



Carer reported experiences

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Carer reported experiences: Supporting someone with a rare disease

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Abstract

This exploratory study aimed to gain an understanding of carer reported experiences derived specifically from persons caring for someone with a rare disease. The survey took place online on the SmartSurvey platform from November 2019 to January 2020. The facilitated workshop took place in Bangor Carnegie Library, Northern Ireland. To be eligible to participate in the online survey respondents had to be adults caring for someone with a rare disease. Fifty-seven respondents took part, 15.8% male, 84.2% female. Thirty-two attendees were part of the facilitated workshop. While carers reported several positive aspects of their caring role, the majority of comments highlighted challenges such as sub-optimal interactions with healthcare professionals, insufficient (or absent) emotional, psychological and social support, lack of financial support and lack of awareness of existing support services. It is important that strategies are put in place to ensure that carers are given the time they need to care for themselves, and that awareness is raised of what support options are available for carers of people with a rare disease(s) from health and social care providers, charities or support groups.

KEYWORDS

awareness, caregiver, focus group, Northern Ireland, public health, questionnaire, rare disease, survey

What is known about this topic and what this paper adds?

- Caring for someone with a rare disease, both formally and informally, can be an extremely demanding role requiring intense and unique care tailored to each individual's specific needs.
- The impact of caring for someone with a rare disease can be seen in many areas of an individual's life including psychologically, economically, physically and logistically.
- This study provides insight into current challenges, and some requested solutions, based on reported experiences from carers of people with a rare disease in the UK and Ireland.
- The use of an online survey promoted flexibility and accessibility for person's unable to attend the in-person workshop.
- The facilitated workshop enabled carers to network, discussing challenges and potential solutions with their peers.

Julie McMullan and Ashleen L. Crowe contributed equally to this manuscript and wish to be known as joint first authors.

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1 | INTRODUCTION

Rare diseases are collectively common and affect a significant proportion of the population (Ferreira, 2019), representing a major public health issue. A rare disease is defined in Europe as affecting less than 5 in 10,000 individuals (Requena-Méndez et al., 2020), and there are at present estimated to be between 6,000 and 8,000 rare diseases making their cumulative prevalence common (Putzeist et al., 2013). However, inconsistencies in how rare diseases are defined leads to impaired diagnosis, challenges researching rare diseases, and treatment (Haendel et al., 2020). Definitions of a rare disease vary by region, as does the prevalence of a given disease. The varying attributes that lead to the diagnosis of a rare disease do need to be better defined (Haendel et al., 2020); however, ultimately these improvements are needed to improve the lives of those who have a rare disease, diagnosed or otherwise.

It is currently estimated that over 450 million persons worldwide (6%–8% world population) have a rare disease (Repetto & Rebolledo-Jaramillo, 2020), with more than four million persons affected across the UK. Support for people living and working with rare diseases is currently inadequate (Kole & Faurisson, 2010; McKnight et al., 2020) with challenges including a lack of understanding by health and social care professionals, delayed diagnosis and difficulties gaining optimal treatment (Crowe et al., 2020; McMullan et al., 2020; Palacios-Cena et al., 2018).

Due to a low prevalence and lack of expertise for many of the >6,000 rare diseases, patients are often forced to become knowledgeable about their own disease state (Crowe et al., 2019). They become 'expert patients' alongside their carer as they seek an empowering and collaborative approach with their clinicians (Pomey et al., 2015). This is a shift from the traditional patient-doctor relationship, with each party revising their role and expectations, which presents many challenges (Budysh et al., 2012; Dudding-Byth, 2015). Caring for someone with a rare disease, both formally and informally, can be an extremely demanding role requiring intense and unique care tailored to each individual's specific needs (Baumbusch et al., 2018). There may be significant merit in peer support from networking with other carers experiencing similar circumstances themselves (Anderson et al., 2013).

The impact of caring for someone with a rare disease can be seen in many areas of an individual's life including psychologically, economically, physically and logistically (Mulroy et al., 2008). The importance of good mental and physical health for the carer is vital to ensure they are able to sustain the essential role which they provide for the individual they care for (Cañedo-Ayala et al., 2020). Neglecting to look after themselves can have devastating consequences on each carer's own health, which directly impacts the person they care for (Williams et al., 2009).

