Understanding the care experiences of people with Marfan Syndrome and Loeys-Dietz Syndrome: A UK Survey

January 2026

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Picker

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Background

Marfan syndrome (MFS) and Loeys-Dietz syndrome (LDS) are rare, inherited connective tissue disorders that can have serious health implications, particularly affecting the cardiovascular system, eyes, and skeletal structure.

There is limited large-scale, person centred data on the experiences of people with MFS and LDS in the UK. Recent policy priorities around personalised and integrated care present an opportunity to address these gaps. To build this evidence base, the Marfan Trust partnered with Picker to deliver a nationwide survey capturing the experiences of individuals with MFS and LDS and their carers. The survey aimed to identify unmet needs, inform policy, and improve care delivery.

Approach

A survey was developed through a scoping review of recent literature, interviews with patients and carers, and cognitive testing to ensure clarity and relevance. The final questionnaire was delivered online and ran for 10 weeks (September-November 2025). It was promoted through the Marfan Trust's networks and social media.

To reach members who were not online or preferred paper correspondence, the Marfan Trust mailed printed questionnaires to a random sample of 200 members.

Data were analysed using descriptive statistics and subgroup comparisons, with thematic coding of free-text responses to complement quantitative findings.

Key findings

395 people completed the survey

84% were diagnosed with Marfan syndrome

64% were female

92% were White

76% completed the survey as a person living with MFS or LDS

24% were carers

Diagnosis and Genetic Testing

Some respondents experienced delays and dismissive attitudes from healthcare professionals before receiving a diagnosis.

- **26%** said that before their diagnosis their health concerns were not taken seriously by healthcare professionals
- **45%** reported being diagnosed within 12 months, but **10%** said it took more than five years
- **86%** said they had received a genetic diagnosis, but only **66%** reported that family members had been offered testing.

Access to Care

Cardiologists were generally accessible, but significant gaps exist in access to other healthcare specialists.

- **81%** were able to access a cardiologist when needed, either *always* or *sometimes*.
- **40%** reported an unmet need for **pain management** services
- **34%** wanted **osteopathy** but were unable to access this
- **25%** indicated an unmet need for **podiatry**
- **69%** of those seeking **mental health support**, did not receive enough support.

Treatment and Monitoring

Most respondents reported taking at least one type of heart medication.

- **59%** received an echocardiogram at least once a year, but a notable proportion were not monitored as frequently as recommended by clinical guidelines.

Care Coordination

The survey revealed poor experiences of care coordination.

- **56%** did not have a named healthcare professional overseeing their care but wanted this support.
- **67%** reported having to *sometimes* or *often* arrange their own care
- **53%** felt different healthcare professionals and services do not work well together.

Care from Healthcare Professionals

- Only **20%** felt that healthcare professionals definitely took their needs and wishes into account
- Only **23%** always had confidence and trust in the healthcare professionals involved in their care.
- **75%** reported not receiving enough, or any, information from healthcare professionals about their condition and ongoing treatment.
- **55%** said they did not receive enough, or any, personalised advice on physical activity.

Quality of Life

- Fatigue, chronic pain, gastrointestinal issues, headaches/migraines, and joint hypermobility were widely reported.
- At least two thirds of respondents said MFS or LDS negatively impacted on exercise, mobility, confidence, mental health, and travel. Nearly half reported employment was affected, with those experiencing more symptoms significantly more likely to be on long-term sick leave or retired due to ill health.
- Awareness of some benefits was low, and the comments showed some respondents faced difficulties accessing financial support.

Recommendations

The survey findings have informed a set of recommendations presented in this report. Key recommendations are outlined below; please refer to the summary section for full details.

Improve Diagnosis and Awareness

Provide targeted training for GPs and non-specialist clinicians to recognise Marfan syndrome (MFS) and Loeys-Dietz syndrome (LDS). Deliver awareness campaigns for healthcare professionals and the public, sharing best practice guidance and patient stories.

Strengthen Care Coordination

Introduce named care coordinators or specialist nurses to oversee multi-disciplinary care and act as a single point of contact. Encourage multi-disciplinary team (MDT) meetings for complex cases.

Expand Access to Specialist Services

Address gaps in pain management, physiotherapy, mental health support, and gastrointestinal care. Offer tailored psychological support for patients and families, recognising the emotional impact of living with a rare condition.

Improve Information and Support

Provide comprehensive, accessible resources for patients and carers, including guidance on exercise, symptom management, and coping strategies. Strengthen signposting to reliable sources of information and practical support.

Address Quality of Life Needs

Offer proactive pain and fatigue management strategies. Provide personalised advice on safe physical activity to reduce fear and inactivity.

Background

The Marfan Trust is a UK-based charity dedicated to supporting individuals affected by Marfan syndrome (MFS) and Loeys-Dietz syndrome (LDS). Their work encompasses providing advice and guidance, funding research, and raising awareness of these rare conditions. Recognising the need for robust evidence to inform care and policy, the Marfan Trust partnered with Picker to design and deliver a national survey aimed at capturing the experiences of people living with MFS and LDS.

Marfan syndrome and Loeys-Dietz syndrome are rare, inherited disorders that affect the body's connective tissue, which plays a critical role in supporting organs and structures. MFS is estimated to occur in approximately 1 in 5,000 to 10,000 individuals in the UK, while LDS is even rarer, with a prevalence of around 1 in 50,000 to 1 in 200,00.(1) Both conditions can have serious health implications, particularly involving the cardiovascular system, eyes, and skeletal structure. Without timely diagnosis and appropriate management, complications such as aortic aneurysms can be life-threatening. LDS shares several clinical features with MFS but often presents additional challenges, including more aggressive vascular involvement and distinctive craniofacial characteristics.

Despite advances in genetic testing and clinical care, people with these conditions often face delays in diagnosis, fragmented care pathways, and limited access to specialist services. Understanding these experiences is essential for improving outcomes and quality of life. The survey was therefore designed to gather data on current care provision, patient experiences, and unmet needs. Insights from this research will inform policy development, guide future research priorities, and support improvements in clinical practice and service delivery across the UK.

Why this research is timely and important

There is currently a lack of large-scale, person centred data on the lived experiences of people with MFS and LDS in the UK. Existing evidence is often drawn from small clinical cohorts or international studies, which may not reflect the realities of UK healthcare systems. At the same time, policy initiatives emphasising personalised care and integrated services create an opportunity to address gaps in provision for rare disease communities. By capturing the voices of those directly affected, this research will provide insight to shape national strategies, improve care pathways, and ensure that future developments in treatment and support are aligned with patient needs.

Methods

Survey development

The following elements contributed to the development of the survey:

- A brief scoping review to identify existing evidence and inform topic areas
- A qualitative phase involving semi-structured, in-depth interviews to capture detailed personal experiences
- Questionnaire testing to ensure clarity, relevance and ease of completion.

Brief scoping review

We conducted a brief review of recent international literature (published since 2019 through Google Scholar) on the care experiences of people affected by Marfan syndrome or Loeys-Dietz syndrome. This informed both the development of the qualitative topic guide and subsequent questionnaire content. Twenty relevant peer-reviewed articles were identified. The main themes across these publications were:

- **Quality of Life (QoL) and Psychosocial Impact**

  Patients with MFS and related conditions (Loeys-Dietz syndrome and vascular Ehlers–Danlos syndrome) experience lower quality of life (QoL) compared to the general population.(2-6) Mental health challenges (anxiety, depression), fatigue, and pain are major contributors to reduced QoL.(2-4,7-12) Psychosocial consequences include loss of self-esteem, health anxiety, stigma, and social isolation.(2-4,13,14)

  There may be impacts on reproductive decision-making, work participation, and satisfaction with life. Many patients leave work early due to health burden; some adolescents and children struggle with school and peer participation.

- **Pain and fatigue**

  Chronic pain is highly prevalent and often undertreated. Pain affects daily activities, work, and leisure; common sites include head, neck, back, knees. Fatigue is extremely common and strongly linked to lower life satisfaction and QoL. Pain and fatigue often reinforce each other, along with sleep problems, creating a cycle of disease burden.(7-10,12,15)

- **Healthcare Experiences and Needs**

  Diagnostic delay can be an issue and/or variations in time since a person's last cardiovascular follow-up or eye examination.(16) Patients value multidisciplinary care, good information, and coordinated appointments.(6,16) Dissatisfaction arises from lack of attention to non-cardiovascular issues (pain, fatigue, mental health).(16) The importance of emotional support and counselling was highlighted in some studies.(4,5,13,14,16,17) There can be inconsistent or contradictory information or advice from health professionals on physical exercise. Patients want individualised, practical guidance on physical activity, coping strategies, and lifestyle.(18,19)

In-depth interviews

Five semi-structured interviews were conducted with individuals living with or caring for someone with Marfan or Loeys-Dietz syndrome. The purpose of the interviews was to explore current care needs, preferences, and experiences, and to identify what matters most to people when receiving care. Participants were recruited with support from the Marfan Trust, which promoted the research opportunity through its social media channels. The sample included a mix of patients and caregivers, selected to reflect variation in demographic characteristics (such as age and sex), condition type (MFS or LDS), time since diagnosis, and geographical location.

The interviews were conducted online and each lasted up to one hour in length. Participants were offered a £30 Amazon or Love2Shop gift voucher to thank them for their time. The interviews were video recorded with the participant's consent.

The interviews were analysed to understand people's experiences of diagnosis, ongoing care and treatment and any impacts of living with MFS or LDS. The key findings across the five interviews were:

- **Diagnosis challenges**: delayed or difficult diagnosis due to lack of GP awareness and dismissive attitudes. Patients often had to self-advocate and research to access genetic testing or specialist care.
- **Knowledge gaps among healthcare professionals**: GPs and some specialists lacked understanding of MFS/LDS, focusing only on isolated symptoms (e.g., heart or eye issues). Patients frequently had to educate their doctors or chase referrals. Some noted contradictory advice on medication and treatment plans due to poor knowledge.
- **Disjointed or poorly coordinated care**: Care can be disjointed, with multiple hospitals and specialists working in isolation with a lack of a central contact point or named specialist for holistic oversight. Notes are not always shared between hospitals, leading to repeated explanations and stress. However, liaison teams and/or proactive geneticists improved coordination in some cases.
- **Emotional and psychological impact**: Shock and fear at diagnosis; feelings of guilt about passing condition to children. Anxiety about deterioration or death, especially after family losses. Limited psychological support - patients often fund or source counselling themselves.
- **Impact on quality of life**: Physical limitations (pain, surgeries, mobility issues) affect daily living, work, and social life. Some had to give up sports or work, leading to financial strain and isolation. Concerns were raised about family planning and reproduction (lack of clear guidance on IVF/genetic testing).
- **Positive experiences**: When care was good, it involved clear communication in lay terms, individualised support (e.g., breastfeeding help post-surgery), quick responses and continuity from liaison teams.

Insights from the literature review and interviews informed the development of the questionnaire content.

Testing the questionnaire

Five cognitive testing interviews were conducted with people living with or caring for someone with MFS or LDS. The aim was to provide insight into how questions were interpreted and to show how the questions performed. Pre-prepared and spontaneous probes were used by the researchers to test each survey item with participants.

The participants were recruited at the same time as the in-depth interviews and again, included a mix of patients and caregivers in terms of demographic characteristics, condition type and time since diagnosis. As with the in-depth interviews, participants were offered a £30 Amazon or Love2Shop gift voucher to thank them for their time.

The testing highlighted where some questions and/or response options were difficult to answer by participants and/or did not adequately capture their experience. These were improved before the questionnaire was finalised to increase the overall quality of the survey data.

Finalising survey content

Findings from the scoping review, in-depth and cognitive testing interviews were used to inform and finalise the survey questions. The survey content was further shaped by Picker's expertise in survey design, the Marfan Trust's insight into the needs of individuals living with MFS or LDS (and their carers), and guided by Picker Principles of Person Centred Care[1]

When developing the survey content, we were mindful that people with MFS or LDS are affected differently, and that features can vary even within the same family who have the condition. The survey was designed so that the wording of questions made sense regardless of how people are affected by the conditions and people were routed past any questions which were not applicable to them.

The final questions were scripted into Qualtrics - an online survey platform. Using Picker's online survey development checklist, two researchers carried out checks to ensure correct scripting, routing logic, and formatting. The survey used Picker and the Marfan Trust branding and included both organisations' logos.

Screening questions at the start of the survey ensured that only eligible participants proceeded. Respondents were eligible to complete the survey if:

- They had a diagnosis of Marfan syndrome or Loeys-Dietz syndrome, or be a parent/carer of someone with these conditions
- Aged 16 or over
- They live in the UK

To maximise accessibility and relevance, routing logic ensured respondents only saw applicable questions. All items beyond the screening questions were optional allowing participants to skip questions if they wished. Online respondents were also asked for consent to use their responses if they exited the questionnaire early.

[1] The Picker Principles of Person Centred care - Picker

Data collection

Online survey

An online survey, hosted via the Qualtrics platform, was the main data collection approach used. The online survey remained open from 6th September to 19th November 2025. The survey was publicised on social media by both Picker and the Marfan Trust. To try to maximise responses, the Marfan Trust used their existing mailing list to share the survey hyperlink via email with 200 members on 14th September. A blanket reminder was emailed on 8th October and 11th November. The survey was also promoted via the Marfan Trust website and the November newsletter. The survey hyperlink was shared with relevant existing networks and stakeholders. Participants could contact the Marfan Trust via phone or email during fieldwork with any queries about their care or the survey. As the survey was distributed via an open hyperlink, participant anonymity was maintained. Where respondents provided an email address to join the Marfan Trust mailing list, this information was not linked to their survey responses.

