with rare diseases may need to meet specialists from various different fields who will be involved in their care. Are you/the person you care for currently receiving treatment from more than one hospital consultant or other medical professional relating to the rare disease?

Yes

No

13. If you answered yes to Question 12 please give details. (How many doctors, whether this is at a hospital or not, the specialism of the doctor)

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14. Please state age at which symptoms/signs were first noticed. (If from birth please state "From birth", if before birth, please state "Before birth")

---

15. Who was the first person to notice the symptoms when they appeared?

Medical professional;

The person with the rare disease

The carer for the person with the rare disease

Other. (Please give details).

16. Have you/the person you care for had any emergency visits/admissions to hospital due to the condition between starting to have symptoms and a diagnosis of rare disease?

Yes

No

17. If Yes, how many?

18. Have you/ the person you care for had to travel outside Northern Ireland for specialist appointments between the onset of symptoms and diagnosis?

Yes

No

19. If you answered yes to Question 18 please state below where you/the person you care for travelled outside Northern Ireland, and how many times

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20. If you answered yes to Question 18, was referral for specialist appointments suggested by a doctor? Or did you refer yourself?

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21. Have you/the person you care for arranged private health consultations for any tests or treatment between the onset of symptoms and diagnosis?

Yes

No

22. If you answered yes to Question 18 please tell us what tests or treatment and where you/the person you care for received it.

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23. Is there one doctor or other health professional in overall charge of your treatment and care or that of the person you care for?

Yes

No

Don't Know

24. Between the onset of symptoms and diagnosis, where did you/the person you care for get information and support? Please tick all that apply.

GP	<input type="checkbox"/>	Specialist Nurse	<input type="checkbox"/>
Practice Nurse	<input type="checkbox"/>	Specialist Counsellor	<input type="checkbox"/>
Social Worker	<input type="checkbox"/>	Voluntary Group/Charity	<input type="checkbox"/>
District Nurse	<input type="checkbox"/>	Internet	<input type="checkbox"/>
Health Visitor	<input type="checkbox"/>	Other	<input type="checkbox"/>
Hospital Consultant	<input type="checkbox"/>		

25. Between the onset of symptoms and diagnosis, which of these was the most important source of information and support?

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26. How long was it between symptoms first appearing and diagnosis? (If waiting for a diagnosis how much time has it been since symptoms first appeared.). Please tick one box.

- Under 3 Months
- 3 Months to a Year
- 1 Year to 5 Years
- 5 Years to 10 Years
- More than 10 Years

27. How many doctors did you/the person you care for see between the first onset of symptoms and diagnosis? (If waiting for a diagnosis how many doctors you have seen so far.) Please tick one box.

- 1-2
- 2-5
- 5-10
- More than 10

28. Between the onset of symptoms and diagnosis, please tell us how many appointments you /the person you care for had to attend. (Write the number in the box)

- GP Appointments
- Hospital Consultant (Outpatient)
- Hospital Outpatient (Tests e.g. X-Ray)
- Hospital Consultant (Inpatient)
- Other

29. Was your condition /or that of the person you care for misdiagnosed before you received the correct diagnosis?

- Yes
- No

30. If more than one misdiagnosis was made please state how many.

31. If a misdiagnosis was made, did you or the person you care for receive treatment and care that was not right for the rare disease?

Yes

No

32. What sort of treatment was this?

\_\_\_\_\_

\_\_\_\_\_

\_\_\_\_\_

**SECTION THREE: AFTER DIAGNOSIS**

**This section asks some questions about the commencement of treatment and care after you received your diagnosis. If you are still waiting for a diagnosis please skip this section and go straight to Section Four.**

33. How long was it from the time you received your diagnosis /diagnosis of the person you care for and commencement of treatment and care? (If you have been diagnosed but are still awaiting treatment, please state the length of time waiting for treatment) Please tick one box.

- One Month
- Three Months
- Six Months
- One Year
- Longer than a Year
- Have not been offered treatment or care

34. Please show below what treatment and care you/the person you care for is receiving.

In each case please tick the relevant box. If you are waiting for treatment, tick the box marked "Waiting" and fill in how long you have been waiting in the last box.

### Medical Treatment

	Receiving	Waiting	Time Waiting
GP	<input type="checkbox"/>	<input type="checkbox"/>	
Outpatient	<input type="checkbox"/>	<input type="checkbox"/>	
Inpatient	<input type="checkbox"/>	<input type="checkbox"/>	

### Allied Health Professional Treatment and Care

	Receiving	Waiting	Time Waiting
Nurse Clinic	<input type="checkbox"/>	<input type="checkbox"/>	
Social Services	<input type="checkbox"/>	<input type="checkbox"/>	
Physiotherapy	<input type="checkbox"/>	<input type="checkbox"/>	
Occupational Therapy	<input type="checkbox"/>	<input type="checkbox"/>	
Speech/Language Therapy	<input type="checkbox"/>	<input type="checkbox"/>	

### Other Services

	Receiving	Waiting	Time Waiting
Counselling	<input type="checkbox"/>	<input type="checkbox"/>	
Benefits Advice	<input type="checkbox"/>	<input type="checkbox"/>	
Home Help	<input type="checkbox"/>	<input type="checkbox"/>	
Respite	<input type="checkbox"/>	<input type="checkbox"/>	
Helpline	<input type="checkbox"/>	<input type="checkbox"/>	
Support Group	<input type="checkbox"/>	<input type="checkbox"/>	

**SECTION FOUR: YOUR VIEWS ABOUT THE SERVICES YOU RECEIVED**

35. Between the time symptoms started and diagnosis, how would you rate the way the health and social care services have responded to your needs? Please tick one box.