There is no consensus scale to capture experiences from formal and informal carers of people with rare diseases across all age groups. For example, the Carer Experience Scale (CES) is an index measure of the caring experience focused on economic evaluations (Goranitis et al., 2014). The ASCOT-carer scale is a self-report

instrument designed to measure social care-related quality of life for family-friend unpaid carers (Rand et al., 2015), while the FAMCARE-2 scale has been used to measure carers' perceived satisfaction with palliative service provision (Aoun et al., 2010). The exploratory approach undertaken in this study aimed to gain an understanding of carer reported experiences derived specifically from persons caring for someone with a rare disease.

2 | METHODS

Multiple methods using qualitative data analysis techniques were chosen (Forbes et al., 2007) with experiences reported by carers supporting individuals with a rare disease(s) sought via a survey and facilitated workshop. All participants provided written, informed consent. Ethics approval was provided by the Faculty of Medicine, Life and Health Sciences, Queen's University of Belfast (QUB) research ethics committee (MHLS 19_08).

2.1 | Patient and public involvement

Patients and public were involved in the design of this survey and helped to promote participation. The project aim was a direct result of previous research within the QUB Rare Disease Team which highlighted the need for research focusing solely on carers. It became apparent that carers are often overlooked in rare disease research and yet they play such a vital role in the life of someone with a rare disease. As part of the dissemination of results, we plan to develop a leaflet to be given to carers at the time of diagnosis. We have sought input from carers and rare disease patients as to how this should be formatted and what information should be included.

2.2 | Online survey

An online survey was hosted by SmartSurvey (SmartSurvey) 6 November 2019 – 31 January 2020 with five sections and 43 questions (Supplementary File S1). Questions were designed using an iterative approach with the Northern Ireland Rare Disease Partnership (NIRDP) and were focused on caring responsibilities, interactions with healthcare and support services, networking, communication and future improvements. Closed and open-ended questions were employed, with the option to save the survey and complete at a later date.

Survey respondents identified as caring for an individual with a rare disease, aged over 18 years old. The survey was promoted via the NIRDP website (NIRDP) and was shared on their Twitter (@NIRDP; 1,168 followers) and Facebook (@NIRDPNews; 1,013 members) sites. Queen's University Belfast also promoted the survey via their websites and Twitter pages. An A5 flyer (Supplementary File S2) was developed to promote the survey displaying a QR code and

a link to the survey and distributed locally across QUB campus and libraries. The survey was also promoted at two rare disease events held locally in Newtownards (December 2019) and Bangor (January 2020), Northern Ireland (NI). Charts and crosstabs were generated using SmartSurvey and MSEXcel to gain frequencies and percentages. The qualitative data from open questions were analysed thematically.

2.3 | Facilitated workshop

A workshop for carers of people with a rare disease was organised in association with the NIRDP (NIRDP) on 16 January 2020 in Bangor Carnegie Library, NI. The workshop was advertised in a similar manner as for the survey. The workshop was an open event, anyone was welcome to attend. It lasted approximately 2 hr. The event was primarily an opportunity for carers to network with one another as well as with multiple carer focused organisations based in NI who were in attendance highlighting their services (Supplementary File S3).

An independently facilitated discussion at the workshop was conducted with all attendees and using small groups. Notes were taken during the discussion and analysed by two researchers (AC & JM). Three key themes were explored during discussions, focusing on each for approximately 20 min:

1. 'Positive aspects of the caring role'.
2. Identifying and prioritising the challenges carers face and to consider how these problems might be overcome.
3. What good support may look like and priorities to improve resources for carers across NI.

3 | FINDINGS

3.1 | Survey

3.1.1 | Respondent characteristics

Fifty-seven respondents participated in the survey, with 56 identifying the person they care for as a family member. Respondents resided in NI, Great Britain or the Republic of Ireland. 80.7% respondents identified as caring for the individual in a voluntary/unpaid capacity, while others identified specifically as being a parent, or wife, and so may also fall under the voluntary role although not explicitly stated; one respondent identified as a paid carer. 87.7% of respondents live with the person they care for, with 35.7% caring for children. Table 1 shows the demographics of survey respondents, with Supplementary File S4 showing specific demographic information.