Paper survey

To reach members who were not online or preferred paper correspondence, the Marfan Trust produced a print-friendly questionnaire and mailed it to a random sample of 200 members with postal addresses on 7th October 2025 with a covering letter and return envelope. Completed paper surveys were manually entered into the live online system by the Marfan Trust.

Analysis

At close of fieldwork, online data were downloaded and cleaned/labelled. Cleaning included removing any participants who had not agreed for their data to be used if they had not fully completed the survey. Some new (derived) variables were created to allow for certain analyses to be undertaken.

Frequency tables for each survey question were generated, showing counts and percentages of responses to each response option. Crosstabulations were produced to compare different subgroups of respondents, for example by demographic characteristics. Suppression rules were applied to frequency tables and crosstabulations, with counts below 5 (overall or by subgroup) suppressed and replaced with an asterisk to protect respondent anonymity and ensure data robustness.

A researcher reviewed all freetext comments and familiarised themselves with the data before coding responses into themes. Illustrative quotes are included throughout the report, and a summary of the main themes from the thematic analysis is presented in Appendix 3.

About the respondents

This section provides an overview of the survey respondents. Due to the approach used to distribute the survey, the number of individuals who received the invitation is unknown, so a response rate could not be calculated.

A total of **395** eligible respondents completed the survey. Most respondents (84%, n=331) were diagnosed with Marfan syndrome. The majority identified as White (92%, n=322) and female (64%, n=224). Respondents' age ranged from 16 to 70 years; nearly half (44%, n=173) were aged between 51 and 70 years old, while only 13% (n=52) were between 16 and 35 years (Figures 1-4).

Figure 1: Syndrome of respondent

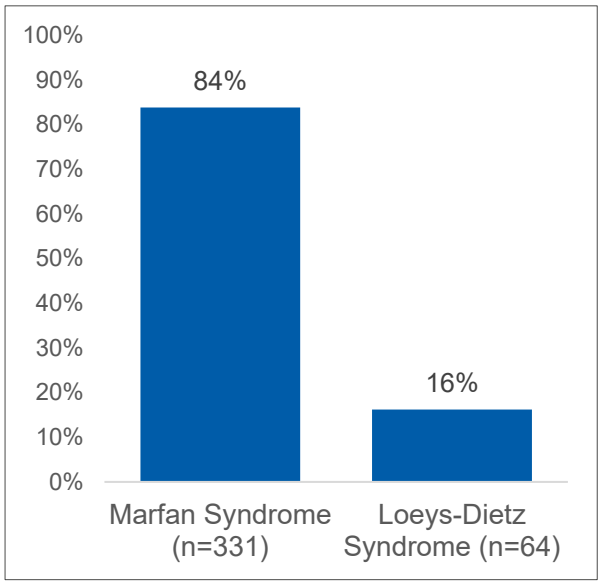


Figure 2: Ethnicity of respondent

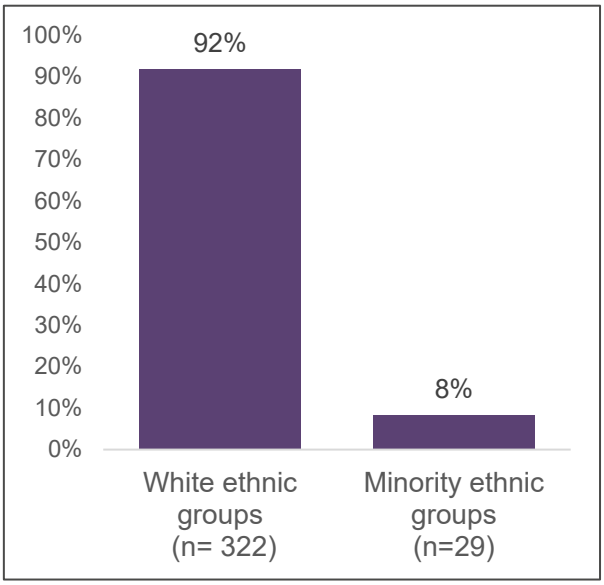


Figure 3: Gender of respondent

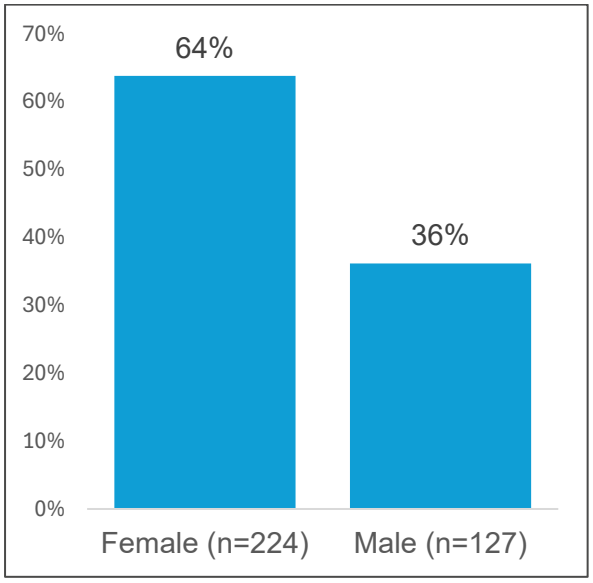
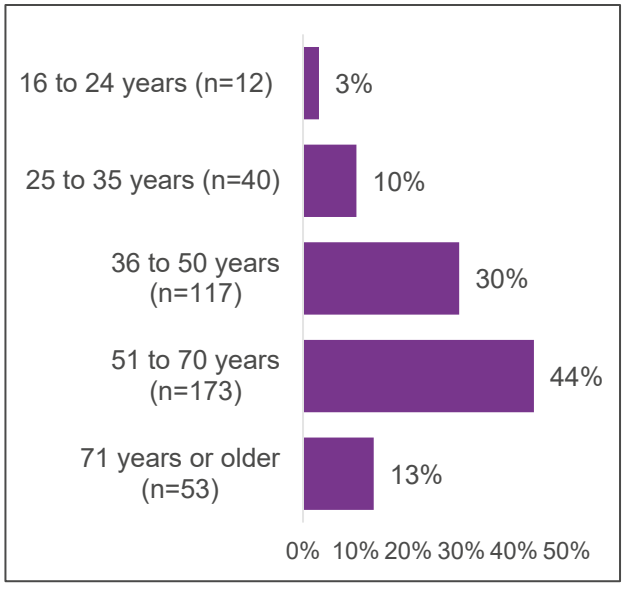


Figure 4: Age of respondent



Most respondents (76%, n=299) answered the survey as a person diagnosed with MFS or LDS. A smaller proportion responded as a parent or carer of a child (18%, n=70) or as a carer of an adult (6%, n=25) with the conditions (Figure 5). Just over one-third (37%, n=144) of respondents were from the south of England (Figure 6).

Figure 5: Respondent type

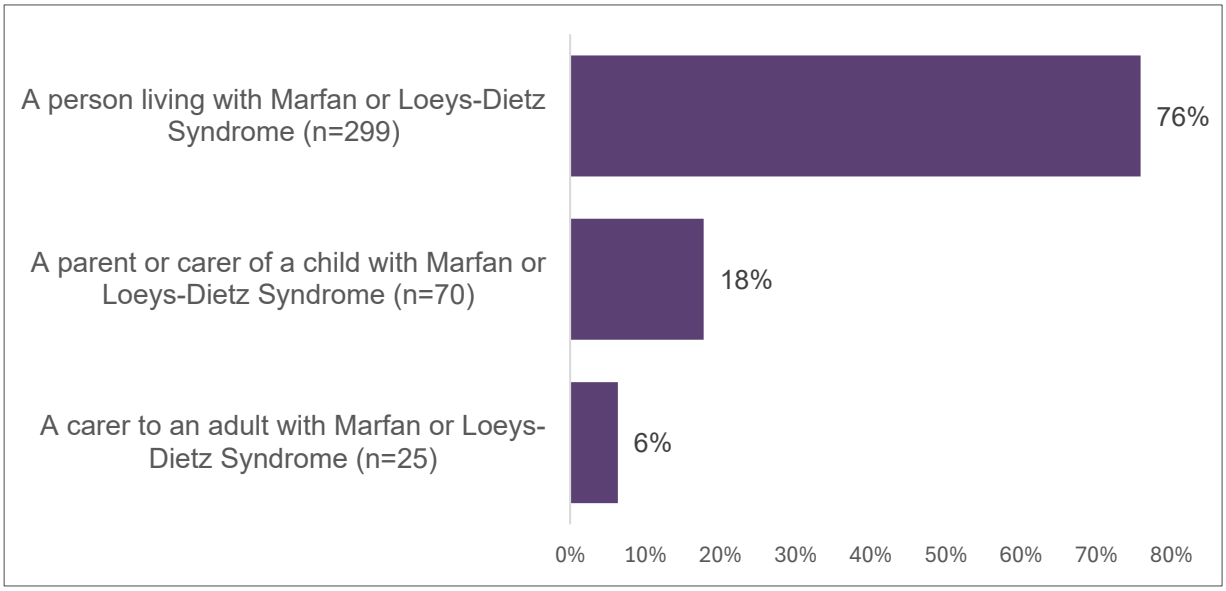
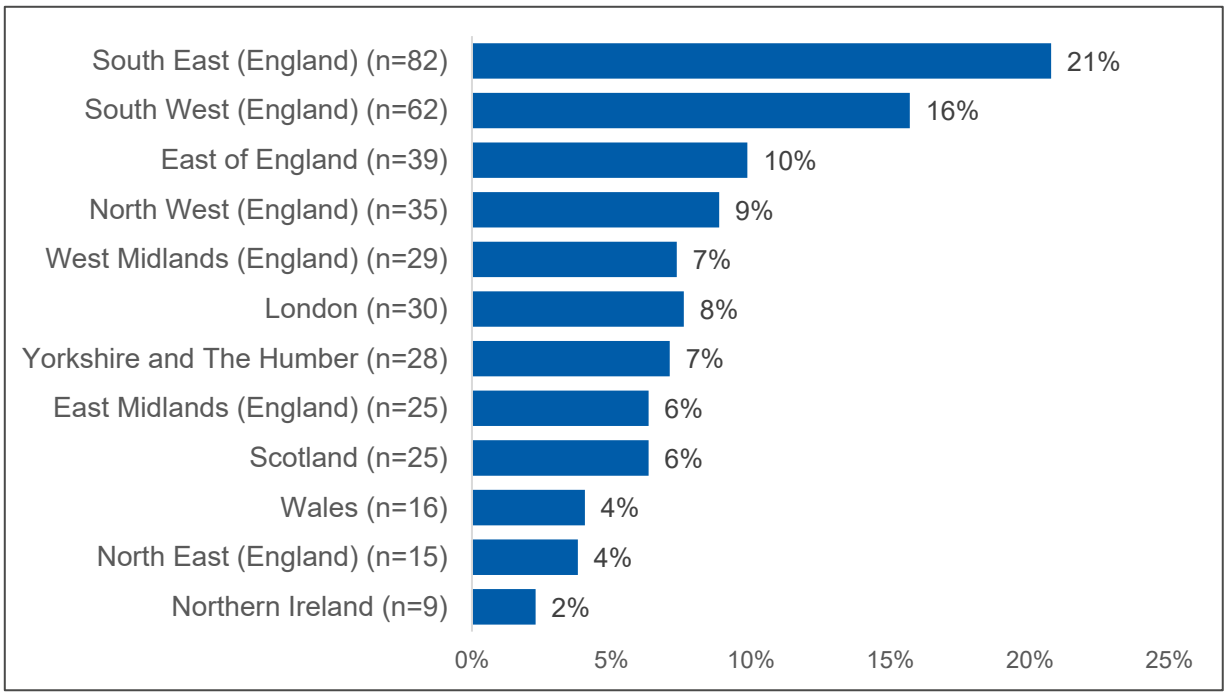


