

# Rare diseases and mental health in the UK - a quantitative survey and multi-stakeholder workshop

**Rosa Spencer-Tansley**

Genetic Alliance UK

**Nick Meade**

Genetic Alliance UK

**Farhana Ali**

Genetic Alliance UK

**Amy Simpson**

Genetic Alliance UK

**Amy Hunter** (✉ [amy@geneticalliance.org.uk](mailto:amy@geneticalliance.org.uk))

Genetic Alliance UK <https://orcid.org/0000-0001-5076-8761>

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## Research

**Keywords:** rare disease, undiagnosed, mental health, health service, patient experience, survey, recommendations

**Posted Date:** September 29th, 2020

**DOI:** <https://doi.org/10.21203/rs.2.19070/v2>

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## Abstract

**Background** Rare disease patients and carers report significant impacts on mental health but this has not been extensively studied. We explored the experiences of UK-based individuals through an online survey, and offer recommendations for policy and practice developed with a multi-stakeholder workshop.

**Results** In total 1,355 patients and 571 carers responded to the survey.

Due to their rare condition, the majority of respondents had felt worried/anxious (95%); stressed (93%); low/depressed (90%); emotionally exhausted (88%). Thirty-six percent of patients and 19% of carers had had suicidal thoughts.

Challenges that are particular to rare conditions and which negatively affect mental health included limited knowledge of the condition amongst healthcare professionals (88%), not being believed or taken seriously by healthcare professionals (80%), and lack of available information about the condition (76%).

Only 23% of respondents felt healthcare professionals considered mental health as equally important as physical health. Almost half of patients (46%) and carers (48%) reported never having been asked about their mental health, or that of the person they care for, by healthcare professionals.

Forty-six percent of respondents had received professional psychological support; the most common reason for not having accessed professional psychological support was that it had not been suggested (41%).

Fifty-nine percent of respondents had accessed sources of additional emotional support, such as patient groups, with the majority (75%) having found this support themselves.

With input from our multi-stakeholder workshop we developed recommendations for healthcare professionals to be supported to effectively and sensitively recognise and address patients' and carers' mental health needs; and for service level coordination of care to integrate professional psychological support with rare disease services.

**Conclusions** Living with a rare disease substantially impacts mental health. Many of the drivers of poor mental health reflect issues specific to managing rare conditions. To meet existing UK government commitments, there should be a focus on empowering healthcare professionals who treat rare disease patients and on integration of mental health support with rare disease services.

## Background

This study aimed to understand the impact of rare disease on mental health, for those affected by or caring for someone with a rare disease in the UK. That living with a rare condition may have negative psychological consequences has been frequently indicated anecdotally, but studies with UK rare disease patients as a collective cohort are restricted to non-peer reviewed reports [1, 2]. The experience of Australian rare disease patients has been somewhat better studied [3, 4, 5].

There is a policy drive to improve mental health services in the UK, with NHS England and the UK government promising sustained investment in order to ensure parity between physical and mental health [6, 7, 8]. The UK Strategy for Rare Diseases [9, 10] – although it does not mention mental health - commits to a person-centred approach for rare disease patients, thus creating a need for more attention on mental health. Our second aim was therefore to use our findings to develop recommendations and facilitate the translation of policy, such as the 'Five year forward view' from NHS England [6], into practice.

A common theme throughout the available literature is that the burden of rare disease is often considerable, and frequently includes a burden on psychological or emotional wellbeing. For example, Rare Disease UK's survey [2] of 1,203 rare disease patients and carers reported numerous psychological consequences for patients and carers, such as increased worry and isolation, yet that the majority of respondents did not feel they received 'sufficient psychological support.' Similarly, of the 30 Australian families affected by rare disease that were surveyed by Anderson et al [3], the majority reported high levels of psychological stress, yet few had received psychological support. Such findings indicate an unmet mental health need amongst rare disease patients and carers.

Further evidence is available from condition-specific qualitative studies, which have used approaches such as interviews or narrative analysis to explore the psychosocial impact of certain rare conditions, and have generally indicated impaired emotional/psychological wellbeing for patients and carers. For example, psychological or psychosocial consequences have been reported amongst individuals, and parents/carers, living with undiagnosed or medically unexplained conditions [11, 12]; and parents/carers of individuals with inherited metabolic disorders [13, 14].

Similarly, quantitative approaches have been employed to attempt to measure the psychological and/or psychosocial impact of specific rare diseases on patients and/or carers. This includes quality-of-life tools, which assess impact of rare disease on a number of variables, including variables related to emotional wellbeing and mental health. These studies have generally indicated considerable impairment on quality of life for rare disease patients, including individuals with oculopharyngeal muscular dystrophy [15], and epidermolysis bullosa [16]; and also parents and carers of individuals with epidermolysis bullosa [16] and CDKL5 disorder [17]. A number of groups have also applied validated tools to screen for depression and/or anxiety amongst rare disease patients, including individuals with myocarditis [18], AL amyloidosis [19] and mitochondrial disorders [20], and have typically reported significant mental health impairments. Additionally, a recent German cross-sectional study reported increased levels of depression and anxiety in a mixed rare disease cohort [21].

The majority of the available evidence is therefore condition-specific, and/or is confined to validated tools that are time-restricted and do not provide information about patients' and carers' past experiences. We wanted to address this gap in knowledge for the UK rare disease population and investigate: the impact on mental health, including the nature of the impact; contributing factors; experiences of support and services in relation to mental health; and steps towards better mental health support.

## Methods

A schematic of the project method is shown in figure 1.

The survey was developed to elicit the mental health experiences of rare disease patients and carers in the UK. The elements in the survey were derived from: a review of relevant literature; a call for relevant information from members of Genetic Alliance UK (over 200 patient groups); and thematic analysis of transcripts from eight interviews with rare disease patients and eight interviews with carers about the impact of rare disease on mental health and their experiences of mental health support [22, 23]. The survey structure reflected the themes identified from the interviews (the emotional impact of rare disease, stressors and promoters of mental health; and experiences of physical health services, professional psychological support and other sources of support). The mixed-methods approach ensured the survey questions were relevant to the research question, and offered support for the survey findings through triangulation of results [24].

The survey also included questions used in a previous study developed by The Neurological Alliance [25] with their permission. an adaptation of the Short Warwick–Edinburgh Mental Well-being Scale (SWEMWBS), (© NHS Health Scotland, University of Warwick and University of Edinburgh, [26]), and items from the Multidimensional Scale of Perceived Social Support (MSPSS) [27].

An advisory group provided guidance on the survey development. The group included rare disease patients and carers, clinicians, a representative from a national mental health charity and a social science researcher.

The majority of the survey questions were quantitative and fixed-choice. Question types included single answers, multiple choice answers, and Likert scale-items. A small number of questions included free-text responses, to give respondents the opportunity to share additional comments for example at the end of each section. Demographic information was collected at the end of the survey.

The survey was piloted with patients and carers and staff members of Genetic Alliance UK (five individuals in total). Volunteers piloted the survey in their own time and provided written or verbal feedback. Volunteers were asked to provide feedback on comprehensiveness, sensitivity, relevance and ease of completion. Additional comments or suggestions were also encouraged. The feedback was discussed with the research team and advisory group and agreed amendments were made.

## Recruitment

A link to the online survey was sent to members of Genetic Alliance UK (over 200 UK-based patient organisations); supporters of Rare Disease UK (over 2,000, including individuals and patient groups); and members of SWAN UK (syndromes without a name) which currently includes more than 1,800 families of children with an undiagnosed condition and an associated network of more than 600 professionals. The online survey was also shared on social media, including Twitter and Facebook, to increase our reach to individuals beyond our immediate networks.

The survey was hosted on SurveyMonkey and was live for a total of 28 days in November and December 2017.

Those eligible to take part were aged 18+, based in the UK, and either a patient with a rare disease, or a parent/carer. The first section of the survey included a set of screening questions to ensure respondents met the defined eligibility criteria, along with information about the study. A consent box was presented, which had to be ticked before the survey could be taken. Respondents had the option to withhold consent for the use of direct quotes from any free-text responses.

## Analysis

Descriptive statistics were used to analyse the data. The results are reported as percentages, and patients' and carers' responses are reported as combined figures for the majority of questions.

The findings of the survey and interviews [22, 23] were discussed at a multi-stakeholder workshop (see figure 1) in order to develop specific recommendations for change in policy and practice.

