RARE VOICES

A National Report on the Experiences of those with Rare Conditions Affecting the Bone Marrow

2023

CONTENTS

FOREWORD	2
EXECUTIVE SUMMARY AND KEY FINDINGS	3
A LOOK AT THE CONDITIONS	6
DEVELOPING THE SURVEY	8
MENTAL HEALTH AND WELLBEING	10
HEALTHCARE SYSTEM AND CONDITION MANAGEMENT	16
PRACTICAL SUPPORT AND DAY-TO-DAY LIFE	22
OUR COMMITMENTS	26
OUR RECOMMENDATIONS	30
APPENDIX	32
ENDORSEMENTS	32
REFERENCES	34

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Alexion AstraZeneca Rare Disease, Pfizer, Roche and Sobi have had no editorial control over the content of this report.

FOREWORD

To be diagnosed with a rare condition is a life-altering moment – an experience that is deeply personal to individuals, but one that everyone within our communities has faced.

Formed in 2019, the *Together for Healthy Bone Marrow Alliance* is the first and only UK alliance for rare conditions affecting the bone marrow that are not caused by cancer. We consist of seven charity partners who have come together to tackle common concerns for our patient groups, all of whom have been impacted by a rare condition affecting the bone marrow.

We passionately believe that rare disease care should be person-centred, and it is this ethos that motivates us to better understand the experiences of people living – or caring for someone – with a rare condition affecting the bone marrow.

Guided by this mission, we launched a first-of-its-kind national survey to gain comprehensive insights from our community of patients, families, and carers. The National Community Survey aimed to improve understanding of patient journeys and lived experiences, identify key concerns and gaps in existing support, and ultimately shine a light on what matters most to our communities.

With our breadth of experience in supporting those living with rare conditions affecting the bone marrow, we understand the challenges that rare diseases can present for our healthcare services.

From the need for more awareness and research, right through to stronger capabilities for psychological and practical support, equity of care and sufficient funding – improvements in these areas will require collaboration from the Government, industry, healthcare providers, policy groups, patient organisations and researchers.

The findings of this survey could not be more timely or important. We know that the Covid-19 pandemic and the ongoing pressures faced by the NHS have placed additional stress on those in our communities. In January 2021, the Government published the *UK Rare Diseases Framework*, a national vision to improve the lives of the approximately 3.5 million people in the UK living with a rare disease.[1] Since its publication, each of the four nations have released their respective action plans, identifying how they will commit to delivering on the Framework. Despite progress in some implementation areas, such as the appointment of both an Adult and a Paediatric Rare Diseases Clinical Lead consultant post in Northern Ireland, and the advancement of a new toolkit for virtual consultations facilitating access to coordinated care of multiple specialists in England, there is still so much more to be done to ensure tangible improvements.[2,3]

This report outlines the key findings from our survey, which identified the following key areas of concern:

- **Mental health and wellbeing**
- **Experiences of the healthcare system and condition management**
- **Navigating life with a rare condition**

It is clear from these results that our communities often feel as though they have no choice but to be the experts and drivers of their own care. The findings provide new impetus to our existing work and priorities and will guide the actions we take and the improvements we advocate for in the future, both as individual charities, and as a collective Alliance.

The Together for Healthy Bone Marrow Alliance

EXECUTIVE SUMMARY AND KEY FINDINGS

What we set out to explore...

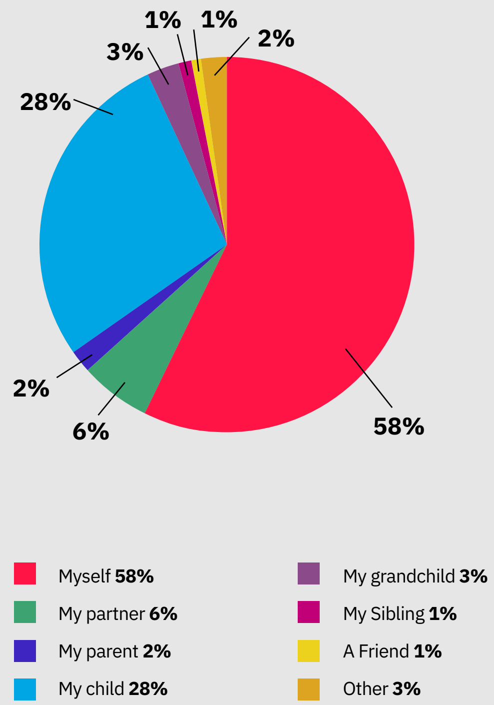
Through the National Community Survey, we wanted to hear from people whose lives are touched, either directly or indirectly, by rare conditions affecting the bone marrow. We set out to uncover:

- The degree of impact rare bone marrow conditions can have on mental health and well-being
- The extent to which people living with or supporting someone with rare bone marrow conditions feel their care is satisfactory and joined-up
- The quality and accessibility of advice and practical support for people with rare bone marrow conditions, and where there may be key concerns and gaps in support
- The level of understanding and awareness of rare bone marrow conditions amongst healthcare professionals and the general public of these rare conditions, and what impact this can have
- The overall impact of living with a rare condition affecting the bone marrow

Who we asked...

People living in the UK whose lives are touched, either directly or indirectly, by rare conditions affecting the bone marrow. This might be as someone who has been given a diagnosis, someone who is living with symptoms, or as someone who supports an individual affected by one of these conditions, for example, a partner, family member, or friend.

Person living with condition



434 responses were received

What they told us...

MENTAL HEALTH AND WELLBEING

Anxiety:

53%
'always' or 'often' feel anxious.

Anxiety is particularly high for those answering the survey as parents of someone living with a rare bone marrow condition, with **31%** 'always' feeling anxious and an additional **46%** 'often' feeling anxious **(totalling 77%)**.

62%
of people affected, either directly or indirectly, by rare conditions affecting the bone marrow, want more information on mental wellbeing.

39%
of those not currently speaking to a psychologist, therapist, or counselor, feel that speaking to one would increase their quality of life.

Loneliness and isolation:

48%
feel lonelier as a result of living with the condition or supporting someone with the condition, and **27%** 'often' or 'always' feel lonely.

33%
A third 'often' or 'always' feel isolated.

Resilience:

45%
Nearly half 'strongly' or 'somewhat' agree that 'when things go wrong in my life, it generally takes me a long time to get back to normal'.

HEALTHCARE SYSTEM AND CONDITION MANAGEMENT

Awareness and understanding:

When asked if they feel that general NHS professionals (outside of their core medical team) have knowledge of their condition, over half **(59%)** disagreed **(31% 'strongly disagree'; 28% 'disagree')**.

Furthermore, respondents feel the level of understanding of their condition among general NHS staff is poor; **34%** think general NHS staff have 'no' or a 'very low' level of understanding, whereas only **6%** think they have 'complete understanding'.

Condition management:

83%
would like access to an up-to-date, verified portal of research about their condition, new treatments, and treatment methods.

53%
Just over half would appreciate some additional support with monitoring symptoms and test results.

PRACTICAL SUPPORT AND DAY-TO-DAY LIFE

Practical support:

65%
would like information on insurance. **62%** would like advice on travelling and associated requirements.

47%
Of those who have tried to access benefits before but haven't been able to, nearly half **(47%)** don't know whether they are eligible for benefit support.

Support for handling public awareness:

50%
would like advice on how to explain the conditions to others, with **54%** describing the level of public understanding of the condition to be 'no understanding at all'.

28%
Nearly a third **(28%)** would like advice on how to speak to their work about the impacts of their condition, and **20%** would like support on how to speak to their, or their child's, education setting.