Figure 6: Geographical location of respondent



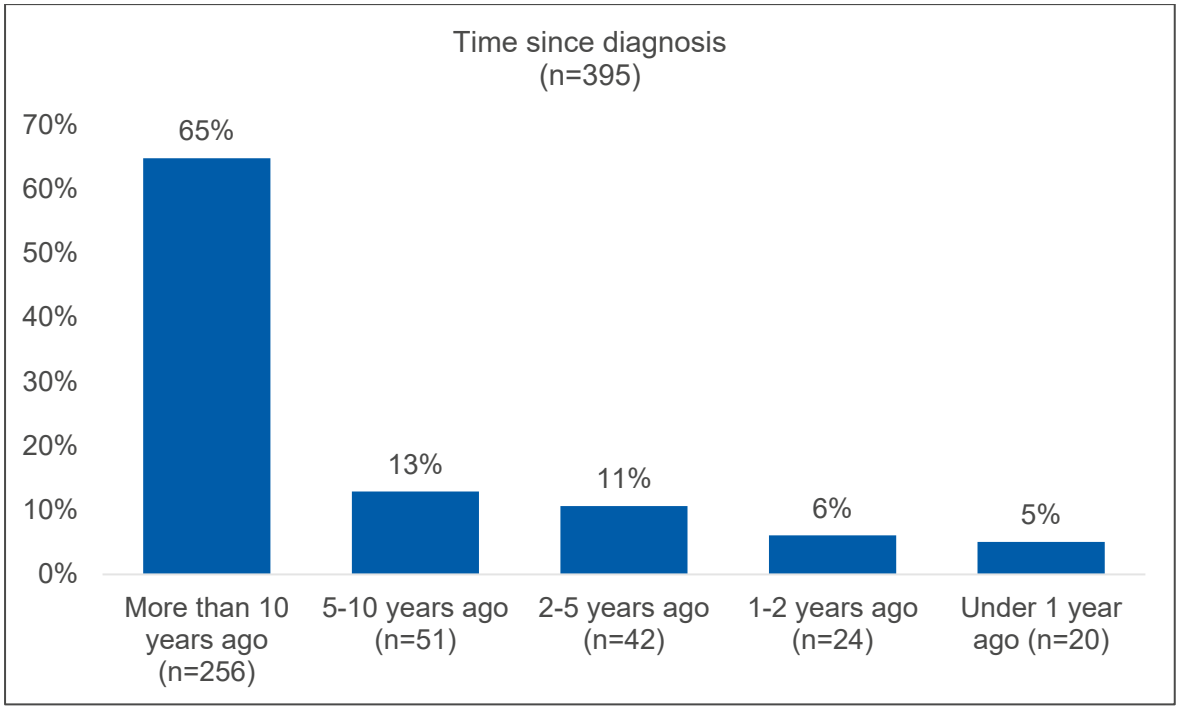
Findings

Experiences of diagnosis

Time since diagnosis

The survey examined the length of time since respondents were diagnosed (clinical or genetic) with MFS or LDS. Overall, 65% (n=256) reported being diagnosed more than ten years ago, 13% (n=51) were diagnosed between 5-10 years ago, and 5% (n=20) had been diagnosed within the past year (Figure 7). This varied significantly by condition: nearly three quarters of those with MFS (73%, n=240) had been diagnosed over a decade ago, compared with only 25% (n=16) of those with LDS (Table 1, Appendix 1). This disparity likely reflects the more recent recognition and classification of Loeys-Dietz syndrome relative to Marfan syndrome.

Figure 7: Length of time since respondents were diagnosed



Respondents were asked about the initial reason they sought medical attention prior to receiving a diagnosis of Marfan or Loeys-Dietz syndrome. Almost one third of respondents (31%, n=122) sought medical attention because of a family history of these conditions. This was even more common among those with LDS, where half (50%, n=32) cited family history, compared with 28% (n=90) of those diagnosed with MFS (Table 2, Appendix 1).

However, more than one third overall (35%, n=138) had not sought medical attention prior to their diagnosis:

- 19% (n=76) reported their diagnosis followed medical attention for another condition
- 6% (n=25) said care was sought on their behalf (such as a medical emergency), and

- 9% (n=37) reported their diagnosis occurred after a medical emergency in a family member.

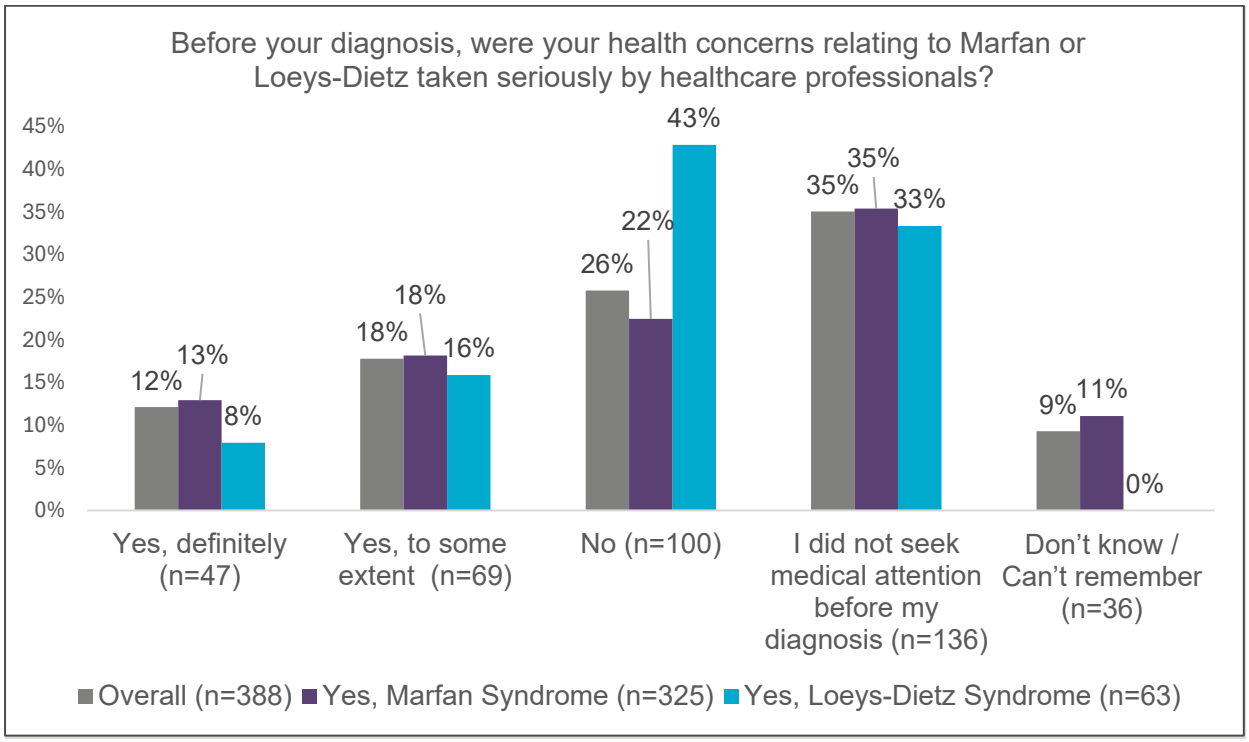
The pattern varied by condition; while 21% (n=70) of MFS respondents were diagnosed following a check-up or care for another health problem, only 9% (n=6) of LDS respondents reported the same.

Time to diagnosis

Timely diagnosis is challenging, as Marfan and Loeys-Dietz symptoms are often overlooked due to limited awareness of these conditions among non-specialist clinicians. The syndromes can also be difficult to diagnose because the signs and symptoms can vary from person to person.

Around one quarter of respondents (26%, n=100) said that before their diagnosis their health concerns were not taken seriously by healthcare professionals. This was especially true for respondents with LDS, 43% (n=27) of whom reported that their health concerns were not taken seriously prior to diagnosis (Figure 8).

Figure 8: Respondent experience of raising health concerns before diagnosis



As perhaps expected, fewer respondents reported that their health concerns were dismissed when the time to diagnosis was shorter. A quarter of those diagnosed within 12 months experienced this (25%, n=42), compared with 45% (n=15) of respondents whose diagnosis took 1-2 years and 44% (n=28) whose diagnosis took more than 2 years (Table 3, Appendix 1)

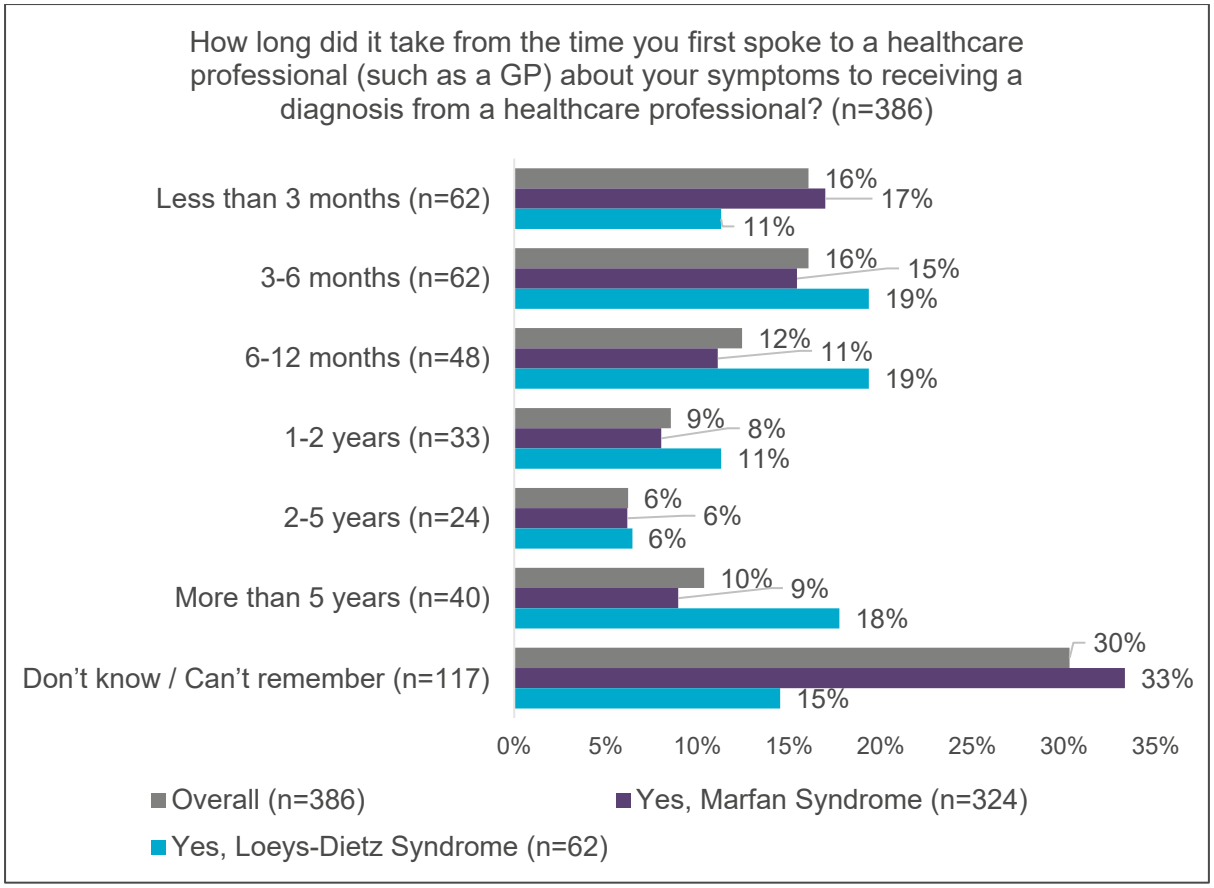
Some respondents shared comments at the end of the survey describing the challenges they faced during their diagnostic journey (See Appendix 3). Examples include:

''I've been trying to get a diagnosis for 10 years nearly as I know I have dural ectasia but due to GP's and physio therapists not knowing what it is. No one has ever listened to me. I might be getting somewhere now but it's most likely leading to a dead end.''

"The lack of knowledge surrounding this syndrome [LDS] within the health care system has caused lasting effects on my body which if diagnosed up sooner I'm sure would be less severe due to medication available and lifestyle changes a lot earlier."

Some caution is required when interpreting the findings relating to the reported length of time to diagnosis (Figure 9). Almost one third of respondents (30%, n=117) were unsure or could not recall the time from first raising their symptoms with a healthcare professional to receiving a formal diagnosis. This uncertainty may reflect the length of time since diagnosis and/or the varied nature of symptoms. For example, some respondents may not have considered instances when they sought medical advice for symptoms that they did not recognise as being related to MFS or LDS at the time.

Figure 9: Length of time to diagnosis



Most respondents (44%, n=172) reported being diagnosed within 12 months, while 10% (n=40) said the process took more than five years. The proportion who reported being diagnosed within three months (16%, n=62) may be surprisingly higher than expected and may indicate differing interpretations of the question. For example, some respondents diagnosed following a medical emergency (or following hospitalisation for a separate condition) may have reported the time from that event to diagnosis, whereas others may have considered the period during which they sought medical attention prior to the

emergency. As noted previously, it is also worth bearing in mind that more than one third of respondents (35%, n=136) said they did not seek medical attention before their diagnosis (see Figure 8).

Among respondents with Loeys-Dietz syndrome, nearly one in five (18%, n=11) waited more than five years for a diagnosis suggesting that delays may be longer for this condition, possibly due to lower awareness amongst healthcare professionals (Figure 9). However, it's possible that respondents with MFS were diagnosed more quickly as the survey indicates they were more often diagnosed following a check-up or medical attention for another condition than those with LDS.

Among respondents diagnosed within the last 12 months, a quarter (26%, n=5) reported receiving a diagnosis within three months. In contrast, a similar proportion (21%, n=4) said the process took more than five years. Although this subgroup is small, these findings suggest that lengthy delays in diagnosis can still occur, even for individuals who have only recently received their diagnosis (Table 4, Appendix 1).