## Sample characteristics

In total 1,926 respondents responded to the survey (1,355 patients, 571 carers); 1,885 respondents of whom were eligible to take part (1,231 patients, 564 carers). Supplementary file 1 shows respondents' characteristics in full. The high proportion of female respondents is more marked for carers (92.4% female, 315/341) than for patients (83.9% female, 778/927). This may be explained in part by the fact that women may be more likely to respond to research requests

of this nature and are more likely to take on the primary care role [28]. The high numbers of white British responses (94.8%, 1,202/1,268), is over-representative in comparison to the population demographics of the UK [29]; whilst black, Asian, and other ethnicities were under-represented in our survey.

We presented an open text response box to collect as much diagnostic information as possible, and manually classified responses using the ERN (European Reference Networks) categories as a framework [30]. (Some respondents were listed under multiple categories, but most fell under just one category). There was a relatively high proportion of respondents representing immunological and neurological conditions, as well as connective tissue, neuromuscular and endocrine conditions. Amongst the carers' sample, there was a high proportion of conditions categorised as congenital malformations and intellectual disability (including undiagnosed conditions with a suspected genetic origin, and chromosomal abnormalities). Some condition categories had little to no representation, including respiratory diseases, urogenital diseases, and paediatric cancer. Such a spread, while introducing a level of bias, is not unexpected given the dissemination method used. Full data are shown in supplementary file 2.

Recommendations were drafted by the authors in response to the survey findings and with the aim of improving services through a focus on both empowering healthcare professionals and service-level coordination. The recommendations were refined through consultation at a multi-stakeholder workshop. The workshop consisted of 16 attendees including a psychologist, a behavioural therapist, representatives of rare disease patient organisations and mental health charities, individual patients and carers, representatives of professional bodies for healthcare professionals, academics, and a member of a government advisory committee.

The findings from the pre-survey interviews and from the survey were presented, followed by the draft recommendations, in order to stimulate constructive engagement. A whole-group discussion was facilitated after each recommendation was presented, ensuring all participants had the opportunity to speak. The discussion was focussed on refining the recommendations to make sure they were achievable and specific. Notes of the discussion were taken by three note-takers. The notes were combined, then checked and refined collaboratively by the authors. The resulting text was marked up as relevant to policy and practice (relating to individual healthcare professionals or to service-level organisation), or out of scope. The recommendations were revised in light of the workshop report.

## Results

### A. Quantitative survey findings

In total 1,926 individuals responded to the survey (1,355 patients, 571 carers). After excluding participants who did not live in the UK, did not provide consent, or did not have a rare condition, the total number of eligible respondents was 1,885 (1,231 patients, 564 carers). Completion rate was 69% for patients (913/1,231) and 60% for carers (340/564). Sample characteristics are reported in the 'methods' section of this paper.

In this paper the term 'carer' is used to represent parents and other non-professional carers of individuals with rare conditions.

## 1. The nature of the emotional impact

### i. Feelings and emotions

Respondents were given a list of thoughts and feelings and asked to rate their overall experience against each in terms of their experience of living with, or caring for someone with, a rare condition. Figure 2 illustrates the range of negative emotions that respondents reported. Almost all indicated that, as a result of the rare condition, they had felt (*some of the time/often/all of the time*): worried or anxious (95%, 1,604/1,688), stressed (93%, 1,578/1,689), emotional (92%, 1,556/1,685), low or depressed (90%, 1,509/1,683), angry or frustrated (90%, 1,514/1,690), emotionally exhausted (88%, 1,485/1,686), or alone (83%, 1,403/1,684). A large proportion of respondents reported they had experienced a sense of loss or grief (74%, 1,246/1,682), feeling at breaking point (70%, 1,183/1,686), and a sense of guilt (64%, 1,078/1,684). Thirty-six percent of patients (434/1,194) and 19% of carers (92/496) had experienced thoughts about suicide (figure 3).

Respondents had also experienced positive thoughts and feelings although this was less marked than for the negative feelings. The majority of respondents reported that, in relation to the rare condition, they had (*some of the time/often/all of the time*): been able to make up their mind (86%, 1,455/1,690), had been thinking clearly (77%, 1,290/1,683), had been dealing with problems well (77%, 1,291/1,688). More than half of survey respondents reported feeling useful (62%, 1,048/1,679), close to others (60%, 1,012/1,684) and optimistic about the future (59%, 996/1,687). Fewer than half of respondents (44%, 747/1,685) reported feeling relaxed; the percentage was lower for carers (34%, 170/498) than for patients (49%, 577/1,187).

### ii) Times that have had a negative impact on emotional health and wellbeing

As shown in figure 4, the majority of survey respondents reported (*agree/strongly agree*) that the following periods of time had negatively impacted mental health: onset of symptoms (84%, 1290/1530), trying to get a diagnosis (83%, 1235/1490), when a diagnosis was given (70%, 991/1418), coming to terms with the condition or diagnosis (83%, 1247/1506), the day-to-day challenges of living with the condition (88%, 1367/1559), and thinking ahead to the future (82%, 1272/1558). The full data are given in supplementary file 3.

### iii) Knock-on impact of poor mental health

Respondents indicated that poor mental health had negatively impacted other aspects of life (*agree/strongly agree*) (see supplementary file 4). This included a knock-on impact on physical health (87%, 1,242/1,420), on work or studies (81%, 995/1,222); and on personal relationships with partners (76%, 984/1,292), friends (71%, 999/1,414), and relatives (69%, 979/1,411).

## 2. Factors affecting mental health

The factors explored in relation to respondents' mental health can be grouped into three categories: i) interactions with healthcare professionals and services, ii) everyday living with a rare condition, and iii) additional factors for carers.

### i) Interactions with healthcare professionals and services

Respondents reported that interactions with individual healthcare professionals had negatively impacted mental health (figure 5 panel A, and supplementary file 5). Many of these factors related to the rarity of the condition. For example, the most frequently identified factor (*agree/strongly agree*) was lack of awareness or understanding of the condition amongst healthcare professionals (88%, 1,308/1,486), followed by not being believed or taken seriously by healthcare professionals when reporting symptoms (80%, 1,158/1,444), and being treated as a medical curiosity (50%, 714/1,433). Additionally, 80% of respondents (1,195/1,487) reported that conversations and interactions with individual healthcare professionals had directly and negatively impacted mental health.

Factors relating to service access and coordination were also reported to negatively impact mental health (figure 5 panel B, and supplementary file 5). This included (*agree/strongly agree*): trying to access health services or treatments (80%, 1,183/1,487), the way care is coordinated or organised between different departments or services (79%, 1,161/1,469), accessing financial support such as disability living allowance (76%, 951/1,254), and accessing other support such as social care or respite care (72%, 945/1,319).

### ii) Everyday living with a rare condition

Factors that are specific to everyday living with a rare condition were also reported to have impacted respondents' mental health (*agree/strongly agree*). This included lack of understanding about the condition amongst the public (90%, 1,313/1,458), feeling uncertain about what the future holds (87%, 1,279/1,469), having to explain the condition to other people (81%, 1,191/1,472), and lack of available information about the condition (76%, 1,096/1,448). Over half of respondents (56%, 792/1,411) identified worrying information they had come across online as a factor affecting mental health. See supplementary file 6 for full data.

For context we asked respondents about other stressors in their lives (see supplementary file 7). A large majority of respondents reported (*agree/strongly agree*) that major life events (86%, 1,196/1,389), financial pressures and worries (80%, 1,059/1,327) and feeling socially isolated (76%, 1,076/1,412) had impacted mental health.

### iii) Additional factors for carers

The survey captured additional factors that had affected the carers' psychological wellbeing. Almost all carers reported that worrying about their child's quality of life (97%, 370/383), and/or worrying about their child's emotional wellbeing (96%, 365/381) had affected their own mental health (*agree/strongly agree*). See supplementary file 8 for full data.

## 3. Evaluation of care

The survey explored respondents' experiences of services and how well they felt their emotional and mental health needs had been met.

### i) Services, parity of esteem and information

Figure 6 panel A shows that, when rating the care and treatment they have received, 62% of respondents (852/1,382) reported that they were satisfied (*fairly satisfied/satisfied/very satisfied*) with services to meet their physical health needs. In contrast, only 39% (532/1,357) were satisfied with services to meet their mental health needs. In response to the statement "I feel my/my child's mental health is considered equally as important as my/my child's physical health by healthcare professionals involved in my/my child's care," the majority of respondents (60%, 814/1,349) chose '*disagree*' or '*strongly disagree*'. A minority of respondents chose '*agree*' or '*strongly agree*' (23%, 316/1,349) (figure 7 panel B).

In addition, approximately half of respondents were satisfied (*fairly satisfied/satisfied/very satisfied*) with the information they were provided about the physical condition (48%, 653/1,367), while fewer respondents (30%, 411/1,366) were satisfied with the information provided about sources of emotional support (supplementary file 9).

