RARE EXPERIENCE 2020

The lived experiences of people affected by genetic, rare and undiagnosed conditions

GENETIC ALLIANCE UK

ACKNOWLEDGEMENTS

Genetic Alliance UK is the national charity working to improve the lives of patients and families affected by all types of genetic conditions. We are an alliance of over 200 patient organisations.

Rare Disease UK is a multi-stakeholder campaign run by Genetic Alliance UK, working with the rare disease community and the UK's health departments to ensure the development and delivery of high quality policy for people affected by rare conditions.

SWAN UK (syndromes without a name) is a patient and family support service run by Genetic Alliance UK. SWAN UK offers support and information to families of children with undiagnosed genetic conditions.

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Thank you to all that responded to our survey and participated in our workshops, without your contribution this report would not have been possible. Thank you to our members who worked with us to disseminate our survey.

Quotes in this report are anonymised contributions from workshop participants and from survey respondents. We use percentages throughout this report to allow for consistent comparisons, we do this even where the denominator is less than one hundred, where we give both sides of the ratio.

The Rare Experience 2020 project has been funded (under the name Patient Experience Report) through a collaboration agreement with Alexion. As part of this agreement Alexion have a non-exclusive licence to use aggregated data from the findings of this work. This arrangement is in accordance with Genetic Alliance UK's Working with Industry Policy geneticalliance.org.uk/working-with-industry-policy/ and Ethical Collaboration Policy geneticalliance.org.uk/ethical-collaboration-policy/.

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FOREWORD

The Orphanet database catalogues more than 6,000 rare conditions[1]. While rare conditions affect 1 in 17 people during their life-time, around 70%[2] begin in childhood and are life-long. Understanding the experiences and preferences of people affected by rare conditions is fundamental to providing care and treatment and to ensuring support, information and services are available and targeted to meet needs.

This is the third comprehensive survey Genetic Alliance UK has undertaken to capture the experiences of those affected by rare conditions. It is seven years since the publication of the UK Strategy for Rare Diseases (the Strategy)[3], and a decade since we published the report of our first comprehensive survey. Over the course of this decade we have seen significant progress in science, medicine and in terms of awareness of rare diseases. Our 2020 survey provides a measure of the extent to which these advances have filtered through to impact on the lives of those affected by rare conditions, how their experiences have changed across the last decade, and a baseline against which we might judge future progress.

Whilst satisfaction remains high amongst those who have access to specialist care and treatment for their condition, we see few improvements for the majority of those who do not. The diagnostic odyssey remains a major challenge; new genomic services have yet to deliver on the promise of early and accurate diagnoses. The scale and frequency at which patients and families experience challenges relating to coordination of care seems to have been unaltered by the passage of time. Despite the commitments made in the UK Strategy, which came without specific financial resources dedicated to its implementation, there remains much to be delivered.

The new Framework for Rare Diseases is due to be published shortly and at a time when it is greatly needed. The force of the pandemic continues to bear heavily on the NHS and on the health and wellbeing of those with rare conditions who are amongst the most vulnerable to Covid-19's impacts. The pandemic has brought interruptions to healthcare and whole-scale disruption of access to education and social care and exacerbated the fundamental challenges for those with rare conditions. But there are learnings from the pandemic experience that could, properly harnessed, serve to accelerate the delivery of yet to be met promises.

It is essential that all those involved in the implementation of the Framework for Rare Diseases recognise the value of the data presented in this report. We encourage those seeking to measure the impact of initiatives, activities and outputs linked to the Framework, to look to the patient experience survey for their key measures of success.

Jayne Spink PhD
Chief Executive
Genetic Alliance UK

INTRODUCTION

POLICY LANDSCAPE

The UK Strategy for Rare Diseases was published in 2013, covering the years to the end of 2020. The Strategy, agreed by all four health departments of the UK, aimed to 'ensure that no one gets left behind just because they have a rare disease'.

Key features of the Strategy included: personal care plans covering health and care services; access to information for patients, their families and carers; better methods of identifying and preventing rare diseases; improving diagnosis; providing better education and training for health and social care professionals; and building on research to improve personalised approaches to healthcare for those with a rare disease.

In the years that followed the devolved administrations published implementation plans to take forward the recommendations of the Strategy. Scotland (2014) was first, followed by Wales (2015), and Northern Ireland (2015). England's implementation plan was split in two between NHS England's responsibilities and the Department of Health and Social Care (covering all other stakeholders in England). Both were published in 2018, more than four years after the launch of the Strategy. A report[4] from the All Party Parliamentary Group on Rare, Genetic and Undiagnosed conditions (for which Genetic Alliance UK provides the secretariat), showed that the community of people living with rare, genetic and undiagnosed conditions felt that this delay contributed to a lack of coordination between the nations and to a reduction in impact of the Strategy. The Strategy and all four implementation plans came with no additional funding with commitments intended to be met by existing funding arrangements.

In February 2019 Baroness Blackwood, then Parliamentary Under-Secretary of State for Life Science with responsibility for rare diseases, announced the development of a UK Framework for Rare Diseases in preparation for the end of the Strategy in 2020. In October 2019 a national Rare Disease Conversation was launched to collect views to inform the new policy. We understand that the model for implementation of the new Framework will mirror the previous Strategy, with individual nations producing their own Action Plan for Rare Diseases in line with their own strategic objectives.

METHODOLOGY

An online survey of 102 questions was carried out in late June to early August 2020. The survey was open to anyone aged 18 or over who considered that they, or the person they care for, has a rare, genetic or undiagnosed condition. The link to the survey was shared widely across Genetic Alliance UK's networks, including to supporters of our Rare Disease UK campaign and membership of the SWAN UK (syndromes without a name) support group for families with children with undiagnosed conditions.

In total there were 1,020 eligible responses: exclusions were for those outside the UK, those who had not given consent and those who completed less than three quarters of the survey. The questions were based on previous surveys undertaken by Genetic Alliance UK, other patient experience surveys (such as the NHS England Cancer Patient Experience Survey), other relevant studies (such as the CONCORD study focussing on care coordination) and the anticipated focus of the expected UK Framework for Rare Diseases. The survey was hosted online using the SurveyMonkey platform and the raw data was then imported into SPSS where the data was cleaned (respondents excluded as outlined above) and descriptive statistics were produced.

Just under 300 different rare / genetic conditions are represented in the survey, with around 190 only being mentioned by one respondent while 18 conditions were mentioned by nine or more respondents. About 10% of respondents (90) mentioned living with more than one rare condition (see tables 1 and 2).

Following the survey, two online workshops took place with respondents who had completed the survey and indicated that they would be interested in taking part in workshops related to the survey. The workshop participants came from all four UK nations and represented both carers and people with a rare or undiagnosed condition (see table 3). The findings from the four main sections of the report were presented and then discussed with the participants. The outputs from the workshops are also included within the report findings.

The survey was broken down into several sections: demographics of the person answering the survey (and the person cared for if a carer was answering the survey), details about the rare / undiagnosed condition, the search for a diagnosis, information and awareness about their condition, coordination of care, access to specialist care and treatments, experiences related to research and use of technology and overall experiences of care. An additional section was added to the end of the survey concerning experiences due to Covid-19, the findings from this work will be published elsewhere. In each section there were fixed response questions as well as open ended questions; quotes have been taken from these questions throughout the report.

The quantitative data in the report has on occasion been broken down for analysis purposes. Breakdowns include whether the person living with the condition is a child or adult, how long ago the person was diagnosed (if they have a definitive diagnosis) and the complexity of the condition. The complexity of someone's condition was defined by how many different aspects of health were affected by the rare or undiagnosed condition; this was then grouped into five categories – 1 or fewer aspects of health affected, 2-3 aspects, 4-5 aspects, 6-7 aspects and 8 or more aspects of health affected.

The survey was answered more often by women (847 – 83%) than men (162 – 16%), a small proportion of respondents said either 'other' or 'prefer not to say' while some skipped this question. People living with a condition made up 82% of the respondents while the other 18% were carers – see table 4 for a detailed breakdown of the demographics of the respondents and the people cared for if a carer completed the survey.


DAM

RESPONDENT PROFILE

Table 1 – How often conditions were mentioned

Number of mentions	Count of number of different conditions
Once	190
Twice	26
Three times	14
Four times	11
Five times	2
Six times	1
Seven times	4
Eight times	6
Nine or more times	18

Table 2 – Number of conditions per respondent

Number of rare conditions mentioned	Number of respondents (%)
1	800* (90%)
2	69 (8%)
3	16 (2%)
4	2 (0.2%)
5+	2 (0.2%)
Total respondents	890[#]

**One person mentioned they did not have a diagnosis*
[#]11 respondents who said that they had a definitive diagnosis did not provide the name of the condition.

Table 3 – Workshop attendees

Demographic	Participants	
Sex	males	4
	females	7
Carer / person with condition	carers	6
	people with condition	5
Region	England	3
	Scotland	5
	Wales	2
	Northern Ireland	1
Age group	25-34	2
	35-44	3
	45-54	2
	55-64	2
	65-74	2
Diagnosed / undiagnosed	undiagnosed	2
	diagnosed	9

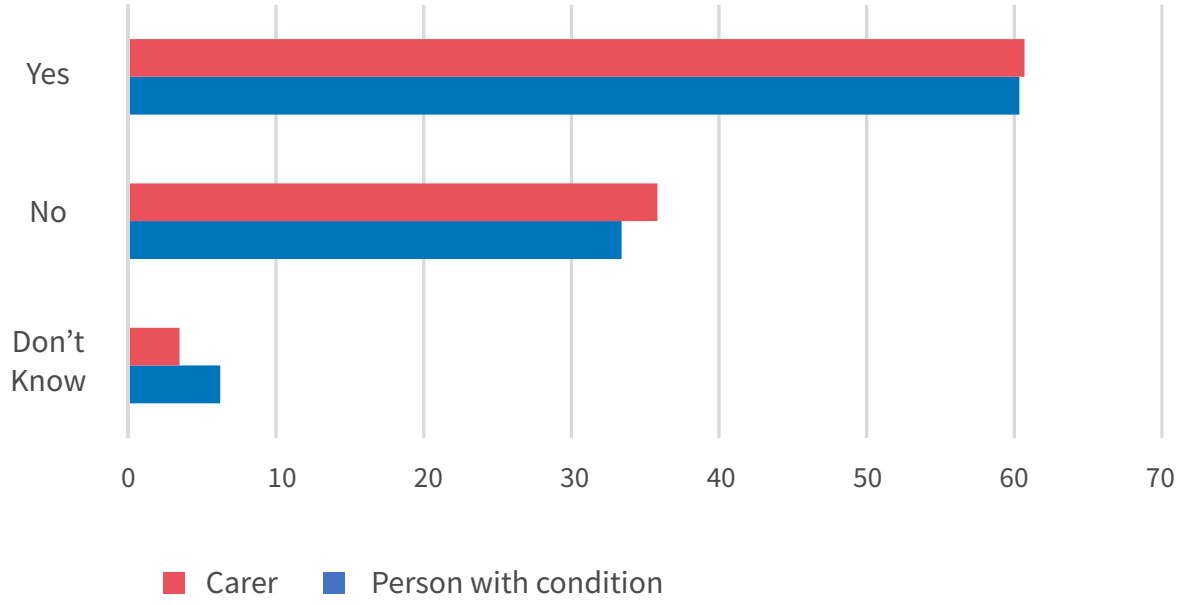
Table 4 – Demographics of the respondents answering the survey and those they care for

		Person with condition answering the survey	Carer answering the survey	Carer describing person they care for
Sex	Male	144 (17%)	18 (10%)	101 (55%)
	Female	680 (82%)	167 (90%)	82 (44%)
	Other	3 (0.4%)	0	1 (0.5%)
	Prefer not to say	4 (0.5%)	1 (0.5%)	1 (0.5%)
Age	Under 18	NA	NA	135 (73%)
	18-24	51 (6%)	2 (1%)	22 (12%)
	25-34	120 (14%)	25 (13%)	11 (6%)
	35-44	159 (19%)	74 (40%)	4 (2%)
	45-54	213 (26%)	53 (29%)	1 (0.5%)
	55-64	176 (21%)	21 (11%)	4 (2%)
	65-74	93 (11%)	9 (5%)	6 (3%)
	75+	19 (2%)	1 (0.5%)	2 (1%)
	Prefer not to say	2 (0.2%)	1 (0.5%)	0
Region	East of England	76 (9%)	10 (5%)	10 (5%)
	East Midlands	54 (7%)	17 (9%)	17 (9%)
	London	71 (9%)	13 (7%)	13 (7%)
	North East & Cumbria	35 (4%)	6 (3%)	6 (3%)
	Northern Ireland	12 (1%)	2 (1%)	2 (1%)
	North West of England	93 (11%)	18 (10%)	18 (10%)
	Scotland	73 (9%)	20 (11%)	20 (11%)
	South East of England	155 (19%)	45 (24%)	43 (23%)
	South West of England	92 (11%)	21(11%)	23 (12%)
	Wales	58 (7%)	10 (5%)	10 (5%)
	West Midlands	45 (5%)	13 (7%)	13 (7%)
	Yorkshire	64 (8%)	11 (6%)	11 (6%)
	Prefer not to say	5 (0.6%)	0	0
Ethnicity	White- English/ Welsh/ Scottish/ Northern Irish/ British	747 (90%)	164 (88%)	168 (91%)
	White- Irish	12 (1%)	5 (3%)	4 (2%)
	White- Any other White background	36 (4%)	11 (6%)	5 (3%)
	Mixed/ multiple ethnic groups- White and Black Caribbean	4 (0.5%)	3 (2%)	3 (2%)
	White and Asian	3 (0.4%)	0	1 (0.5%)
	Any other mixed/ multiple ethnic background	4 (0.5%)	0	1 (0.5%)
	Asian/ Asian British- Indian	3 (0.4%)	0	0
	Pakistani	2 (0.2%)	1 (0.5%)	0
	Any other Asian background	1 (0.1%)	0	0
	Black/ African/ Caribbean/ Black British- African	2 (0.2%)	0	0
	Caribbean	2 (0.2%)	0	0
	Any other Black/ African/ Caribbean background	0	0	1 (0.5%)
	Other ethnic group- Arab	1 (0.1%)	0	2 (1%)
	Other ethnic group- Any other ethnic group	4 (0.5%)	0	0
	Prefer not to say	9 (1%)	2 (1%)	0

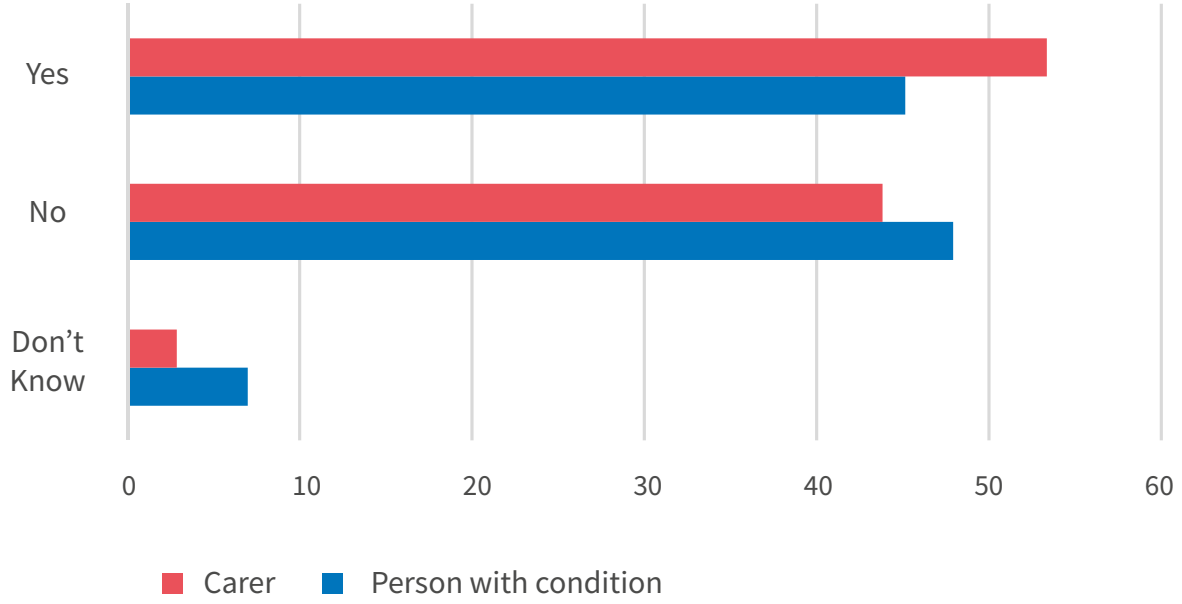
		Person with condition answering the survey	Carer answering the survey	Carer describing person they care for
Work / study status	Work full time (35 hours per week or more) in paid employment (this may include working from home)	172 (21%)	34 (18%)	3 (2%)
	Work part time (less than 35 hours per week) in paid employment (this may include working from home)	141 (17%)	59 (32%)	2 (1%)
	Self employed	56 (7)%	15 (8%)	2 (1%)
	Do agency/supply work (this may include working from home)	7 (1%)	2 (1%)	0
	Do voluntary work (this may include working from home)	72 (9%)	21 (11%)	2 (1%)
	Not working or unemployed	280 (34%)	54 (29%)	30 (17%)
	Recently furloughed as a result of the Covid-19 pandemic	33 (4%)	8 (4%)	0
	Recently made redundant as a result of the Covid-19 pandemic	11 (1%)	2 (1%)	0
	Attend school/college/university (may be currently learning from home)	52 (6%)	5 (3%)	92 (51%)
	Attend special needs school/college/university (may be currently learning from home)	3 (0.4%)	0	38 (21%)
	Retired	117 (14%)	5 (3%)	4 (2%)
	Carer	3 (0.4%)	11 (6%)	0
	Prefer not to say	11 (1%)	2 (1%)	1 (0.6%)
				Pre-school age 11 (6%)
Ability to work affected	Yes	436 (52%)	105 (57%)	48 (26%)
	No	252 (30%)	62 (33%)	16 (9%)
	Don't know	43 (5%)	6 (3%)	7 (4%)
	Not applicable	101 (12%)	13 (7%)	113 (61%)
Ability to study affected	Yes	189 (23%)	56 (30%)	70 (38%)
	No	197 (24%)	46 (25%)	78 (42%)
	Don't know	34 (4%)	3 (2%)	6 (3%)
	Not applicable	413 (50%)	81 (44%)	31 (17%)