A LOOK AT THE CONDITIONS

Our charities support people affected by a range of rare bone marrow conditions, including those which are genetic, inherited or acquired during a person's lifetime. The conditions covered include:

APLASTIC ANAEMIA (AA) – a rare and life-threatening blood disorder caused by the bone marrow not functioning properly, that can be acquired or genetic. In people with AA, the bone marrow fails to produce enough of all three types of blood cells: red, white and platelets. The word 'aplastic' means the body's inability to create new cells, so that tissue cannot grow or regenerate.[4]

CONGENITAL DYSERYTHROPOIETIC ANAEMIA (CDA) – a group of inherited anaemias that reduce the number of healthy red blood cells in the body. All CDAs are passed down through families. This typically results in anaemia (low red blood cell count) and too much iron in the body. Over time, CDA can cause organ damage.[5]

DIAMOND-BLACKFAN ANAEMIA (DBA) – a genetic rare blood condition where the bone marrow fails to produce enough red blood cells. DBA is usually diagnosed before the age of two and patients require treatment to compensate for their lack of red cell production. DBA can cause fatigue, poor growth, lack of appetite and a pale complexion. In about half of cases patients also suffer associated congenital abnormalities.[6]

DYSKERATOSIS CONGENITA (DC) AND RELATED TELOMERE BIOLOGY DISORDERS (TBD) – inherited conditions which cause premature ageing of cells and organs due to repair abnormalities in the telomere (the tips of chromosomes). These conditions can lead to bone marrow failure – which causes anaemia, low white blood count and platelet-blood clotting problems – lung fibrosis, liver cirrhosis and other conditions. It can severely affect children, as well as adults later in life.[7]

FANCONI ANAEMIA (FA) – a rare, genetic cancer-predisposingdisorder manifested by a variable presence of congenital anomalies in up to 70% of cases, a progressive bone marrow failure in childhood (usually leading to haematopoietic stem cell transplantation (80% chance), a predisposition to acute myeloid leukaemia (10% chance), and in particular, oropharyngeal/anogenital squamous cell carcinoma in early adulthood (over 50% in post-bone marrow transplant survivors).[8]

PAROXYSMAL NOCTURNAL HAEMOGLOBINURIA (PNH) – an acquired condition where blood cells missing certain proteins are vulnerable to be attacked by a particular part of the body's immune system called "the complement". The process by which the red blood cells are destroyed is called haemolysis and is responsible for many of the symptoms of the disease.[9] Some people have both PNH and AA and those with PNH are most often diagnosed as adults.

SHWACHMAN-DIAMOND SYNDROME (SDS) – a rare inherited condition that affects many parts of the body, particularly the bone marrow, pancreas, and bones. The multi-system disease is characterized by poor growth due to difficulty absorbing food, bone marrow dysfunction leading to impaired production of white blood cells and leukaemia predisposition. The complications of SDS can affect several other parts of the body, including the liver, heart, endocrine system (which produces hormones), eyes, teeth, and skin.[11]

SICKLE CELL DISORDER – Sickle cell disease is the name for a group of inherited health conditions that affect the red blood cells. People with sickle cell disease produce unusually shaped red blood cells that can cause problems because they do not live as long as healthy blood cells and can block blood vessels. Sickle cell disease is a serious and lifelong health condition, although treatment can help manage many of the symptoms.[10]

DEVELOPING THE SURVEY

Launched in November 2022, our survey was designed to be completed by patients, families, carers, and anyone whose lives are impacted by rare conditions affecting the bone marrow.

Co-creation was a core principle during the survey build, enabling the community to play a full part in its design. To achieve this, a phased development process was implemented:

A **Community Panel** was formed to act as a forum, bringing together representatives affected by the conditions. Members of the panel were invited to share their own experiences and had the opportunity to comment on the survey question ideas.

A **Call for Evidence** was issued to gather relevant information, such as scientific papers, literature, local and national guidelines, patient and healthcare professional questionnaires (past or present), and any additional resources that could be relevant.

An **Insights Paper** was developed based on the evidence submitted through the call for evidence and the community panel feedback. This paper had two main aims. Firstly, it 'took the temperature' on the volume, depth, and focus of the evidence that already exists for these conditions to ensure the survey would uncover new data. And, secondly, it informed us about what people living with each of these conditions felt would be valuable to know, to ensure that the views of the community were fully reflected in the survey design.

The process was led and governed by a **Steering Group** made up of representatives from the seven charities of the Alliance. Depending on the terms of their support, some industry partners were also given the opportunity to submit questions to be considered for inclusion in the survey, however they had no editorial control over the content of this report.

Methodology:

An online survey was distributed through community networks, social media, and other channels. The survey was open to anyone affected by a rare condition affecting the bone marrow.

Qualitative, **one-to-one interviews** were conducted to bring the survey results to life and explore any specific issues in further detail.

All respondents were living in the UK at the time of completing the survey or interview and, after providing their consent, were given the option of providing further demographic details such as age, gender, ethnicity, and employment status. All respondents were asked which condition they are living with or, if they are connected to a person living with this condition, how they would describe this connection, for example partner, parent, child, grandchild, sibling, friend or other.

The online survey was open from 15th November to 18th December 2022, and the one-to-one interviews took place in January 2023. In total, 434 responses were received for the online survey, and seven people were interviewed for the qualitative survey – each representing a different condition.

Timeline:

MENTAL HEALTH AND WELLBEING

Living with a rare condition, or supporting someone who is, will impact how someone thinks, feels and acts. In addition to the medical needs and consequences for the patient, the emotional and psychological burden brought about by any disease, and especially one which is rare, can have a significant impact on a person's life.

Through our research leading up to the survey, we found consistent evidence of the negative impact that rare conditions affecting the bone marrow have on mental health and wellbeing, as well as the need for improved psychological support. The survey was an opportunity to delve deeper into this area, to try to understand how people are feeling and coping in relation to living with, or supporting someone living with, one of these conditions.

> It makes me worry about my child's future and to what extent it will impact her life and by how much it will shorten it.
>
> **Anon**

Anxiety and isolation

> I can't stop worrying about what the future has in store for my daughter and how that impacts her and my family.
>
> **Anon**

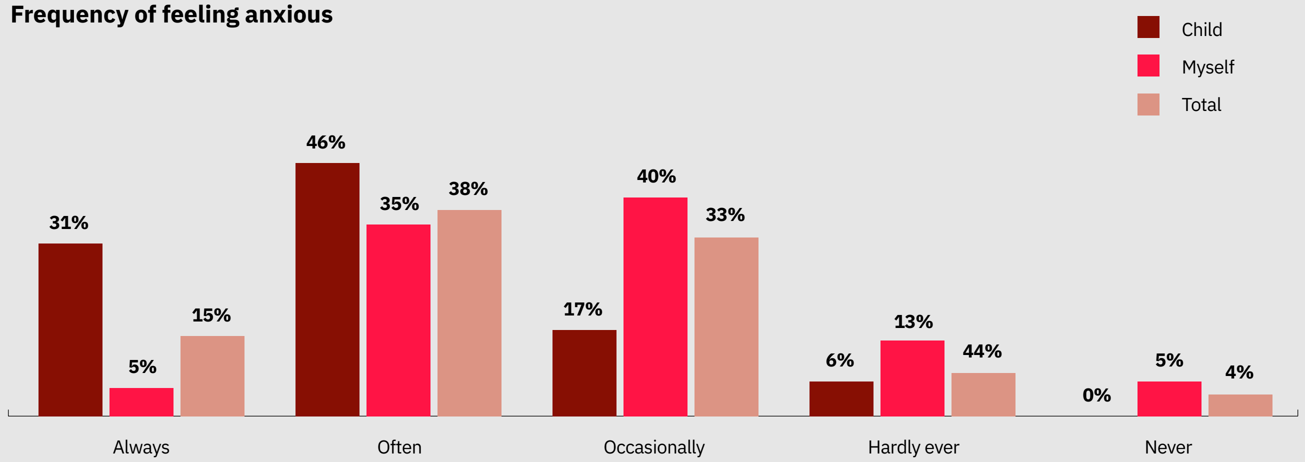
> I worry for the day my son's bone marrow changes – the pandemic has had a massive impact on his mental health and that of our family.
>
> **Anon**

Feelings of anxiety are widespread amongst these communities, with 53% of respondents reporting always or often feeling anxious. This is particularly relevant for parents of someone living with a rare bone marrow condition, with 31% always feeling anxious and an additional 46% often feeling anxious – totalling 77%. When asked to explain in their own words, respondents indicated that parental feelings of anxiety are significant when it comes to the future of their child, with one parent reflecting, ***"It's heart-breaking, the lack of control...we don't know whether we're coming and going some days [with] no clear outlook to the future. Everything is overshadowed by this condition. I have never known anxiety like it..."***.