## ii) Healthcare professionals asking about mental health

Respondents were presented with the statement “healthcare professionals ask about my/my child’s mental and emotional wellbeing’. Almost half of patients (46%, 454/988) and carers (48%, 173/362) chose *‘never’*, and just 8% of patients (81/988) and carers (28/362) chose *‘often’* or *‘always’*.

In a separate question for carers only, 57% (208/363) reported they had *‘never’* been asked about their *own* mental health by healthcare professionals involved in their child’s care. Full data are given in supplementary file 10.

A proportion of respondents reported (*agree/strongly agree*) having had positive experiences when healthcare professionals had discussed mental health and wellbeing with them (figure 7): some reported that the discussions were handled sensitively (35%, 377/1,068), that they felt genuine (34%, 369/1,086) and that they had a positive impact on emotional wellbeing (24%, 252/1,066).

Conversely respondents also reported (*agree/strongly agree*) having had negative experiences when discussing mental health and wellbeing with healthcare professionals (figure 8), including that the discussions made them feel anxious (44%, 468/1061); uncomfortable (36%, 386/1060) or made them feel worse (34%, 356/1059).

For full data see supplementary file 11.

## iii) How to improve care to better support wellbeing and mental health

Our findings, given in full in supplementary file 12, show that a large majority of respondents felt that the following would improve their mental health (*agree/strongly agree*): greater awareness of the emotional challenges of living with a rare condition amongst healthcare professionals (91%, 1,184/1,308), greater sensitivity amongst healthcare professionals (85%, 1,090/1,284), and being asked more frequently about mental health and wellbeing by healthcare professionals (81%, 1,056/1,296).

Respondents also reported that easier access to emotional support would improve their mental health; the majority felt (*agree/strongly agree*) that easier access to professional psychological support (85%, 1,069/1,262) and better signposting to alternative sources of emotional support (86%, 1,105/1,283), would benefit their mental health.

# 4. Experiences of professional psychological support

## i) Access and barriers to professional psychological support services

Just over half (54%, 719/1323) of respondents had not accessed any professional psychological support. Of those who had accessed professional psychological support, just 7% had accessed it through a specialist clinic for their condition (41/588) and only 2% of respondents (14/588) had been offered professional psychological support at the time of diagnosis of the rare condition. More respondents had been referred by their GP (48%, 280/588) or clinicians at their hospital (21%, 123/588). A proportion of respondents (18%, 106/588) had accessed private psychological support (i.e. ‘arranged and paid for it myself’). Of these respondents, over half (55%, 58/105) had paid more than £500 in total and approximately one quarter (26%, 27/105) had paid between £100 and £500.

Respondents identified several factors that had prevented them from accessing professional psychological support (full data are given in supplementary file 13). The most commonly reported factor was that it had not been suggested by healthcare professionals involved in their care (41%, 527/1,285), followed by not being able to afford private psychological support (29%, 372/1,285) and that the waiting lists for mental health services were too long (23%, 299/1,285). Carers in particular cited too much pressure on their time as being a barrier (22%, 77/350).

## ii) Evaluation of professional psychological support

Respondents who indicated that they had accessed professional psychological support were asked to evaluate the support received (figure 8). Around half of these respondents (49%, 282/570) had found it was helpful (*agree/strongly agree*), but fewer (37%, 209/571) felt it was tailored to their needs (*agree/strongly agree*).

When asked about ease of access, over half of respondents (56%, 319/571) reported (*agree/strongly agree*) they did not have to travel far to access the support. A smaller proportion felt that the support was available when needed (31%, 179/578) or had been easy to access (27%, 155/580). Fewer than half

(34%) were confident they would be able to access professional psychological support again if they needed it (198/580) (*agree/strongly agree*).

## 5. Support from other sources

Fifty-nine percent of respondents (759/1,292) reported they had accessed additional emotional support such as peer support online (81%, 617/759) or face-to-face (36%, 274/759) and support from charities or the community sector (25%, 192/759).

The majority of respondents (75%, 534/713) who had accessed such support reported having found it themselves. A minority had been signposted to the support by a healthcare professional (18%, 125/713). and the most frequent reason for not accessing such support was not knowing how to access it (41%, 210/516). Eighteen percent of respondents believed there was no such support available to them (92/516). See supplementary file 14 for full data.

The vast majority of respondents indicated (*agree/strongly agree*) that such sources of support were helpful for a variety of reasons (figure 9).

Over half of respondents (67%, 862/1,282) reported that their family 'really tries to help them' (*agree/strongly agree*) but fewer felt that they get the emotional support they need from their family (48%, 612/1,281). Similarly, a proportion of respondents (47%, 588/1,256) reported that their friends really try to help them (*agree/strongly agree*) but fewer felt that they get the emotional support they need from friends (37%, 462/1,257).

### 1. Multi-stakeholder workshop recommendations

The survey findings, and recommendations for changes to policy and practice drafted by the authors, were discussed at a multi-stakeholder workshop. The recommendations were subsequently refined as follows:

## Recommendations for the empowerment of healthcare professionals:

1. Healthcare professionals should be provided with the skills, knowledge and capacity to:
  - demonstrate awareness of the challenges of living with a rare disease,
  - handle discussions about mental health sensitively.
2. Healthcare professionals should routinely signpost patients and carers to sources of support.

### Recommendation for service-level coordination:

3. Coordinated rare disease services should include assessment of mental health needs and access to mental health services. This should be extended to carers.

## Discussion

### The nature of the emotional impact of rare disease

Data documenting the impact of rare disease on mental health are scarce. The current study is significant in that it is not restricted to a specific rare disease type and has a broad scope of investigation that includes the nature of the impact of living with a rare disease, what factors affect mental health, experiences of support and services in relation to mental health, and what patients and carers judge would improve mental health support.

Our findings demonstrate that rare disease has a substantial negative impact on mental health for both patients and carers. This reflects existing grey literature such as a survey of UK- and US-based respondents which reported high levels of 'worry,' 'anxiety and stress' and 'depression' amongst rare disease patients and carers [1] and a Europe-wide survey that found 38% of respondents felt they needed psychological support [28]. It is also consistent with findings from condition-specific studies which used diagnostic tools to measure anxiety and/or depression in rare disease patients and/or carers [18, 19, 20] and impact on quality of life [15, 16, 17].

Our findings show that the negative impact on mental health can be chronic, and can occur or recur at different time periods such as during the 'diagnostic odyssey'; when a diagnosis is given; and while coming to terms with the condition. This is valuable as it highlights those periods when additional emotional support may be particularly important. The impact of the 'diagnostic odyssey' is concerning: it can last a long time, with one in four waiting longer than five years for a diagnosis [2] and many will not see the end of this phase, as the diagnostic yield of genome sequencing remains significantly lower than 100% [31, 32]. This means that the mental health of rare disease patients is affected before they are categorically defined as such, and before they might access support through coordinated services or disease-specific patient support groups. Other groups have asserted that receiving a rare disease diagnosis may be a particularly emotionally challenging time [3], in which patients and families experience feelings such as shock, prompting the authors to call for routine

psychological support at the time of diagnosis. It may seem counterintuitive that both not having a diagnosis, and receiving a diagnosis, can lead to poor mental health, but both offer challenges; for example, the anxiety of not knowing what is 'wrong' on one hand, and the shock of a poor prognosis on the other. Moreover, our study also indicates that patients and carers need ongoing support in addressing the emotional challenges that a rare condition can present. Future research could usefully explore individuals' experiences during these periods, and use validated tools to facilitate the assessment of interventions.

Notably, whilst almost all of our survey respondents reported negative emotions in relation to their condition, many also reported having experienced positive thoughts and feelings, and they described a variety of coping mechanisms during an earlier qualitative study [22, 23]. It would be valuable to explore further what factors promote positive adjustment and coping in people living with rare diseases and whether there are any differences to those living with common chronic conditions.

## Factors affecting mental health

An important finding from this study is that many of the drivers of poor mental health that were identified by respondents are associated with challenges related to managing a condition that is rare. This includes a lack of understanding about the condition amongst the public, feeling uncertain about what the future holds, and having to explain the condition to other people. Similarly, many contributing factors arising from interactions with health and other services reflected issues associated with the rarity of the condition. For example, a lack of awareness amongst healthcare professionals, not being believed or taken seriously by healthcare professionals, and being treated as a medical curiosity. Factors related to service access were also likely related to the rarity of the condition, in particular the way care is coordinated; it is well established that care is often poorly coordinated for rare disease patients, many of whom have complex needs and are usually under the care of several doctors from different specialties [2].