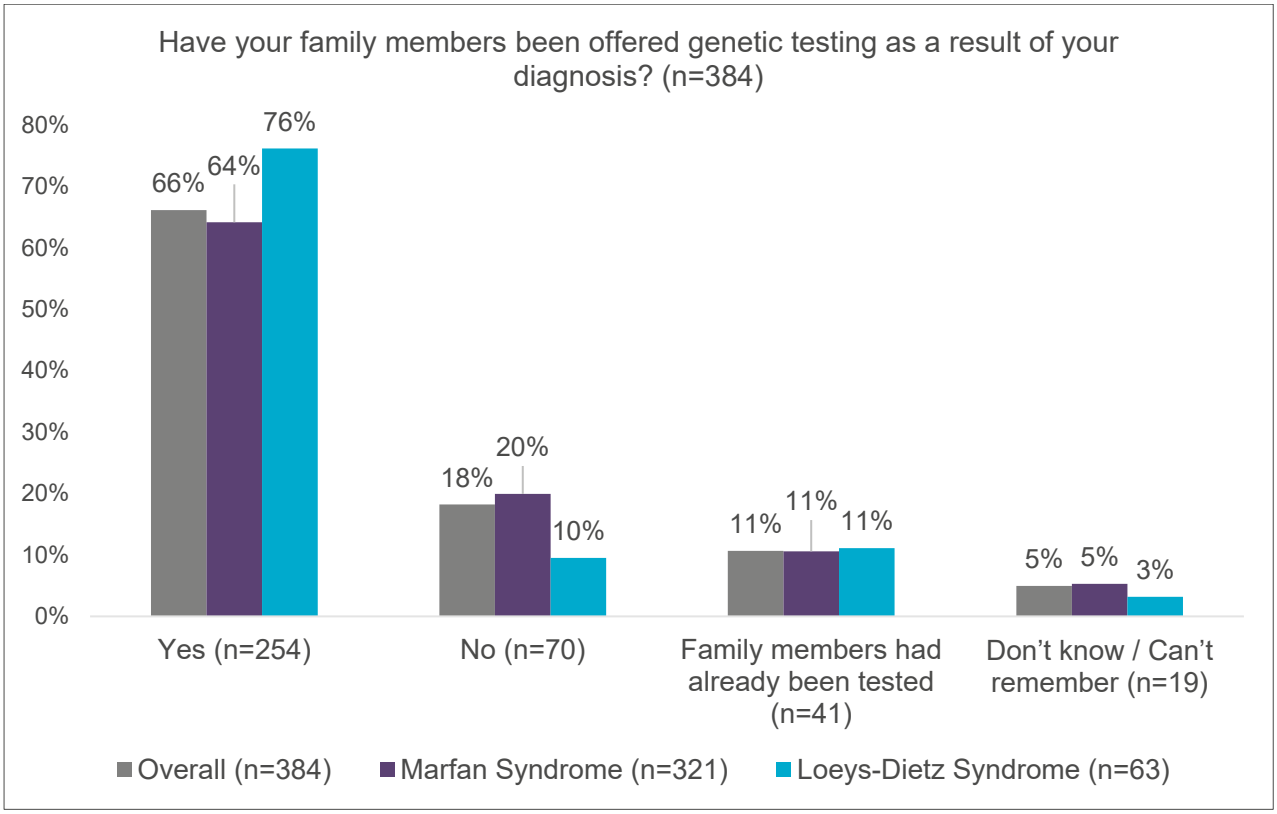
Genetic testing

Most respondents (86%, n=328) reported receiving a genetic diagnosis of Marfan or Loeys-Dietz syndrome. This figure was even higher among those with Loeys-Dietz syndrome, where almost all (95%, n=60) had a confirmed genetic diagnosis (Table 5, Appendix 1).

Genetic testing for family members was less common. Two-thirds of respondents (66%, n=254) said their relatives had been offered testing following their diagnosis (Figure 10). There were differences between conditions: 76% (n=48) of Loeys-Dietz respondents reported that family members had been offered testing, compared with 64% (n=206) of those with Marfan syndrome (Figure 10).

Notably, one in five people with Marfan syndrome (20%, n=64) said that their family members had not been offered genetic testing.

Figure 10: Proportion of respondents reporting genetic testing for family members



Examples of written comments regarding genetic testing include:

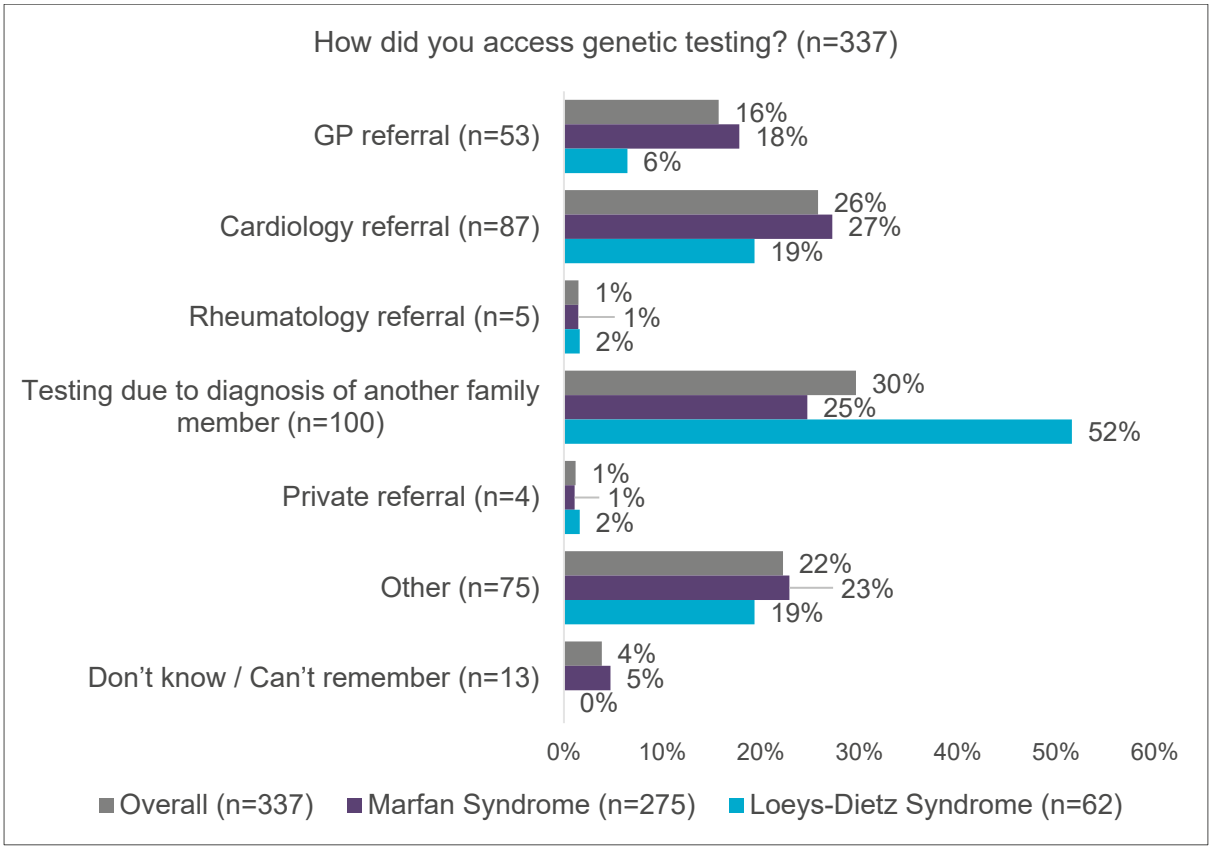
"I was diagnosed with Marfans syndrome at birth but my parents were never offered genetic testing. My father was then diagnosed over ten years later when he suffered an aortic aneurysm."

"I had an aortic dissection which wasn't diagnosed until after 7 months. My aorta was dissected from the root to my abdomen. I was given a genetic test after I was discharged from hospital, the test was positive for defect on the fribillin 1 gene. All my children were given tests."

"I was only formally diagnosed via genetic testing in 2014 when I turned up to the GP where I lived in the West Midlands saying 'I'm pregnant, I have suspected marfans syndrome, the internet says I should tell you asap'. Referrals flew everywhere and I was diagnosed within 3 months!"

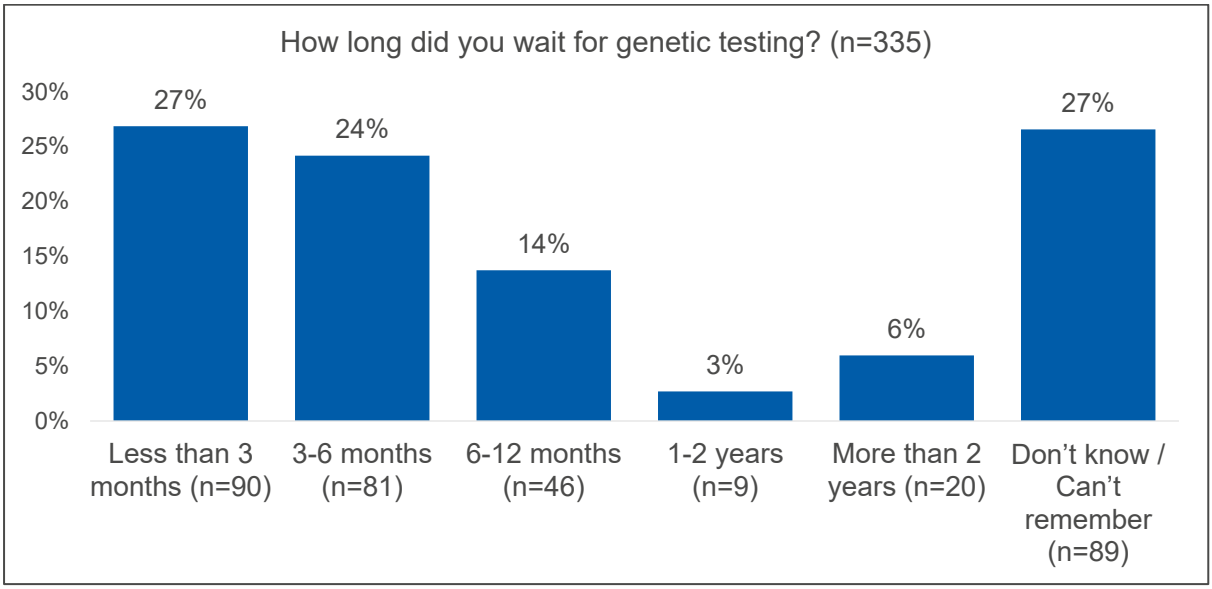
The survey explored how people accessed genetic testing, revealing differences between conditions. Half of those with Loeys-Dietz syndrome (52%, n=32) were tested following a family member's diagnosis, compared with 25% (n=68) of those with Marfan syndrome (Figure 11). For Marfan respondents, cardiology referrals were slightly more common (27%, n=75) than for Loeys-Dietz syndrome (19%, n=12). A sizeable proportion of respondents overall (22%, n=75) reported that they had accessed genetic testing another way. A review of respondent's freetext comments provided at this 'other' response included orthopaedic referrals, paediatric referrals and referrals via ophthalmology.

Figure 11: Routes by which respondents accessed genetic testing, by condition



About half of respondents (51%, n=171) received genetic testing within six months of referral, with over a quarter of these (27%, n=90) waiting less than three months. However, delays were experienced by some: 14% (n=46) waited over 6 months, and 9% (n=29) waited more than a year (Figure 12).

Figure 12: Length of time between referral and genetic testing



Among those respondents who were diagnosed within 6 months, most (82%, n=87) waited less than 6 months for genetic testing, with half of these (50%, n=53) waiting less than 3 months (Table 6, Appendix 1). This suggests that quicker diagnoses are associated with more timely access to genetic testing.

Access to care and support

Overall

Symptoms of Marfan and Loeys-Dietz syndrome can vary widely between individuals, leading to different healthcare needs.

The survey found that at least half of respondents did not want or need access to certain specialists in the past two years, including Ear Nose Throat (ENT) specialists, Gastrointestinal (GI) specialists, mental health professionals, neurologists and occupational therapists.

Regarding cardiologists, only 6% (n=23) said they did not want or need access to a cardiologist during this period. Around half of respondents (51%, n=190) reported they were *always* able to access a cardiologist when needed, with a further 30% saying this was the case *sometimes*. However, more than one in ten respondents (12%, n=46) said they were unable to access a cardiologist when they wanted or needed care. Over half of respondents with MFS (53%, n=165) said they could *always* access a cardiologist when needed, compared with just 40% (n=25) of respondents with LDS (Table 7, Appendix 1)

The survey revealed that the most common gaps in access to healthcare professionals (Figure 13) were for pain management services, reported by 40% of respondents (n=143), and osteopathy, required by 34% (n=122).