53%

of respondents reporting 'always' or 'often' feeling anxious. Feelings of anxiety widespread amongst these communities

Frequency of feeling anxious



Gaps in psychological support

Our findings reveal a noticeable disparity between people's emotional experiences and the level of support they are receiving.

62%

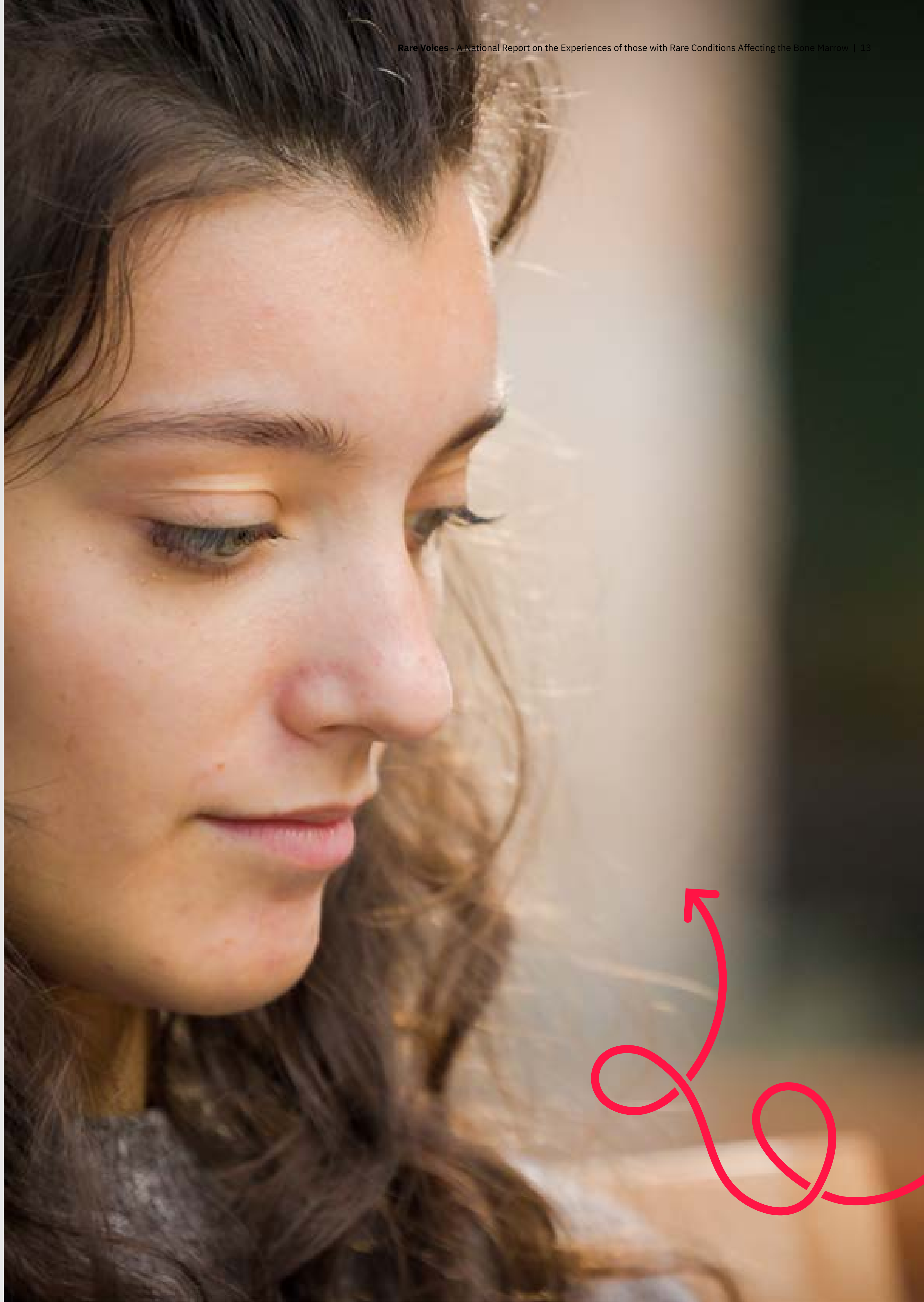
of people affected, either directly or indirectly, by rare conditions affecting the bone marrow, want more information on mental wellbeing.

39%

feel speaking to a professional would increase their quality of life. (Of those not currently speaking to a psychologist, therapist, or counsellor).

This is further evidenced by all the qualitative interviews, which confirm that support from a psychologist or counsellor would be welcome: some cited experiences of depression, while others recognised more broadly that they would benefit from speaking to a professional. However, people reported challenges with knowing what type of counselling they would need, where to access it and how the costs would be covered.

> "I had to BATTLE and plead in desperation to get psychological support for my son. This was one of the biggest challenges I went through. Support for patients and families in gaining access to psychological support would be invaluable."
>
> **Anon**



Loneliness and contact with others

Feelings of loneliness and isolation also emerged in the findings. **64%** report some degree of loneliness, with **27%** saying they often or always feel lonely. This is significantly higher (over four times) than the **6%** of respondents to a UK Government Community Life Survey 2021/22 who said they often or always feel lonely.[12] This implies that people living with rare bone marrow conditions are over four times more likely to be lonely than the general population.

48%

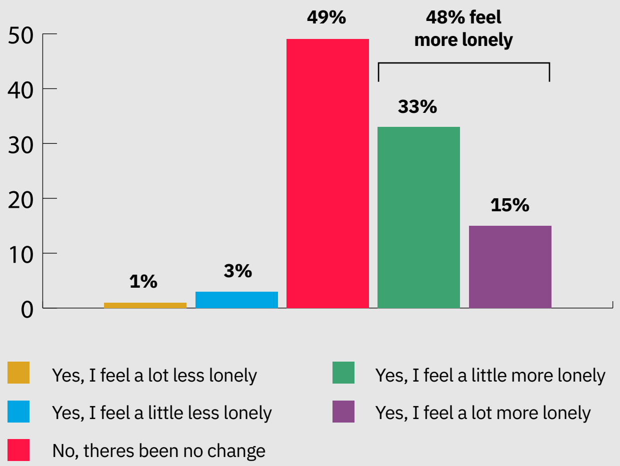
feel lonelier as a result of living with the condition or supporting someone with the condition, and **27%** 'often' or 'always' feel lonely.

33%

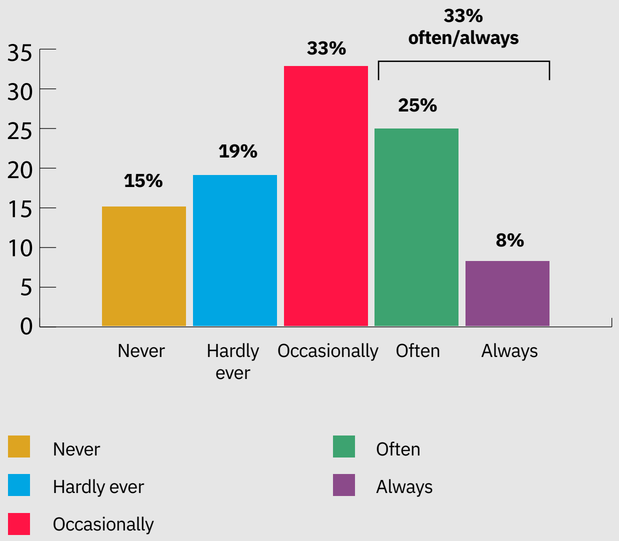
A third 'often' or 'always' feel isolated.