Our findings are consistent with previous research into the experiences of Australian families affected by rare diseases from Zurynski et al [5] who found that delay in diagnosis was associated with anxiety, frustration and stress, and that the perceived lack of knowledge amongst healthcare professionals was associated with increased parental stress and anxiety. Furthermore, Zurynski's group reported that parents perceived the lack of knowledge amongst clinicians to be a leading cause of diagnostic delays, highlighting an interrelation between two emotional stressors. Similarly, interviews conducted with caregivers of children with inherited metabolic conditions [14] identified emotional challenges associated with poor coordination of healthcare services and there is anecdotal evidence about the burden of time and energy required to manage numerous appointments across different specialisms [33]. Having symptoms questioned by healthcare professionals, or being mislabelled with a psychological health issue when trying to seek help for physical symptoms, which 80% of our respondents agreed had impacted mental health, have also been previously been reported as triggers of poor mental health in specific patient populations [11].

It is known that long-term conditions significantly increase the risk of mental health issues. For example, previous analysis has indicated that a third of individuals with a long-term physical health condition also have a comorbid mental health issue [34, 35]. Such associations have been the subject of policy-level initiatives in the UK, such as the UK Government's 2011 'No Health without Mental Health' report [36]; and the 'Five Year Forward View on Mental Health' report [6, 7] which emphasised the need to address the link between physical and mental health. The implication of our findings is that those affected by rare conditions may face additional stressors, on top of those associated with managing chronic health conditions, because of the rarity of their condition. As such they may be particularly vulnerable to experiencing mental health issues.

The interrelation between physical and mental health is complex [37, 38]. As well as evidencing the impact on mental health of living with a rare disease, our findings reflect the worsening of physical health that can occur as a result of mental health challenges. Rare disease patients are not immune from this kind of downward spiral.

It is also important to remember that patients do not live in a vacuum and may be balancing multiple psychosocial issues along with those directly associated with their condition. Our survey highlights that patients and carers face the challenges of everyday life such as financial pressures, major life events, social isolation, and work and employment issues. It may be that such factors are exacerbated by the rarity of the condition. For example, social isolation is likely to be a particularly pertinent issue for rare disease patients and carers [2] due to factors such as poor understanding of the condition amongst the public and peers; whilst previous research has highlighted the considerable financial costs, associated with managing rare conditions, that patients and carers face [39].

## Practice implications based on our findings

The recommendations arising from this study, around the empowerment of healthcare professionals and service-level coordination, are targeted at specific government departments, advisory bodies, professional medical bodies and medical schools [22]. Alongside other patient organisations, healthcare professionals and NHS policy makers, Genetic Alliance UK is directly contributing to the successor to the UK Strategy for Rare Diseases, which will be a new policy framework for rare conditions. We are therefore able to use these recommendations to guide future policy in this area.

## Recommendations for the empowerment of healthcare professionals:

1. Healthcare professionals should be provided with the skills, knowledge and capacity to:
  - demonstrate awareness of the challenges of living with a rare disease,
  - handle discussions about mental health sensitively.
2. Healthcare professionals should routinely signpost patients and carers to sources of support.

### Recommendation for service-level coordination:

3. Coordinated rare disease services should include assessment of mental health needs and access to mental health services. This should be extended to carers.

In their 2016 paper, Molster et al [4] advocated for greater awareness of rare disease amongst clinicians, to improve the diagnostic process and ensure informational needs are met. We propose, based on our findings, that this should also extend to greater awareness of the emotional challenges of rare disease. Healthcare professionals could show greater sensitivity when delivering a diagnosis to patients and carers, for example [5]. Our findings also indicate a need for greater sensitivity when interacting with patients without a diagnosis (80% of our respondents agreed being misbelieved or not taken seriously had impacted their mental health). This is consistent with calls from previous studies exploring the experiences of patients living without a diagnosis [11, 12], as well as anecdotal evidence [33].

Our respondents were clear that they would benefit from healthcare professionals asking more frequently about mental health, with the majority reporting that they are never or infrequently asked about mental health. To our knowledge this is the first study to assess the frequency of such conversations and the finding is reflected in a recent policy report that suggests patients with rare neurological conditions are less likely to be asked about their mental health than neurology patients with more common conditions [40]. Given that many rare disease patients have frequent contact with healthcare professionals [2], this is alarming. It is vital that healthcare professionals are provided with training so they are aware of the emotional challenges faced by their rare disease patients, they are able to communicate with sensitivity, and are equipped to handle discussions around mental health. This is underscored by our finding that while some respondents had positive experiences when discussing mental health with healthcare professionals, for others the conversations had directly caused increased levels of anxiety.

It may be that healthcare professionals would feel more comfortable raising the subject of mental health if they know that appropriate professional psychological support is available should it be indicated. Healthcare professionals need to be confident about how to make such referrals, but the current level of mental health service provision might be a barrier even for professionals who feel personally equipped to ask their patients about their mental health; recent cuts to funding of psychological support services have resulted in high thresholds for access and long waiting times [41].

Additional sources of emotional support, such as peer support and support from patient groups, has been shown to be important to rare disease patients and carers by other researchers [3,5] and patient organisations [2]; our findings support this and add detail about why this is. However, fewer than half of our respondents had accessed such support, a finding reflected by previous studies [2, 3]. Our survey indicates this is largely because of not knowing how to find it and that healthcare professionals are not routinely signposting to additional sources of support, possibly due to a lack of knowledge [1]. Training and professional development should therefore incorporate information about signposting.

There are resources available to help healthcare professionals signpost to support in the UK, for example on the website of Genetic Alliance UK [42]. It is important to note however that the small numbers of individuals affected by each rare condition may mean condition-specific support is not always available but there is a growing number of online communities, including on Facebook and through platforms such as Rare Connect, connecting individuals across geographical areas [43]. Notably, amongst our survey respondents, online support was by far the most frequently utilised of all additional sources of support.

Our data indicate problems with access to effective professional psychological support, and a lack of parity of esteem between physical and mental health. Given the high need for support that our study has evidenced, we recommend that coordinated rare disease services should include assessment of mental health needs and access to mental health services, and, crucially, that this should be extended to carers.

We found that patients and carers face multiple barriers to accessing professional psychological support, such as it not being suggested by healthcare professionals, and a large majority of our respondents also felt that easier access to professional psychological support would improve their mental health. Our study echoes findings from research in Australia [4], the UK [2] and across Europe [28] that found few patients and carers felt they had received sufficient psychological support. Our findings also indicate that when professional psychological support is accessed it could be more effective and better tailored to patients' needs. This is consistent with a recent survey of UK-based individuals with neurological conditions [40].

Parity of esteem is a principle in which mental health is given equal priority to physical health, and in 2014, 'genuine parity' was listed by NHS England [6] as an ambition for achievement in the National Health Service by 2020. Our data have uncovered an apparent lack of parity of esteem for patients and carers living with a rare disease. We propose that assessment of mental health and a care pathway to support services, as part of coordinated care, be instigated for rare disease patients and carers. Very few of our respondents received psychological support via specialist rare disease centres. NHS England and the UK government have recently made several commitments to make mental health a key healthcare focus, with promises of sustained investment, in order to ensure parity between physical and mental health [6, 7, 8]. The UK Strategy for Rare Diseases [9, 10] has also seen commitments from the UK government to integrate health and social care services, and a person-centred approach for those affected by rare diseases. Our recommendation aligns with these national commitments and underscores the need for them to be implemented. The gold standard for rare disease care is generally held to be coordination through multidisciplinary team working. Indeed, previous studies in Australia have recommended that psychological support is "embedded in RD services" [5] and that psychological support is routinely available following diagnosis [3].

There is a similar need for national plans and mainstream policies to improve mental health support for rare disease patients and their carers in other nations. While 25 of the EU member states have national plans, a commitment to improved psychological support is still lacking and European governments have recently been called on to address this [44]. National policy in North America, Asia and Australia is largely focussed on research and treatment innovations, although the Canadian Organization for Rare Disorders has developed a national strategy and is calling for its implementation [45, 46]. Change in all regions may be encouraged by the recognition of rare diseases by the World Health Organization as a priority disease area, and its identification of psychosocial care as an area of unmet need [47].

A key strength of this study is that it was not restricted to particular rare diseases, addressing a significant gap in knowledge as the majority of research conducted previously was condition-specific, and/or did not explore the impact on mental health in depth. The value of undertaking research into patients' and carers' experiences across rare diseases as a collective, in a healthcare system that is oriented towards more common diseases, has been highlighted by previous researchers [4], as it will "raise the profile of rare diseases and is likely to result in less duplication of efforts and resources across the range of rare diseases."

We did not set out to address the question of whether there is under-diagnosis of mental health conditions amongst rare disease patients and carers. Our approach has allowed for a deeper understanding of patients' and carers' experiences of their mental health now and during their past. An additional strength is the inclusion of stakeholders throughout the project, on the project advisory group and through the multi-stakeholder workshop.