Half of carers (~50%) look after an individual full time/24 hr a day. The majority of respondents (86.0%, $n = 49$) do not have any

TABLE 1 Demographics of survey respondents

Demographics of Respondents	%	Frequency ($n = 57$)
Gender		
Male	15.8	9
Female	84.2	48
Age, years		
25–34	14.0	8
35–44	24.6	14
45–54	49.1	28
55+	12.3	7
Country of residence		
Northern Ireland	96.5	55
Great Britain	1.8	1
Republic of Ireland	1.8	1
Number of individuals currently cared for^a		
1	77.2	44
2	10.5	6
3	10.5	6
4	1.8	1
Relationship to the person cared for		
Family	98.2	56
Friend	0	0
Other (started as client, now a friend)	1.8	1
Age of the person(s) cared for^a		
Less than 18	35.7	20
18–24	16.1	9
25–34	12.5	7
35–44	7.1	4
45–54	14.3	8
55+	19.6	11
Prefer not to say	1.8	1
Capacity of care for the person		
Voluntary / unpaid	80.7	46
Paid-employed	0	0
Self-employed	7	4
Other	12.3	7
Live with the individual cared for		
Yes	87.7	50
No	12.3	7
Geographic setting		
Urban	47.4	27
Rural	52.6	30

^aPlease note that the number of individuals cared for is 78, but that a person may have two or more persons cared for in separate age categories.

respite care available. Of the 14% ($n = 8$) who do have respite care available 75% ($n = 6$) of them have used this respite facility. Prior to caring for the individual with the rare disease 54.4% ($n = 31$) of the respondents had not heard of a rare disease, and 75% ($n = 42$) of the respondents had not had any interactions with someone with a rare disease.

3.1.2 | Training for carers

Only 14.3% ($n = 8$) of the respondents had received formal training to assist with their caring for the individual with a rare disease. Completed training included medication administration, hoisting, tube feeds, wheelchairs, oxygen administration, first aid, manual handling, safeguarding, working with people with learning difficulties, maintaining airways, Multi Agency Public Protection Arrangements (MAPPA) training. When asked if they would like to receive formal training as a carer 25.5% ($n = 14$) said they would like to receive in person training, 25.5% ($n = 14$) said they would like a workshop, 7.3% ($n = 4$) said yes to it being delivered online at a specific time, and 38.2% ($n = 21$) said yes to it being delivered online but accessible in their own time. Of the respondents only 20% ($n = 11$) said they would not like to receive formal training as a carer.

3.1.3 | Understanding of genetics and multi-omics

More than $\frac{4}{5}$ rare diseases have a genetic cause (Health, 2013), yet only 3.5% ($n = 2$) of respondents identified as having a confident understanding of genetics. Approximately one-quarter of respondents would like to know more about genetics (26.3%, $n = 15$) and multi-omics (24.6%, $n = 14$).

3.1.4 | Mental health

All respondents (100%, $n = 57$) stated that caring for a person with a rare disease impacted their mental health. The impact reported was overwhelmingly negative with respondents comments such as,

(Respondent 1) 'Exhausting while mentally and physically draining', (Respondent 2) 'We all feel lost and helpless sometimes', (Respondent 4) 'Suffocating at times', (Respondent 37) 'It's very stressful and causes anxiety'. Only 12.3% ($n = 7$) of respondents stated that they had received psychosocial support in relation to their role as a carer. When asked what they had received or what services they had accessed one individual described a brief conversation with a GP, another spoke of self-referred to counselling. Others had used cognitive behaviour therapy, a pain management, mental health course, landmark worldwide (<https://www.landmarkworldwide.com/>) and one person said 'Always have nurses or doctors to call if worried about anything 24/7'.

3.1.5 | Medical appointments and healthcare professionals (HCPs)

In relation to medical appointments, dealing specifically with rare disease, 30.2% ($n = 16$) described them as being a positive experience and 69.8% ($n = 37$) as a negative experience (see Figure 1a,b). The majority of respondents (80.7%, $n = 46$) did not feel that medical professionals had sufficient knowledge to look after those with a rare disease (Figure 1b).

3.1.6 | Networking

Half of respondents (54.4%, $n = 31$) were involved in one or more support groups, with a similar number having contacted a rare disease group (52.7%, $n = 29$), and significantly less a carers group (12.3%, $n = 7$). Reasons for contact included seeking peer support/feeling isolated; information about policy; experiences from other families; about the disease after getting diagnosis; what services and help are available; offering help; to seek out medical experts on the field; to seek diagnosis and to raise funds. Quotes included:

Respondent 39 'To see if anybody could help as we feel we are on our own and been to everyone around the world to see if anybody could help us'.