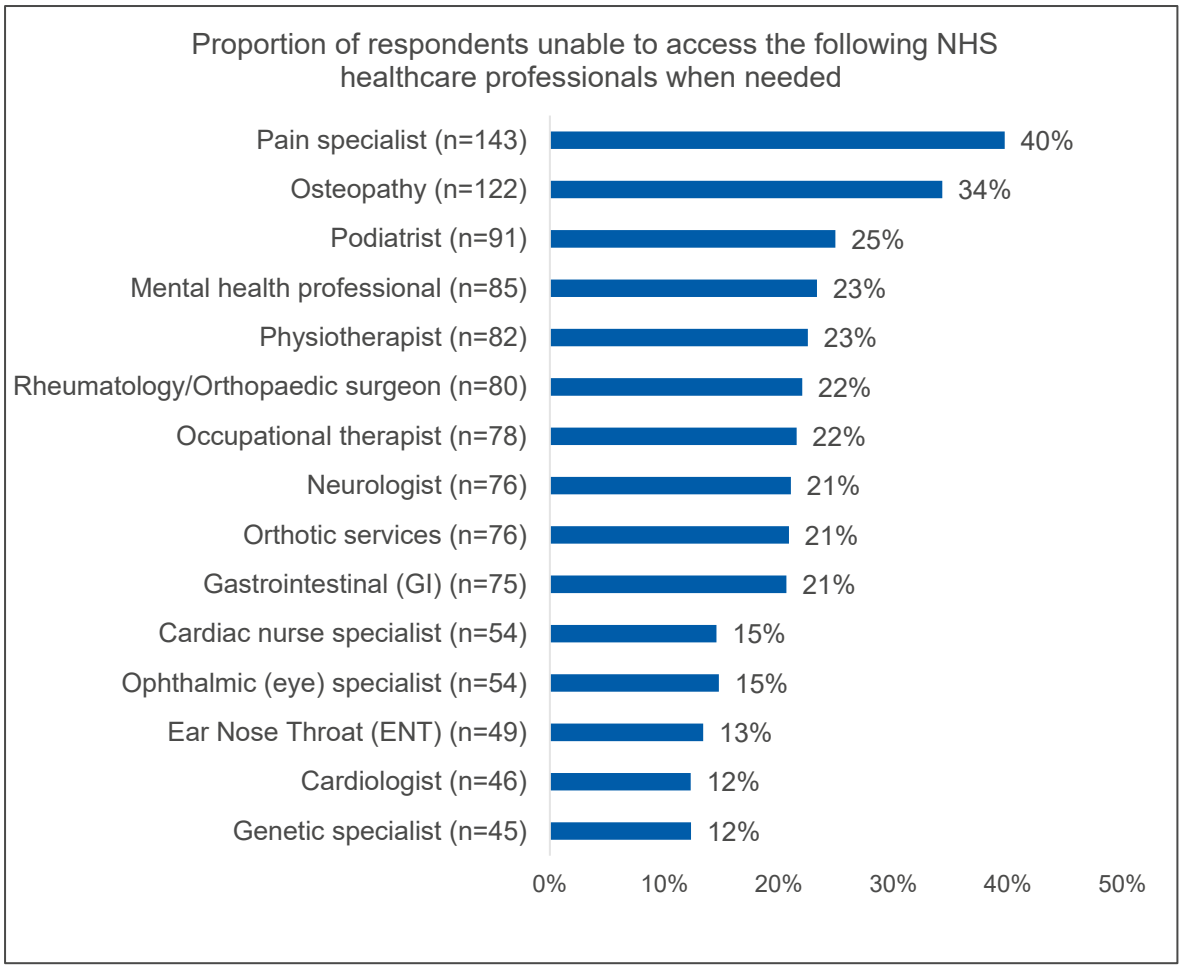
Several respondents shared written comments at the end of the survey highlighting the challenges they faced in accessing certain healthcare services (see Appendix 3 for details). Examples include:

“The cardiology care I have received in the West Midlands has been amazing and I feel very safe with them. I still find it very hard to access any other forms of care. It took me 4 years to see a specialist for pelvic organ prolapse as my referral kept getting lost … and many years of fighting to see a neurologist for my migraines. But I find once you do see a specialist, they are brilliant. It's just a long, hard fight to get there.”

“I felt frustrated that care for Marfan Syndrome focuses entirely on the heart. I have had to find my own support for other symptoms of the syndrome.”

“No regular gastrointestinal checks as an adult with [condition name] and a risk of Crohns.”

Figure 13: Proportion of respondents reporting unmet need for healthcare professionals



A minority of respondents reported paying privately to access healthcare services. However, among those with Marfan syndrome, some did so for specific treatments: 12% (n=37) accessed osteopathy, 11% (n=32) saw a podiatrist, and 8% (n=24) used private physiotherapy (Table 8, Appendix 1). This suggests that gaps in NHS provision for certain services may lead some people to seek private care, particularly for therapies that support mobility and pain management.

"My physical disabilities are getting so much worse now I'm in my mid 50's and I just don't know where to turn for help. Everything takes months/years to get appointments, by which time I've paid a fortune for private help."

"Local authority has long waiting lists currently on waiting list for pain clinic (2 years) and on waiting list for wheelchair referral. I have access specialists privately at much cost."

"The impact of perimenopause with adhd (also linked to Marfan) on my memory and attention has been debilitating and in order to support this I am going to have to spend money accessing private healthcare to get testosterone prescribed."

Just over half of respondents (52%, n=194) reported needing to travel to more than one hospital or clinic to receive the care they required, but said this did not bother them. In

contrast, one quarter (25%, n=92) found travelling to multiple hospitals or clinics burdensome (Table 9, Appendix 1).

As expected, respondents with more symptoms affecting daily life were more likely to travel to multiple hospitals or clinics for care. Among those experiencing more than ten symptoms, almost all (98%, n=37) reported needing to attend more than one hospital or clinic, compared with 57% (n=11) of respondents whose symptoms did not affect day-to-day life (Table 10, Appendix 1). Furthermore, just over half of those with more than ten symptoms (53%, n=20) found this travel burdensome, whereas only 20% (n=6) of respondents without symptoms impacting daily life reported the same. The findings suggest that while many patients are willing to travel for specialist care, a significant minority experience this as a burden - particularly those with more symptoms affecting daily life - highlighting the need for more integrated care.

"My specialist center in [City] is amazing - but they only have cardiologist, genetics and rheumatologist there (Dr [name]) it's frustrating that for gastro, neuro, orthopaedic I have to go to other hospitals and that there is no communication between hospitals and I have to constantly repeat myself and advocate wherever I go."

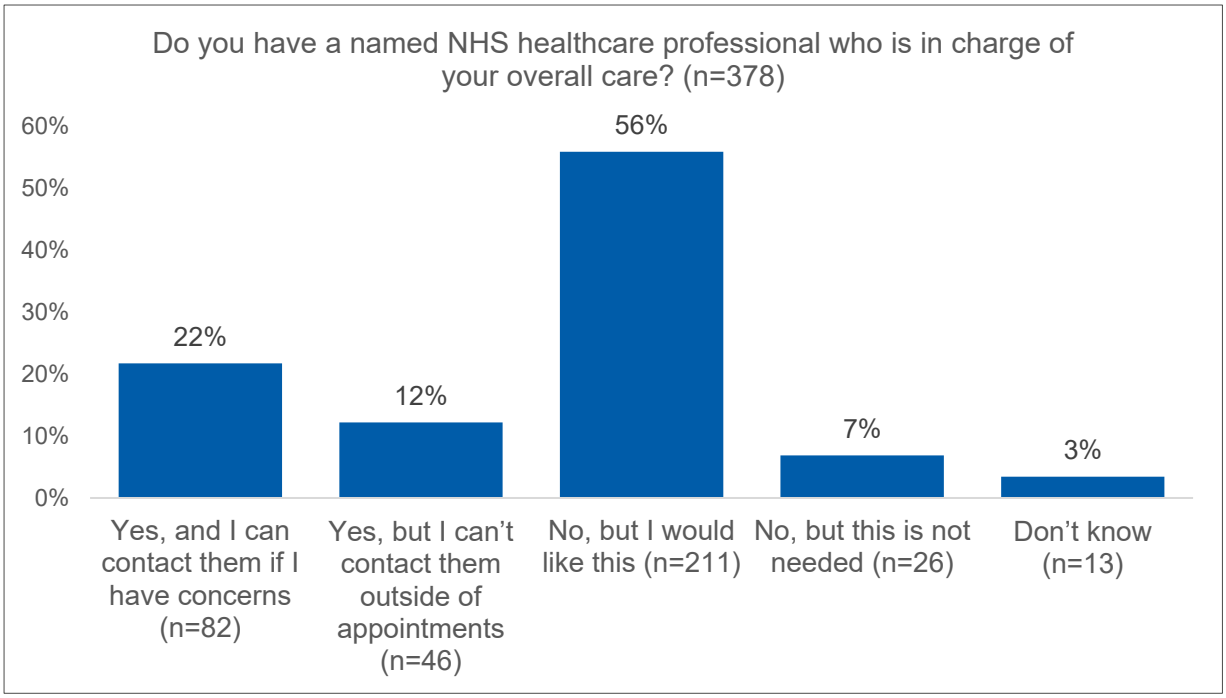
It is worth noting that one interview conducted during the survey development phase indicated that the family chose local hospital care because travelling to a specialist hospital would have posed too great a barrier. This highlights how geographical distance and travel challenges can significantly influence care decisions, potentially limiting access to specialist expertise and comprehensive multidisciplinary support.

Coordination of care

Effective coordination of care is essential for people with MFS and LDS as these conditions affect multiple body systems requiring input from a wide range of healthcare professionals.

Around one third of respondents (34%, n=128) reported having a named healthcare professional responsible for overseeing their care (Figure 14). Of these, 12% (n=46) reported being unable to contact them outside of appointments if they had concerns. More than half of respondents (56%, n=211) said they would like to have a named NHS healthcare professional to coordinate their care. This highlights a significant gap in care coordination and suggests that many patients feel unsupported in navigating complex, multi-specialist pathways.

Figure 14: Proportion of respondents with a named NHS healthcare professional to oversee care



Written comments about a lack of care coordination were common from respondents (see Appendix 3 for further examples):

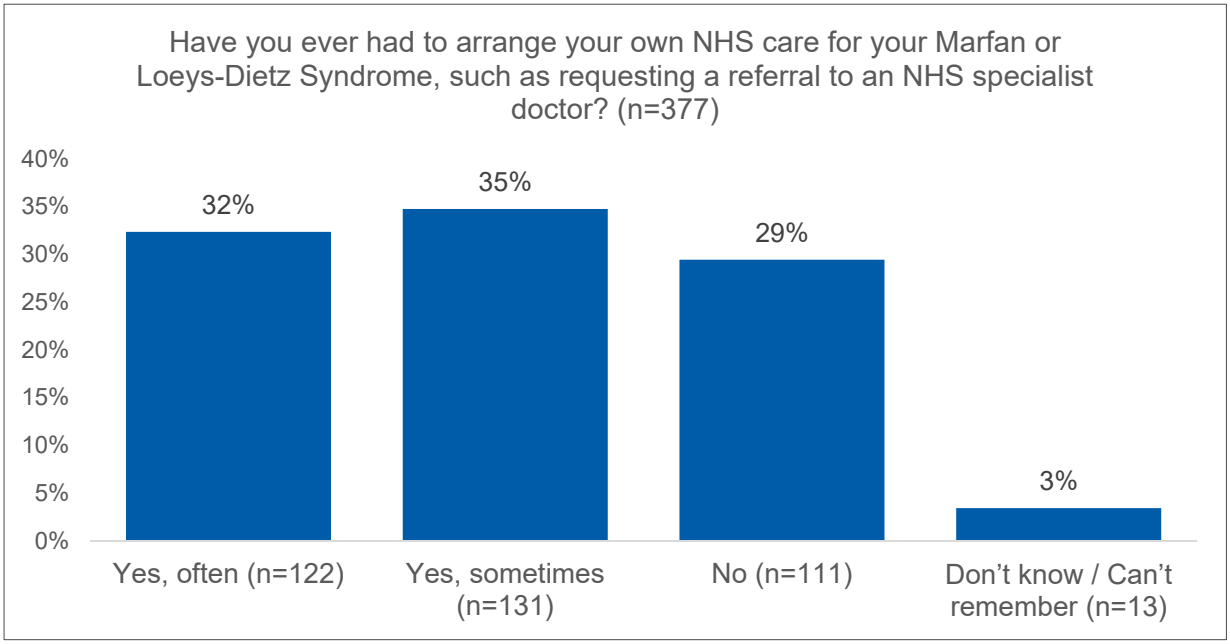
"It's been very difficult working with so many different departments within the hospital and having to find out how often each appointment needs to be. It's very disjointed without someone liaising with all relevant departments."

"There is no one overall expert who oversees treatment and necessary referrals. I feel as a parent to a child with LDS I do my own research and do a lot of initiating and requesting the correct referral. The services although individually are excellent they are disjointed, no holistic approach. A MDT meeting would be extremely beneficial to talk through all the issues and each specialist takes on whatever is relevant or links up with other specialists for a holistic approach"

"There doesn't appear to be any joined up medical treatment with medical services being siloed and not communicating with each other across the disciplines. This makes managing any health issues difficult because there isn't a single point of contact. This lengthens the time taken to access any medical treatment because each issue requires a fresh GP referral which then means many months of NHS waiting lists (e.g. it took over 2 years for me to access competent podiatry services!). My GP appears disinterested and often advises to go to A&E with any issues, which I feel isn't right."

Only 29% (n=111) of respondents said they did not need to arrange their own care, such as requesting referrals to specialist clinicians. In contrast, almost one-third (32%, n=122) reported that they *often* had to organise their own care, and a further 35% (n=131) said this was *sometimes* required (Figure 15). These findings indicate that a substantial proportion of patients are actively managing complex care pathways themselves, which may increase stress and risk of gaps in treatment.