The responses about loneliness were given further context by additional responses, with a third **(33%)** only meeting up with others outside the home once a fortnight or less, and nearly half **(48%)** seeing friends and family members less regularly as a result of the condition. The reasons behind this were found to be multifaceted, from the impact of fatigue to the experience or prospect of pain causing an inability to plan ahead, as well as feeling uneasy about social situations where there may be a lack of support or even judgement.

Has the feeling of loneliness changed as a result of the condition/supporting someone with the condition?



How often do you feel isolated from others as a result of the condition/supporting someone with the condition?



"I can no longer plan for the future, I have shrunk my world to family and mainly two friends, I am fearful of going out, and I don't talk about my illness much to anyone.

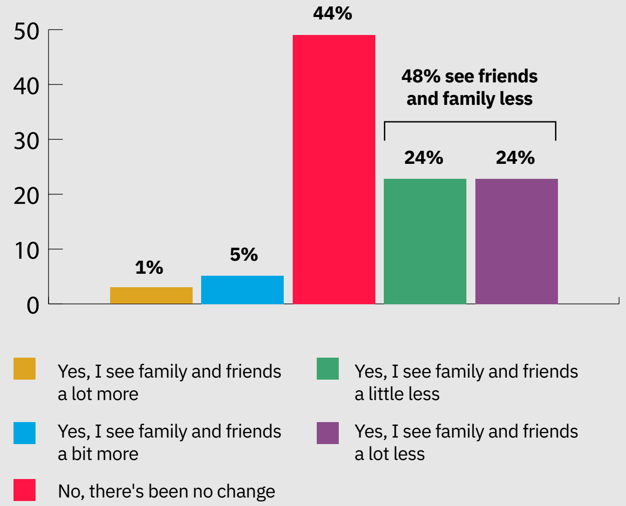
Anon"

Lesley Howells is Maggie's Lead Psychologist, Consultant Clinical Psychologist, and Lead Clinical Psychologist on the Better Together for Healthy Bone Marrow Project, working with six of the Alliance charities to improve psychological provision for our communities. Commenting on the survey, Lesley said, *"The results from this project provide a rich evidence base for the unique psychological needs of people living with these ultra rare conditions. The need is wide ranging and cross-generational. From my own work with these conditions, I know that, depending on the bone marrow condition, the focus for support may be on: the family adjusting to uncertainty during a child's infancy; a child and their parents as they become a teenager, where peers become more important but the medical regime required for their condition needs to be maintained; the psychosocial impact of stem cell transplant as a young adult; and living with a 'watch and wait' approach to treatment whilst wanting to plan a family or build a career".*

"I sometimes feel like the seriousness of the condition is not understood, and I find myself having to justify my decisions in relation to things like social settings, as people don't understand why I have to be more cautious than most.

Anon"

Impact on regularity of seeing friends



HEALTHCARE SYSTEM AND CONDITION MANAGEMENT

People living with a rare condition face many hospital appointments and other touchpoints with the healthcare system. In our survey and interviews, we asked several questions to get a better idea of people's experiences of, and feelings towards, the healthcare system. We included questions around appointments, ease of access and joined-up care, through to levels of awareness and understanding from both the core medical team directly responsible for managing and treating a person's condition, as well as healthcare professionals more generally.

Navigating the healthcare system

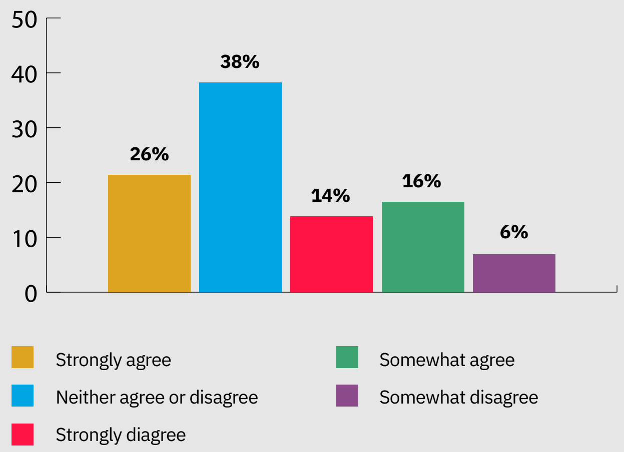
The survey results clearly demonstrate that navigating the healthcare system appears to be a struggle for many. This links to a wider theme which came through, that some feel as though they have no choice but to be the experts and drivers of their own care. While most **(83%)** know where to access the information they need to help manage their condition, over half of people surveyed **(53%)** would like to receive additional support with keeping track of information, such as symptoms and test results. Furthermore, only around half **(54%)** of respondents think test results are easily accessible for medical staff when needed. One respondent reflected, ***"I've built up various 1–4-page summary guides to hand to new consultants"***, while others raised the utility of a symptom tracking app. Factors such as condition, perceived stability of condition, experience, personal organisation and technological experience, all impact on how people manage their care personally. Although it is reassuring that overall **64%** feel confident navigating the healthcare system in relation to their condition, more than one fifth **(22%)** of the 434 respondents do not feel confident doing this, with **54%** indicating they would welcome more advice and support.

53%
would like to receive additional support with keeping track of information, such as symptoms and test results.

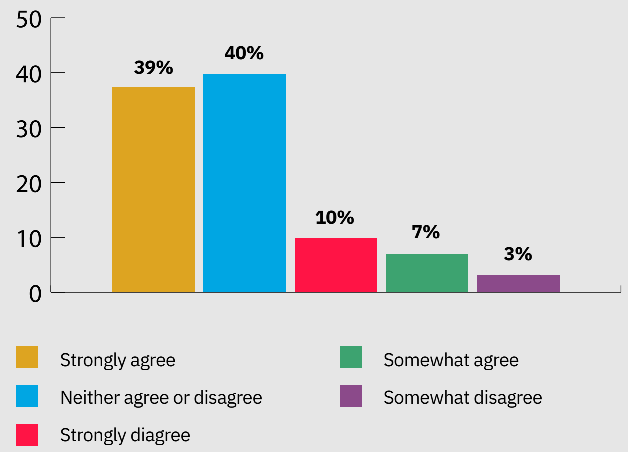
54%
think test results are easily accessible for medical staff when needed.

54%
indicating they would welcome more advice and support.

I feel confident navigating the healthcare system in relation to the condition.