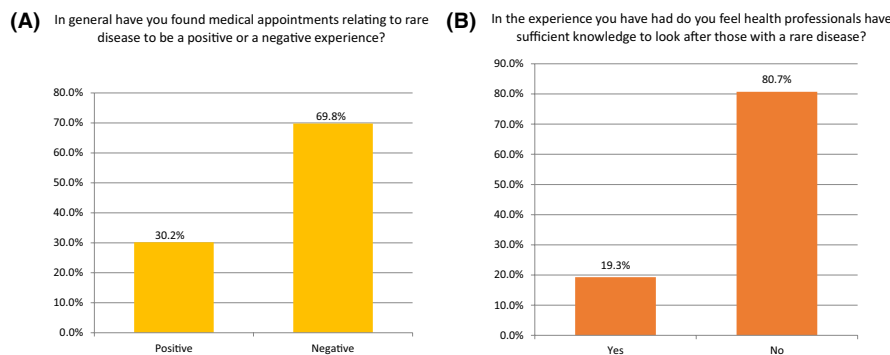


FIGURE 1 (a and b) Experience of healthcare appointments and healthcare professionals

Respondent 24 'For support, help and to meet other people with similar problems. To get the right information to educate me and family'.

Of the respondents who had contacted a charity/support group, 70.6% ($n = 24$) said they had their need met, for example,

Respondent 11 'The support has been greater online from people going through the same thing than health professionals'.

Respondent 12 'It worked well. And a place you feel others understand your struggles'.

Respondents reported communicating with other carers most frequently *via* internet forums (45.9%, $n = 17$) and in person (29.7%, $n = 11$). Many stated that communicating with other carers reduces isolation and keeps them in a positive mind set. When asked if networking with different sets of people would be of interest the majority of respondents answered yes (Supplementary File S5).

Only 1 respondent thought that sufficient support is available for those who care for someone with a rare disease with several suggestions made to improve support (Figures 2 and 3). Quotes included:

Respondent 1 'Financial support!!!! It is incredible that we don't get any recompense for caring roles, not even a small pot of funds to allow a long weekend away once a year'.

Respondent 22 'More information and support on how to cope and help'.

Less than half of respondents (40.4%, $n = 23$) had attended a rare disease conference/event. Of those who did attend, 18 (78%) found it very useful, 4 (17%) said it could have been better, and 1 (4%) said they would not go back. Many carers were unable to attend such events due to lack of time, lack of respite care and insufficient resources including financial. The challenges experienced by carers and priorities for carers are summarised in Figure 4a,b.

3.2 | Facilitated workshop/discussion

The first topic for discussion focused on the positive aspects of being a carer. Carers reported feeling a sense of pride; being able to help the person to enjoy life; giving life purpose and fulfilment; and gaining an increased medical knowledge. This was followed by discussion of the biggest challenges for carers. Interactions with HCPs was highlighted as a major hurdle and carers stressed their need to be taken seriously in such settings. Ideas such as a feedback system for HCPs and raising awareness of the importance of treating individuals with rare diseases, and those caring for them, with dignity and respect were suggested. Insufficient emotional, psychological and

social support were highlighted with an advice helpline suggested as an approach to help.

Financial support was discussed with many carers explaining that carers allowance is not sufficient and many are not considered eligible to receive it. There was a consensus that carers should be recognised and adequately paid for their role.

Carers requested information on services such as respite and where to access specific equipment. They reported regular feelings of guilt due to the lack of attention they give siblings because of the demands put upon them by their caring role for a child. Several participants suggested that respite services would be welcomed to enable taking other family members for a short break. The majority explained that current respite services are generally inadequate for their needs and suggested having respite carers who understand individual medication needs; good communication would help improve this service. Figure 5 displays the complex considerations needed to enable effective respite care. Many improvements carers suggested included online training, websites which link relevant resources, organisations and psychosocial support.

4 | DISCUSSION

Those with a rare disease have requested changes in the health-care systems they use, improved interactions with healthcare professionals, more public awareness, and better information access around rare diseases specifically, but also support (Crowe et al., 2019; McMullan et al., 2020). However, if the voice of the carer is ignored then improvements made based on these requests will not have the same impact, as a part of the story is missing. Research focusing on rare disease often does not include carers yet given the unique role they have in the person's care it seems crucial that their views are heard (Currie & Szabo, 2019a, 2019b; Niemitz et al., 2019; Wu et al., 2020). Furthermore, with approximately half of all rare diseases affecting children (Batshaw et al., 2014), there is often a focus on the parents as carers of those with a rare disease, which although absolutely necessary, leaves many others who are caring for adults with a rare disease unheard. Our research emphasises challenges faced by carers supporting a range of individuals with a rare disease and highlights areas where support is lacking.

4.1 | More Support Required (including finances, training, mental health and respite)

The majority of attendees highlighted their financial struggles as carers for individuals with a rare disease. This was in both priorities of what would help most and in biggest challenges. This was in line with what the literature states of other carers, where for example, there are significant additional costs associated with raising a child with a disability (Sloper & Beresford, 2006), with 38% of parents having

Financial support

"Financial support!!!! It is incredible that we don't get any recompense for caring roles."