Impact of rare conditions on respondents' lives

Ability to work affected:



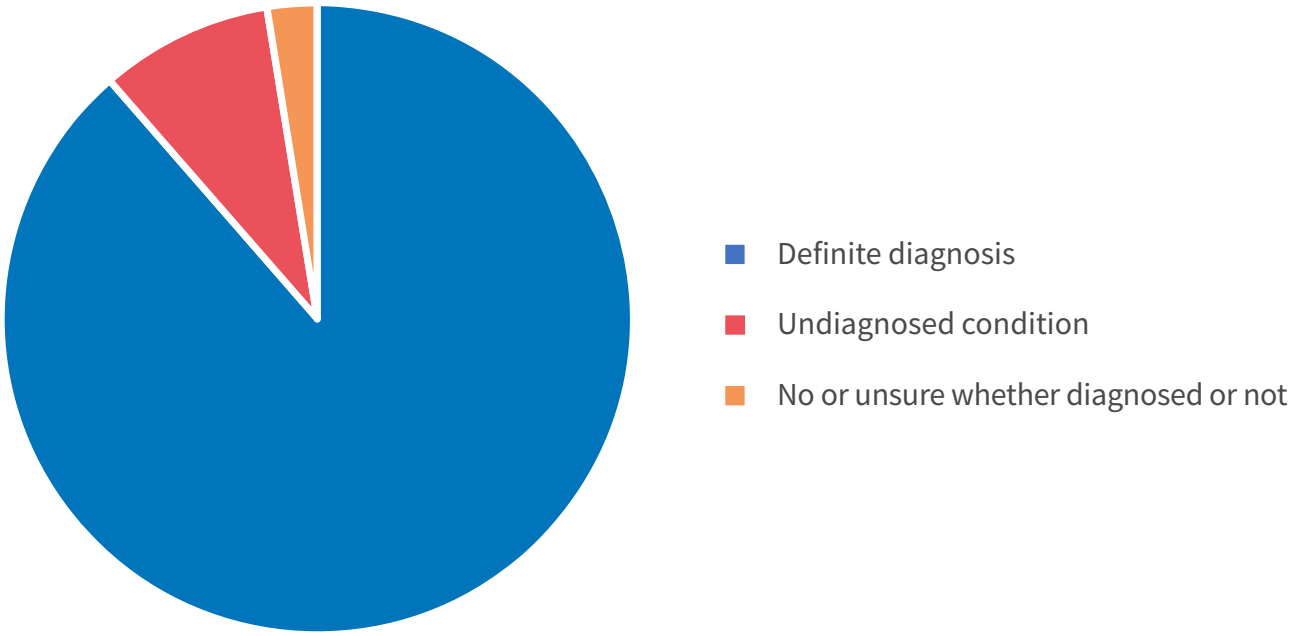
Ability to study affected:



For those who said it was applicable, around 60% of carers and people with a rare condition have had their ability to work affected, and a slightly smaller proportion had their ability to study affected.

DIAGNOSIS

Whether person with lived experience has been diagnosed or not



The majority, 901 (88%), of respondents had a diagnosis, compared with 90 (8.8%) who were undiagnosed.

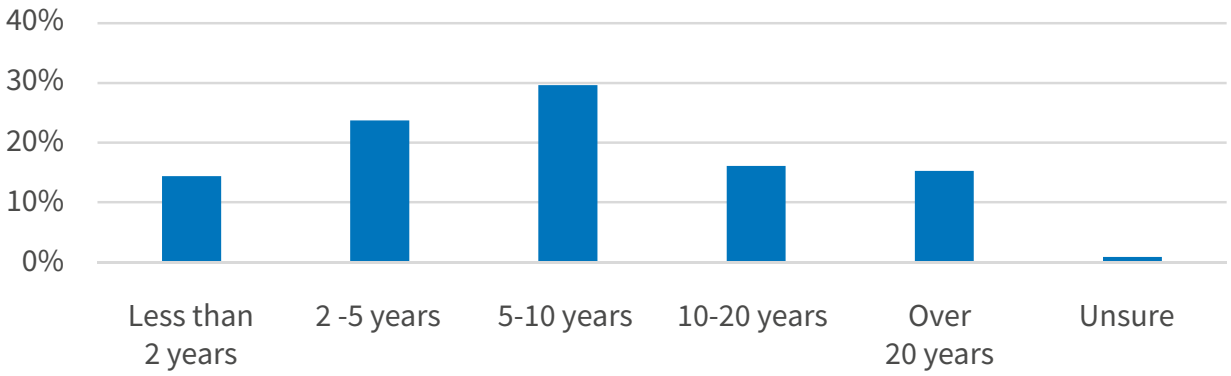
There are two components of the undiagnosed group:[5]

- People who are living with an identifiable rare condition. They could be diagnosed if the right diagnostic tool was applied or they met the appropriate clinician to make a diagnosis
- People who are affected by a condition which has not been properly identified and characterised either through identification of the causative gene or through a clinical definition by a clinician – a syndrome without a name. They cannot be diagnosed until this condition is characterised.

Both of these groups are on a diagnostic odyssey, and while they are on their journey it is difficult to separate the two. To serve them both well, it is necessary to acknowledge that they can be either group, and not make an assumption either way. To assume the former could delay access to appropriate treatment of symptoms for a family with a child with a syndrome without a name. To assume the latter could mean that someone receives generic care for their symptoms and does not access a specialist clinic to address their condition.

In the past there has been a tendency for those affected by a condition that is not properly identified to accept diagnoses that are descriptions of symptoms, such as 'global developmental delay' or 'behaviour disorders'. The advance of genomic diagnostic tools may have lead to more members of this group searching harder or longer for a diagnosis, as it becomes better understood that there may be a genetic cause for their condition.

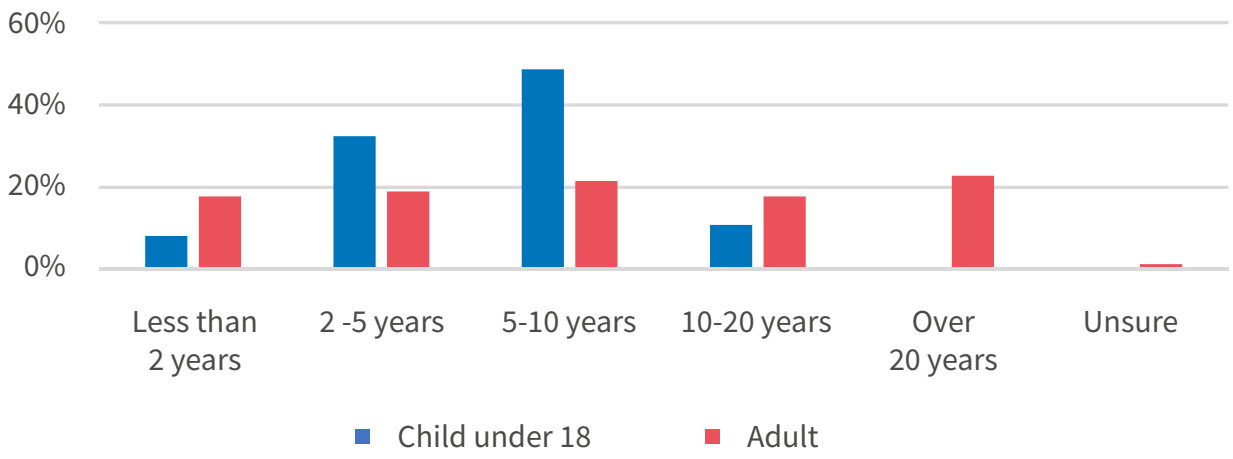
For those who are undiagnosed – how long it has been since they first consulted a doctor about the onset of symptoms



Please state how long it has been since you/they first consulted a doctor about the onset of symptoms

In total 62% of respondents without a diagnosis have been undiagnosed for more than five years since they first contacted a healthcare professional about the onset of their symptoms.

For those who are undiagnosed – how long it has been since they first consulted a doctor about the onset of symptoms x whether person with condition is now a child or an adult



This graph would appear to show that a significant portion of people who are still searching for a diagnosis after five years since first displaying symptoms were children. Of the 37 undiagnosed children 22 (59%) have searched for a diagnosis for more than 5 years. Of the 79 undiagnosed adults 49 (62%) have searched for 5 or more years. Of course, a portion of the adults in this category will have been children when they first displayed symptoms.

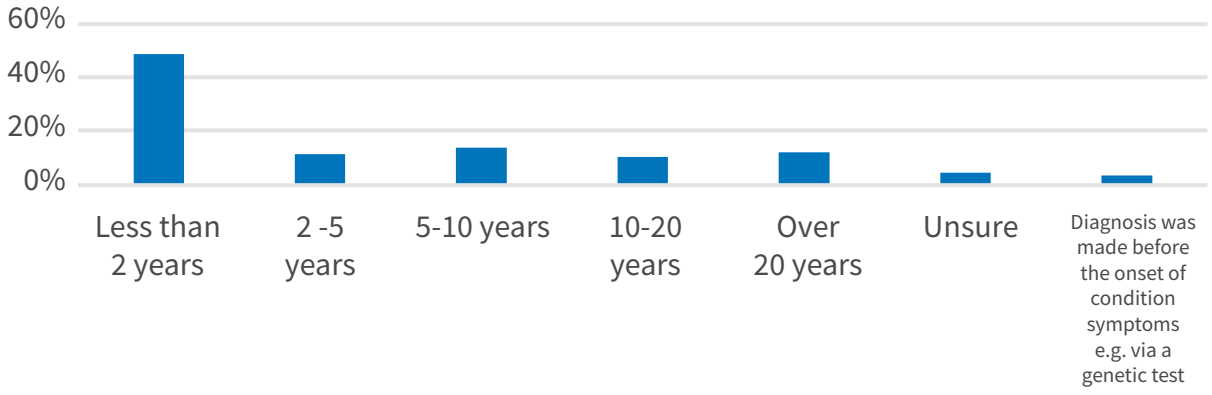
This community is the focus of the SWAN UK (syndromes without a name) support network run by Genetic Alliance UK. This project aims to deliver the same support that condition specific member groups of Genetic Alliance UK supply to people diagnosed with rare and genetic conditions.

Journey to a successful diagnosis

For respondents who have a diagnosis, we can see that their journey has not been smooth.

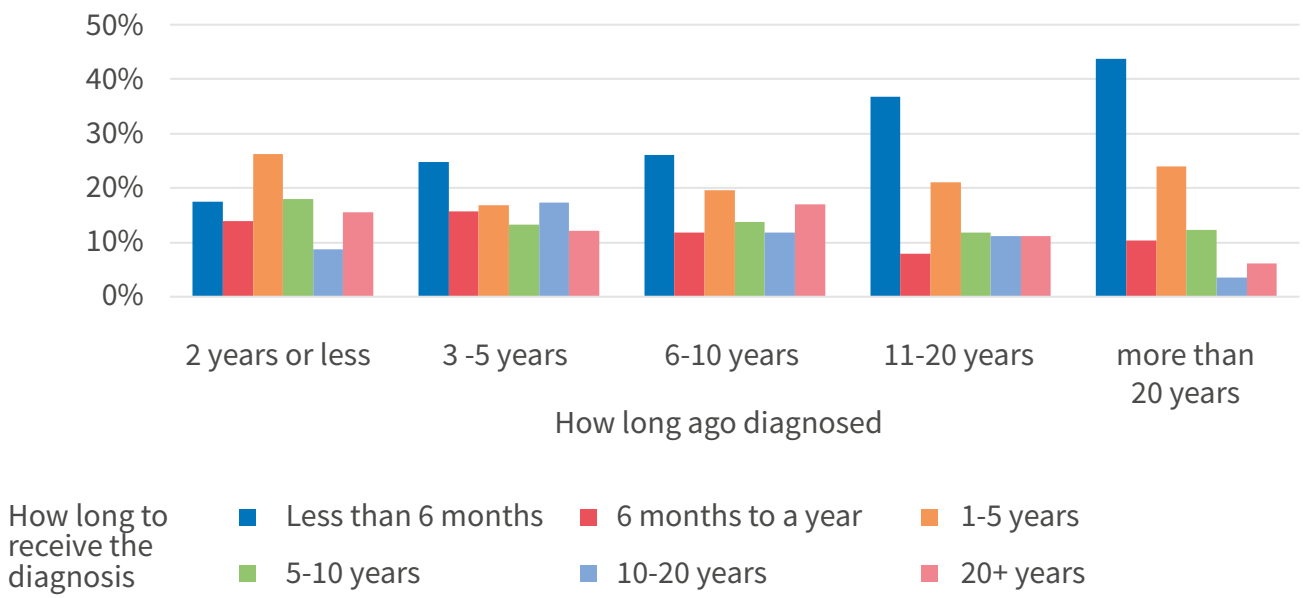
'Ill from childhood, not diagnosed until over 40 years old, still being neglected.'
(Respondent 391)

How long did you/they have to wait for a definitive diagnosis after first consulting a doctor about the onset of symptoms?



Nearly two-fifths (38%)of respondents received a diagnosis within a year, a further fifth (20%) between 1 year and 5 years, and a third (35%) waited more than 5 years.

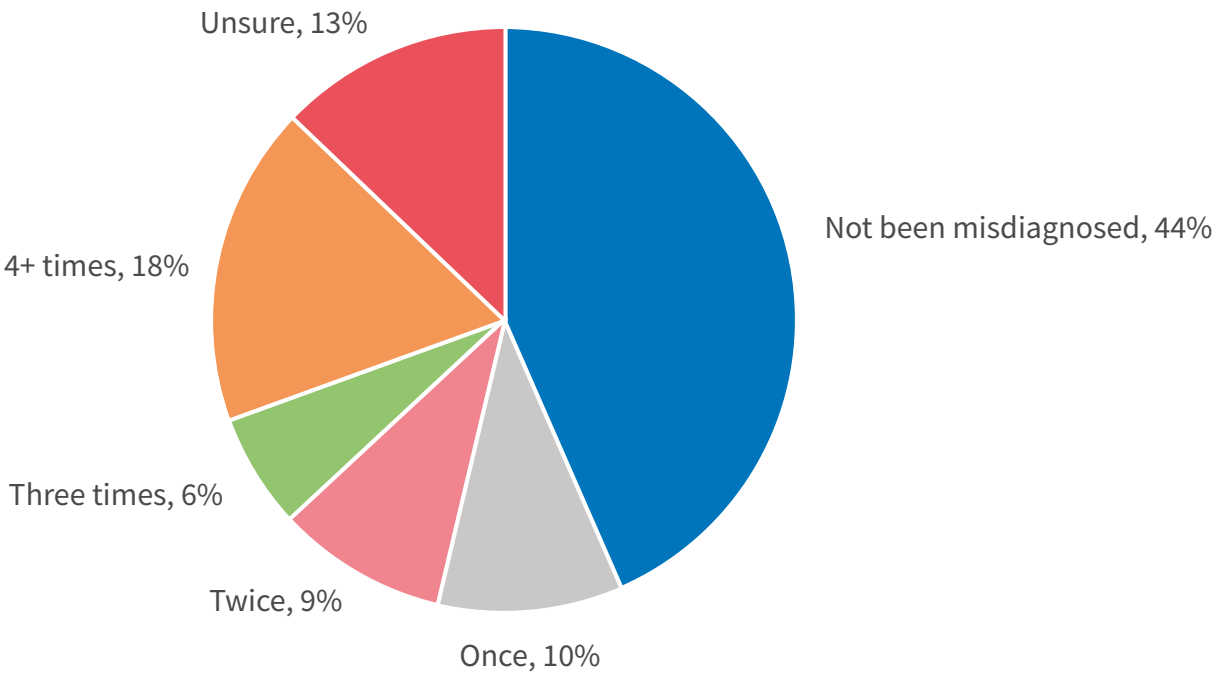
How long ago diagnosed x how long it took to receive the diagnosis



This graph paints a complex picture – within the sample of people who took part in the survey it appears that the length of time to get diagnosed has increased over time. For respondents who were diagnosed 20 years ago, 44% received their diagnosis in less than 6 months after first consulting a doctor about the onset of symptoms. This compares with those who were diagnosed within the last 2 years, where only 18% received their diagnosis in less than 6 months.

This is a concerning signal, but there are many factors at play here. It could be possible that conditions that were previously diagnosed within 6 months, are now taking longer due to slower progress of patients through the system. Genomic medicine is not certain to increase the rate of diagnosis within 6 months, as the majority of people accessing this diagnostic tool will pass through a non-urgent pathway, which may take longer than 6 months from start to finish. We should not forget confounding factors such as the life-limiting nature of rare conditions, which unfortunately means that there will be fewer people around who are thirty or forty years into their journey with a rare condition.