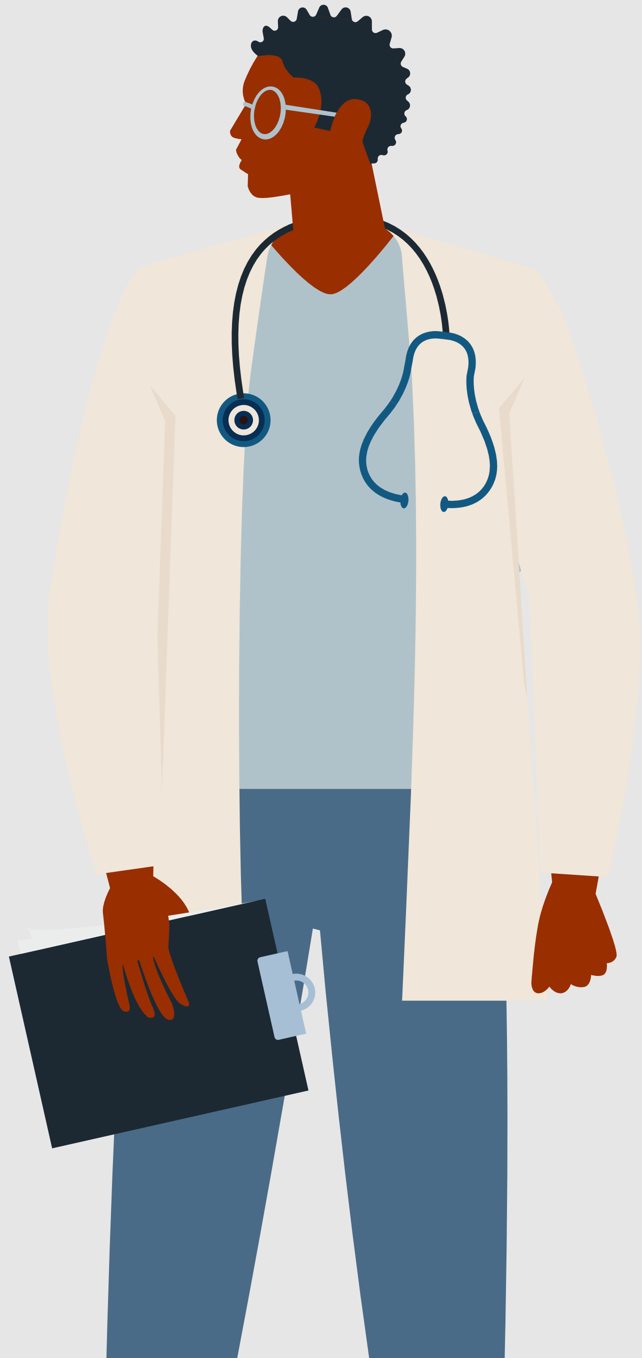
I feel confident advocating for my health/the health of the person I support, with healthcare professionals.



Satisfaction and joined-up care

Joined-up care is identified as a central theme in the NHS Long Term Plan, and is described as bringing together and coordinating different health, care and other services to meet the needs, choices and aspirations of the individual.[13] While satisfaction with people's core medical teams is generally positive, with **61%** of those surveyed very satisfied, and a further **27%** quite satisfied with this care, lower satisfaction tended to be related to not feeling that there was a main contact or specialist available to speak to. Moreover, in addition to the finding that only **54%** of respondents think test results are easily accessible for medical staff when needed, just **51%** strongly agree that the core medical professional team managing the condition have direct access to specific medical notes.

Meanwhile, the logistics around attending appointments can be challenging for many; **19%** believe appointments do not take place within a reasonable number of locations and a quarter **(25%)** feel as though the locations of their appointments are not a reasonable distance from home. It is clear that respondents want coordinated care that treats the patient more holistically, whether they are engaging with the health service in relation to their rare condition or for other health concerns. One respondent highlighted that ***"specialist teams are only interested in one condition but when you have many you need a coordinator to oversee you as a whole person."***



The need for a specialised service

While we speak with a unified voice, it's worth reiterating that each condition is unique and lived experiences will be influenced by a wide range of factors. A significant disparity exists between these conditions, for instance PNH has a funded National Service which is renowned worldwide for the high quality knowledge and expertise in managing patients with PNH. As PNH is a rare disease, there is a concentration of experience within the Service that cannot be matched elsewhere in the world. The Service, which is based in Leeds and London, is a designated 'Highly Specialised Service' (PNH HSS) which provides support and access to treatment to people living with PNH. The PNH HSS was established following a successful application to the then National Commissioning Group for Highly Specialised Services in England, and there are agreements in place with Healthcare Commissioners in Scotland, Wales and Northern Ireland for this Service to provide support to PNH patients from the rest of the UK. ***The Together for Healthy Bone Marrow Alliance*** recognises the disparity between the patient experience of people with access to the PNH HSS compared to those without, and understands that the latter experience is fragmented, and lacking multi-disciplinary care.

Indeed, this funded Service has improved the quality of care and experience of the healthcare system for PNH patients. In some places, the survey results supported this; for instance, **50%** of PNH respondents know where to access the information needed to help manage the condition, compared to **39%** of all other condition respondents (percentages for 'strongly agree'). The survey has also shown that a higher proportion of people with PNH **(77%)** feel supported by services (healthcare and/or charity/patient groups) than people living with the other rare conditions affecting the bone marrow **(63%)**. Thinking about care more broadly, **66%** of PNH respondents report being very satisfied with the level of care they receive from their core medical professional team for their condition, compared to **60%** for all other conditions.

Personal accounts also demonstrate the need for specialised services for these rare conditions.

"When my father became very ill with bone marrow failure, lung fibrosis and cirrhosis of the liver, he saw at least 10 different specialists before finding a doctor who took our concerns that the problem could be a rare Telomere Biology Disorder seriously. The complex nature of the condition meant that he needed to attend hospital almost every week for appointments with different specialists or for blood transfusions in several different locations. Better education for doctors would have meant a faster diagnosis and access to expert, multi-disciplinary care in one location would have helped him and our family enormously."

Anon

"I didn't get any support from anybody other than my core medical team, as my illness began in 2001 when little was known. The ongoing problems I have post-transplant are dismissed by GPs who have no understanding of my previous diagnoses and excessive treatment plans."

Anon

50%

of PNH respondents know where to access the information needed to help manage the condition.

77%

of PNH respondents feel supported by services (healthcare and/or charity/patient groups.

66%

of PNH respondents report being very satisfied with the level of care they receive from their core medical professional team for their condition.

"The PNH service consider it a privilege to be able to offer patients a gold standard service for rare disease. With a centralized team available 24 hours a day in case of emergency, and a wider team available during working hours, patients are supported by telephone in between appointments by staff with specialised knowledge. Travelling to 8 different locations around the UK to try and reduce travel for patients when accessing expert care, has been appreciated by patients and clinicians. The model of care delivery is considered an example of excellent care by other PNH services worldwide, and can be used as a template for other rare diseases with similar patient numbers as PNH." –

Dr Morag Griffin, Consultant Haematologist

"The PNH National Service has established a highly specialised service for the individuals affected by the condition, their families and the broader healthcare system providing a positive and transformative impact with significant improvement to quality of life. This initiative has demonstrated focused expertise, with consistency of access to medical and nursing staff who understand the intricacies of the diagnosis and its management, providing tailored treatment and care. Working in a shared care approach with local healthcare teams means we are able to deliver highly individualised and specialised care which supports better management of symptoms. The service has become a hub for patient support and advocacy, offering resources and guidance to individuals and their families. This has eased the emotional and psychological burden of dealing with a rare condition and has created a sense of community among those affected. It has offered a catalyst for research which has led to the development of new treatments, contributing to the advancement of research in other areas. The service has gained international recognition as a model for addressing rare disease – inspiring others and helping develop networks in the community. Most importantly the service has inspired hope, resilience and determination in the rare disease community, showing that dedicated efforts and resources can make a substantial difference." –

Louise Arnold, Trainee Advanced Clinical Practitioner, PNH National Service

The need for increased awareness of rare diseases

The National Community Survey has helped shape our understanding of how people living with, or supporting someone with, a rare condition affecting the bone marrow access, navigate and interact with the healthcare system. The UK Rare Diseases Framework highlights the need for better awareness of rare conditions among healthcare professionals through appropriate education and support, identifying that a patient's journey will often start with their GP and they will likely have various touchpoints with paramedics in their lifetime.[1] It is reassuring that most people surveyed are satisfied with the core medical team managing their care; however, when asked if they feel general NHS professionals (outside of the core medical team) have knowledge of their condition, the majority of respondents **(59%)** disagreed **(31% 'strongly disagree'; 28% 'disagree')**, with **34%** feeling as though general NHS staff have no or very low understanding of their condition, and only **6%** thinking they have complete understanding. One patient commented, ***"It just seems so overwhelming. I feel every step of the way I have had to try and explain the complexities all over again".***