"Most support is for more well known causes such as cancer. Parents of children with cancer for example receive much emotional support as well as financial support and even family respite breaks. There is nothing available for families and parents of children with rare diseases even though they put whole families under permanent and long term strain."

Resources and information

"The carer knows the signs of when the individual is unwell or exhibiting new symptoms. They should be listened to. Given support when patient is emotionally low. Given advice re finance, where to access aids. Too much time spent looking for help alone."

"There doesn't seem to be the resources / knowledge about the disease for anyone to be interested in us."

Support network

"I don't think other carers understand RD carers, they don't understand that things change unpredictably. They think we just don't bother to turn up Now that childhood has passed & my daughter is now an adult, I realise there's not much for us at conferences, discussion still revolves around children. I'd like to know who else is in my boat and how do they deal with things. Can I do things in a better way, even re provision of equipment & services, and the nights without sleep, who else is up at night too? How do they deal with this."

"I think that if I had received support when my child was first diagnosed and if the support was continuous that would have helped rather than being left on our own to try to figure it out."

Respite

"Access to a database of professional carers to help is lacking."

Mental health

"Because there isn't enough people who would suffer the same illness as myself in Northern Ireland which makes it very hard to deal with as you are on your own."

Diagnosis

"More carers information given at diagnosis. This is the single most important thing."

Awareness raising

"Again more awareness and education needed so patients and their families can be properly and adequately informed prepared and supported."

Health service

"Better healthcare provision with patients details/diagnosis highlighted without the continuing need of trying to explain the diagnosis to each and every care provider."

"More awareness of various disease. And if the gp doesn't understand the illness they should seek appropriate training or at least look it up."

"Better training for doctors, physios, occupational therapists. Would help if there was an overall specialist to help manage the rare disease within Northern Ireland rather than having to travel to London."

"For a start for professionals and health systems work with carers. Appointments are designed to suit systems, often run late, exorbitant parking. Why can't we do more via Skype/phone appointments/out of hours. Don't assume Carers live near the one they care for - or have nothing else to do except be a carer. I work, have 2 kids under 5 and live 50 miles away. Also consider Carers are trying to work in the best interests of the cares for - so if they ask lots of questions or query treatment or side effects it's because they KNOW that cares for individual better than the professional."

FIGURE 2 Responses when asked what could be done to help

FIGURE 3 What would you like to see improved regarding support for carers?

<p>Mental health</p> <p>"More help learning to cope with living from this condition."</p>	<p>Voice for carers</p> <p>"A stronger body of people to have some say in policies and to lobby for themselves."</p>
<p>Respite</p> <p>"An understanding that Carers have lives and suffer burnout. By caring they are saving the government EEEEE."</p>	<p>"Letting carers have a voice so carers and the ones they care for are listened to... the doctors/nurses put you out of the room when talking to the patient (sometimes family or carers knows more about the condition or do's or don'ts that can cause injury to that person). Give RESPECT to the carers they are just important as the nursing staff or doctors."</p>
<p>Awareness and understanding</p> <p>"More understanding of the family as a whole living and caring for someone with this illness."</p>	<p>Psychosocial support</p> <p>"More help, counselling support, more funding for 'extras', more information on disease and where to get help generally."</p>
<p>Resources</p> <p>"More money, more resources, more support, more recognition."</p>	<p>Support</p> <p>"That it's recognised as a full time job, that there is more support made available and practical and emotional support."</p>
<p>Training for carers</p> <p>"Training on how to perform basic medical tasks so that we're not waiting for health professionals to arrive."</p>	<p>"When support organisations contact us and offer help and information like when contact for families with disabled children did to ask if I could fill out this survey."</p>
<p>Knowledge and information</p> <p>"Better advice when patient diagnosed and or being discharged from hospital. Signposting from specialist team. Don't just discharge a child with no plan of care."</p>	<p>Financial</p> <p>"Some benefits so I can be home more instead of working full time to pay the mortgage"</p> <p>"Direct payments scheme often works ideally to help match clients and personal assistants."</p>

(A) : Biggest challenges when caring for someone with a rare disease.



(B) : Themes of priorities that would help most in caring for someone with a rare disease.