Our sample was self-selecting therefore it is not possible to determine the generalisability of our findings, although the large sample size provides some confidence. There could be a bias toward those with more difficult experiences of mental health.

Further consultation with patients, carers and service providers, with experience of different types of rare disease, is needed to tailor guidelines for implementation of our recommendations. We expect that our recommendations, through feeding into national policy work, will provide the foundation for this progress to be made.

## Conclusion

To our knowledge this is the largest UK-based study to systematically explore the impact of living with a rare disease on mental health. Our findings indicate a substantial impact on mental health for both patients and carers and that many drivers of poor mental health reflect issues that are specific to managing a condition that is rare. Based on our findings and discussion with stakeholders we recommend that healthcare professionals should be provided with the skills, knowledge and capacity to demonstrate an awareness of the challenges of living with a rare disease; and that healthcare professionals should routinely signpost to sources of support. We also recommend that coordinated rare disease services should include assessment of mental health needs and access to mental health services, and that this should be extended to carers. If effectively implemented, these changes could do much to address some of the mental health issues patients and carers in the UK currently face; and to ensure that mental health is considered as important as their physical health. Since our data were analysed, the covid-19 pandemic has been a significant additional stressor for the mental health and wellbeing of rare disease patients and carers, making improvements in support at a national level even more urgent [48].

## List Of Abbreviations

ERN: European Reference Networks

## Declarations

## Ethics approval and consent to participate

Approval for this study was granted by the Social Care Research Ethics Committee, reference 17/IEC08/0028. Consent to participate in the online survey was secured via an obligatory tick box ahead of the survey questions. Participants had the option of withholding consent for use of direct quotes from any free text responses.

## Consent for publication

## Availability of data and materials

The datasets used and/or analysed during the current study are available from the corresponding author on reasonable request. The full survey is also available on request.

## Competing interests

The authors have no competing interests to declare.

## Funding

The study was funded by public donations to Rare Disease UK, the national campaign for people with rare diseases and all who support them. Rare Disease UK is a campaign run by Genetic Alliance UK, the national charity of over 200 patient organisations, supporting all those affected by genetic conditions.

## Authors' contributions

RST and AH designed the study and survey. RST analysed the survey data. NM and FA led on the recommendations development; all authors contributed to the workshop output analysis. RST and AH wrote the paper, with advice from AS and feedback from the other authors.

## Acknowledgements

We would like to acknowledge the members of the advisory group and the multi-stakeholder workshop for their input, all the patient groups who helped disseminate the survey and the individuals who participated, and Sarah Gilbert for supporting the workshop.

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## Figures

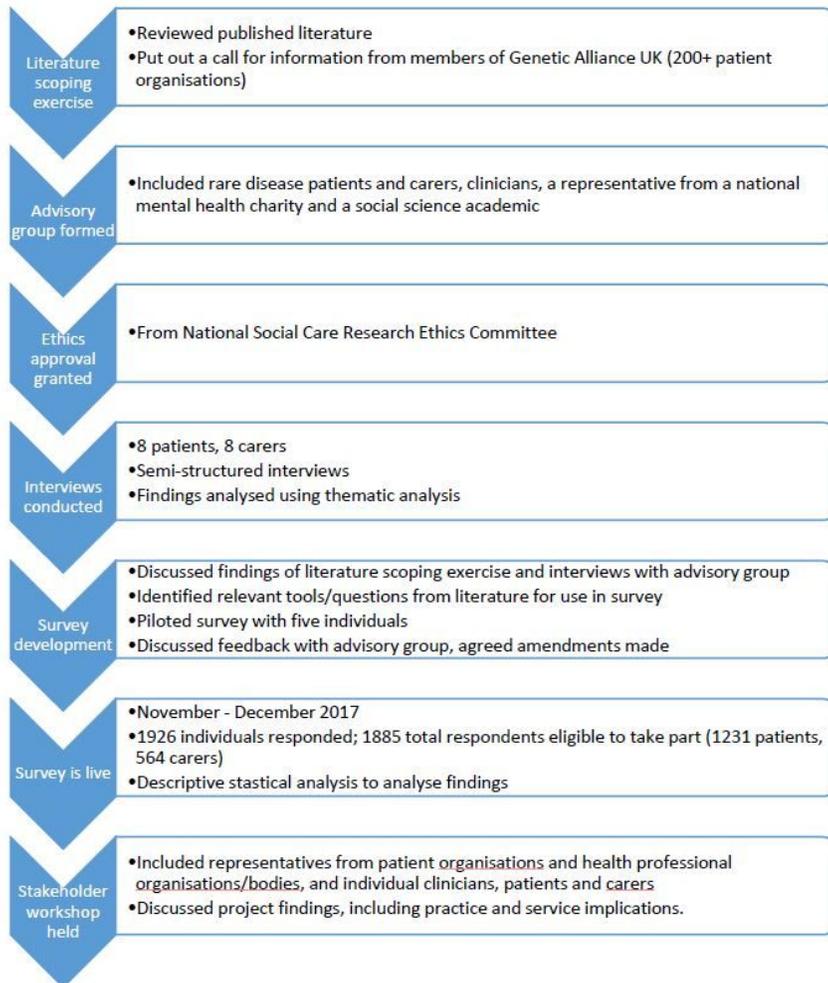
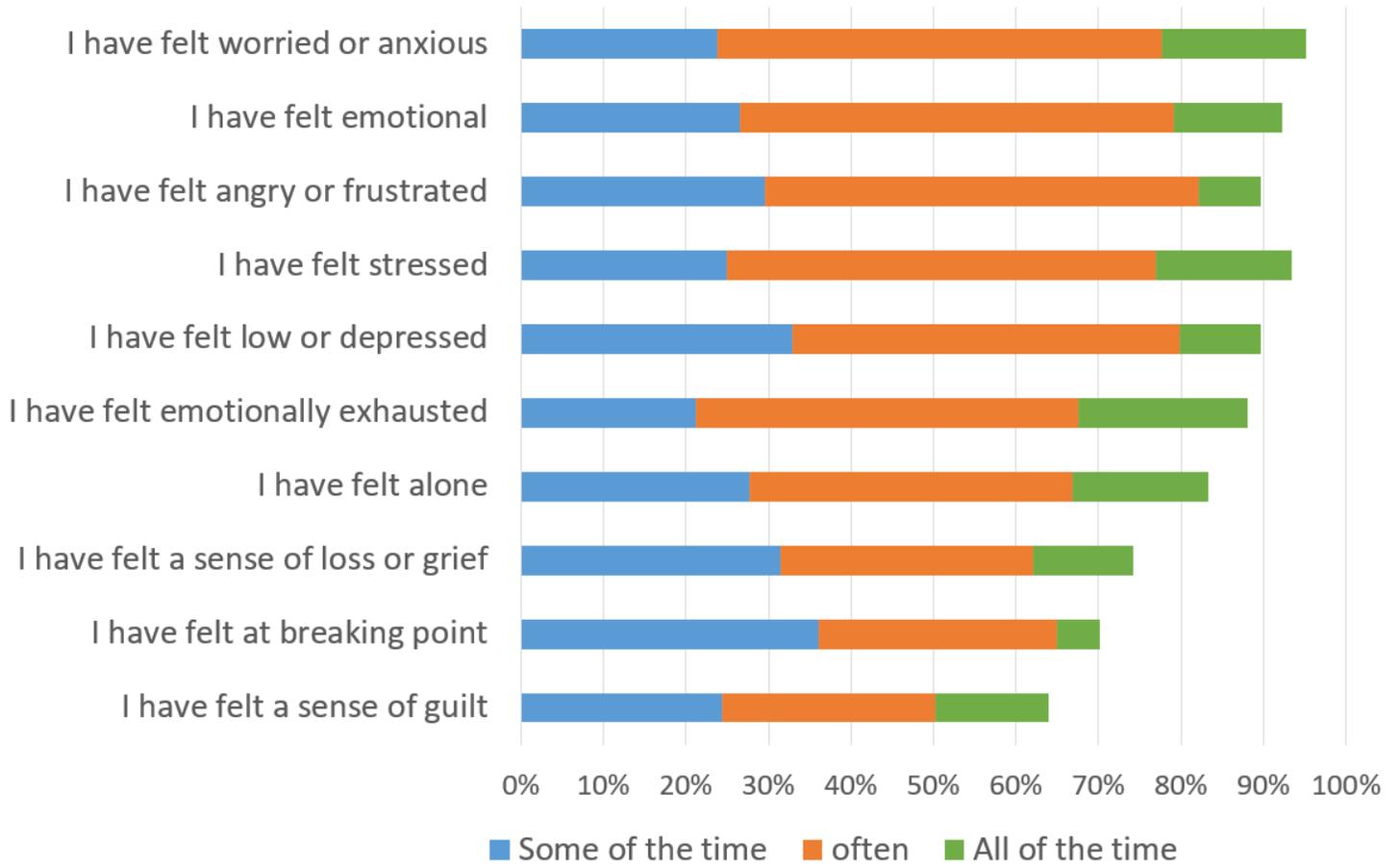


Figure 1

Flow chart of study methods



**Figure 2**  
Emotions attributed by respondents to living with a rare condition. Percent of respondents choosing some, often or all of the time. Patients and carers combined.