Number of times misdiagnosed - those with definitive diagnosis



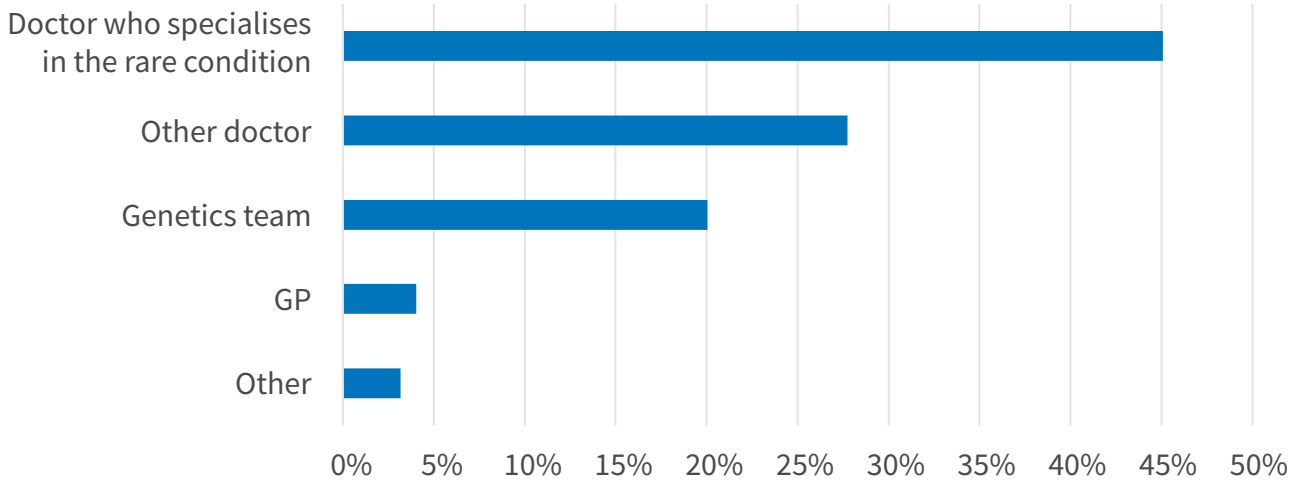
Misdiagnoses appear to be a normal part of the diagnostic odyssey for people with rare conditions. Around a third (33%) of people who had a definitive diagnosis had 2 or more misdiagnoses. These may have contributed to longer times to get a final diagnosis, and indeed indicate that at least a portion of the final diagnoses reported may be misdiagnoses.

There appears to be an issue with patients with rare conditions being misdiagnosed with mental health conditions, as some respondents indicated that their misdiagnosis was sometimes due to physical symptoms being treated as psychological symptoms. This kind of misdiagnosis is the most worrying to find, as this results from a failure of healthcare professionals to properly engage with ill people seeking help. Some respondents mentioned they have been labelled as 'drug-seekers' or 'nuisances'.

'A lot of my symptoms were dismissively blamed on anxiety rather than investigated properly. I have anxiety because of symptoms, not the other way around.'
(Respondent 399)

It is difficult to be critical of the overall pattern of misdiagnoses though, as these come from clinicians working to find a diagnosis for a condition, which is of course a shared aim with our respondents. Perhaps a more interesting measure for future analysis would be the time between misdiagnosis and their corrections.

Which healthcare professional diagnosed the condition

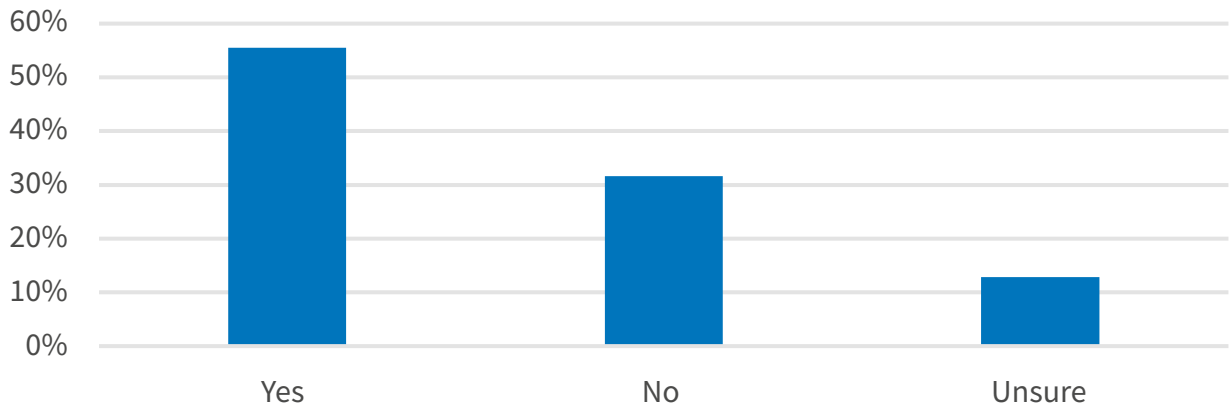


Nearly half (45%) of diagnoses for our respondents were made by doctors who specialise in specific rare conditions. This demonstrates the importance of the referral pathway. For these diagnoses to be made the primary and secondary healthcare providers need to continue the referral chain to the specialist clinicians who are able to make these diagnoses.

Respondents meet many healthcare professionals during their journey to find a diagnosis. This can be a very positive experience, however for some respondents the relationship with some of the healthcare professionals they meet can be quite challenging. The challenges faced include not being listened to, being dismissed, not being treated holistically, a perceived lack of knowledge from the healthcare professionals with some admitting to the person seeking help that they don't know what to do.

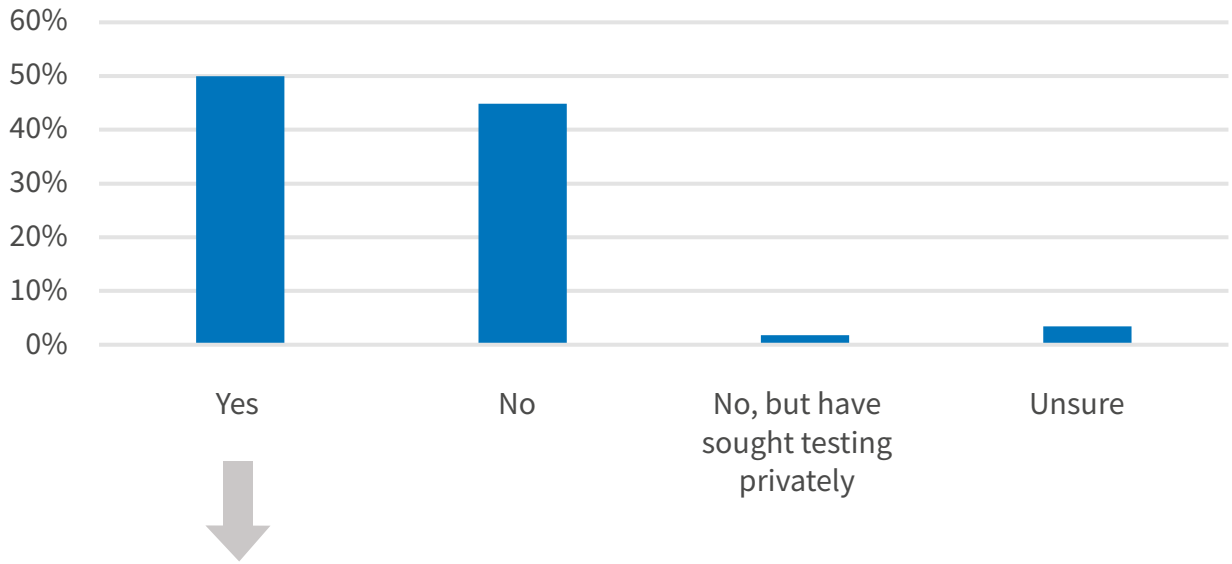
'In over 20 years, and after living in 3 different towns, I have only met 3 doctors who did not dismiss me or even laugh in my face.' *(Respondent 217)*

For those without a definitive diagnosis, have you/they been misdiagnosed in the past?

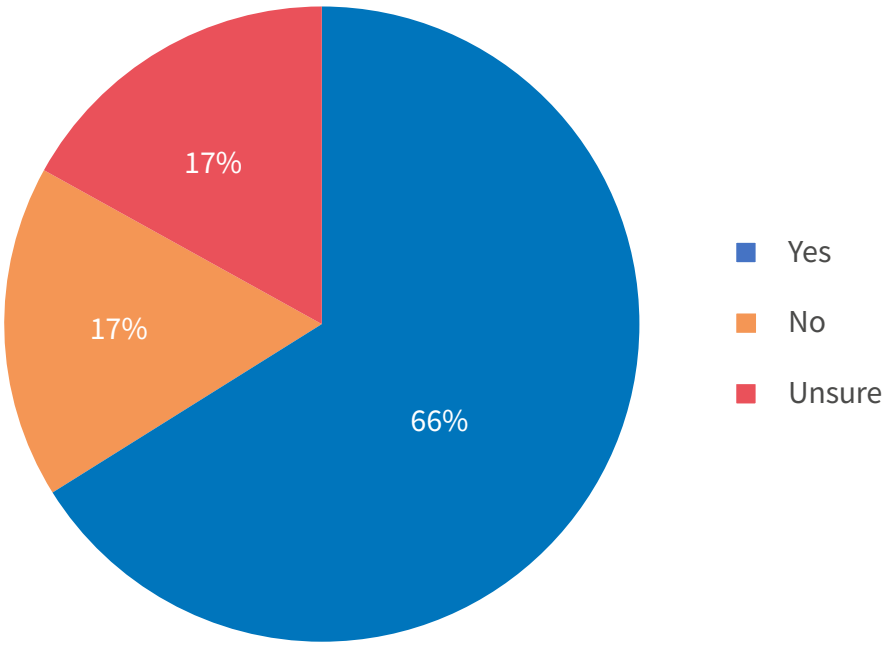


Misdiagnosis appears as part of the diagnostic odyssey for those yet to reach the end of it, with over half (56%) of respondents who did not have a definitive diagnosis stated that they had been misdiagnosed in the past.

For those undiagnosed, have you/they been offered genetic or genomic testing?

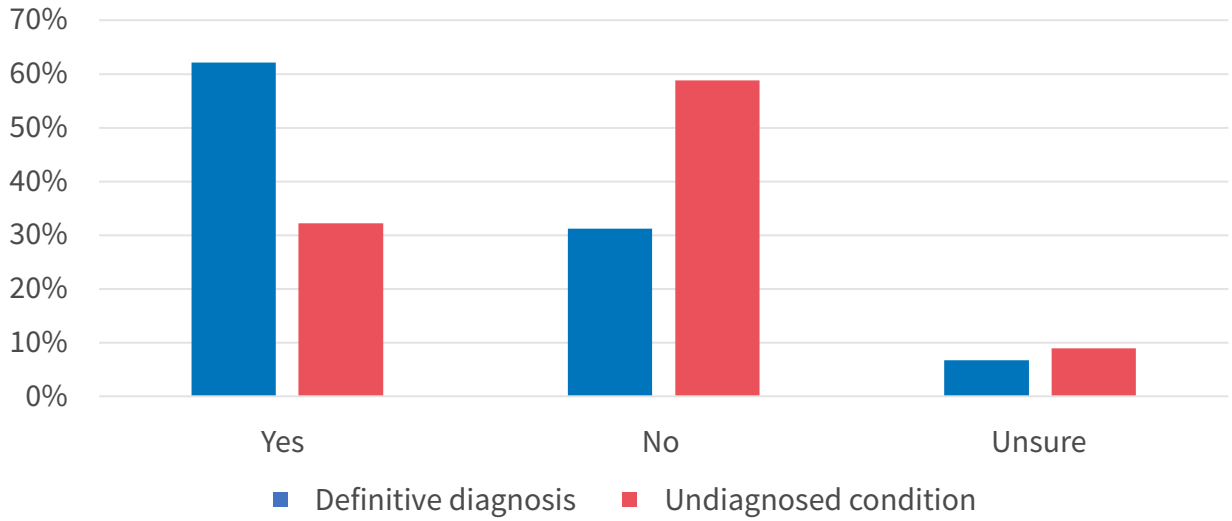


Offered whole genome or exome sequencing



There appears to remain a significant portion (45%) of those without a diagnosis who have not had the benefit of genetic testing to identify the condition that affects them.

Whether have a HCP they can ask questions of by whether diagnosed or not

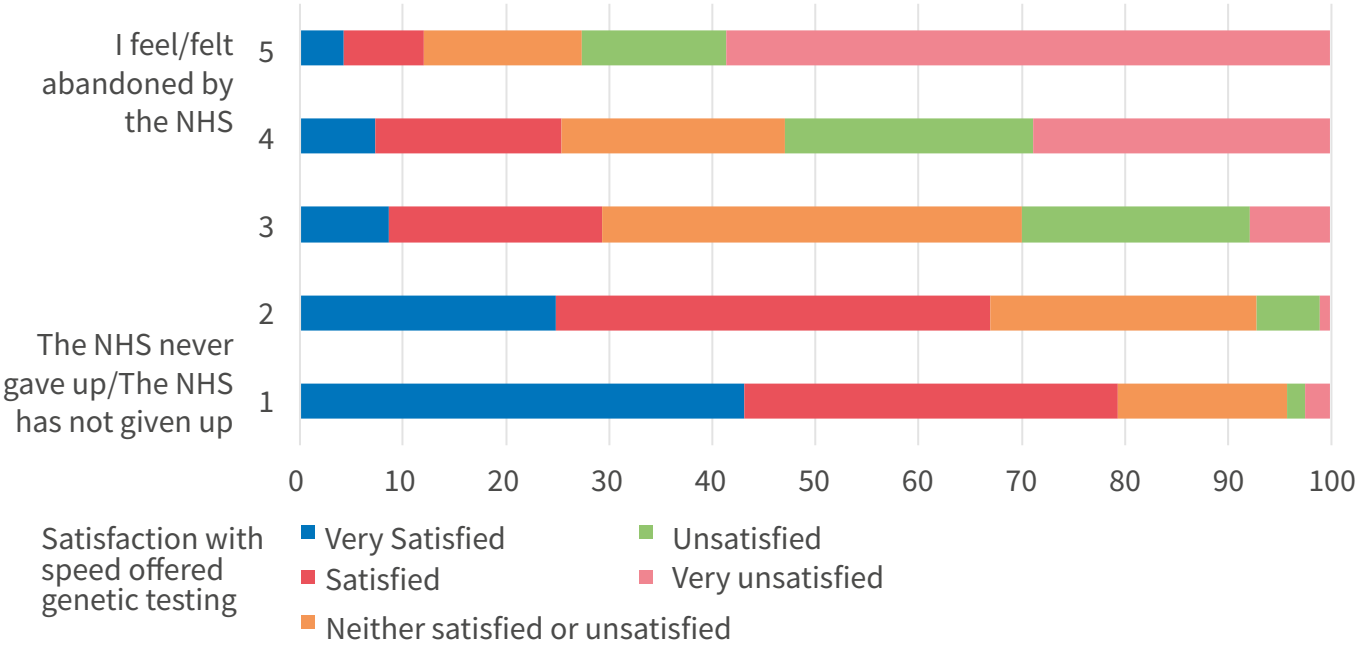


It is clear that it is not possible to access specialised care for a specific condition without a diagnosis; less than a third (32%) of those without a diagnosis had a specific healthcare professional they can go to with questions, compared with nearly two thirds (62%) of those with a diagnosis. It is important though that people are treated throughout their diagnostic odyssey, especially as we see how long it can be. The respondent's comments here are some of the starkest, indicating the extent to which people can feel abandoned when they are stuck in a diagnostic odyssey.

'I feel like no-one cares or considers it important to find a diagnosis.'
(Respondent 1,391)

'I have spent years from a small child trying to get a diagnosis.
The impact of a late diagnosis has been irreversible … I feel lonely and abandoned.'
(Respondent 65)

Satisfaction and response from the NHS



There appears to be a strong relationship between the speed at which people are offered a genetic test and their perception of the NHS and its attitude towards helping them to a diagnosis. Those who were more satisfied with the speed with which they were offered testing had a more favourable attitude towards the NHS in their search for a diagnosis. People will have many experiences along their journey to diagnosis and genetic testing would have only accounted for part of that, however, this relationship may reflect a perception within the community of the power of genetic testing, and its status as one of the most powerful tools in reaching a diagnosis.

'The search was led by me but once we found the right doctor they worked very hard to find an answer. Things changed as genetic testing improved.'
(Respondent 153)

Other experiences which may have impacted upon how someone rated their experience of searching for a diagnosis include the relationship they have with the healthcare professionals they meet. Respondents said they felt that they had not been heard or believed and dismissed by doctors which subsequently led to them losing trust and confidence in the NHS generally and some doctors specifically.