Figure 15: Frequency of respondents arranging their own care



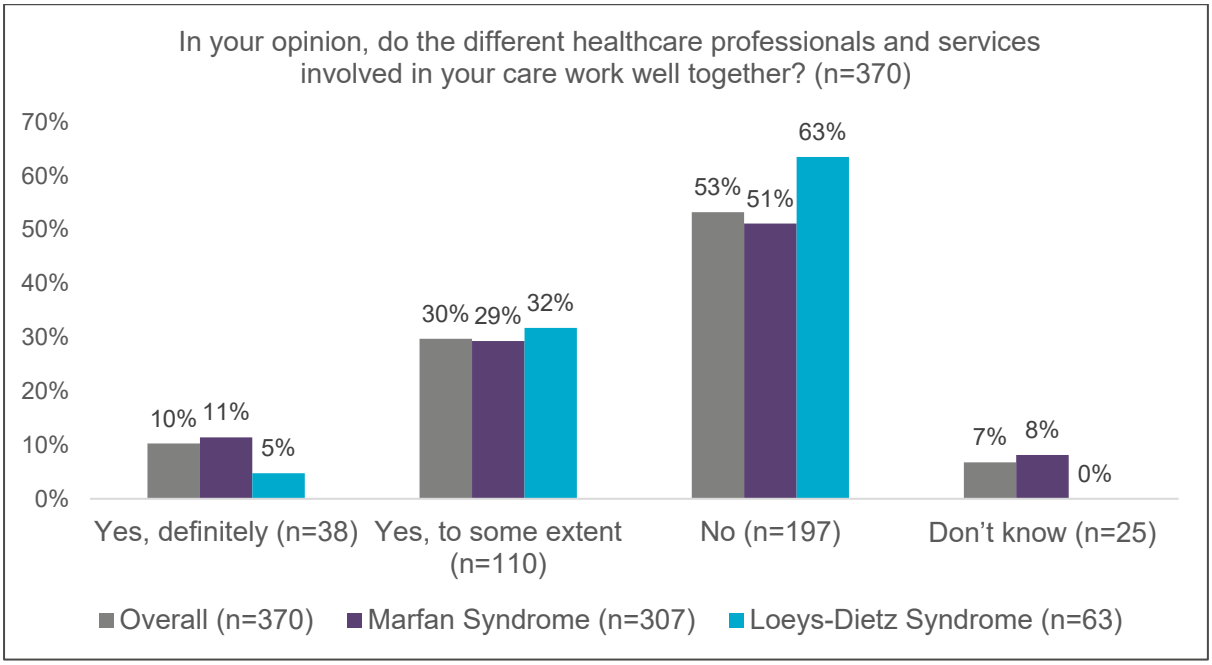
Several respondents described the challenges of navigating fragmented care pathways and the absence of coordinated support (see Appendix 3 for further examples):

"I have no one person in the NHS overseeing my health, and recently realised it had been almost 4 years since my last echocardiogram, and had to request one."

"I find very difficult to have to fight all the time too be seen for each specialist. I am missing a specific clinic that it will help to [address] all the symptoms at once."

It is therefore perhaps unsurprising that over half (53%, n=197) felt that different healthcare professionals and services do not work well together (Figure 16). This proportion was higher among respondents with LDS (63%, n=40) compared to those with MFS (51%, n=157) (Figure 16). This finding underscores the fragmented nature of care for people with Marfan and Loeys-Dietz syndromes and highlights the need for integrated, coordinated care models to reduce patient burden and improve continuity.

Figure 16: Respondent views on how well services work together



Cardiology care and scans

Most respondents (61%, n=229) reported seeing a cardiologist at least once a year, and over a quarter said they attended appointments every two years. However, one in ten respondents (10%, n=38) indicated they do not regularly see a cardiologist (Table 11, Appendix 1).

Differences between conditions were found: 70% (n=44) of respondents with Loeys-Dietz reported seeing a cardiologist at least once a year, compared with 59% (n=185) of respondents with Marfan syndrome (Table 11, Appendix 1). This disparity likely reflects the more severe and early-onset cardiovascular manifestations associated with Loeys-Dietz syndrome, which may require closer monitoring.(20,21)

"…the ongoing care is difficult to manage. I'm supposed to have annual cardiac check-ups for example at [Hospital] but these often extend to 18-24 months between appointments and involve having to chase and cajole the cardiac service to obtain an appointment ... I have the impression that if we didn't chase appointments then I'd never hear from the NHS again!!"

"There is no specialist cardiologist around where I live. I am seen by a general cardiologist in [town], Scotland who does not know about Loeys-Dietz Syndrome and how to care for me. I have to travel to Aberdeen (1.5hours) for an MRI. I have been on waiting lists for nearing a year, for a DEXA scan and ophthalmology. My GPs Google Loeys-Dietz Syndrome in front of me. Nothing I say is taken seriously. If I mention something that is bothering me and my own research results, I am accused of hypochondria or told the internet is a bad place."

Current guidelines recommend that people with Marfan or Loeys-Dietz syndrome undergo regular monitoring with an echocardiogram (heart ultrasound) at least once a year.(1) The survey found that just over half of respondents (59%, n=214) have this scan every six months or annually (Figure 17), suggesting that some patients may not be receiving the

recommended level of monitoring. Some respondents offered written comments illustrating these difficulties:

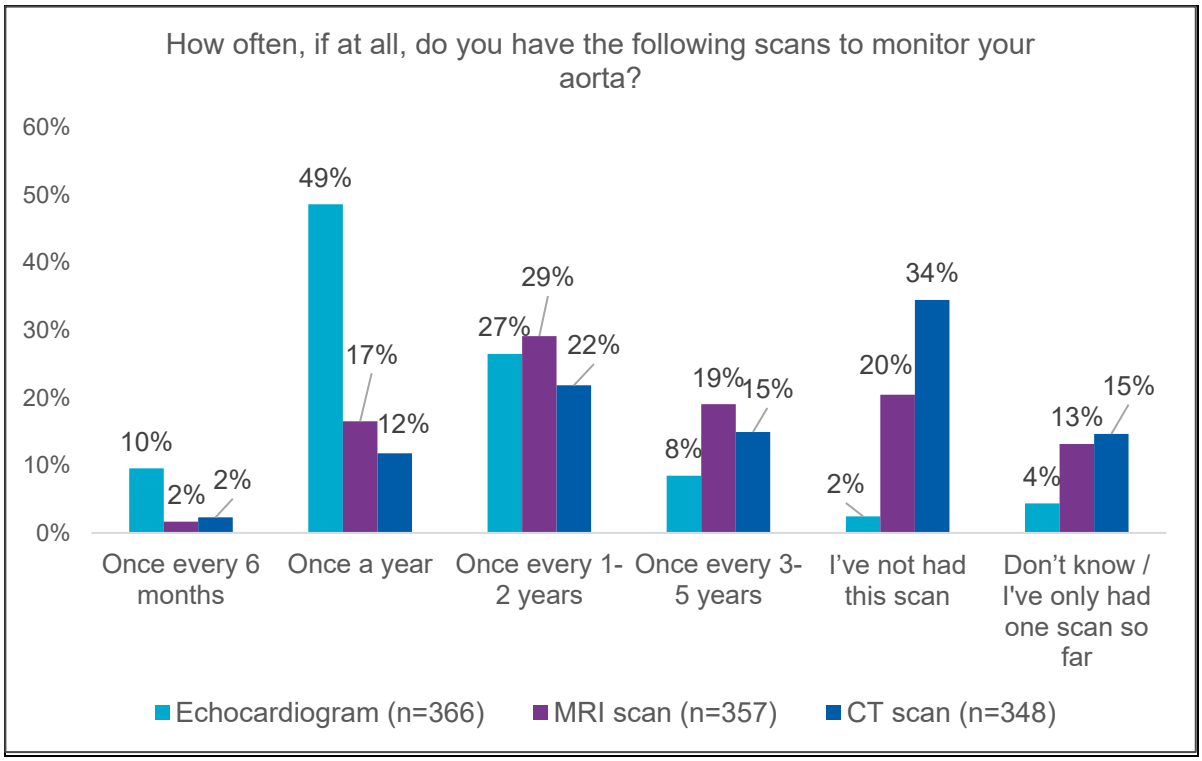
"I had a very big battle with a cardiologist about my regular heart scans. He discharged me with out informing me 2 times and attempted a 3rd time. Twice I contacted the genetics team who emailed him to say I needed regular scans."

"I cannot access the recommended yearly scans for Loeys-Dietz syndrome and am very worried my health is at risk as a result."

"Up until 10 years ago I had an ultrasound and saw a cardiologist dr every year. Then I was moved to 2 yearly ultrasounds and haven't seen a specialist since. I just get a letter saying 'no change, we'll send you for another scan in 2 years'. I have tried fighting this, but get told that 2 yearly checks are standard now. Despite me knowing they aren't."

Lower proportions of respondents reported regularly receiving an MRI or CT scan when compared with an echocardiogram (Figure 17). This would be expected, as the guidelines recommend that vascular imaging via an MRI or CT scan is conducted at first evaluation and then every 3-5 years if stable for MFS and every 1-3 years for LDS.(1)

Figure 17: Frequency of respondents receiving an echocardiogram, MRI scan and CT scan



In patients with MFS, treatment with either a beta blocker or angiotensin receptor blocker (ARB) is recommended to reduce the rate of aortic dilatation.(1) Most respondents reported regularly taking at least one type of heart medication: 59% (n=220) take a beta blocker and/or 48% (n=179) have been prescribed an angiotensin receptor blocker and/or 30% take another type of heart medication. However, more than one in ten respondents (13%, n=48) said they had not been offered any heart medication (Table 12, Appendix 1). Differences emerged by time since diagnosis: among those diagnosed within the past year, 44% (n=7)

had not been offered any heart medication, compared with 9% (n=22) of respondents diagnosed more than 10 years ago (Table 12, Appendix 1). This suggests that newly diagnosed patients may experience some delay in accessing appropriate heart care medication.

Support with mental health

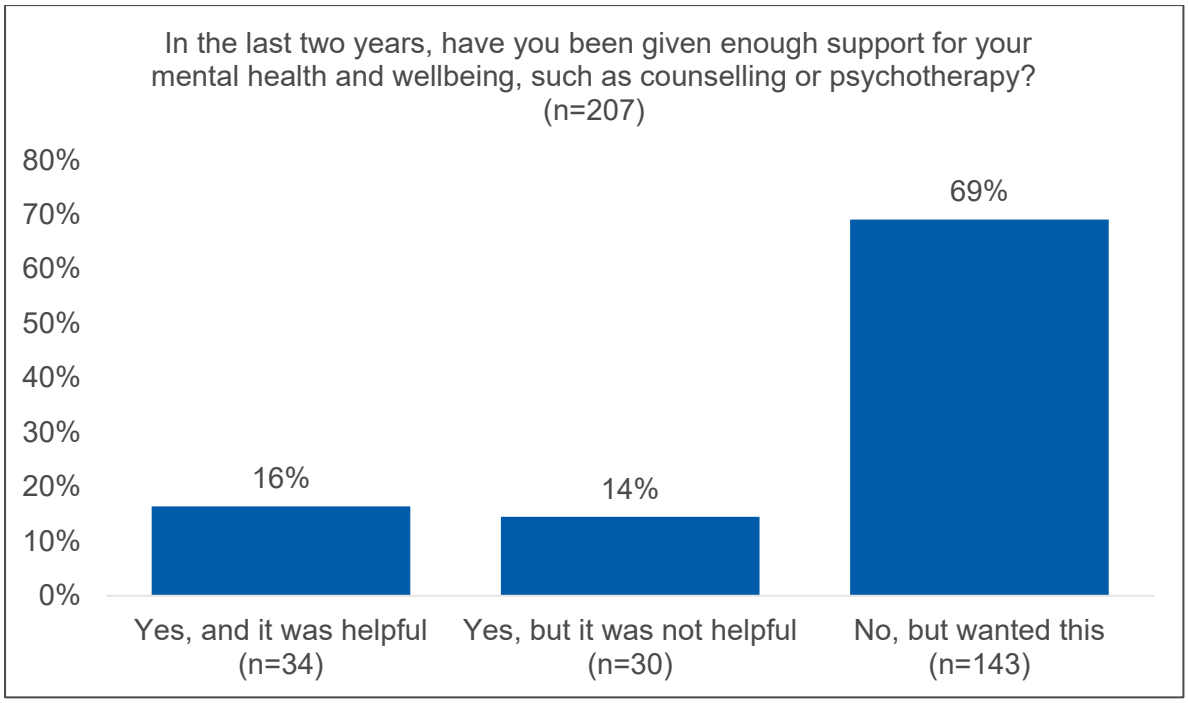
Forty-five per cent of respondents (n=170) felt they did not want any support in the past two years for their mental health and wellbeing, such as counselling or psychotherapy (Table 13, Appendix 1). Of those respondents who wanted this, a large proportion (69%, n=143) said they did not get enough support for their mental health (Table 14, Appendix 1).

Of the respondents that were given support for their mental health and wellbeing, only 16% (n=34) found it to be helpful with the remainder 14% (n=30) reporting it was not helpful (Figure 18).

"I receive yearly check ups of my heart and an MRI every two years but nobody ever asks how I am mentally or if I have pain other than my heart. I would like help with my mental health but don't no how to go about it."

"Been waiting for my spinal surgery from before COVID. Been pushed from pillar to post and the surgeon retires. My fusion has collapsed and my curvature continues to get worse. I am in pain and can't walk far distances. No one understands the impact this has on my mental health."