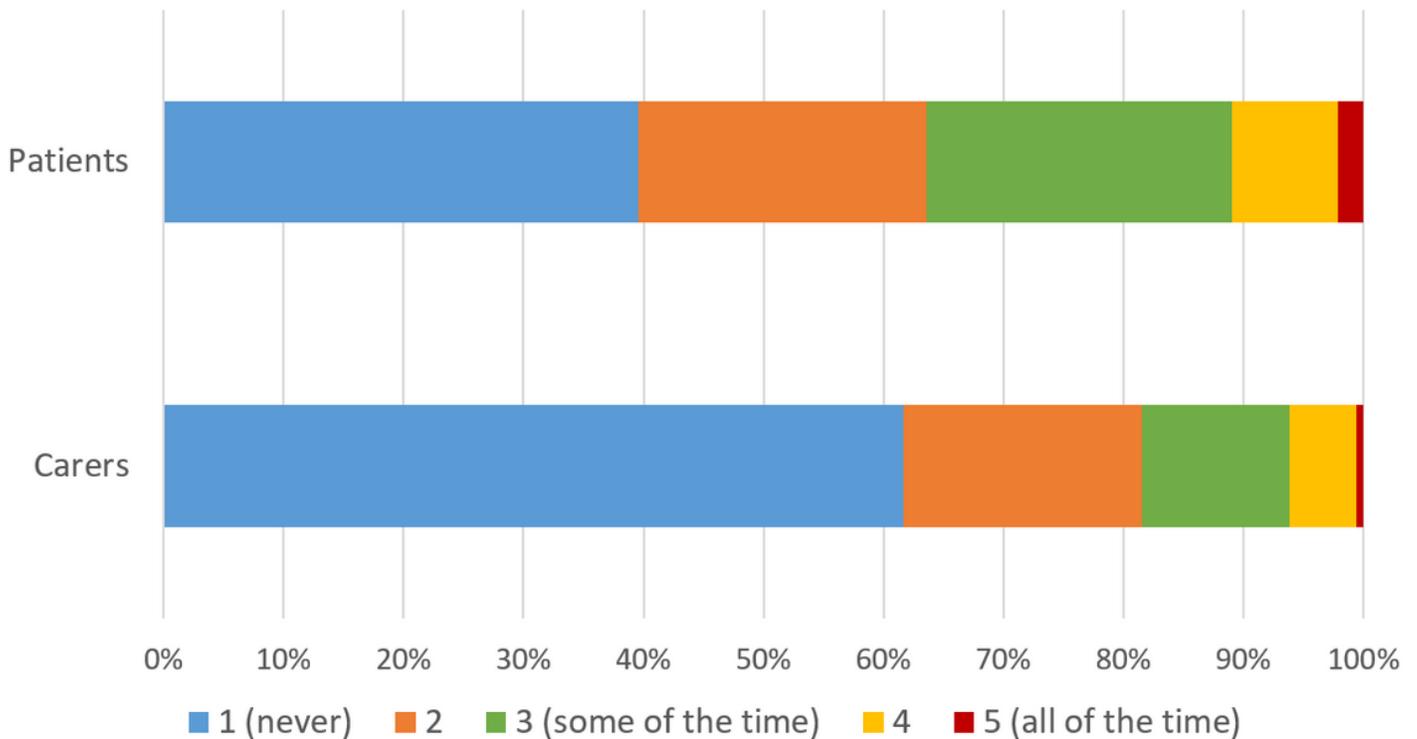


Figure 3

"I've had thoughts about suicide", attributed to rare condition.

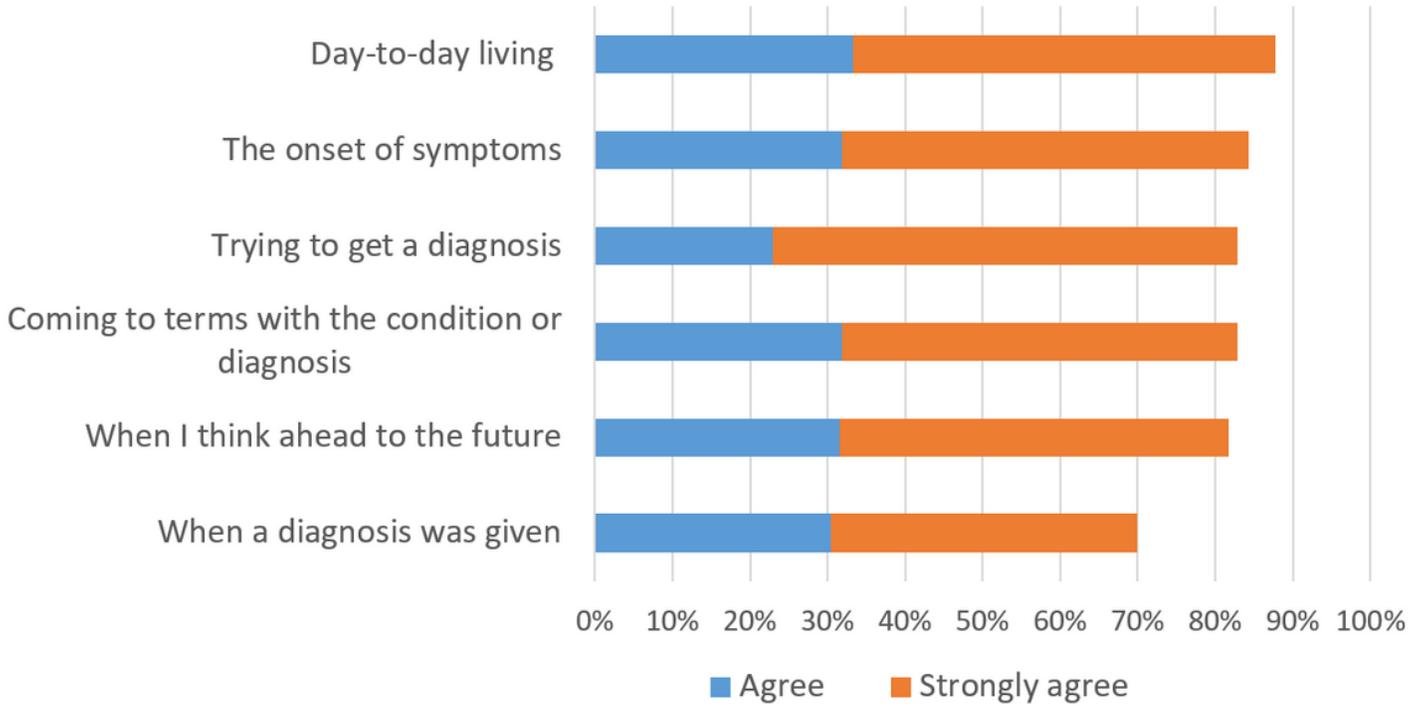


Figure 4

Periods reported to have a negative effect on mental health Percent of respondents choosing agree or strongly agree. Patients and carers combined.