'It feels as though you're shouting "help" but no words are coming out…. We've been let down by not just one hospital but all of them and the over 20 doctors we've seen.' *(Respondent 57)*

'I finally fought for a diagnosis at 24 but the damage of having been labelled a lazy liar since childhood despite my severe chronic pain, still leaves psychological scars. I doubt myself to this day due to negative and dismissive comments from these doctors and other adults. They let me down.' *(Respondent 1,253)*

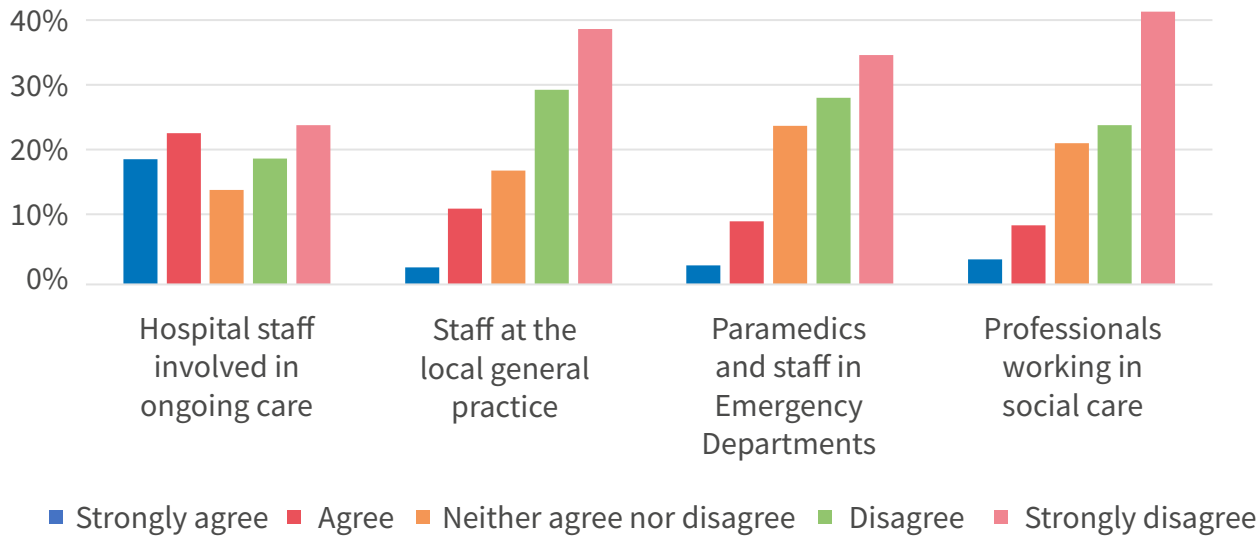
AWARENESS OF GENETIC AND RARE CONDITIONS AMONG HEALTHCARE PROFESSIONALS

People affected by rare conditions meet many healthcare professionals along their journey to find a diagnosis and beyond while they live with their rare condition. For some this can be a positive experience, for others this can be particularly challenging. Respondents describe not being listened to whilst others felt they were being dismissed. Some healthcare professionals admitted to the person seeking help that they don't know what to do.

'Doctors should not assume patients are lying just because their knowledge or ability to diagnose is limited. The lack of belief or rudeness and dismissal by doctors who did not have knowledge to diagnose was the worst part of the journey.'
(Respondent 1,137)

'When I entered the room where I was diagnosed at age 40, having shown symptoms since I was 3 days old, the consultant said, "Whenever I receive a referral letter that rude I know I am about to meet a very resilient person.'
(Respondent 227)

Healthcare professionals' knowledge – Extent agree that professionals have sufficient information about their condition

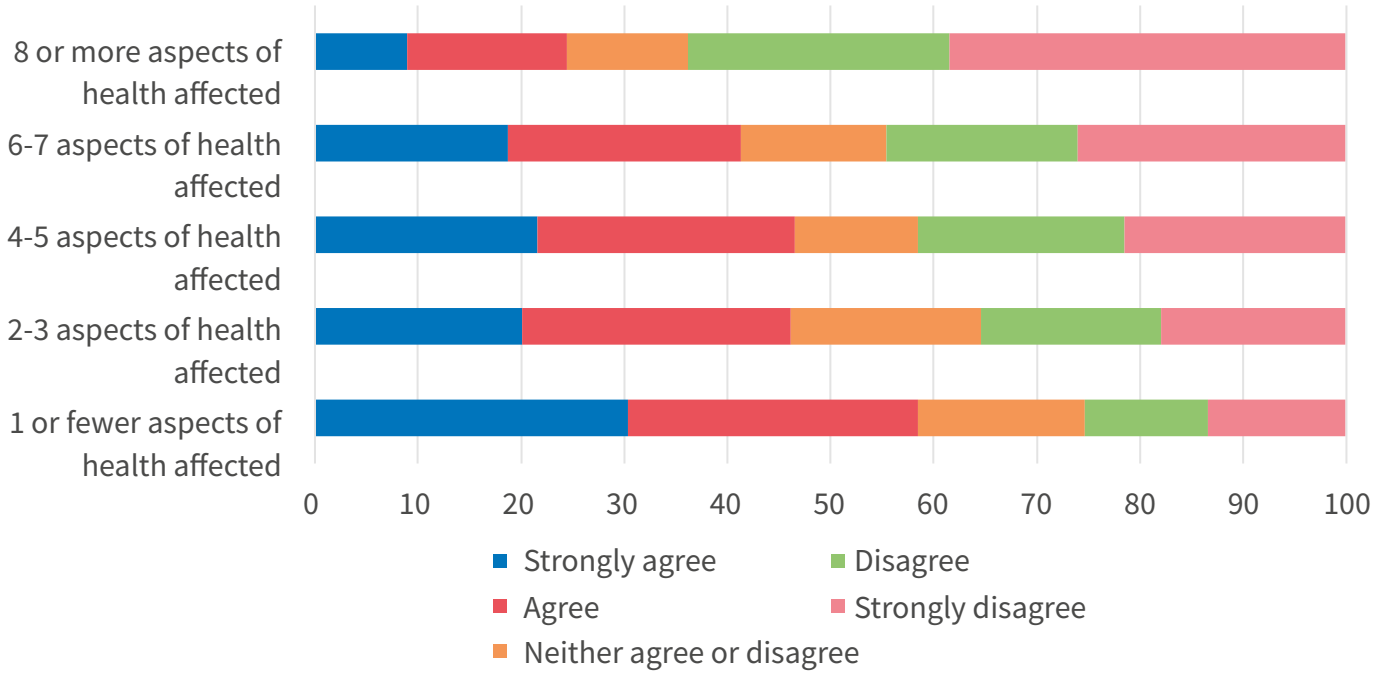


On the whole, respondents felt that healthcare professionals did not have sufficient information about their rare condition. In particular, people reported a lack of knowledge of healthcare professionals outside of the hospital system. For example, 42% of respondents agreed or strongly agreed that hospital staff involved in ongoing care had sufficient information about the condition. Whereas only 12% agreed or strongly agreed that paramedics and staff in emergency departments had sufficient information about their condition.

'You can always tell when someone has googled his condition (HSP) … my son's particular form of the disease is really rare and has different challenges.' *(Respondent 38)*

'I have been refused the medication I need by ER staff due to them misunderstanding my conditions and symptoms.'*(Respondent 1,218)*

Extent agree that professionals have sufficient information about their condition - Hospital staff involved in ongoing care by complexity of condition



The extent to which respondents agreed that they think hospital staff involved in their ongoing care have sufficient information about their condition varied by how complex their conditions were. The more complex the condition the lower proportion of respondents agreed that hospital staff involved in ongoing care had sufficient information (59% of people with only one aspect of health affected compared to less than a quarter (24%) of people with 8 or more aspects of health affected). Respondents said they sometimes had a variety of experiences across hospitals or within hospitals across different departments.

'There's a lot of difference between departments in the same hospital. In one instance a rheumatologist told me she didn't believe in my condition, she referred me to a physiotherapist in the same hospital who is amazingly knowledgeable.'
(Respondent 469)

People affected by rare conditions indicated that they are concerned when they are dealing with healthcare professionals who are not aware of their condition as they feel their health is potentially at risk if an inappropriate treatment or medication is administered. This could be mitigated if the NHS implemented the use of alert cards.

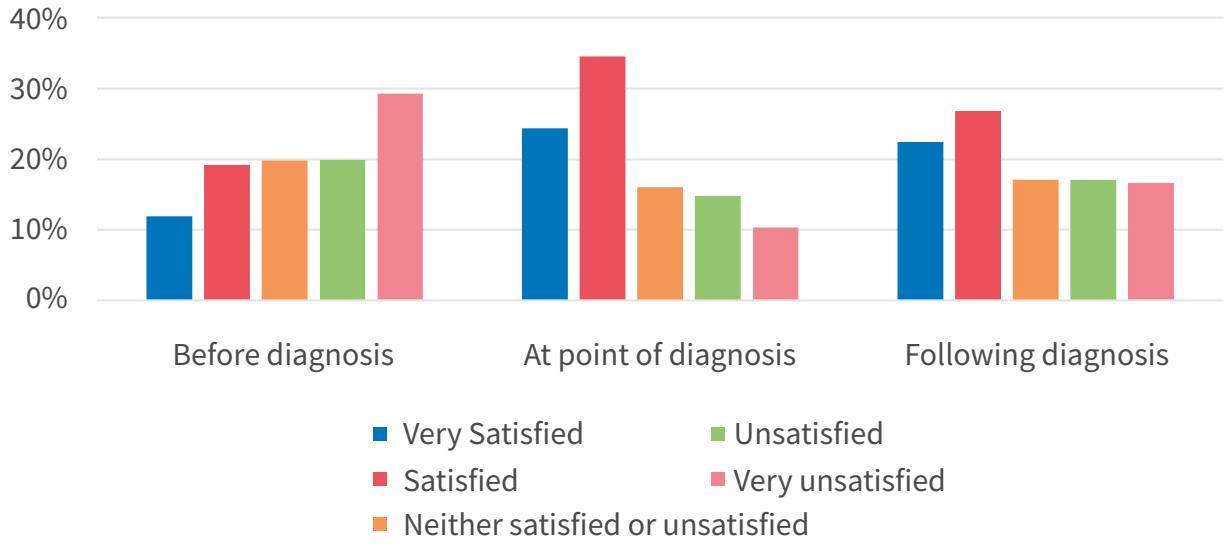
An alert card usually lets others know that the bearer of the card has an important medical condition that might require special care or attention. The purpose of the card is to alert medical staff to important health information if the bearer is unable to communicate, whether that be because they are unconscious, injured, or simply want to backup what they are saying with a card. Medical alerts have the potential to improve care and treatment and save lives in emergency situations. Genetic Alliance UK has called on the NHS to implement alert cards across the UK[5].

'I've had a paramedic google my current diagnosed condition.'
(Respondent 52)

'Most doctors haven't even heard of my condition, or like to assume I am lying when I say I have them.' *(Respondent 112)*

'Rare and unknown conditions not being taken seriously by medical staff without prior knowledge of them is dangerous and I rarely feel safe with a care provider who is not aware of my condition, or who is making medical or medication decisions that are not appropriate for someone who has said condition.'
(Respondent 304)

Information provided by healthcare professionals



People show the lowest level of satisfaction with the information they received from healthcare professionals before a diagnosis, with less than a third (31%) saying they were satisfied or very satisfied with the information provided. In contrast, people were most satisfied with the information they received from healthcare professionals at the time of diagnosis (59% stating they were satisfied or very satisfied). This number drops following the diagnosis, with only 49% of respondents stating that they were satisfied or very satisfied with the information received from healthcare professionals.

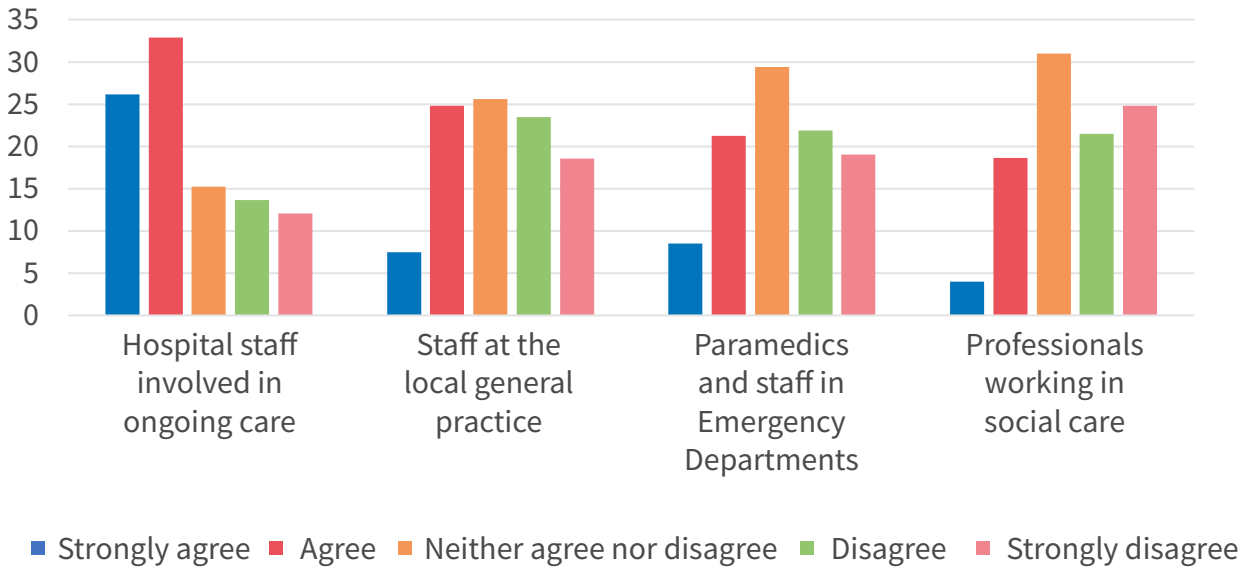
Only 11% of respondents chose their GP as their main source of information and support, with some indicating in their comments the knowledge of their GPs was limited.

'My GP doesn't know my condition or how serious it is, so I mostly rely on my specialist and specialist nurses.' *(Respondent 178)*

'I have to make all the decisions about my care as my GP knows nothing about my rare diseases and I'm not under the care of any consultants.'
(Respondent 34)

For some, overcoming the diagnostic odyssey and receiving a definitive diagnosis is just one of many hurdles in their journey to understanding their condition and the implications that this will have for the future.

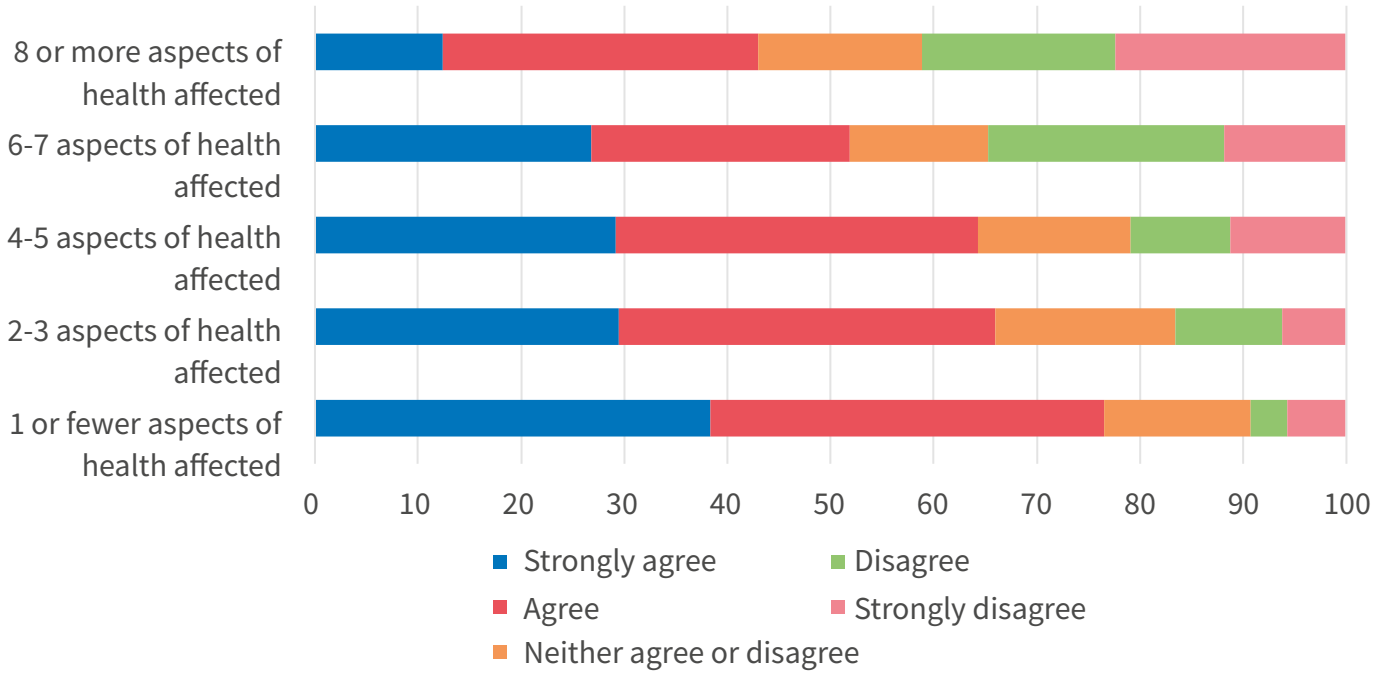
Trust in healthcare professionals



When talking about hospital staff involved in ongoing care 59% of respondents agreed or strongly agreed that they had confidence and trust in these professionals; this was the highest ranking professional group from the options available to respondents.

For staff at the local general practice just under a third of respondents (32%) agreed or strongly agreed that they had confidence and trust. There was a similar proportion (30%) when asked about paramedics and staff in emergency departments. The professional group with the lowest proportion of respondents agreeing or strongly agreeing that they had confidence and trust was for professionals working in social care – less than a quarter (23%).

Extent that agree that have confidence and trust in the hospital staff involved in ongoing care by complexity of the condition



There seemed to be a negative relationship between trust in hospital staff and how complex someone's condition was, with confidence and trust decreasing as complexity increased . For example, over three quarters (77%) of people with only one aspect of health affected agreed or strongly agreed with the statement that they had confidence and trust in the hospital staff involved in ongoing care. In comparison, only 43% of people with more complex conditions (8 or more aspects of health affected) agreed that they had confidence and trust in the hospital staff involved in their ongoing care.

'There was no multidisciplinary working. We were left to attend lots of different clinics/specialists with little joining up of information. Nobody had an overview of her symptoms.' *(Respondent 10)*

'I wasn't treated as an individual, but as a nuisance. This has affected my mental health seriously.' *(Respondent 236)*

'All the signs were there, but nobody was searching.' *(Respondent 1,184)*

Learning from people affected by rare conditions

The vast number and complexities of rare diseases means that it is not possible for a single healthcare professional to know about every rare condition; the rare disease community is accepting of this. However, people affected by rare conditions felt it was important for healthcare professionals to gain knowledge from the patients and to take opportunities to learn.