> "We haven't been able to find support to solve symptoms which are outside the core symptoms but have a huge detrimental effect on our child's life."
>
> **Anon**

Some respondents felt it cannot be expected for healthcare professionals to be familiar with every rare condition. Some expressed frustrations about the lack of awareness amongst wider healthcare professionals, feeling as though the only way to experience a form of joined-up care is to take responsibility for joining it up themselves. Our findings demonstrate the need for increased awareness of rare bone marrow conditions among healthcare professionals and we support the Government's call for improved training to support quicker diagnosis and better patient care.[1]

> "More of a timeline about what to expect and when for post stem cell transplantation. Maybe this is impossible as it will be different for everyone, but I feel that I am just living from one appointment to the next."
>
> **Anon**

> "I'm made to feel like I'm exaggerating the condition and that my daughter is fine because she looks fine. It makes me feel like never confiding in anyone again about the seriousness of it."
>
> **Anon**

PRACTICAL SUPPORT AND DAY-TO-DAY LIFE

Being diagnosed with a rare condition affects every aspect of someone's life, from social life to education to employment. For this reason, effective support is fundamental to ensuring an individual's overall wellbeing and quality of life. This is especially important for someone living with the challenges presented by a rare condition as, when compared to more common conditions, various forms of support may not be as accessible or developed.

Effective support can take a range of forms. Through this project, we set out to better understand the gaps in existing support for our communities, beyond their condition management. We explored peer support between people connected by their conditions, support from wider networks such as families, friends and charities, and professional support. We also explored practical support and suggestions for assisting with the impacts and challenges faced as a result of living with, or being affected by, a rare bone marrow condition.

Portal of relevant information

Our findings showed the need for current, reliable resources that are readily available for patients to view and use as needed. 83% called for access to an up-to-date verified portal of research about their condition, new treatments, and treatment methods. As one of the most concerted results of the survey, this finding highlights the importance of a centralised and regularly updated hub of relevant information with which people can engage. Research updates and results, including industry research, should be communicated in a timely manner internationally, as well as with study participants and communities affected as a priority.

"Due to the condition being so rare, I naturally turned to Google to try and find out more about what my diagnosis would mean for me, and what my prognosis might be. However, the information I came across on Google was sparse and alarming – PNH sounds very scary when you look it up. I would have really valued being pointed straight away in the direction of an up-to-date, comprehensive hub of information about my condition, to improve my understanding about what I was going through and manage my expectations."

Jovita Fawcett, living with PNH

"Patients and carers are often left with the burden of keeping a clear history of both medication and test results due to the limited exchange of information across multiple disparate clinical systems. Not having this information can often lead to unnecessary duplicated tests and other potential delays and impacts to treatment."

Kapil Gotecha, SDS UK

Alongside this, our findings show that people would value other forms of practical support and improved information and resources.

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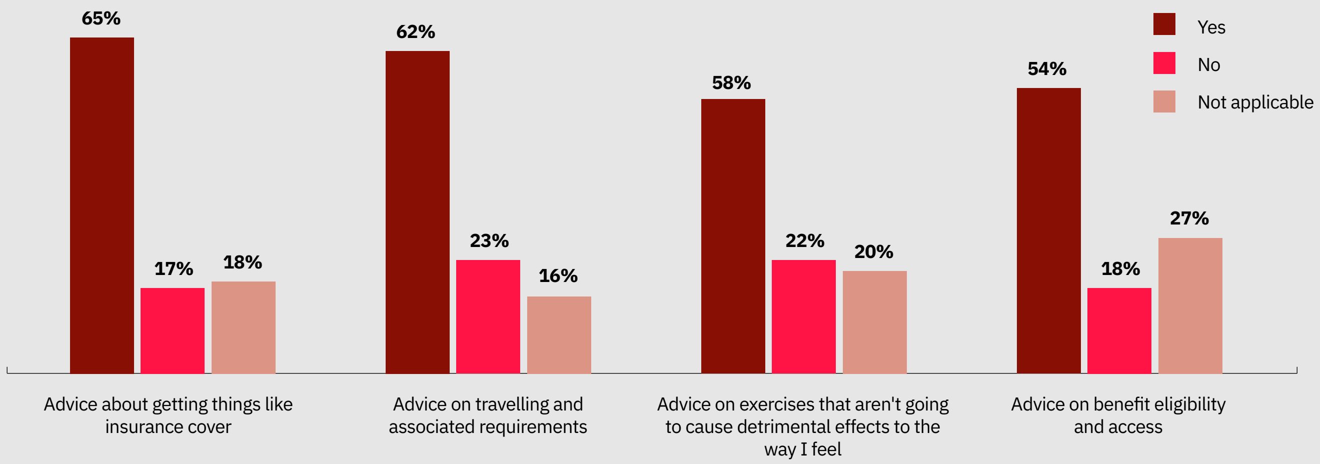


Practical support

This project identified that our communities need better access to information and research around navigating things such as insurance cover **(65%)**, travel and associated requirements **(62%)**, ways to exercise **(58%)** and benefit eligibility **(54%)**.

65%

need better access to information and research such as insurance cover.

Benefit system support

Looking more closely at accessing benefit support, **18%** were unable to access the benefit system despite wanting to. Of this group, **47%** were unable to access the system due to not knowing if they are eligible, and **15%** were unable to find the support to help them do it.

18%

were unable to access the benefit system despite wanting to.

15%

were unable to find the support to help them do it.

> "I was rejected by PIP* (after previously being on DLA*) as they didn't understand my condition and how it affected me."
>
> **Anon**

Living with a rare condition can make everyday practicalities more challenging, and there are clearly huge opportunities for improved information and resources to support with these challenges. Lesley Howells, Maggie's Lead Psychologist, commented, *"These results highlight a desire for better information about the condition, treatment and research into the condition. This could have a tangible effect on the high levels of anxiety reported, and provide a more hopeful vision of the future. For someone struggling with the financial impact of a diagnosis, access to benefits advice and support with applications will have a bigger impact than a session on how to manage the stress."*

* PIP: Personal Independence Payment
DLA: Disability Living Allowance

Public awareness

Alongside practical assistance, half **(50%)** of patients, families, and carers would welcome advice on how to explain their rare condition to others, with the majority **(82%)** describing the level of public understanding of the condition to be very low, or to be non-existent. Although it is regarded as reasonable that the public have a lack of understanding about these bone marrow conditions because they are so rare, this leads to compounded feelings of disheartenment and isolation. For instance, some described the lack of public awareness as **"exhausting"**, with another recounting ***"it makes us feel isolated and hard to talk to others... as no one understands"***. Other respondents explained ***"it's very frustrating to have to explain it over and over again"***, and there can be ***"no understanding of how exhausted you are constantly simply because you look ok"***. In particular, people would appreciate support in explaining the condition to their workplace **(28%)** and to their, or their child's, education setting **(20%)**. The interviews conducted as part of this project reinforce that the 'hidden' nature of these conditions can make them more difficult to communicate about effectively.

> Misunderstood. Having to explain that my condition is more than just anaemia. Whilst I appear well and I have to continue to work to live, it is difficult for individuals to understand how serious my condition is.
>
> **Anon**

> Isolated and under-supported generally in comparison to those patients with cancer/leukaemia.
>
> **Anon**

> It makes me feel concerned that if my daughter is taken ill when I am not around it is worrying as no one will understand her condition. It makes us feel isolated and it's hard to talk to others about as no one understands.
>
> **Anon**

> Makes you feel less connected as others don't understand the challenges you face supporting someone with the condition.
>
> **Anon**

The value of charities

The findings suggest that support and connection are mainly found via charities or online groups set up for people with the same condition, with one person stating that ***"they are a lifeline for families"***. There was a general feeling that support groups are particularly helpful in the early period of diagnosis, with some people wishing they had reached out to their relevant charity sooner. Another theme is the importance of these groups for families, and for lessening feelings of loneliness.