Figure 18: Respondents experience of support for mental health



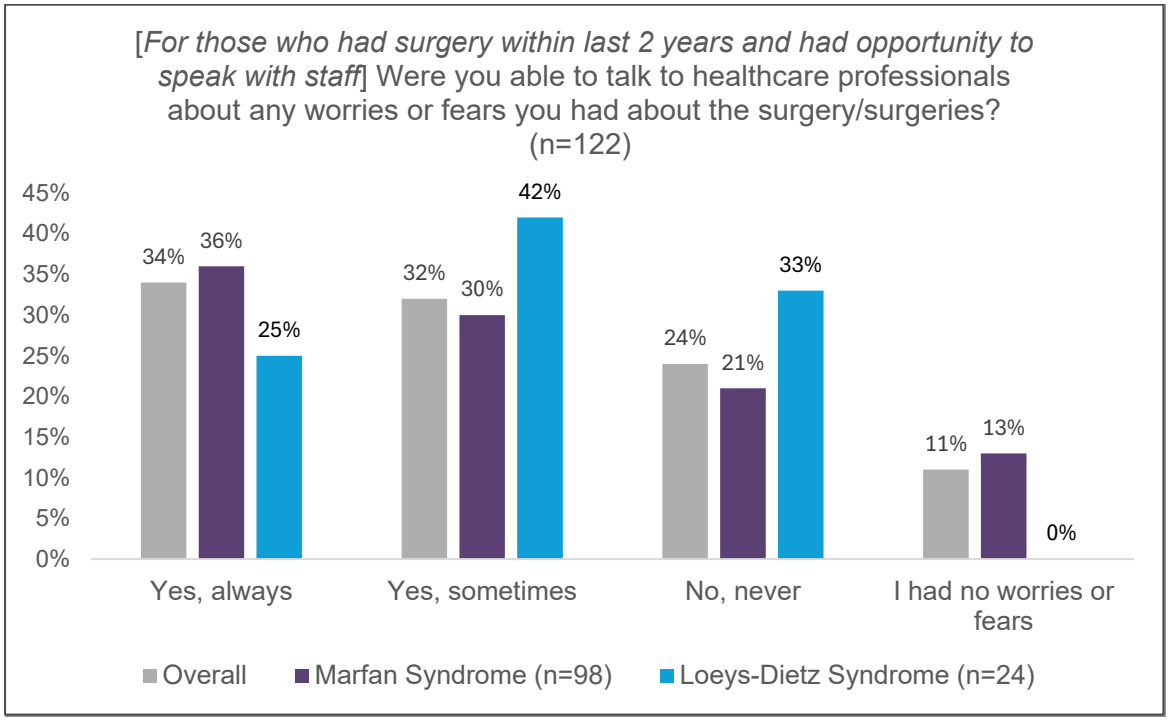
Regional differences were observed in respondents' experiences of mental health support, though these findings should be interpreted cautiously due to small sample sizes. The proportion of respondents reporting insufficient support ranged from 25% (n=2) in the North East of England to 93% (n=13) in the West Midlands (Table 14, Appendix 1).

Surgery

The majority of respondents had undergone at least one surgical procedure related to MFS or LDS, only 21% (n=80) reported never having surgery. The most common procedures were aortic surgery (43%, n=159), heart valve surgery (27%, n=101) and eye surgery (25%, n=93). See Table 15, Appendix 1 for details.

Among those who had surgery within the last two years and were not admitted as an emergency (i.e., had the opportunity to speak with staff), one-third (34%, n=41) said they were *always* able to discuss any worries or fears with a healthcare professional, with a further 32% (n=39) said they could do so *sometimes*. However, almost a quarter (24%, n=29) reported never having the opportunity to talk to a healthcare professional about their concerns. This proportion was higher among respondents with LDS, with one third (33%, n=8) reporting no opportunity to discuss surgery concerns with staff (Figure 19). This highlights a gap in emotional support during the surgical process, which may affect patient confidence and overall experience of care.

Figure 19: Respondents experience of support with surgical concerns



Care from healthcare professionals

Relational aspects of care

The survey revealed mixed experiences regarding relational aspects of care. Only one in five respondents (20%, n=74) felt that healthcare professionals *definitely* took their needs and wishes into account, and a similar proportion (23%, n=85) reported *always* having confidence and trust in those involved in their care. In contrast, more than half said their needs and wishes were considered only *to some extent*, and that they only *sometimes* had confidence and trust in healthcare professionals (Figures 20 and 21).

Figure 20: Respondents views on whether staff took their needs into account

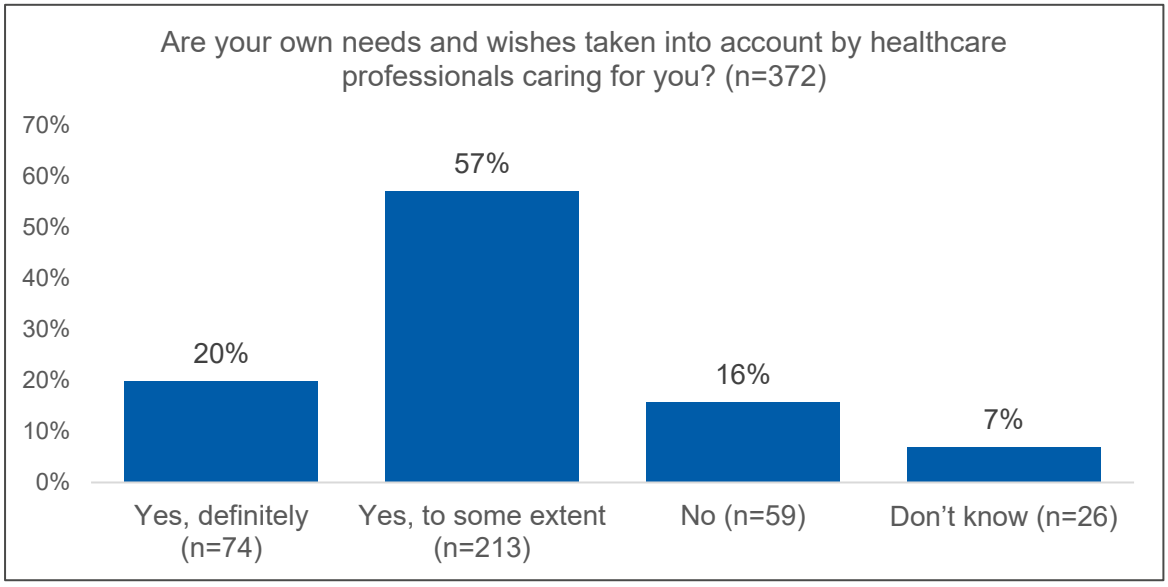
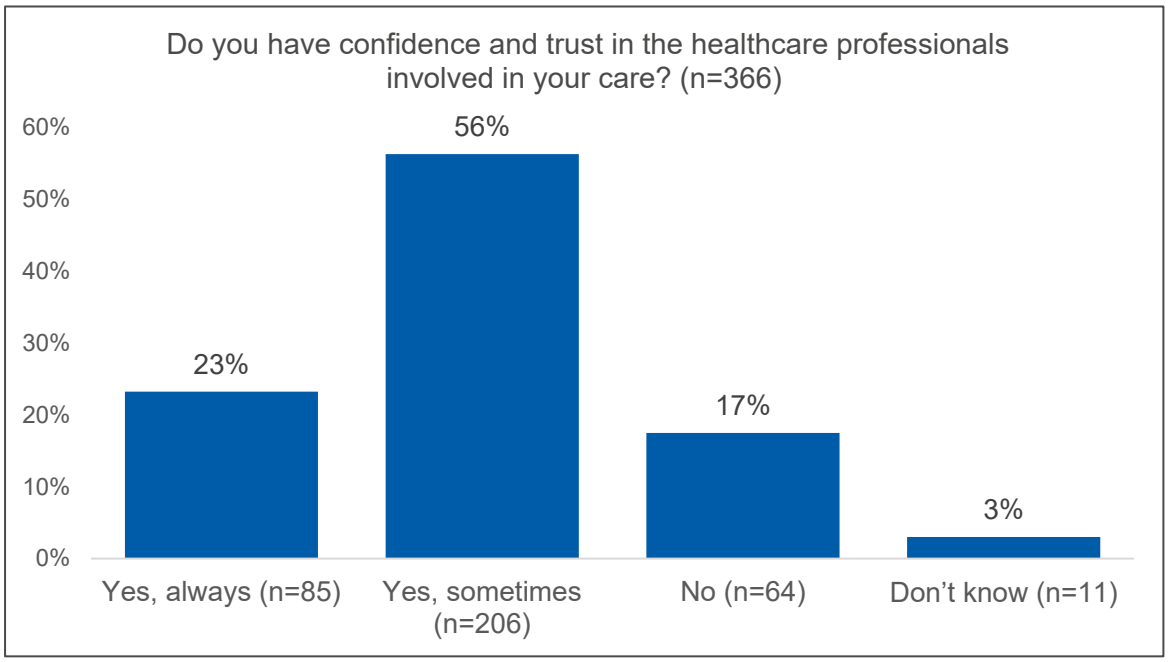


Figure 21: Respondents confidence and trust in healthcare professionals



These findings suggest a potential disparity between experiences with different healthcare professionals. Interview participants highlighted that while they often trusted some

specialists with greater knowledge of MFS or LDS, they lacked confidence in professionals unfamiliar with these conditions. Survey comments echoed concerns about limited awareness and understanding among healthcare professionals, highlighting the need for improved training to ensure patients feel understood and supported throughout their care journey (See Appendix 3 for additional comment examples).

"GP response to my multiple requests for help have been dismissive, rude, belittling. The overall impression is that they do not believe in Marfan Syndrome despite it being diagnosed by a genetic test […] Told by GP that she knew other Marfan people who went to the gym and fixed their issues. No understanding of the immense range of presentations. Extremely upsetting to be met with that response. I now try to research my own referrals and know that the GP will not help me."

"Our 3 year old has been diagnosed with Loeys diets for nearly a year now - yet there is no answers from our hospital on whether she should be on medication on or not (I believe she should be from what other hospitals are doing). I worry things in her care will be missed due to lack of knowledge from the healthcare professionals."

"Not many health care professionals that I come across are aware of the internal effects of Marfan - too often I get the comment that I don't look like I suffer from it. The link between my aortic problems, heart failure and Marfan don't seem to be flagged. I have to do a lot of explaining."

Information provision

Information from healthcare professionals

Just over one quarter of respondents (27%, n=105) said the information they received from healthcare professionals at diagnosis was *definitely* helpful, with a further 37% (n=144) finding it helpful *to some extent*. However, almost one quarter (24%, n=94) reported that the information was either not useful or that they received none at all. Although experiences did not vary significantly by syndrome type or time since diagnosis, the survey indicated that a shorter time to diagnosis was associated with more positive views of information provision at the time of diagnosis. Among those diagnosed within six months, 41% (n=51) rated the information as definitely helpful, compared with only 22% (n=14) of those whose diagnosis took more than two years (Table 16, Appendix 1).

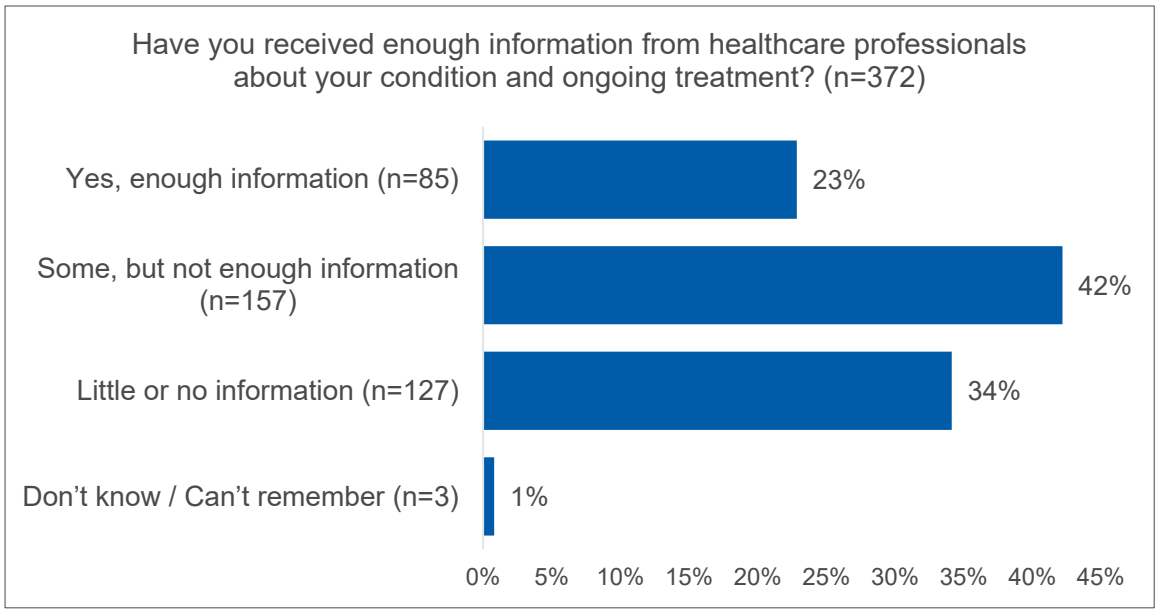
"Not enough information or support given to you upon diagnosis, this is a heartbreaking illness and carers patients, dependants all need more information and advice and support."

"The genetic consultant that told me I have Marfan's didn't really know too many details and just referred me to the Marfan's trust and other websites to look up my condition. There is no further support in Scotland. I've had more support from the Marfan's nurse on the Marfan's trust website that I found on Facebook."