FIGURE 4 (a) Biggest challenges when caring for someone with a rare disease. (b) Themes of priorities that would help most in caring for someone with a rare disease

to reduce their paid employment hours or leave work all together (Pelentsov et al., 2016). Many adult rare disease patients employ carers under the 'direct payments' scheme; spouses and children living in the same household are not eligible to receive 'direct payments' funding for their caring roles (Direct, 2020), despite the frequent need for 24/7 care. Although there is sometimes a small amount of funding available from the government as a carer's allowance, it is not nearly enough to cover costs of medical treatment and travel (Anderson et al., 2013; Zurynski et al., 2008).

Unanimously carers experienced insufficient support, which is a common issue internationally (Anderson et al., 2013; Aubeeluck et al., 2012; Lagae et al., 2019; Pelentsov, Fielder, Laws, et al., 2016; Van Groenendael et al., 2015). For instance, in a palliative care intervention study by Lyon et al., it was found that over 80% of the family

caregivers would value more support with knowing what to expect in the future when caring for their child, more support around financial, legal, work issues and support around getting help to allow for time for themselves (Lyon et al., 2019). Several respondents from our survey perceived there was more support available for more common diseases, with so little resources available for rare diseases. For example, it was said that, (Respondent 8) 'Most support is for more well-known causes such as cancer'. Parents of children with cancer, for example, receive much emotional support as well as financial support and even family respite breaks. There is nothing available for families and parents of children with rare diseases even though they put whole families under permanent and long-term strain.

Provision of additional support, or improved signposting towards existing resources would minimise some of the strain described by

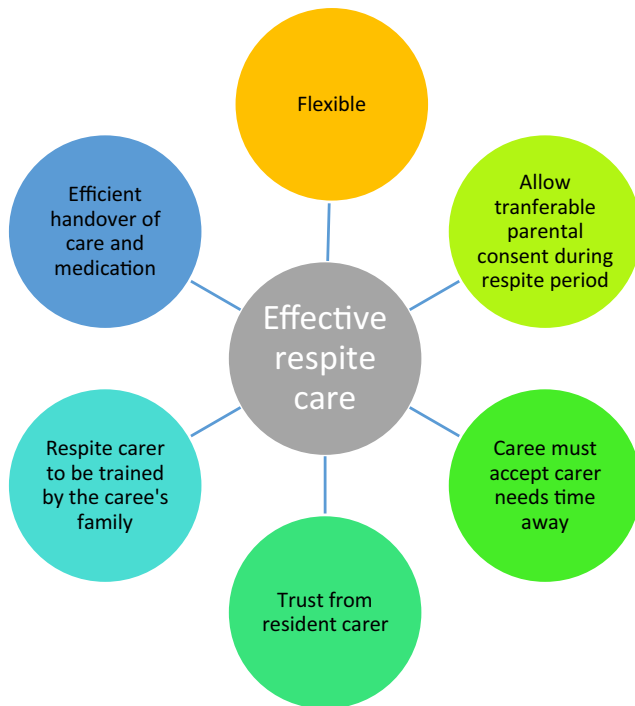


FIGURE 5 What is needed to enable effective respite care

carers. A particular area highlighted in both the survey and facilitated discussions was additional support requested for carers and siblings of children with a rare disease(s):

(Respondent 46) 'Support and help with childcare for siblings, because it is not possible to bring siblings to certain clinics in the hospital because of infection control. Also, when the sick child is an inpatient, you cannot leave them in the hospital, and it is often very difficult to get any help with the siblings. Juggling family life is very difficult'.

Lagae et al. (2019) described how 91% of carers feel caring makes family life difficult, stating that 46% of siblings had missed a leisure opportunity in the past 2 weeks due to parental caring responsibilities.

A request for accessible respite was raised many times in this study, which complements international reports where respite support is not adequately provided and would be highly valued to help with day to day management of their loved one's symptoms (Kasparian et al., 2015; Lyon et al., 2019; Mori et al., 2017; Mutch et al., 2017; Palacios-Cena et al., 2018). Mutch et al. in particular found that more support would be valued in regards accessing respite (Mutch et al., 2017), and Mori et al. observed that mental health outcomes did not necessarily improve with respite care having been provided, suggesting that it was not adequately done so (Mori et al., 2017). Practical support is very important to minimise excess strain on carers, particularly when carers feel their friends and family cannot empathise with their situation (Wiblin et al., 2017).

Formal training for carers is lacking - our survey confirmed that less than a fifth of the respondents had received formal training as carers. Parents of those with a rare disease have expressed that they would value receiving training to assist them in their caring needs (Somanadhan & Larkin, 2016) as would young carers (Kavanaugh et al., 2019). Training for carers would enable improved, safer care and may facilitate better relationships with HCPs. Providing training to primary carers for people with complex care needs may result in cost-savings for healthcare providers by minimising adverse events and unplanned hospital admissions. More than a third of the respondents preferred training delivered online to flexibly work with their caring commitments.