Organised events were identified as a welcome way of sharing information and meeting people with the same condition, and one patient called for ***"more opportunities to hear families' stories and network with other families"***, which charities can facilitate.

> Fully embracing the lower intensity, but no less impactful, psychological resource the communities themselves provide in the sense of belonging they create and the vital role-models of people with a lived experience that they offer, and the readiness of clinicians to offer clear and compassionate communication at, for example, family conferences.
>
> **Lesley Howells, Lead Psychologist**

OUR COMMITMENTS

Together, we have the ability to amplify the voices of our communities. What's become apparent through this project, is just how aligned the issues of our communities are. As organisations, we are committed to using these findings to inform our focus moving forwards.

1

Developing mental health support

The survey clearly evidences the mental health challenges of living with these rare conditions. We wish to use these key findings to obtain and/or strengthen and maintain psychological support for patients, carers and family members particularly at the point of diagnosis, plus the facilitation of continued face-to-face peer support to reduce feelings of anxiety, loneliness and isolation.

Due to the variation in treatments available for the different conditions covered in this report (for instance, curative vs. lifelong maintenance), the psychological support needs of the patient and family networks will differ. This is also true for genetic vs. acquired conditions. Furthermore, it's important to remember that mental and well-being support can often look like practical support to ensure basic needs are being met; a stepped care approach is needed.

Thanks to support from the National Lottery, six of the charities of the Alliance will be working with a psychologist over the next two years to deliver psychologically-informed support in a stepped care approach, from trained volunteers answering the helpline to listen and signpost people to useful resources, to increased access to specialist psychological support. However, structural change is needed to address this unmet need in a sustainable way, and we're calling for psychological support to be part of the care plan for each rare disease patient.

Lead Clinical Psychologist on the **Better Together for Healthy Bone Marrow Project**, working with six of the Alliance charities to improve psychological provision for our communities, Lesley Howells commented ***"Our response must be versatile to respond to the diverse need of the rare bone marrow community. Through my work with these communities, I have found the impact of a diagnosis on a person's close family to be very clear, particularly when a child or parent is diagnosed. I have highlighted a need for expert psychological support, including clinical psychology provision, as well as a Family Therapist with specific skills in systemic and relational psychological approaches for all ages. Critically, there is also a need for the resource of an experienced nurse, knowledgeable in bone marrow failure and equipped with additional psychological training, to provide an easily accessible multi-faceted approach, similar to the Maggie's unique hybrid role of Cancer Support Specialist.***[15]

"But, in the long term, this need must be met in a sustainable way, ideally through an integrated psychological therapies and support framework which utilises the shared skills and resources of both the NHS and third sector providers, similar to the framework introduced in 2022 by the Scottish Government for cancer.[16] ***The scale may be smaller, but the same shared and integrated vision is essential.***

"What is great about the report as a whole is that it reveals more potential, practical ways to support emotional wellbeing and resilience. For example, by providing specific advice on how to explain the rare condition to others (which half of respondents have said they would welcome), and by connecting people with others who have experience of the condition, small organisations like ours can have a real impact on the feelings of isolation reported."

Noémi Roy, Consultant Haematologist, Oxford University Hospitals and Trustee, Congenital Anaemia Network (CAN)

2

A renewed focus on improving support for accessing benefits and insurances

The demand for support with benefits advice was no surprise to our Alliance, who know the financial impact living with a rare condition can have, especially in the cost-of-living crisis. Those affected by these rare conditions, unlike cancer, do not receive automatic access to some sources of financial support. The Aplastic Anaemia Trust (AAT) has developed a partnership with Maggie's Cancer Care charity, which enables referrals to specialist benefits advisors. Last year, the AAT saw an increase in demand for this service of 42%, with the biggest increase when the cost of energy bills rose in the Autumn.

Six Alliance charities will be working together on a pilot project in 2024 to strengthen our support to people who need help understanding and accessing the benefits system. This project will look at improving existing charity provision, and creating support for the conditions where it does not exist currently. We expect this project, funded as part of the three-year National Lottery Community Funded Better Together Project, will highlight how the existing system is not addressing financial support needed by those with rare conditions. We hope this will inform recommendations for system change and provide a useful template for other rare conditions.

It is our view that the UK's benefits system should appropriately recognise people diagnosed with rare conditions, many of which are invisible. Our organisations can provide feedback and insight into how the current benefits system is not fit-for-purpose as it is excluding people with rare conditions from financial support that is less difficult to access and obtain for people with more common conditions.

3

Supporting the development of a repository of current research developments, clinical studies treatments which is accessible to patients and families

The findings demonstrate a clear and unified call for a portal of accurate and detailed information about these conditions that would be accessible to and understandable by patients, carers and healthcare professionals. The charities will continue to look at this issue closely, for instance the PNH Global Alliance (PNHGA), of which PNH Support belongs, strives to upload a list of clinical trials in PNH, and is planning to develop a webpage for accessible research results.

"It's widely recognised nowadays that patient involvement in research is important, and The AAT is keen to provide more opportunities for researchers to update patients about their work and findings. We'd love researchers and clinicians to recognise the link between this desire for research information and the high levels of anxiety felt by our communities, particularly regarding concerns for the future. In my years working closely with aplastic anaemia patients and their families, it is evident that this anxiety can be partly due to a lack of up to date information about the conditions and the prognosis for people to be able to find online. Providing this sort of information about research, trials and other relevant health developments can offer certainty about the current environment and hope for a future with better outcomes."