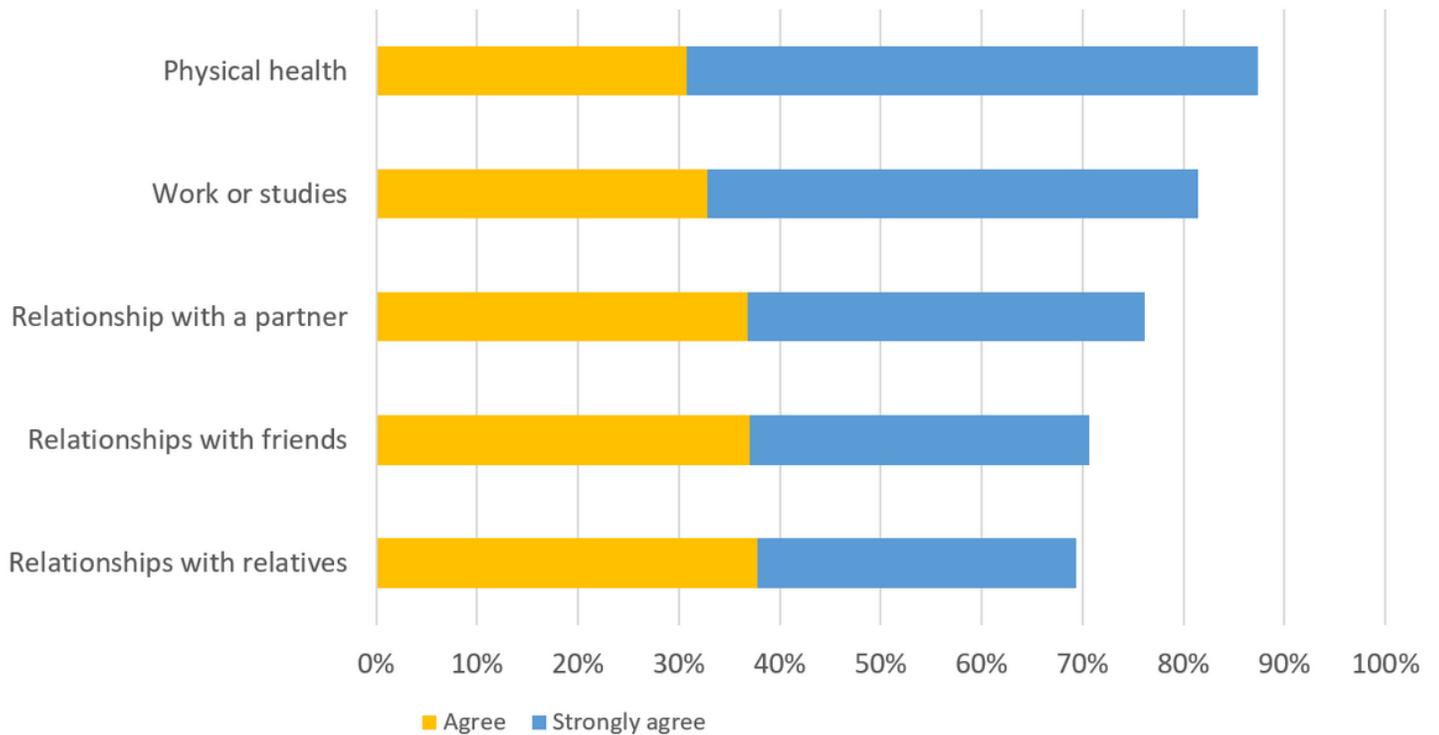


Figure 5

"The following things have had a negative impact on my emotional health and wellbeing". Panel A shows the extent to which respondents agree that interactions with healthcare professionals have had a negative impact (percent of respondents, patients and carers combined). Panel B shows the extent to which respondents agree that interactions with services have had a negative impact (percent of respondents, patients and carers combined).



**Figure 6**  
 Evaluation of physical vs mental health services, and parity of esteem Panel A shows the rating of physical vs mental health services. Panel B shows the level of agreement with physical and mental health having parity of esteem. Both panels show percent of respondents, patients and carers combined.

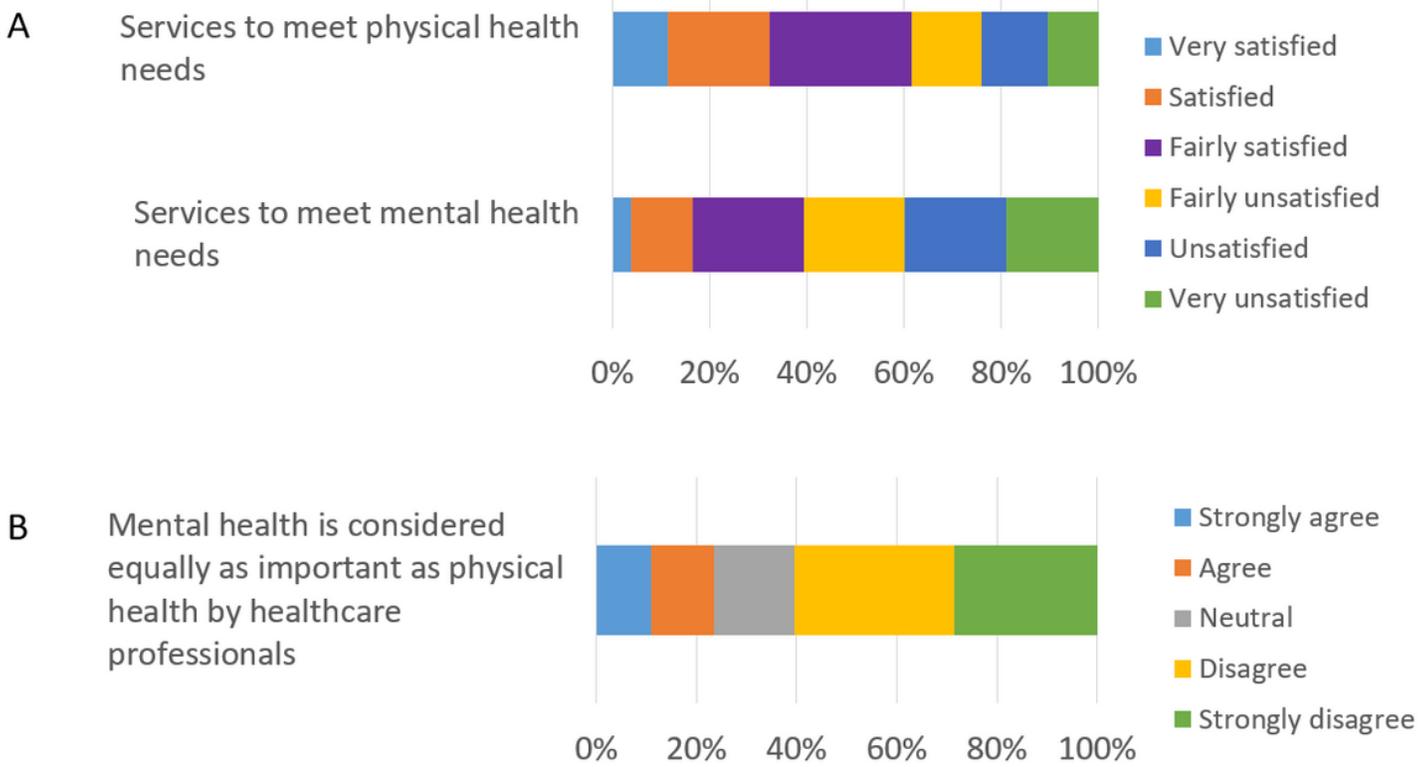


Figure 7

"Discussions about mental health and wellbeing with healthcare professionals...". Percent of respondents that agreed or strongly agreed with the statements, patients and carers combined.