All (100%) of the carers responding to the survey reported that caring has affected their mental health. Limited support for emotional/psychological/social wellbeing was also raised many times during the discussions. Additional practical issues reported in the literature included the limited help they receive for things like transportation, shopping and chores (Roscoe et al., 2009). With recent findings showing 84% of carers in the UK feel more stress due to their caring role (State of Caring, 2015,) it is a common problem which needs to be addressed. A survey by Pelentsov (2016) reported 37% of carers for someone with a rare disease were being treated for depression, 41% for anxiety and 10% for other mental illnesses (Pelentsov, Fielder, Laws, et al., 2016). There is a clear need for improved psychosocial support for carers of people with a rare disease(s).

4.2 | Awareness and information

Awareness and information were mentioned repeatedly by respondents seeking '*more information*'. A lack of awareness was highlighted for the impact that caring for someone with a rare disease has on the life of each carer and their family. A respondent (*Respondent 33*) stated that '*Again more awareness and education needed so patients and their families can be properly and adequately informed prepared and supported*'.

Internationally, carers of those with a rare disease have asked for educational resources for friends/family (Kasparian et al., 2015; Pelentsov et al., 2015). Providing accessible educational resources for families and the community as a whole would be a practical step to help raise awareness of the role of carers and common impacts when caring for someone with a rare disease. A discussion area in this project that we did not find featured in the literature was a need for a lobbying body for carers of those with rare disease.

A lack of appropriate information and communication for rare diseases has been highlighted with individuals seeking more information about rare disease diagnoses, implications of long-term care plans, and community support teams to provide clarity for carers and help them feel more supported (Crowe et al., 2019; Lyon et al., 2019; Pelentsov et al., 2016; Pelentsov et al., 2015). Carers often have incomparable personal knowledge of how a rare disease affects the person they support. In spite of this knowledge, carers are rarely

provided with sufficient practical or medical information to assist them in their role as a carer (Mori et al., 2017; Pelentsov et al., 2015). Information particularly sought in this study focused on information about diagnosis, treatments, support and finances. Focused information at the time of diagnosis, (*Respondent 16*) 'More carers information given at diagnosis. This is the single most important thing.', was not something commonly reported in the literature, though it was raised in response to the survey, although more timely diagnosis and better access to diagnosis were referred to several times (Palacios-Cena et al., 2018; Pelentsov, Fielder, Laws, et al., 2016; Pelentsov et al., 2015). Although information at the time of diagnosis may not be the single most important thing to all carers, it would support carers to make choices. Ultimately, carers of those with a rare disease are living in an information vacuum where so many of us take the access and ease to 'common' information for granted.

4.3 | The experts (Healthcare professionals and carers)

Sub-optimal interactions with HCPs have been highlighted repeatedly in studies across the globe; our workshop and survey, predominantly derived from NI, showed no exceptions and indeed poorer experiences reported than observed internationally. For example, one experience was that (*Respondent 1*) 'Healthcare professionals rarely know anything about any of the rare diseases...it's frustrating to repeat the same thing to multiple consultants and they don't appear to talk to each other. Better coordination of care would be good'.

International results for example show 81% of carers felt their HCP did not have enough knowledge to treat their rare disease, compared to 54% of parents reporting their HCP lacked adequate knowledge (Pelentsov, Fielder, Laws, et al., 2016). And this is in fact opposite to an Australian study which reported 73% of carers felt their GP had adequate knowledge of their rare disease (Anderson et al., 2013). Many carers were dismayed feeling left to figure out the disease themselves due to the lack of understanding by HCPs, which echoes other findings (Baumbusch et al., 2018). A lack of knowledge by some HCPs can have detrimental effects, including delaying diagnosis (Bendixen & Houtrow, 2017), which is already a stressful event (Palacios-Cena et al., 2018). Where relevant, carers being recognised as experts in a rare disease is necessary, but not often the case (Kesselheim et al., 2015; Syed et al., 2015). Carers are keen to have equality with patients when it comes to healthcare services 'The same respect for my needs as that which the disabled person is entitled to'. Carers being recognised and treated equally would streamline the care of the patient, where the healthcare system relies on carers to carry out so much of the day to day functions necessary for a patient's wellbeing. Collaboration with healthcare professionals is mentioned in the literature as something of value and which has a positive impact on carers (Kesselheim et al., 2015; Shapiro et al., 2019; Syed et al., 2015). Carers from the survey were adamant as they would like to receive

more respect, and experience improved communication between HCPs with both patients and carers, particularly with respect to co-developing care plans.