Stevie Tyler, The Aplastic Anaemia Trust

4

Facilitating improved opportunities for peer-to-peer support

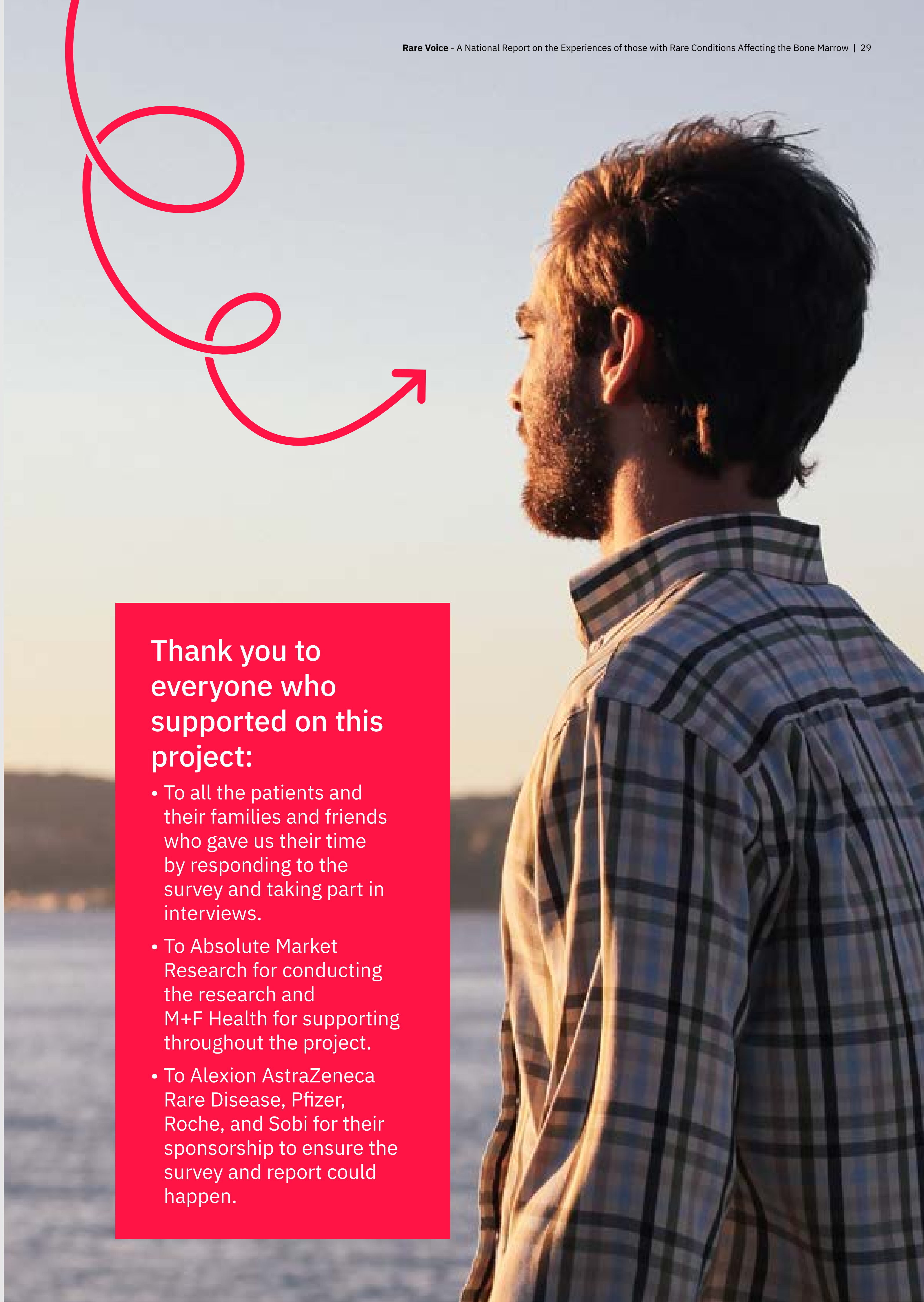
Rare diseases can have an isolating impact on patients and their families, as demonstrated in the mental health findings of this report. Our charities have first-hand experience of the importance of opportunities for people with the same or similar conditions to meet and share their experiences. For several of our conditions, our own large Family Days provide the only opportunity for patients with the same condition to connect in person. Based on survey feedback from three Family Days run by CAN, Fanconi Hope, and PNH Support respectively, 84% would recommend the Family Day to a friend or colleague in a similar situation, based on a scoring system where above 80 is 'world-class'.[17] In this same survey, following the event 85% reported feeling better informed, 95% feel more connected to others who may be experiencing similar things, 92% feel more connected to sources of support, and 91% feel less isolated. These events also provide clinical experts the opportunity to deliver the information about treatments and research that are so appreciated by our communities. An important focus for many of our charities is to ensure the sustainability of these events, which are vitally important to the wellbeing of our community, but which are reliant on a huge voluntary effort from a small number of people who are concerned about how they might be replaced to sustain these events in the future.

Other peer-to-peer support opportunities offered by our organisations include moderated groups to successfully connect people online, walking events, one-to-one buddy systems, and patient meetings and coffee mornings. We are exploring ways to expand and iterate this support to reach as many people affected as possible. Our charities are well-placed to provide this form of peer-to-peer support but are limited by resource and funding.

5

Using our voices to call for improved disease awareness for clinicians, teachers and the public

Each of our small communities faces many different challenges, but the findings of this project highlight common issues that unite us, and give us more reason to stand together. Our hope is that by collaborating on this and future projects, our organisations can amplify these common issues and themes, and provide evidence to drive decision making individuals and organisations such as the NHS to create lasting change. We will be highlighting these findings over the next year, to lobby the government and the NHS to effect change. Six of the charities are also working on providing new resources to help explain the condition to others, and will be using the findings of this project to inform this focus.



Thank you to everyone who supported on this project:

- To all the patients and their families and friends who gave us their time by responding to the survey and taking part in interviews.
- To Absolute Market Research for conducting the research and M+F Health for supporting throughout the project.
- To Alexion AstraZeneca Rare Disease, Pfizer, Roche, and Sobi for their sponsorship to ensure the survey and report could happen.

OUR RECOMMENDATIONS

This report has provided a much-needed platform so that the voices of those impacted by rare conditions affecting the bone marrow can be heard. We set out to better understand the impact of living with, or supporting someone living with, one of these conditions and the findings will have a legacy – shaping future campaigns and strategies of our individual charities, as well as our collaborative efforts.

Our charities know the many areas where our communities face common challenges and impacts, and this project has reinforced and evidenced this further. We believe that sustained support from government and the NHS is crucial here, without which significant improvement to life, care and support for those living with a rare disease will be challenging. **We will be lobbying government and health providers for improved access – crucially, equitable access – to multi-disciplinary care in specialist centres.**

There are clear, tangible opportunities across mental health provision for significant change to be implemented; findings have revealed specific areas of concern when it comes to mental health and wellbeing, and we are now much more aware of how people struggle and where support could really make a difference. We also have a clearer picture of the precise types of support needed, with people calling for wellbeing advice and psychological support in particular. The findings further highlight the scale of anxiety felt by parents as a group, and the pressing need to tackle the loneliness and isolation felt by all. Whilst we are ensuring that our charities use our resources to support mental well-being of our communities, **we need resource and expert support to ensure patients and their families are able to access psychological support when they need it.** Importantly, we also need psychological support to be an integrated element part of all care plans so people living with these conditions can access the support they need.

The extent to which people feel they have to be the experts and drivers of their own care, and the challenges found in navigating the healthcare system is also clear and there are opportunities for practical solutions and support with these. Implementing joined-up, integrated care is about improving people's lives and delivering better outcomes which should be an area of continuous focus. The UK Rare Disease Framework includes better coordination of care and improved access to specialist care, treatments and drugs as two of their key priorities. **As an Alliance, we are calling for these rare conditions affecting the bone marrow to have designated specialised commissioning services in the way that is the case for PNH.** The PNH HSS is an example of how this can effectively work in practice, and we believe this paves the way for future services, providing a strong blueprint for commissioners to work from.

Lastly, findings highlight the specific types of lifestyle and practical support that could make a difference to people's lives. Notably, there was almost complete consensus in people calling for **access to up-to-date research about these conditions, new treatments, and treatment methods**. This is the type of insight that wouldn't have been possible without this work being undertaken, and we hope to be driven forwards.

As a collective, we have the power to amplify these voices and bring about positive changes to the lives of those within our communities. The survey and this report are the first, important steps. We've listened, and now we need to act.

ENDORSEMENTS

This timely report highlights the complexity and needs of supporting the well-being of those affected by different subtypes of bone marrow failure. It shows clearly the gaps that exist in living with, or supporting someone living with, one of these subtypes of bone marrow failure. This report will hopefully act as a springboard to shape and improve future services within the NHS for bone marrow failure patients and their families.

Professor Inderjeet Dokal FMedSci
Chair of Child Health and Honorary Consultant in Haematology
Queen Mary University of London and Barts Health London, England, UK

Many patients with rare bone marrow conditions will go on to receive a stem cell transplant, which is an intense treatment. The survey results show that unfortunately many patients and families still don't get the psychological support they need to help them navigate treatment and the long-term impact of their disease. It's brilliant that charities are coming together to help address this gap, but to make equitable progress we need psychological and mental health support to be seen as a fundamental aspect of all rare bone marrow and stem cell transplant services.

Yasmin Sheikh
Head of Policy & Public Affairs
Anthony Nolan

APPENDIX

The National Community Survey was conducted by Absolute Market Research.

The online survey was open from the 15th of November to the 18th of December 2022, and the one-to-one interviews took place in January 2023. In total, 434 responses were received for the online survey and Seven people were interviewed for the qualitative interviews – each representing a different condition.

For further information or support from any of the charities, you can visit their individual websites:

- Congenital Anaemia Network (CAN)
- DBA UK
- DC Action
- Fanconi Hope
- PNH Support
- SDS UK
- The Aplastic Anaemia

The Together for Healthy Bone Marrow Alliance

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