'Generalist practitioners cannot know about all conditions but they do need to know where they can access information when presented with a patient with a rare condition. They also need to be able to be taught by the patient.'
(Respondent 173)

'Stop teaching doctors they are the experts in rare conditions, they aren't. They study them for a matter of hours during qualification, and GP's not at all thereafter. Patients who suffer for decades are the experts.' *(Respondent 510)*

'Although at first glance my rare diseases don't overlap, the treatment does. I check PubMed for papers... a PubMed portal should be in all hospital departments.' *(Respondent 138)*

'It's impossible to access the specialists I want to see, it simply is not offered to be seen by them. Instead you end up in front of an ignorant specialist who has no or little knowledge of my condition....I know more about EDS, which bruises their ego and they are rude, unhelpful and dismissive.'
(Respondent 391)

In contrast, when healthcare professionals asked people about their rare condition and tried to learn and further their knowledge, respondents reported positive interactions.

'This condition is rarely known by most professionals, however the team that have been dealing with me have really tried to understand and get their heads round the complete picture of my diagnosis to enable them to treat me adequately. I am very happy with them.' *(Respondent 95)*

'Once during a hospital admission a nurse asked me all about one of my conditions as she had never heard of it and wanted to learn. I have taken opportunities to educate doctors who have wanted to learn more too.' *(Respondent 363)*

'The specialists I see are always looking to improve their knowledge, taking part in research and trying to gain a better understanding of the syndrome.' *(Respondent 2)*

'Some GPs need to listen more and not make assumptions. The best doctor I had was a locum with an open mind.' *(Respondent 1,219)*

COORDINATION OF CARE

Coordination of care involves working together across multiple components and processes of care to enable everyone involved in a patient's care (including a team of healthcare professionals, the patient and/or carer and their family) to avoid duplication and achieve shared outcomes, throughout a person's whole life, across all parts of the health and care system, including:

- Care from different healthcare services (e.g. different medical disciplines – medical, mental health, behavioural, health promotion)
- Care from different healthcare settings (including primary and secondary; community settings e.g. social care) and locations (e.g. rural/urban)
- Care across multiple conditions, or single conditions that affect multiple parts of the body
- The movement from one service, or setting, to another

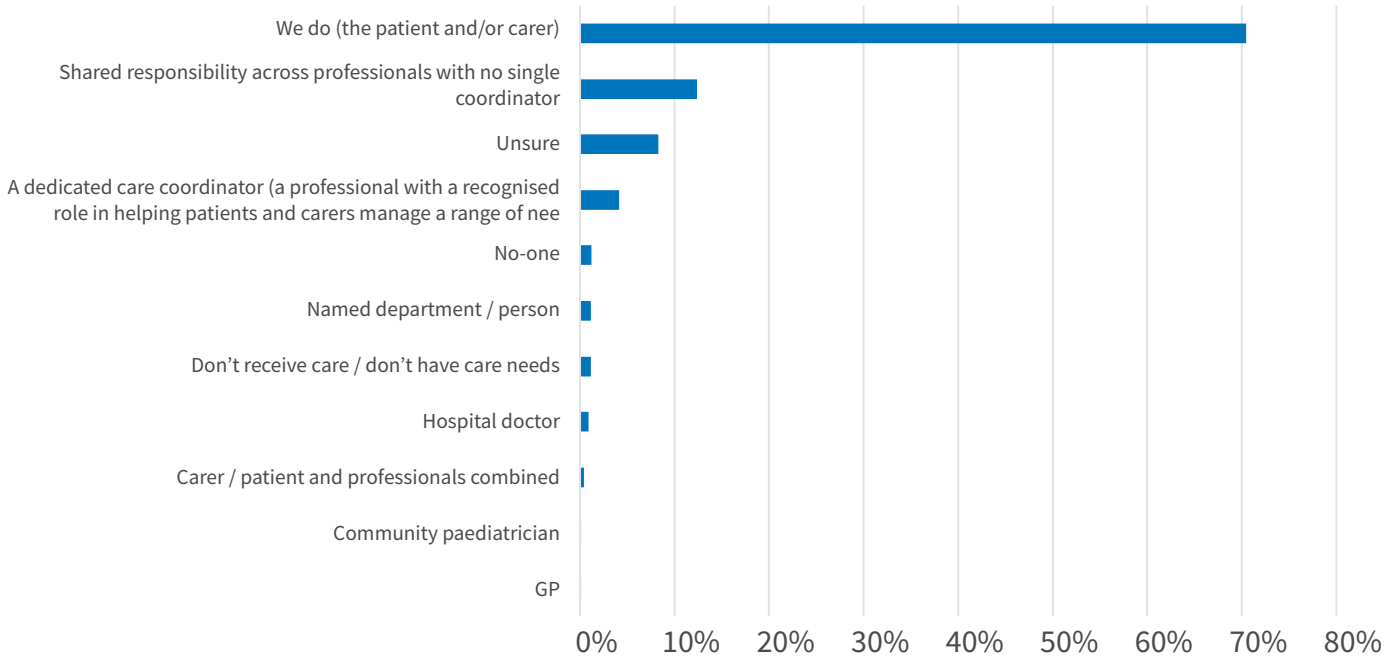
Coordination of care should be family-centred, holistic (including a patient's medical, psychosocial, educational and vocational needs), evidence-based, with equal access to coordinated care irrespective of diagnosis, patient circumstances and geographical location.[7]

This approach can bring the benefits of:

- ensuring all clinicians caring for someone with a rare condition consider each others' proposed interventions, ensuring there is synergy in the patients' overall care and treatments do not interact negatively or adversely affect other body systems
- reducing the burden of a high frequency of clinics by scheduling them efficiently and with account of how results of tests and scans can feed into all aspects of care
- ensuring primary and secondary care providers understand all aspects of care and can play their role appropriately, involving specialist teams appropriately.

The COordiNated Care Of Rare Diseases (CONCORD) study[8] which began in 2018 will deliver a comprehensive taxonomy of care coordination.

The current picture of care coordination - Who coordinates (or organises) the majority of their care



The patient / carer is the one who coordinates the majority of care for 71% of the respondents. The next most common coordinator for our respondents was 'shared responsibility across professionals with no single coordinator' – 12%. Taken together, at least 83% of respondents did not have a single care coordinator provided by the health service.

Only 4% stated that they had 'A dedicated care coordinator' which was defined in the survey as 'a professional with a recognised role in helping patients and carers manage a range of needs between different professionals or across care settings. They may be a full-time coordinator or may coordinate care as part of their main role, such as a GP'.

Respondents stated that they often did not have a choice over who coordinated their care and sometimes the carer / person with the condition had to take over the role even if it had been assigned to someone else.

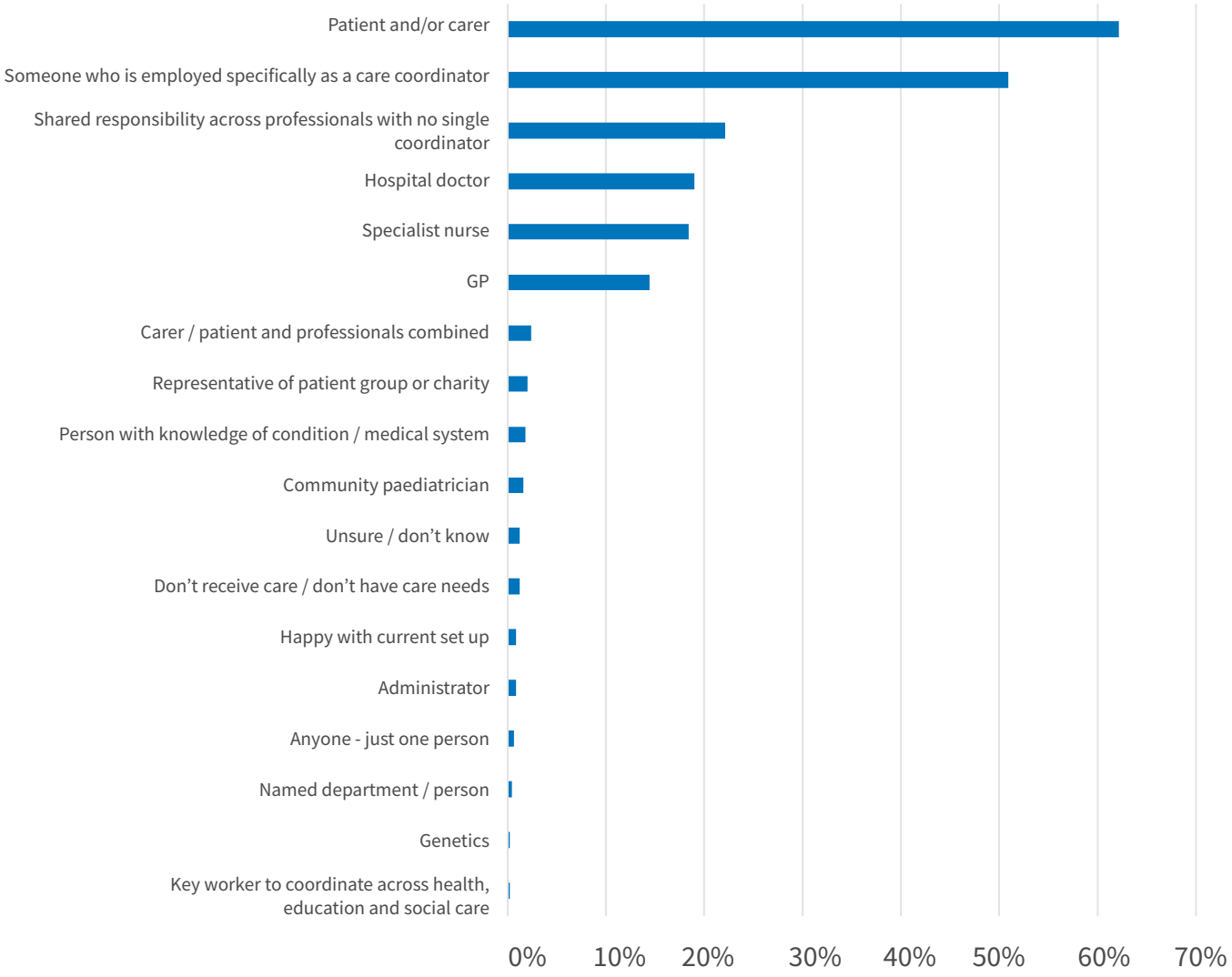
'Spent 10 years being called a liar and/or a hypochondriac. So now I feel like I have to self-manage my symptoms and don't trust that I will be taken seriously if I see a doctor.' *(Respondent 48)*

'I was fobbed off by my GP and hospital specialists. I'm still reluctant to visit GP with ongoing symptoms.' *(Respondent 1,177)*

'At annual review, I see a number of health care professionals ie doctor, physiotherapist and have all relevant tests eg ECG, breathing tests within the one appointment which is very well organised.' *(Respondent 818)*

'Having to have multiple clinical appointments with different disciplines on different days and taking time off for each visit.' *(Respondent 490)*

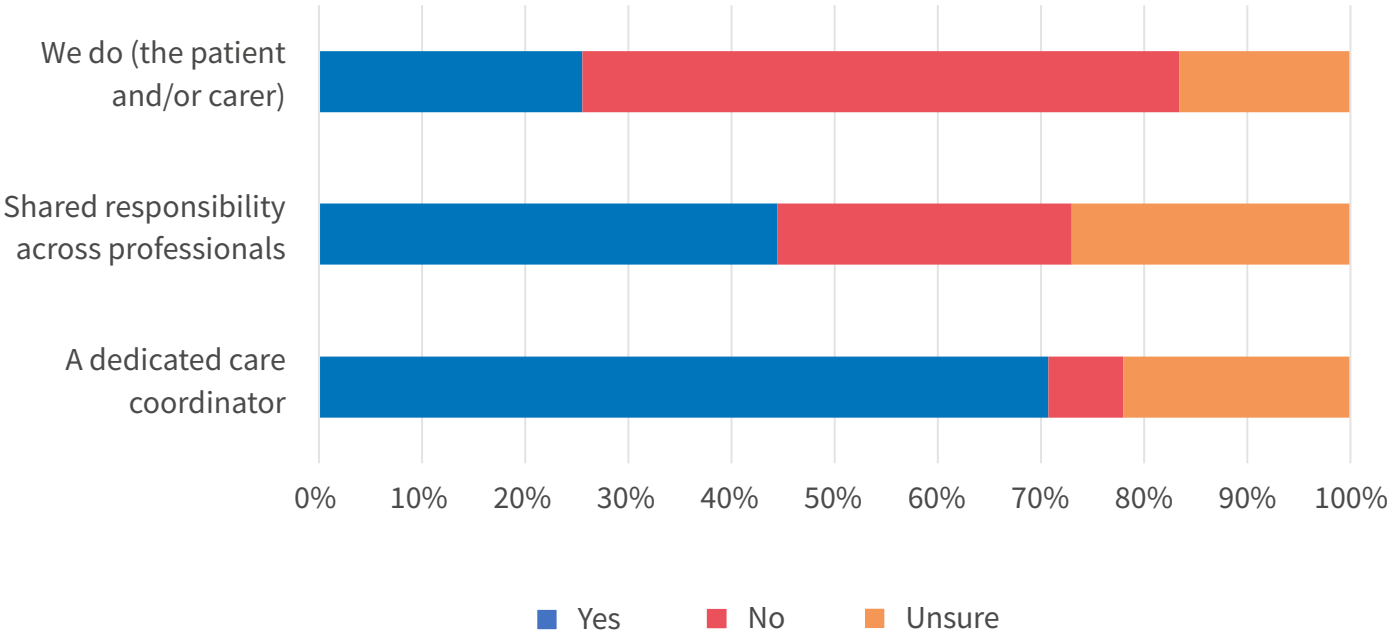
Ideally who would they like to coordinate their care



When asked who would they prefer to coordinate their care, nearly a third (31%) of respondents wanted to coordinate their own care, around a quarter (26%) wanted someone specifically employed as care coordinator. Though, of those that already coordinated their care, a greater proportion – 41% ideally wanted to coordinate their own care and a quarter (25%) wanted someone specifically employed as a care coordinator. On the other hand, of those whose care is coordinated across professionals, just over a third (35%) ideally wanted someone specifically employed as a care coordinator, and just 7% had a preference for 'patient and / or carer' to manage care coordination.

Respondents mentioned the advantages of having their care coordinated by someone else. For some there was a sense that they were on 'opposing sides' to the medical team and they would have preferred a joint approach. Respondents also said that care coordination could be quite burdensome, especially for those having to cope with an illness also. Another reason for wanting a care coordinator (other than themselves) was 'to ensure nothing was missed'. Professional care coordination does not appear to be a preference for everyone, though some of the differences in view based on their current situation indicate that of those that are currently experiencing professional care coordination, the value is clearer.

Whether care is effectively coordinated x who coordinates care



When the perceived effectiveness of coordination of care is considered against who is doing the coordination, the difference is clear. For those who had a dedicated care coordinator 71% felt that their care was effectively coordinated, only 7% thought it was not. When the person themselves or their carer was the one who coordinated care around a quarter (26%) felt that this care was effectively coordinated, over half (58%) thought it was not.

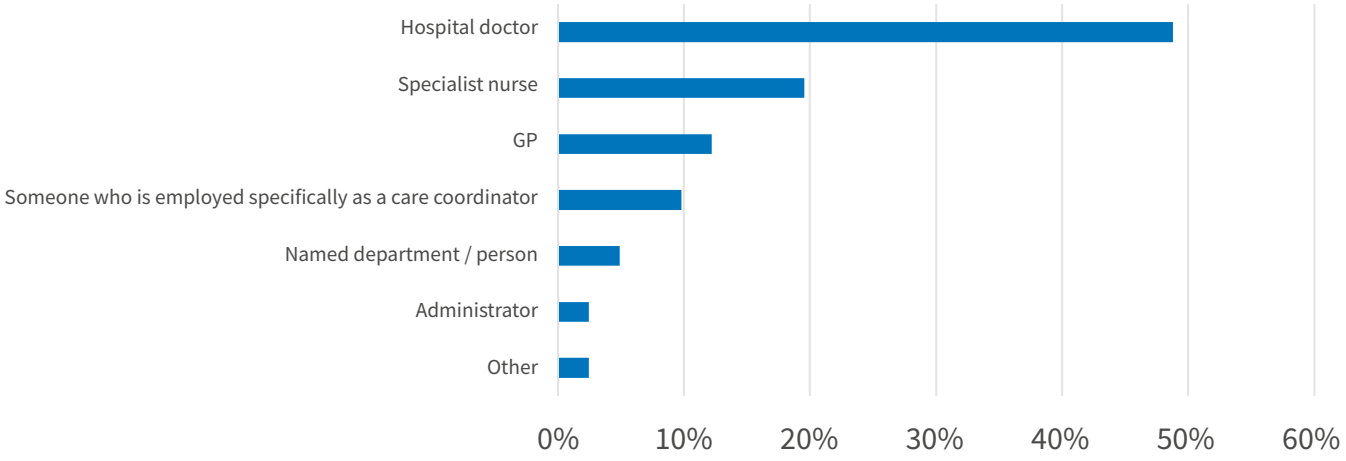
The preference for those that already deliver their own care coordination to continue doing so may be the consequence of previous negative experiences or a lack of confidence in the health service to deliver such an important job. It is certainly important that in future, when care coordinators are introduced, the benefits of professional care coordination are clearly communicated to allow informed choice in its uptake.