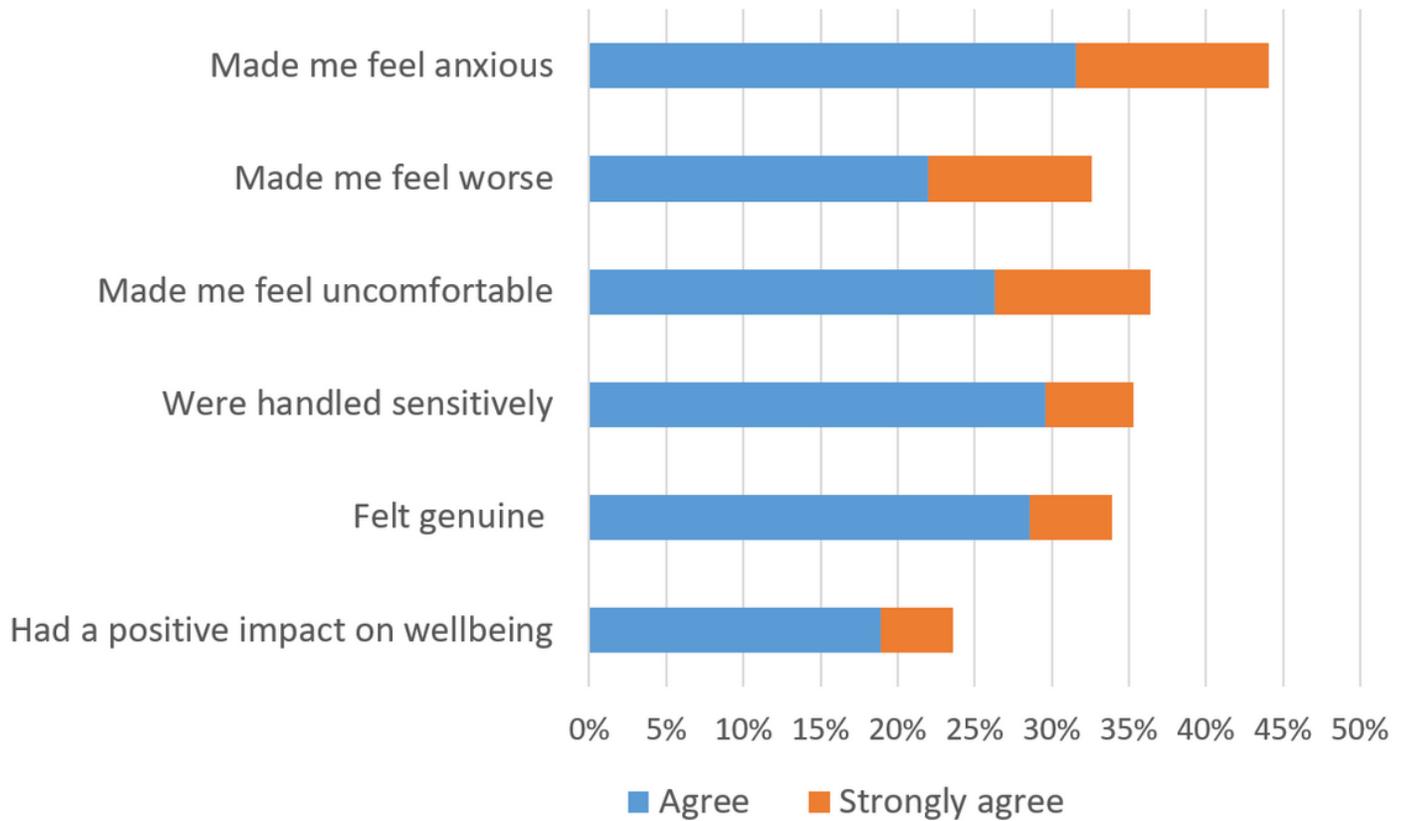


Figure 8

Evaluation of professional psychological support received. Extent to which respondents agreed with statements about the support received. Percent of respondents, patients and carers combined.

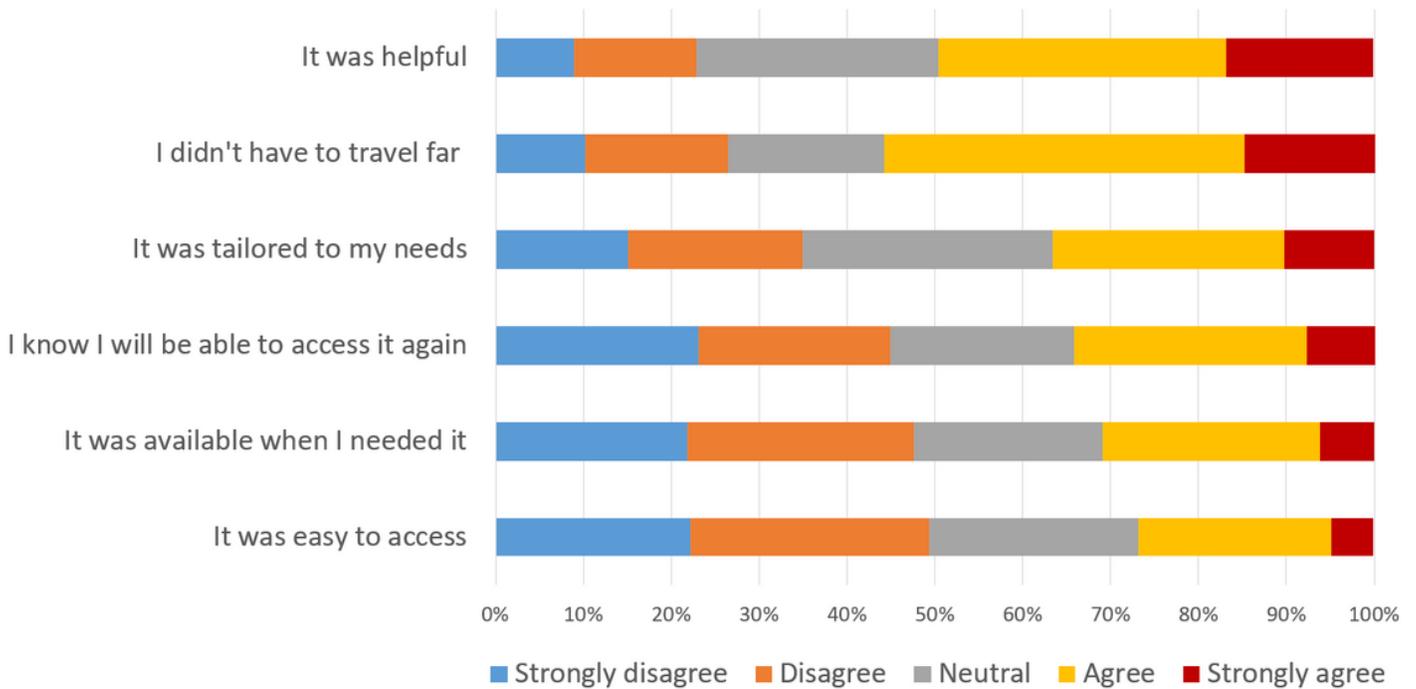


Figure 9

Ways in which other sources of support are helpful. Percent of respondents who agreed or strongly agreed with answers to the question "How has this support been helpful to you?". Patients and carers combined.

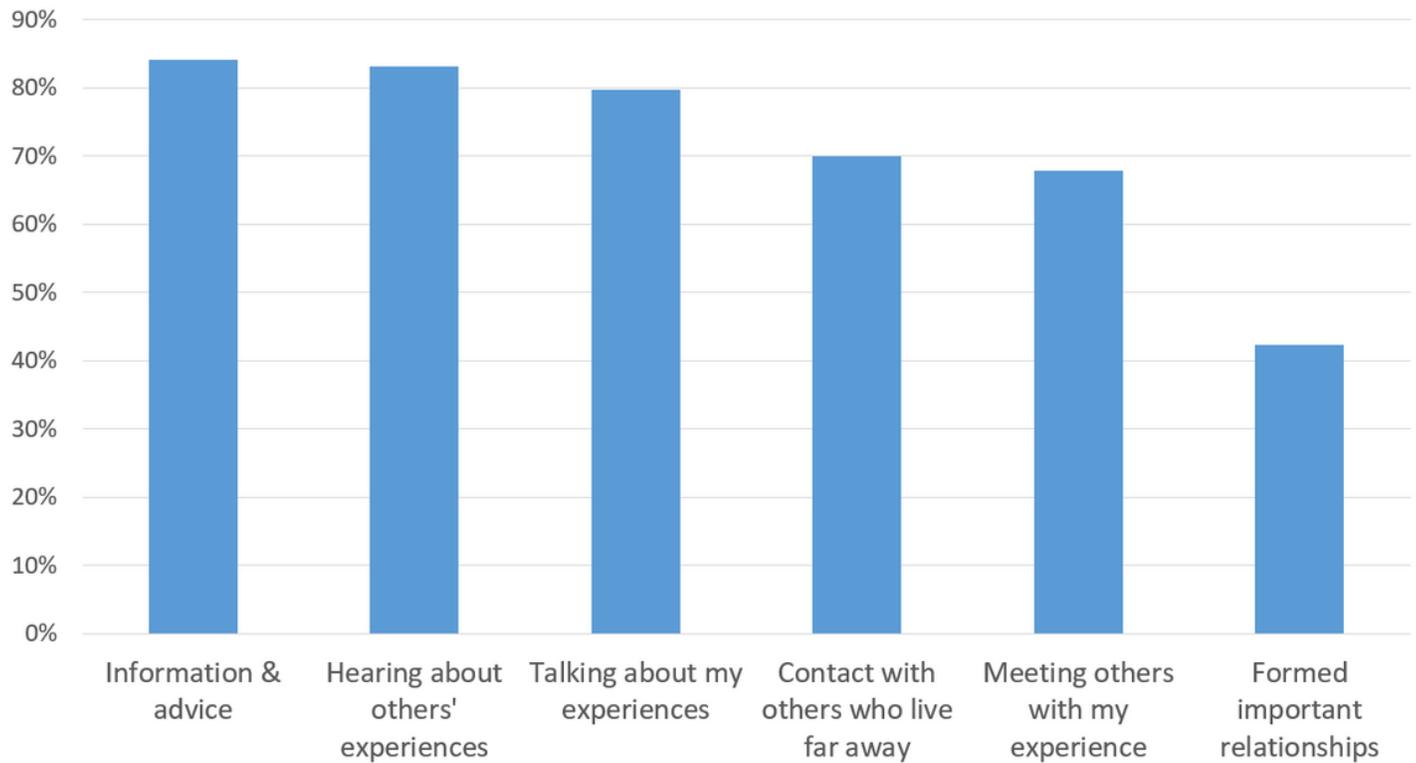


Figure 10

## Supplementary Files

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