Excellent

Very Good

Average

Poor

Very Poor

36. How would you say you feel about the treatment and support you have received? Please tick all that apply to you.

Disappointed

Relieved

Frustrated

Reassured

Anxious

Comforted

Angry

Supported

37. Have you raised concerns with the people responsible for treatment and care relating to the care you/the person you care for received between the onset of symptoms and diagnosis?

Yes

No

38. Have you felt concern relating to the care you/the person you care for received between the onset of symptoms and diagnosis, and not voiced those concerns to the people responsible for treatment and care?

Yes

No

39. When diagnosis/suggested diagnosis was made, did a medical professional explain it to you?

Yes

No

40. Thank you for answering all our questions. The space below is for you to use to tell us anything about your experience of diagnosis you would like to. We are interested to hear any part of your story.

**Optional**    If you want to keep in touch about this project or rare disease in general, please give your contact details below

**Name:** \_\_\_\_\_

**Address:** \_\_\_\_\_  
\_\_\_\_\_

**APPENDIX 2  
RARE DISEASES  
LIST OF RESPONSES**

	<b>Name Of Condition</b>	<b>No. of People</b>
1.	Albinism	3
2.	Achromatopsia	1
3.	Agammagobulinemia	1
4.	Motor Neurone Disease (All Types)	13
5.	Aniridia	1
6.	Arnold Chiari Malformation	1
7.	Niemann-Pick Disease (All Types)	1
8.	Brittle bones (Osteogenesis Imperfecta)	1
9.	Bulbar Palsy	1
10.	Cerebellar Ataxia	2
11.	Children's Interstitial Lung Disease	1
12.	Chronic Myeloid Leukaemia	1
13.	Cobalamin C Deficiency	1
14.	Cohen Syndrome	1
15.	Achromatopsia (Cone –Rod)	1
16.	Cone Rod	1
17.	Congenital Central Hypomyelination Syndrome	1
18.	Corticobasal Degeneration	1
19.	Cowden's Syndrome (Multiple Hamartoma Syndrome)	2
20.	Duchenne Muscular Dystrophy	1
21.	Dyspraxia	1
22.	Ependymoma Tumour	1
23.	Essential Thrombocythemia	1
24.	Fabry's Disease	1
25.	Fanconi Anaemia	1
26.	Fragile X	1
27.	Friedreich's Ataxia	2
28.	Wegeners Granulomatosis Vascular	5
29.	Group B Strep Meningitis	1

30.	Haemochromatosis	1
31.	Huntington's Disease	8
32.	Idiopathic Intracranial Hypertension and Chiari	1
33.	Idiopathic Late Onset Cerebellar Ataxia	1
34.	Nystagmus	2
35.	Kartageners Syndrome	1
36.	Myalgic Encephalitis	7
37.	Morquio's Syndrome	1
38.	Myasthenia QRA	1
39.	Niemann Pick Disease Type C	2
40.	Neuromyelitis Optica	1
41.	Non Alcoholic Chronic Pancreatitis	1
42.	Optic Nerve Hypoplasia	1
43.	Osteogenesis Imperfecta	4
44.	Pemphigus Vulgaris	1
45.	Pernicious Anemia	1
46.	Perthes Disease	1
47.	Polycystic Kidney disease	1
48.	Polymyositis	1
49.	Prader-Willi Syndrome	2
50.	Progressive Supranuclear Palsy	3
51.	Relapsing Polychondritis	1
52.	Rett Syndrome	1
53.	Sarcoidosis	1
54.	Schizencephaly	1
55.	Dravet's Syndrome	1
56.	Septo-optic Dysplasia	1
57.	Spinal Muscular Atrophy	5
58.	Spinocerebellar Swan	1
59.	Syringomyelia	2
60.	Tarlov Cyst Disease	1
61.	Thalassaemia H&H Disease	1
62.	Transverse Myelitis	1
63.	Undiagnosed Genetic Disorder	1

64.	Undiagnosed Neuromuscular Disorder	1
65.	WAGR Syndrome	1
66.	Sloan's Disease	1

**APPENDIX 3**  
**Table Showing Number of Responses per Question**

Question Number	Number Of Responses To This Specific Question
1	127
2	125
3	127
4	112
5	62
6	65
7	66
8	62
9	72
10	39
11	114
12	7
13	113
14	65
15	109
16	99
17	108
18	48
19	106
20	20
21	20
22	108
23	24
24	104
25	96
26	90
27	106
28	106
29	103
30	92

31	102
32	30
33	60
34	27
35	101
36	72
37	3
38	69
39	53
40	49
41	18
42	95
43	96
44	96
45	88
46	30
47	96
48	70
49	80
50	65

Remember you can contact your local office on

**Telephone 0800 917 0222**

or email [info.pcc@hscni.net](mailto:info.pcc@hscni.net)

**Southern Area**

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Craigavon BT66 8BB

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Tyrone and Fermanagh Hospital  
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**South Eastern Area**

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25-27 Wellington Place  
Belfast BT1 6GD

**Belfast Area**

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Belfast BT1 6GD

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Broughshane  
Ballymena BT42 4NJ

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