'There is no coordination of my daughters care by anyone other than myself and her and frankly we are floundering around in the dark.' *(Respondent 1,251)*

'My specialist doctor keeps my GP informed and works with the NeuroMuscular Centre. It's working well so far.' *(Respondent 901)*

'Many members of my support team consult with each other which makes a huge difference.' *(Respondent 262)*

Who is the care coordinator if have a dedicated care coordinator



Of the small number (42) of respondents who had a dedicated care coordinator, 20 (49%) said their coordinator was a hospital doctor, 8 (20%) had a specialist nurse, 5 (12%) a GP, 4 (10%) someone who was employed specifically as a care coordinator.

'My understanding is that under the last UK rare diseases strategy we were all due to get care co-ordinators and this certainly hasn't happened for me. Personally, given patient expertise, I would prefer to be paid to coordinate my care rather than have to train up a paid coordinator but either way this is an area for improvement in the next iteration of the UK rare diseases strategy.' *(Respondent 11)*

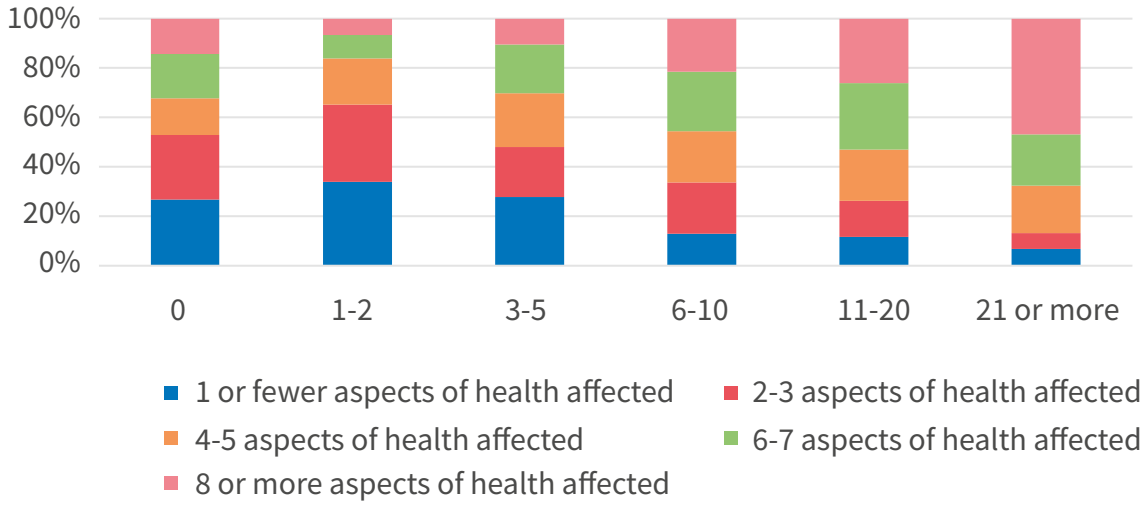
'Although in a residential care home that is supposed to coordinate all needs, this is having to be done remotely by me as a parent.' *(Respondent 184)*

'My specialist nurse tells me exactly where I need to go for appointments and when, what I need and what I need to do. I'm blessed and grateful for such good care.' *(Respondent 178)*

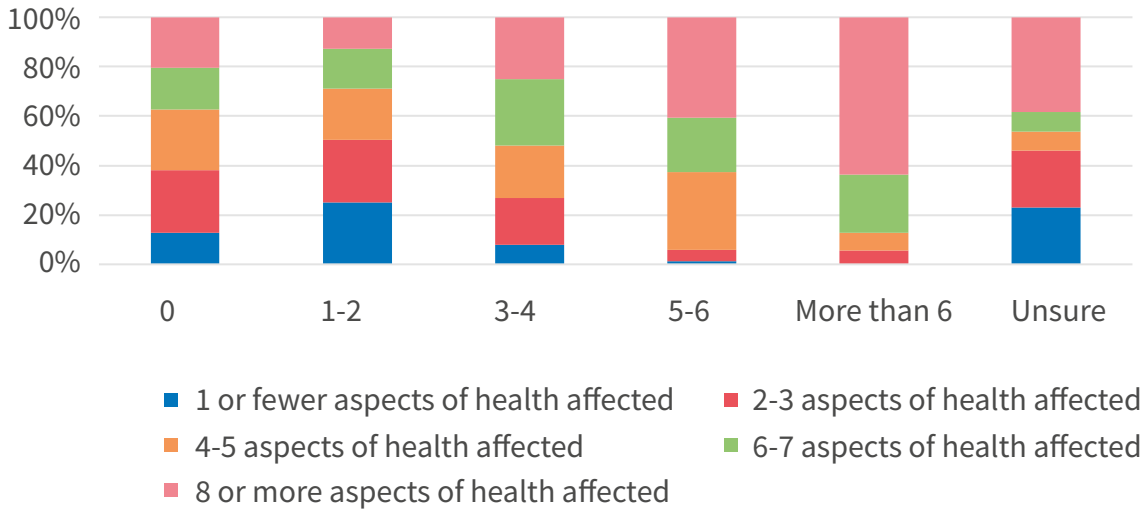
'All my care is organised by me and is private – there is nothing available on the NHS for my condition.' *(Respondent 353)*

Where might care coordination be most necessary?

Frequency of health service use in relation to their rare / undiagnosed condition x complexity of the condition



Number of different clinics attend in relation to their rare / undiagnosed condition x complexity of the condition

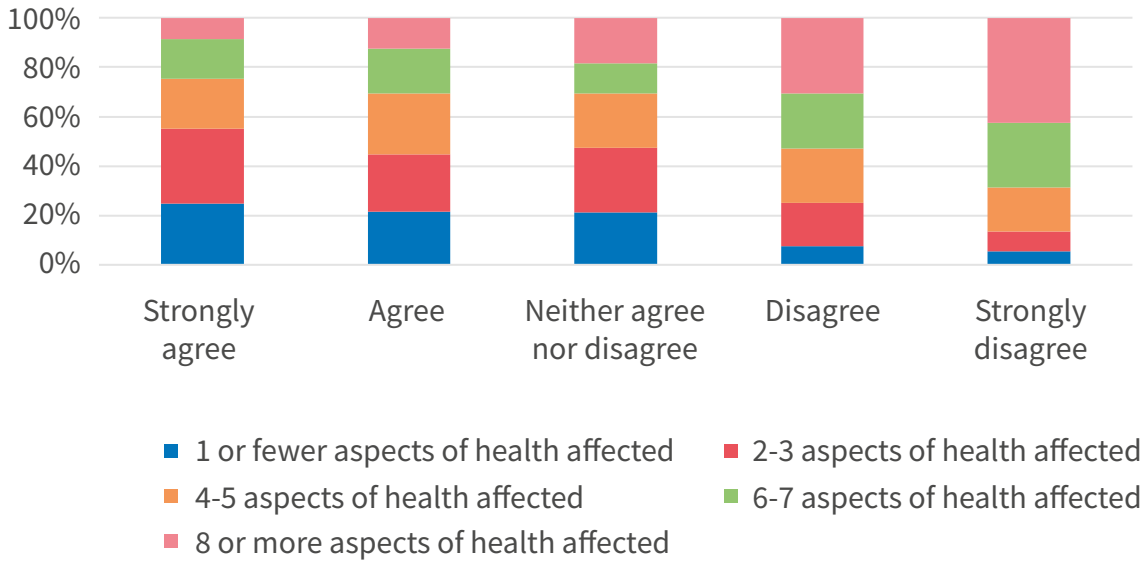


There seemed to be associations both between the number of aspects of health affected (complexity of the condition), the frequency of appointments for any service, and the number of different clinical services people attend. The more complex the condition the higher the number of times the person has contact with the health service over 12 months and the more complex the condition the higher the number of different clinics attended.

Respondents mentioned that appointments were sometimes cancelled or the yearly appointments were less frequent – this implies that for some respondents the number of contacts per year would be higher if they could access the health system as much as they wanted to. For those who said they do not attend any clinics there seems to be little relationship with the complexity of their condition; 38% had three or fewer aspects of health affected and 37% had 6 or more aspects of health affected. It is possible that for those people able to attend specialist centres they are able to have many appointments in one day and therefore potentially fewer attendances overall with the healthcare services.

‘Different departments do not seem to coordinate, so don’t tie up symptoms from one specialist area to another (eg linking cardiology to gastroenterology even though my symptoms have the same root cause).’
(Respondent 230)

Extent agree that professionals caring for them work as team by condition complexity



There seemed to be some association between how strongly respondents agreed with the statement that the professionals caring for them worked as a team and the complexity of their condition with fewer aspects of health affected being more likely to agree or agree strongly with the team working statement.

We could not discern how efficiently the health service was treating these respondents with high frequency care. It may be that this frequency is appropriate for some, but for others this is likely to be an inefficient approach to scheduling appointments and coordinating clinics. Inefficiency like this will have a big negative impact on the lives of individuals, threatening work and education opportunities. Given the relationship found between condition complexity and both use of the health service and the quantity of different clinics attended, it would appear that more complex conditions should be considered a high priority for delivery of care coordination.

‘A personal care coordinator who is on my side (it sometimes feels like I’m on opposing sides with the medical team) to sort things out instead of me battling away just to get basics, would be great.’
(Respondent 138)

‘It’s chaotic & relies on me organising and chasing, which is exhausting & dispiriting, particularly as I also suffer from fatigue as part of my condition. There is nothing I would love more than a dedicated person to coordinate my care for me.’
(Respondent 974)

‘A specialist nurse would improve patient care and relieve the workload as well as coordinating care with GPs and other areas.’
(Respondent 658)

‘I would really appreciate someone with knowledge of my son’s condition overseeing his care to ensure nothing is missed.’
(Respondent 736)

ACCESS TO CARE AND TREATMENT

Specialist centres provide expert advice on diagnosis, assessment and treatment of a particular condition. Centres are made up of a team of different specialists, sometimes also including scientists and researchers. Specialist centres support patients across the UK, not just in their local area.

Respondents who were able to access specialist centres spoke very positively about them. They valued being able to see multiple healthcare professionals in one trip. Having access to doctors who are experts in their rare condition also meant that they then do not have to repeatedly explain their condition.

‘The centre has physio, social as well as medical care available at the same time.’
(Respondent 969)

‘Staff are experts, take time, listen, coordinate tests and results during visits. Treat patients as equals.’ *(Respondent 124)*

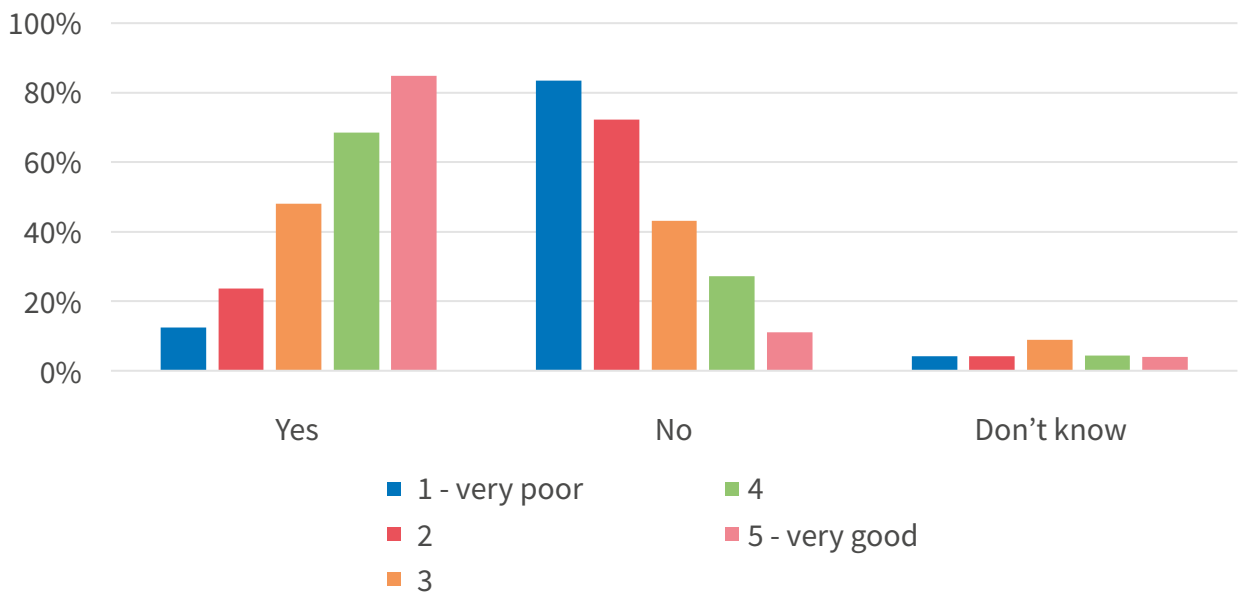
‘I don’t have to explain everything over and over again.’
(Respondent 674)

Respondents described being listened to and the centres being responsive to their needs. Another positive aspect of the specialist centres was being treated holistically.

‘They listen and provide any help I need. Without going into too much detail, they have arranged counselling for me at the moment.’ *(Respondent 446)*

‘They are responsive whenever I raise a concern and (after learning I’m not a hypochondriac!) listen to me whenever I contact them with a new symptom or issue.’ *(Respondent 54)*

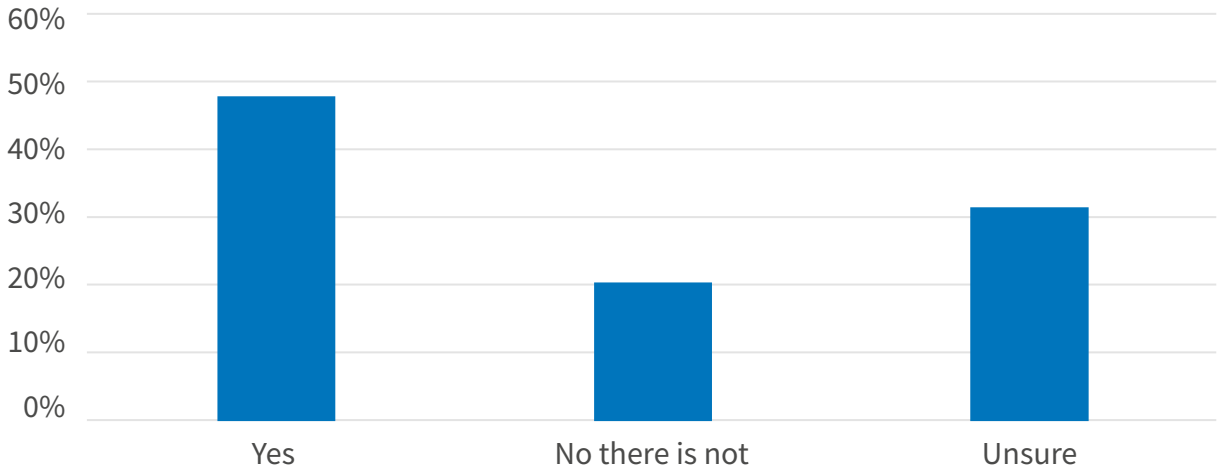
Access the specialist centre by how they rate their overall care for their rare / undiagnosed condition



There is also a strong association between accessing a specialist centre and how someone rates their overall care. For those who rated their care as '5-very good', more than 4 in 5 (85%) accessed a specialist centre, 11% with the highest rating scale did not access a specialist centre. For those who rated their care as '1-very poor', only 12% accessed a specialist centre while 84% of those with the poorest care rating did not access a specialist centre.

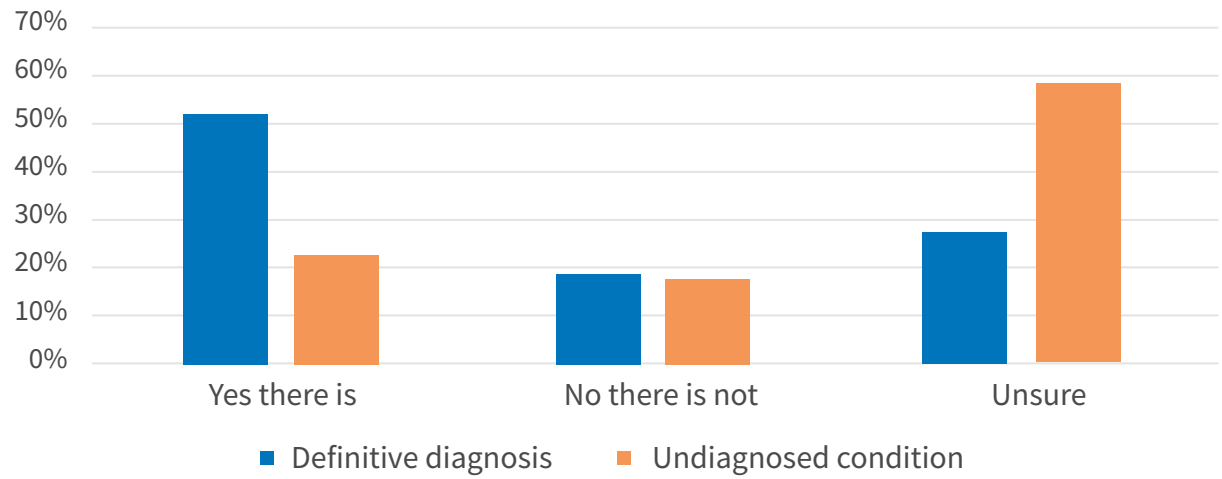
As discussed earlier, the chance of reaching a diagnosis improves when people have access to specialists with better knowledge of rare conditions. Only 4% of people with a definitive diagnosis were diagnosed by a GP compared to 45% of people who were diagnosed by a doctor who specialised in the rare condition.