4.4 | Social isolation

Social isolation was frequently reported by carers with many experiencing no free time for themselves while caring for individuals up to 24 hr a day. In the survey, 86% of participants reported that there is no available respite for the person they care for, which is inevitably going to affect the social life of the carer and in turn their mental health. Respondent 19 said 'we need care packages to take account of respite needs, we don't want traditional respite options, we need those we care for to be able to go away for a weekend with a PA fully funded'. Similarly Pelentsov and colleagues observed 58% of carers losing friends since they started caring for someone with a rare disease (Pelentsov, Fielder, Laws, et al., 2016) while 80% of carers said caring made socialising difficult (Lagae et al., 2019). Online support groups have been documented in the literature to help with mental health effects of caring (Aubeeluck et al., 2012). Of our survey respondents, 70% of those currently in communication with other carers used the internet or social media to communicate. The flexibility in regards location of an online platform makes it easy for carers who cannot arrange alternative care for the person with a rare disease whom they support. When the respondents were asked about networking the predominant result was that there was a preference of networking with healthcare professionals and other carers rather than charity workers or patients. Increased capacity for networking would facilitate connection and improve information dissemination and sharing.

5 | STRENGTHS AND LIMITATIONS

This research study is based on the experience of carers of people with a rare disease in the NI, Great Britain and the Republic of Ireland. It highlights the challenges that they are facing and allows for them to put forward their own solutions and suggestions for support, understanding and information. Using an online survey was deemed the most efficient way to reach a large number of carers in a short time period. The hope was to get a range of views from a wide geographical spread such as from both urban and rural settings, as well as between age ranges of carers. Carers often find attending events a challenge due to their caring responsibilities, so making the survey available online made it more accessible (Schumacher et al., 2014). Online surveys offer flexibility as they can be completed at a time suitable to the individual and have a function to enable it to be completed in stages (Davies, 2016), saving progress as they work through the questions. They also allow anonymity and the opportunity to skip questions that they are not comfortable answering (Davies, 2016).

Given that no appropriate carer scale survey could be found in the literature, the questions of this survey were designed specifically for this study using an iterative approach, with input from the NIRDP (NIRDP). The survey focused on the challenges carers' face, the support they receive and how they feel they could be helped.

The workshop participants may have been a biased sample as all were self-motivated to attend, and the survey respondents were self-motivated to complete the survey. Therefore, as researchers we have presented these results transparently, maintaining an awareness of the respondents and participants motivation for participating in this study and the implications this may have on data interpretation. Both carers of children and adults with a rare disease are included here, and the survey was open to paid and voluntary carers.

The workshop also provided a valuable opportunity for those caring for people with a rare disease to network, which is valuable to carers (Shapiro et al., 2019), and to meet representatives from voluntary organisations. The carers surveyed were based predominately in NI, which provides an important local evidence base for policy and practice implications.

6 | CONCLUSION

Carers of those with a rare disease are asking for better access to psychosocial support, better financial provision for their substantive role, improved access to helpful information to give clarity for the future, training to assist them in their role, and options for respite care. Improved interactions with HCP's and primary carers that encompasses both understanding and recognition of carer's crucial role in the life of whom they care for. A carer experience scale is required for carers of people with a rare disease. While many themes are common across all carers, some are unique to rare disease carers, for example, the difficulties experienced at appointments with healthcare professionals and the lack of awareness and information surrounding rare diseases. Future surveys for 'carers' should include a question asking if they care for someone with a rare disease so that their unique needs can be identified. The time taken for carers to provide such detailed and valuable responses to this study demonstrates how much those caring for someone with a rare disease need improvement both for their own quality of life, and to enable them to enrich the life of who they care for by having better provisions to facilitate that care.

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CONFLICT OF INTEREST

The authors have no conflicting interests to declare. AJM is a former board member and HM an existing board member of the NIRDP.

AUTHOR CONTRIBUTIONS

AJM and JM conceived of the project, designed the survey with input from patients, healthcare professionals and voluntary groups. JM and AC drafted the manuscript. JM and KD collected the survey data. AC, JM, and KD analysed the data. All authors contributed to data interpretation, manuscript revision and agreed the final version for submission.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available on request from the corresponding author. The data are not publicly available due to privacy or governance restrictions.

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SUPPORTING INFORMATION

Additional supporting information may be found online in the Supporting Information section.

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