Awareness of specialist care



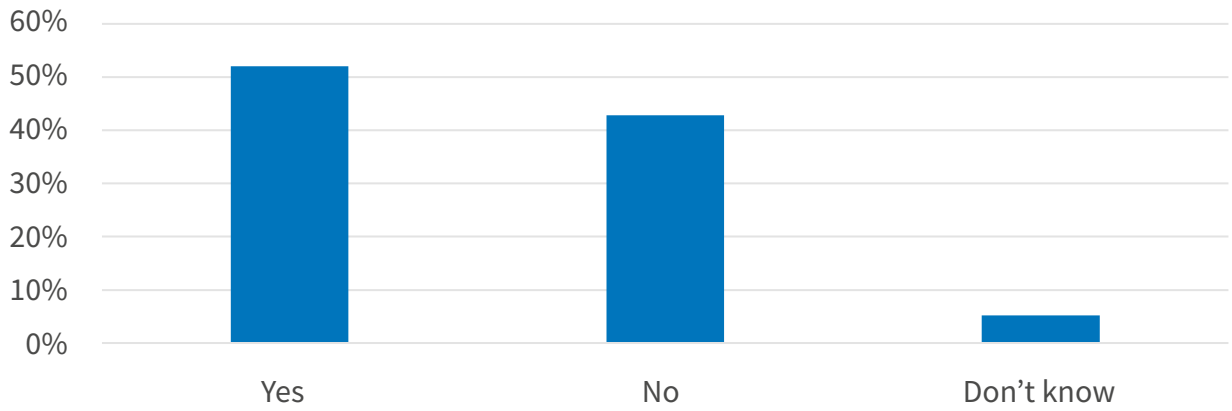
Nearly half (48%) of respondents said there was a specialist centre for their condition, around 1 in 5 (21%) said there was not and nearly a third (32%) were unsure.

Is there a specialist centre by whether have a diagnosis or not



Whether there is a specialist centre or not is influenced by whether the condition has been diagnosed or not, with a far lower proportion of those without a diagnosis being aware of a specialist centre. For those without a diagnosis less than a quarter (22%) said there was a specialist centre, around 1 in 5 (19%) said there was not and over a half (59%) were unsure.

Do you/they access the specialist centre for the condition?



Of the respondents who said there was a specialist centre for their condition, just over half (52%) said they accessed the centre, 43% said they did not and 5% were unsure. Some respondents said they had difficulty accessing specialist centres describing long waiting lists to try to get an appointment because of the limited numbers of centres available.

'Accessing specialist care and treatment is, basically a nightmare – it feels like I have to put up and shut up with the very "unspecialist care" I manage to get. They make me feel guilty for hoping and needing a bit more than this.'
(Respondent 1,152)

'There are so few specialists and specialist places for my condition, so waiting lists are incredibly long and this has a huge impact on your physical and mental health. It also means you have to travel long distances to be seen and can only be given the bare minimum of tests/treatment/support because there aren't enough services for all the people that need them.'
(Respondent 932)

Respondents also highlighted difficulties getting referrals to be able to access the specialist centre whether that was from primary care or secondary care.

'Apparently our local GPs can't/won't refer to the specialist centre.'
(Respondent 438)

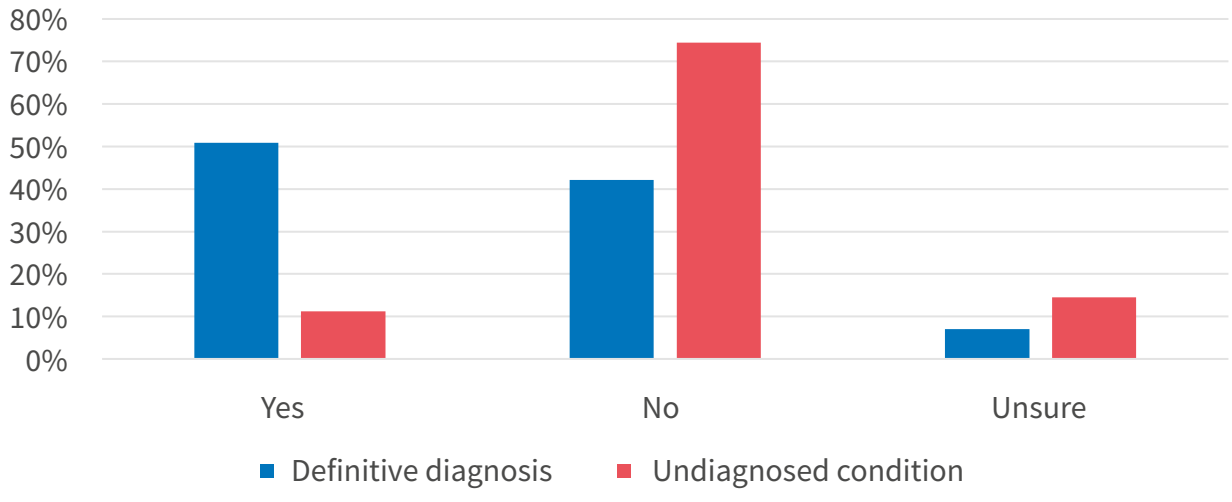
'The NHS centre requires a referral from a Rheumatologist and I have NEVER been referred to a NHS rheumatologist despite requesting it.'
(Respondent 302)

Respondents said there were not enough specialists and too little funding. Some people said they had difficulty accessing specialists and had to pay privately to see expert doctors.

'Specialist care and knowledge has only been open to us having gone private. Immunology is severely underfunded in the UK, not enough expertise is available.' *(Respondent 288)*

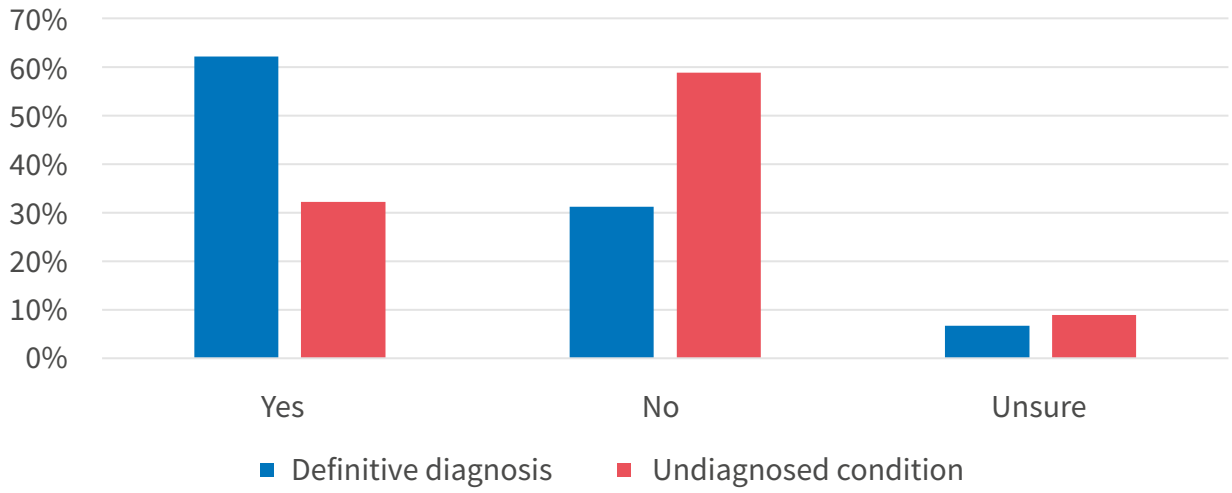
'There is clearly insufficient specialist care for rare conditions. We have really struggled to find anyone expert in EDS or indeed any information other than via a support group.' *(Respondent 209)*

Have an expert doctor by whether have been diagnosed or not



Without a diagnosis it can be difficult to access specialist care and treatment. Just over half (51%) of respondents with a definitive diagnosis had a doctor who was an expert in their condition. Whereas only 11% of those undiagnosed had an expert doctor.

Whether have a HCP they can ask questions of by whether diagnosed or not



Furthermore, there is a clear distinction between those with and without a diagnosis in terms of whether they have a specific healthcare professional they can go to with questions about their rare or undiagnosed condition. Nearly two thirds (62%) of people with a definitive diagnosis have a specific healthcare professional they can go to with questions compared with less than a third (32%) of people who are undiagnosed. Of respondents who had someone they could ask questions of, nearly two thirds (65%) said that this was a specialist or consultant.

Specialists are key providers of reliable information, and around a third (34%) of respondents said a specialist was their main source of support and information. However, people voiced concerns that despite having access to specialist care, they do not have the opportunity to see them very often.

'The physiotherapists at my genetics clinic are always helpful and ready with suggestions to help me try to live a better life, but I get to speak to them once a year.' *(Respondent 875)*

'Many people have not heard of ataxia. My GP surgery gives me no support the only consultation is once a year with neurology.' *(Respondent 687)*

Travelling to access specialist care

Respondents often said they were happy to travel long distances to access specialist care and some respondents received specialist care in a different nation to the one they live in. Unfortunately, some people described difficulties in accessing specialist care in a different nation.

'The majority of all other appointments are fairly local. We obviously don't mind travelling that distance once a year to get extremely specialist support.' *(Respondent 104)*

'I would HAPPILY travel to Centres of Excellence in England, ie Bath, Birmingham or London. I want expertise!' *(Respondent 654)*

'It's outside my area and therefore costs more. I'm not going to die of my condition, so my GP considers specialist help unnecessary for me.' *(Respondent 185)*

'I travel to London for specialist care, since luckily I can and as I know it to be better than that available locally. When I get sicker I don't know what will happen.' *(Respondent 169)*

Where someone lived could also impact on whether they were able to access a specialist centre with some respondents describing how they were unable to go outside their local area.

'There are specialised centres in [the] UK but not in the East Midlands.' *(Respondent 883)*

'If you are living with the disease for the rest of your life I think you should be given an invitation to attend the specialist centre at least once even if it is not local.' *(Respondent 351)*

For some even if the care was within the same country, the distance of travel was prohibitive. Some respondents reported difficulties getting to specialist centres and others were unable to attend.

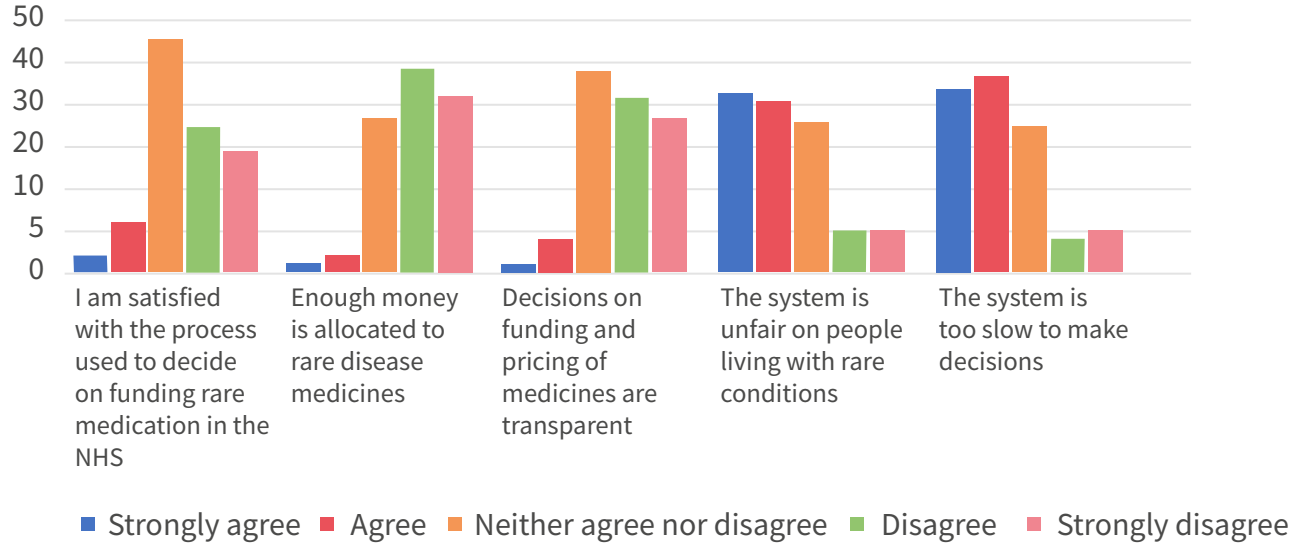
'The orthotics department is located in a hospital that is half a mile from the nearest bus line, so I have to walk half a mile to get the orthotics that I need to walk. Why is there no bus service to the hospital that provides disability support?' *(Respondent 429)*

'[Don't attend] Because it is a 7-8hr drive away from me.' *(Respondent 207)*

Access to medicines for rare conditions

The International Rare Disease Research Consortium (IRDiRC) counts 393 new indications for rare disease medicines between 2010 and 2019. This demonstrates massive progress towards the development of medicines for rare conditions, but it remains true that for the vast majority of rare conditions there is no medicine. Nearly half (48%) of respondents were not aware of any medications for their condition, 12% were unsure.

Extent agree with the below statements:



Respondents were quite concerned about the funding environment for medicines for rare conditions in the UK. The attitudes displayed by respondents matched those expressed in Genetic Alliance UK's Action for Access report launched in 2019. Nearly three quarters (71%) of respondents disagreed with the statement 'Enough money is allocated to rare disease medicines', 64% of respondents agreed with the statement 'the system is unfair on people living with rare conditions', and 66% agreed that 'the system is too slow to make decisions'.

'There are therapies available, but I do not qualify for them at the moment. I do not know how decisions about access to therapies are made, but of course the NHS is underfunded in all areas.' *(Respondent 816)*

'It can take a long time to start new therapy/treatments for rare diseases. It may be that a treatment is recommended but then you have to wait for funding to be approved.' *(Respondent 550)*

'The allocation of funding for conditions within the NHS is hugely unfair. Some conditions are emotive & strongly funded via the NHS… This is not at all the case of rarer diseases which fade into the background but can have devastating lifelong consequences for those who suffer with the condition and their loved ones.' *(Respondent 939)*

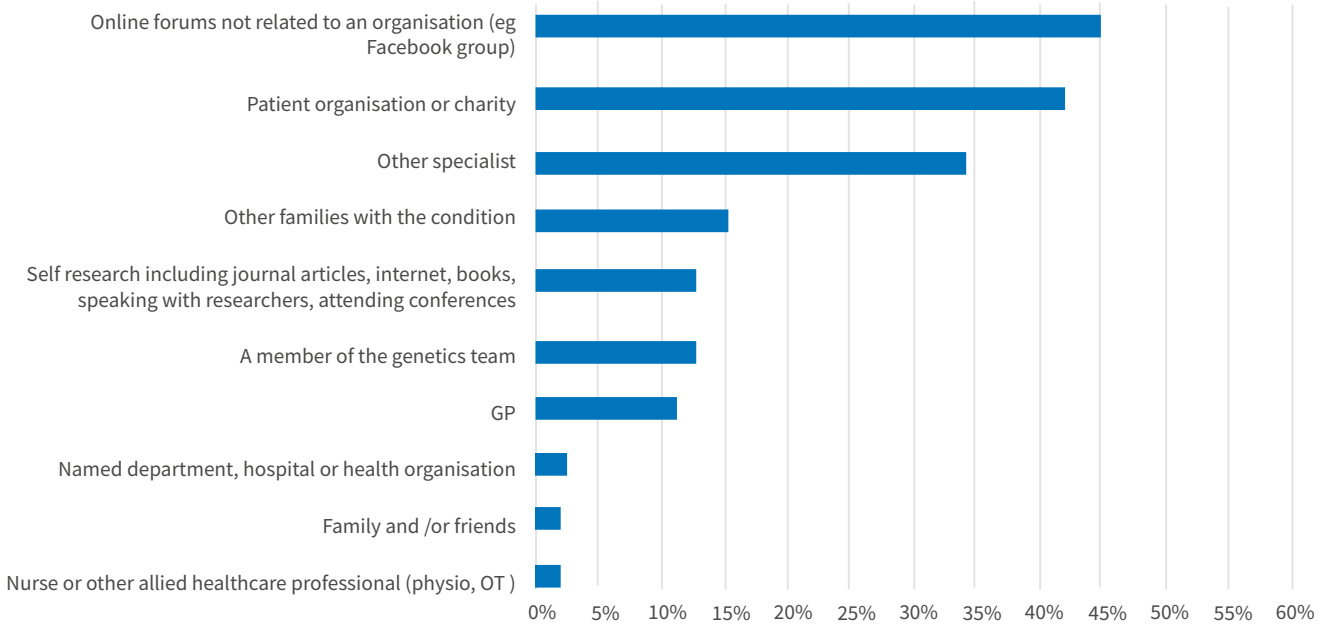
'It feels as if our condition isn't severe enough to get as much funding as major conditions.' *(Respondent 1,083)*

'The treatment for my condition has been declined by NICE. I understand why but with a rare disease the trial groups are small so do not always give statistically significant results. This can swing things so that it appears that the treatment is not effective.' *(Respondent 173)*

'It should be about the patients' needs not about the cost of the medication.' *(Respondent 63)*

PATIENT VOICE

Who have been the main sources of support and information?



It is clear that patient organisations and the rare condition community are key sources of information and support about rare conditions. Online forums (a source for 45% of participants) and patient organisations (41%) were the most frequently used sources of information. Around a third (34%) of respondents said a specialist was their main source of support and information. A significant portion of respondents (13%) looked for information themselves.

'We were lucky that our community paediatric consultant was very open to listening to what we discovered via a global Facebook support group and referred us to more specialists as a precaution.' *(Respondent 727)*

RESEARCH

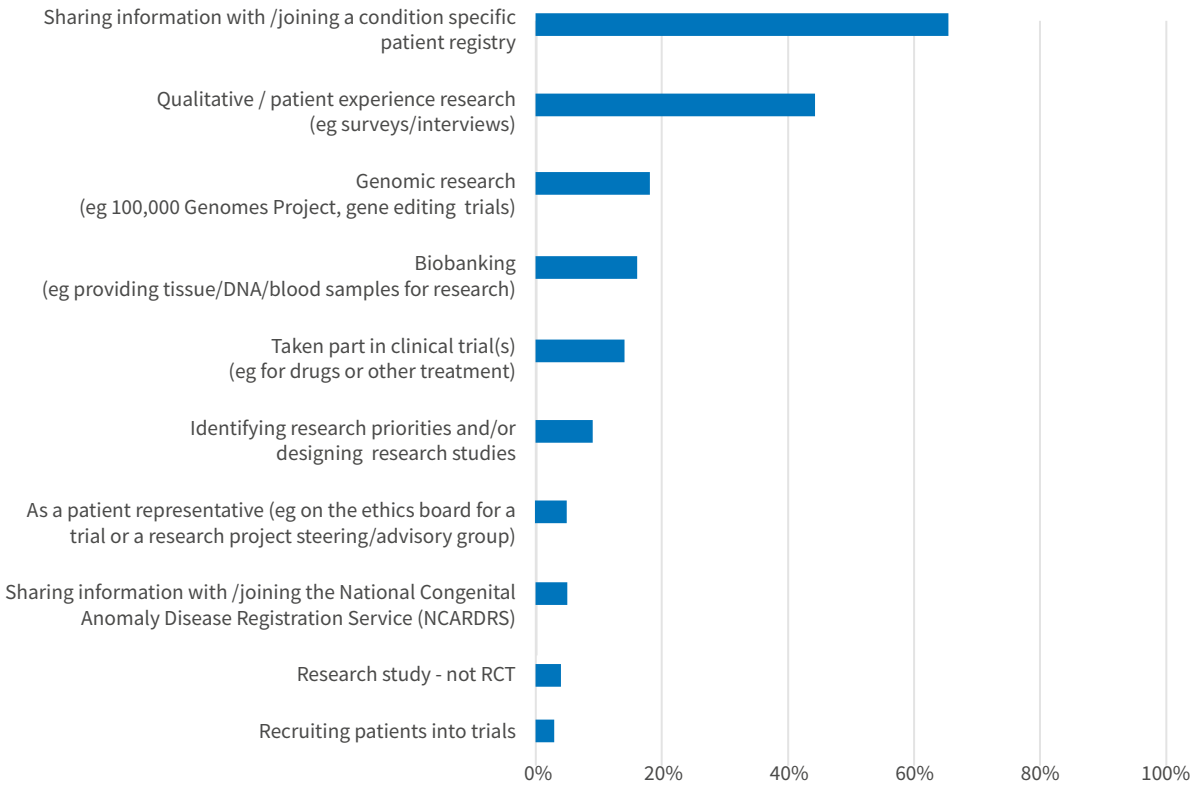
People affected by rare conditions place a high value on research and the ability to take part in research. Research can help identify a condition, provide information about how a condition will progress, and can lead to the development of better care and treatment.

Many respondents reported being involved in research projects to help raise awareness and improve knowledge of rare conditions. Some respondents who want to take part in research have been unable to because of a lack of opportunity or because they have not been made aware of any research projects.

''I would like to be more involved in research to help others with my condition I have volunteered but not been selected so far.' *(Respondent 768)*

'We are so busy coping we are not aware of research opportunities we can participate in.' *(Respondent 33)*

Types of research have participated in (if have participated in research)



'Research is necessary and essential to understand/learn more of the disease.' *(Respondent 893)*

The most common way which respondents identified as having been involved in research was through sharing information with/ joining a condition-specific patient registry' (65% of respondents). A significant number (44%) also reported being involved in research by participating in qualitative and patient experience research (eg surveys and interviews). Other research activities where more than 10% of respondents who had participated in any research had taken part were: genomic research – 18%; biobanking – 16%; clinical trials – 14%. Around a third (32%) of respondents had not participated in any research.

Some respondents described their frustration in the lag between taking part in research and seeing the benefits. Others described how research should be based on the issues that are important to those affected by rare conditions.

'I've been ill for decades and taken part in lots of research. However, I never seem to see anything come of it. I'm still receiving no care, there are still no good treatments and no proper diagnostic tests.' *(Respondent 34)*

''Implement the research that already exists, there's so much information available about a number of rare disorders but that information is not trickling down to GP services, A&E etc.' *(Respondent 288)*

Others described how research should be based on the issues that are important to those affected by rare conditions.

'Research questions based on what's important to patients should be explored.' *(Respondent 638)*

'Some UK research into MdDS would be very welcome. It is embarrassing that there has only been one paper about MdDS published here and that its focus was on stigma and illness intrusion etc.' *(Respondent 11)*

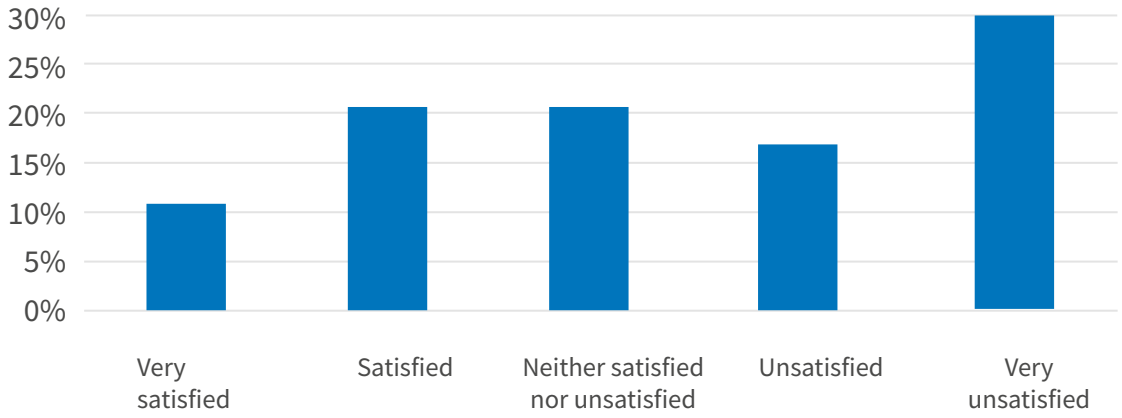
Respondents on the whole wanted to take part in research but they wanted it to be easier to find out about research projects and to be made aware of the results when the research was complete.

'They should share results with patients. I have a science degree, not telling me stuff is another level of torture.' *(Respondent 1,281)*

'Just make sure that these are publicised to patients including Facebook groups in an easily accessible way. Make it easy to find out about what research is going on.' *(Respondent 106)*

TRANSITION

For those who had transitioned between child and adult services, how satisfied were they?



On the whole respondents who had experienced transition between child and adult services were not very satisfied with the experience. Just a third (32%) of these respondents were either satisfied or very satisfied with transitioning between child and adult services, almost a half were unsatisfied or very unsatisfied with the experience. For those who had transitioned between child and adult services there was a sense that they were having to 'start all over' again.

There was also potential confusion when different clinics used different age cut-offs to decide when someone was no longer a child.

'Some clinics treat 16 as [an] adult whereas others it is 18, so very disjointed.' *Respondent 32)*

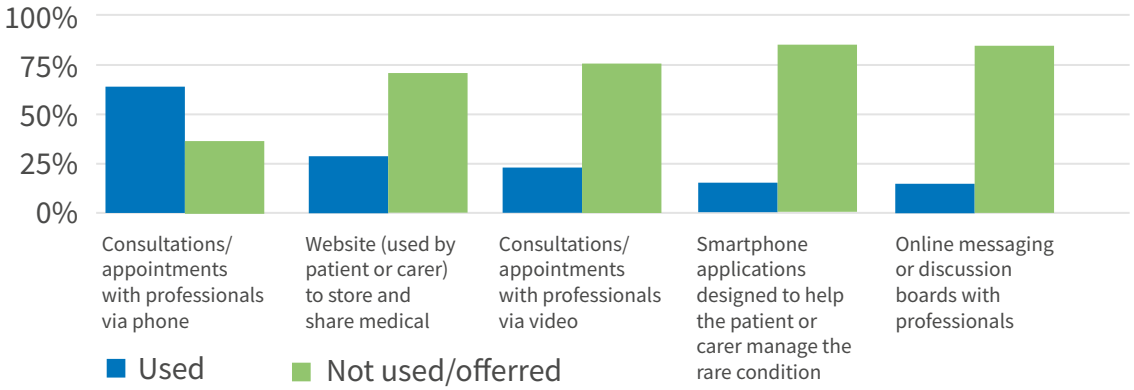
'No coordinated transition from child to adult care. Had to start all over.' *(Respondent 35)*

'Transition from early years to school system has not been good. Support recedes, have to reapply.' *(Workshop participant)*

'Experts keen to diagnose at first, felt listened to and had confidence. But since daughter reached 18 confidence very low, despite ongoing complex and serious issues. No interest from HCPs.' *(Workshop participant)*

DIGITAL, DATA AND TECHNOLOGY

Technology - used vs not used/offerred



There was great variation amongst respondents about their use of technology. The most common (63% of respondents) way in which respondents had used technology was 'Consultations / appointments with professionals via phone'. The next most common (29% of respondents) way was 'Website (used by patient or carer) to store and share medical information'.

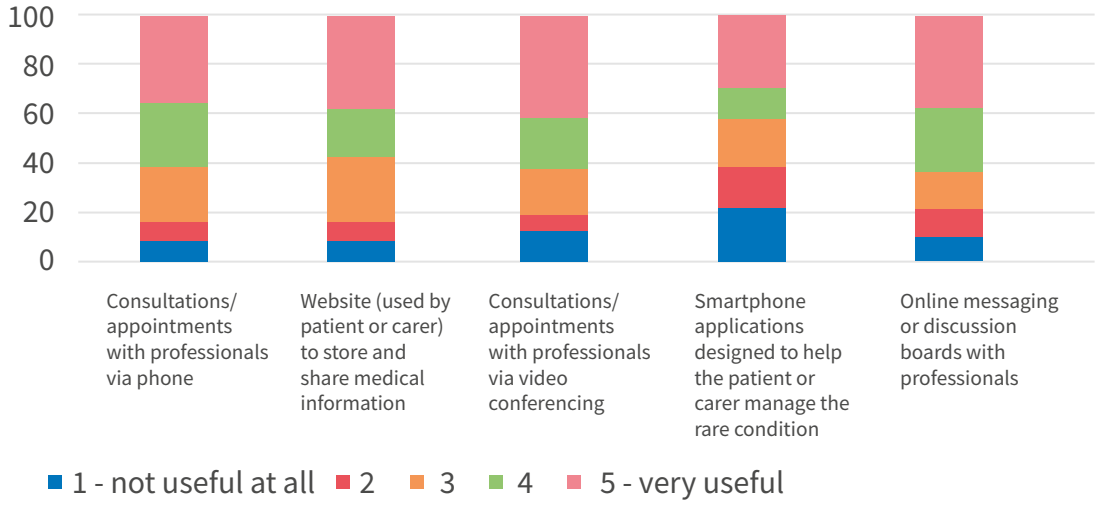
Respondents' negative experiences with technology centred around phone or video conferences not being as suitable for them as face to face consultations, especially if any physical examination was needed. Respondents also highlighted how technology did not always work and using some technology could lead to increased levels of stress or anxiety.

'It is very difficult for his paediatrician to provide support when she can't see him.' *(Respondent 1,311)*

'I have stopped using a fitbit as it made me become rather anxious and obsessive.' *(Respondent 169)*

'I find over phone it's a lot easier for the patient to be interrupted or cut off when they are trying to explain something.' *(Respondent 472)*

Usefulness of technology used



Generally, for respondents who had had the opportunity to make use of various technologies, they considered them more useful than not.

The least useful of the technologies seemed to be 'smartphone applications designed to help the patient or carer manage the rare condition', with less than half (42%) giving a rating of 4 or 5, with 5 being 'very useful'.

Respondents' positive experiences with technology centred around how phone or video conferences saved time and money and avoided potentially difficult travel. Respondents also highlighted how technology meant they were able to do things from their own home which many found more comfortable.

'Phone calls with specialist nurse is useful so I don't have to travel.' *(Respondent 888)*

'Wearables to track location, falls detector, emergency SOS.' *(Respondent 248)*

'Emailing consultants directly is of huge benefit.' *(Respondent 1,245)*

'Online prescription renewal is one of my favourite things.' *(Respondent 429)*

'Patient network has provided some Webinars with experts. Really good.' *(Respondent 567)*

DISCUSSION

Main message from this report

There are more than 6,000 rare conditions, a diverse range of conditions, affecting a diverse range of people in the UK. This diversity is reflected in the findings from this study. These are not easy findings to read. Each negative experience is an encapsulation of someone, or a family, who has had to face major challenges not just because of their rare condition, but in accessing the support for their condition that they are entitled to.

Other clear messages from this report

People with rare conditions are insufficiently served by the system

Time to diagnosis, access to specialised services, coordination of care, satisfaction with the NHS, access to medicines, awareness of healthcare professionals, access to information – every topic we covered showed a wide range of experiences. There is a lot of work to be done to improve outcomes and experiences for people living with rare conditions.

Diagnosis is crucial

Our findings have underlined the importance of diagnosis for people living with rare conditions. A fast diagnosis can mean quick access to specialised care and avoids the diagnostic odyssey and potential misdiagnoses.

Awareness of rare conditions among professionals is valuable and needs improving

No one can be aware of all rare conditions individually, but ignorance of rare conditions generally among healthcare professionals leads to slower referral, slower diagnosis, misdiagnoses and slower access to appropriate specialised care.

Professional care coordination is rare but necessary

We found that most people with rare conditions have to coordinate their own care, but those that have experienced professional care coordination prefer it. We saw the complexity of so many peoples healthcare, and how many of our respondents are unable to work or learn because of their condition.

Pockets of excellence demonstrate the system can deliver

Throughout this study, we have found positive experiences. They have rarely been the majority experience, but they demonstrate that with resource and coordination, it is possible to deliver excellent care to people living with rare conditions.

Impact of rare disease policy in the UK

This is not the place for a full review of the UK Strategy for Rare Diseases (2013-2020), but it is clear that the need for good quality policy making for people living with rare conditions has not gone away. The priorities of the Strategy are endorsed by our respondents, and it is a shame that none of those priorities appear to have been solved.

There is much great work that has been done in the era of the Strategy, and a lot of this has not yet borne fruit. Genetic Alliance UK is aware of a great deal of talent and activity in areas such as genomic

diagnosis, research to identify new conditions, the registration of existing conditions, creation of repositories of data on rare conditions for research, the examination of models of care coordination, programmes to deliver care for the undiagnosed community and many others.

Progress in addressing the challenges that rare conditions pose is always likely to be slow – systems can take time to develop and implement, and the process of developing research findings is a long one. This makes it all the more important that work underway to serve people living with rare conditions in the UK is visible and measurable. It is unfortunate that the Strategy did not have this visibility and measurability built into it from the very beginning. It would have allowed a platform for accountability, but importantly given people who are waiting on its progress a better understanding of how their world might improve and when.

The Strategy has not been a failure, but nor can we mark it as a success. The ultimate beneficiaries of the Strategy are people living with rare conditions, and this report shows that there is work to be done to improve their experience of care in the UK.

The future of rare disease policy in the UK and some lessons from the past

The UK Framework for Rare Diseases is expected to be published this month (December 2020). Its structure has been informed by the National Rare Disease Conversation in 2019, giving the focus to diagnosis, awareness of rare diseases among healthcare professionals, care coordination and access to therapies. Our findings reinforce those from the Conversation making these four topics the key priorities.

The need for overarching UK policy for rare conditions is arguably greater than ever before. The UK is leaving the European Union, potentially disconnecting from European Reference Networks, the growing platform for sharing expertise on rare conditions across borders. We will disconnect from the medicine and medical devices regulation programmes of the EU. We will very soon have national genomic medicine services running in the UK. These have enormous potential to increase the proportion and rate of diagnoses of rare conditions — creating a growing population of people with diagnosed conditions. These people will need care, care pathways, and care coordination. Many will only be able to have these with targeted research and data collection. Genome UK (2020) commits the UK to 'increase life science industry research and development spend in the UK by identifying new opportunities for innovative and cutting-edge industry partnerships'. A glance at any clinical trial registry today shows that those opportunities are in rare conditions — and it follows that this work cannot happen in isolation. What is good for people living with rare conditions is good for innovation: a comprehensive care system that can identify and support everyone living with a rare condition.

The Conversation and our own Patient Experience Report (2015)[10] are the only two times during the life of the Strategy that the views of people living with rare conditions were examined. A more regular examination of their views, perhaps mirroring the structure of the NHS England Cancer Patient Experience Survey[11] would provide a much needed signal on how the new policy is affecting people's experiences of care for rare conditions in the UK.

A regular rare condition experience survey is a key starting point for an important and necessary element of forthcoming policy missed in the Strategy: metrics to measure implementation. These were missing in two dimensions previously, implementation and impact. It has not always been clear whether recommendations have been fully acted upon, let alone whether their intended impact has been achieved. A national survey could deliver a significant portion of the latter dimension. Focussed metrics and a dashboard of implementation would fill the gaps.

The timing of the publication of implementation plans of the 2013 Strategy increased the challenge of implementation. The plan for England, with 84% of the population of the UK and a majority of

specialised NHS service providers for rare conditions, came years after the other three nations, and meant that the major opportunities for a collaborative approach to implementing the Strategy were gone. Genetic Alliance UK urges the four nations of the UK to work together to develop their action plans for the implementation of the UK Framework for Rare Diseases to ensure all opportunities for collaboration are properly explored. These need to be delivered quickly. Once 2020 ends there will be a gap that needs to be filled by action plans.

The final lesson from the 2013 Strategy is that progress is rapid and policy dates fast. We hope to see regular review built into the commitments made by the devolved administrations.

Genetic Alliance UK will publish its detailed discussion of the UK Framework for Rare Conditions in time for Rare Disease Day 2021 (28 February). This will contain more detailed recommendations for the action plans we hope to see in 2021.

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