Fewer than one in four respondents (23%, n=85) felt they had received enough information about their condition and ongoing treatment, with the majority (42%, n=157) reporting they had received some, but not enough. Over one third (34%, n=127) said they had received little or no information at all (Figure 22). The survey indicated that respondents experiencing more symptoms affecting daily life were less likely to feel they had received sufficient information about their condition. Only 16% (n=6) of those reporting ten or more symptoms

said they had received enough information, compared with 41% (n=13) of those without symptoms impacting day-to-day life (Table 17, Appendix 1)

Figure 22: Respondent views on information provided about their condition and treatment



"My energy levels and pain have become a lot worse in the last two years. My Cardiologist wondered if it could be linked to perimenopause but suggested I find out from other people on the Marfan Trust Facebook page. I can't find an answer and just feel I have been left to cope and just learn to live with it. I can't get any answers."

These findings highlight a significant gap in information provision by healthcare services, suggesting that many patients feel under-informed about their condition and treatment, which may hinder their ability to manage their health effectively.

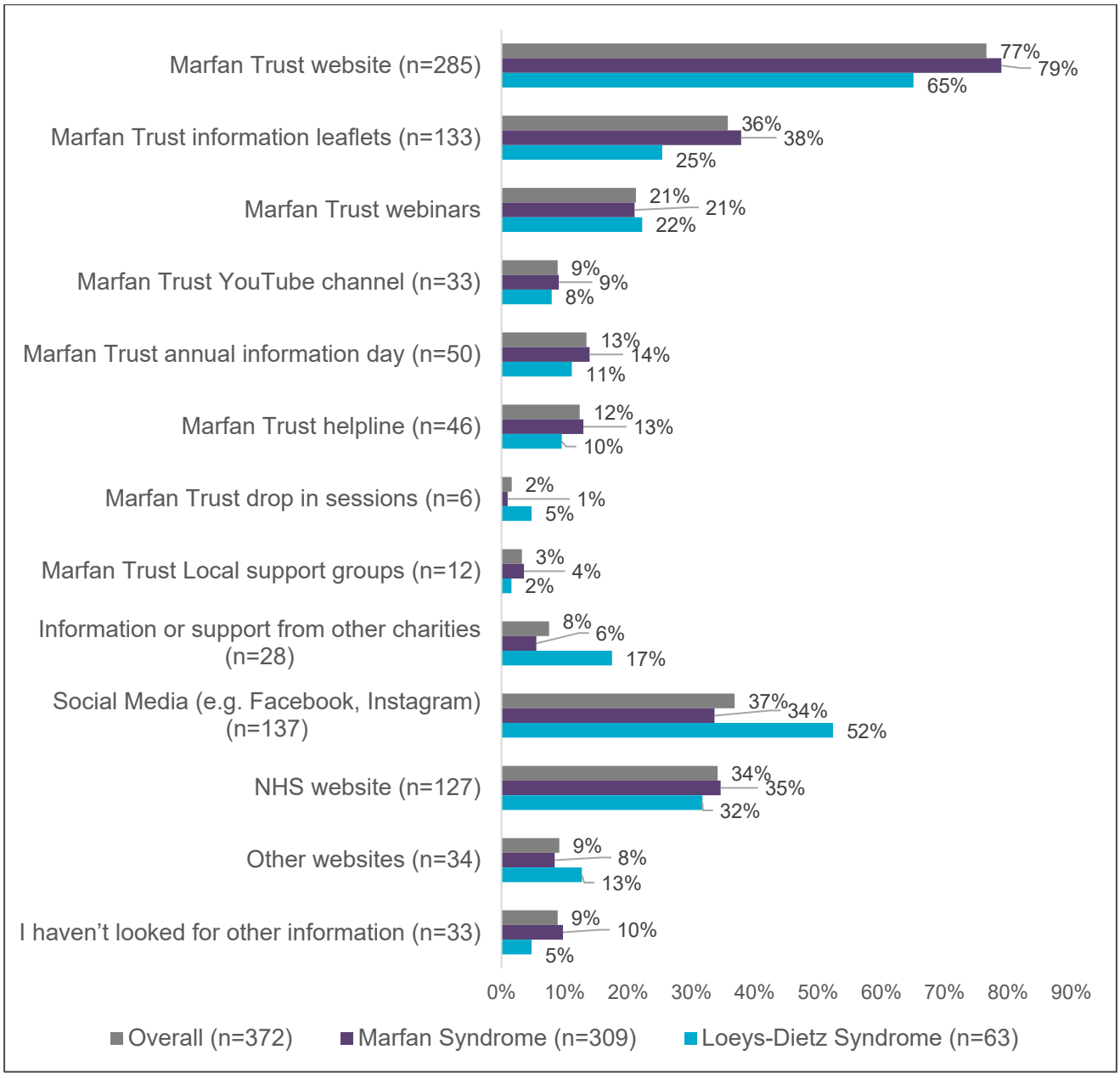
Information from other sources

As shown in Figure 23, the Marfan Trust website was the most common source of information with just over three quarters of respondents (77%, n=285) using it for additional support or information about their condition.

"The Marfan trust are a brilliant organisation and have helped my family lots! I always point people in their direction on social media sites or if I meet someone in person who needs Marfan related help and advice."

Other commonly used sources included social media (37%, n=137), Marfan Trust information leaflets (36%, n=133) and the NHS website (34%, n=127). Differences emerged between conditions: respondents with MFS were more likely to use the Marfan Trust website and its leaflets, whereas a higher proportion of those with LDS reported using social media (Figure 23).

Figure 23: Additional information sources used by respondents, by condition



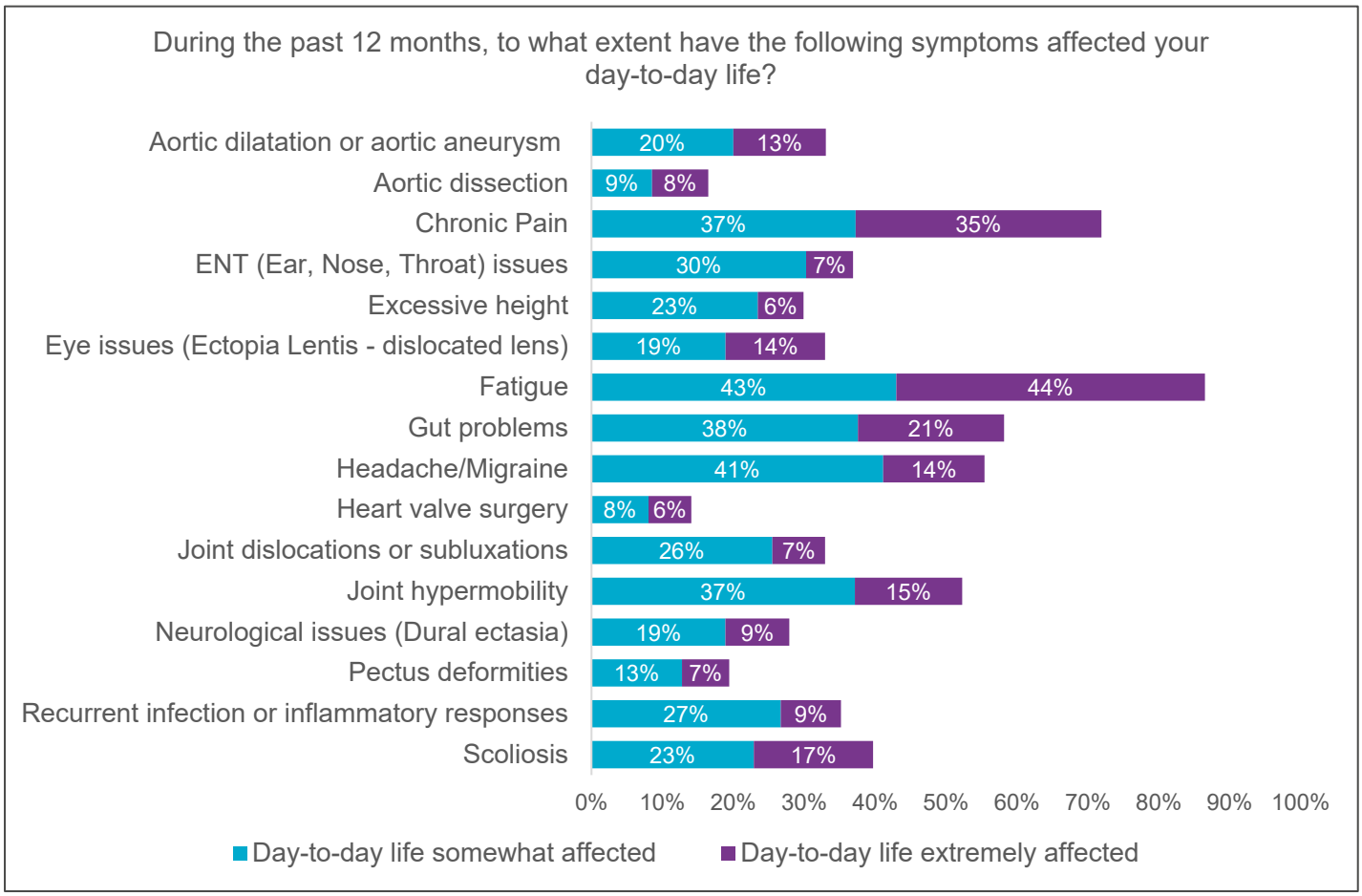
Impact on quality of life

Symptoms affecting daily life

Respondents were asked which symptoms had affected their day-to-day life over the past 12 months. As shown in Figure 24, more than half reported that the following symptoms had either *somewhat* or *extremely* affected their daily life:

- Fatigue (87%, n=308)
- Chronic pain (72%, n=251)
- Gut problems (58%, n=206)
- Headache / migraine (55%, n=194)
- Joint hypermobility (52%, n=183)

Figure 24: Proportions of respondents reporting symptoms which affect daily living



These findings align with previous research showing that fatigue and chronic pain are commonly reported symptoms among people diagnosed with MFS and LDS.(5,7,8,10,11,15)

"Pain and fatigue are the worst aspects to cope with. Pain starts within 20-30 minutes of getting out of bed and lasts until bed time. I am on pain clinic advised pain relief to try and cope. Fatigue means I can do one short task per day. Holidays involve one day active then one day of rest."

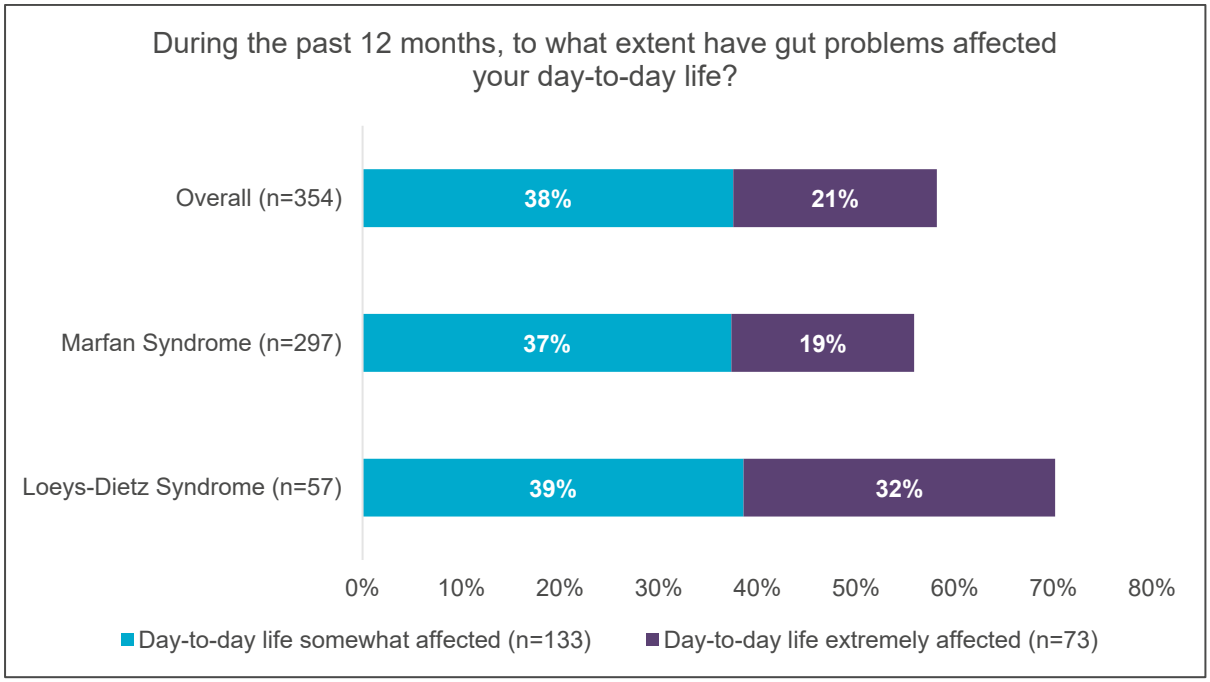
Only 8% (n=20) of respondents felt that healthcare professionals *always* did everything possible to help manage their pain. Just over half (51%, n=129) said this happened *sometimes,* while more than one third (35%, n=87) reported that healthcare professionals *never* did everything they could to control their pain (Table 18, Appendix 1). These findings suggest a significant gap in pain management for people with Marfan and Loeys-Dietz syndromes. As noted previously, 40% (n=143) of respondents were unable to access a pain specialist in the past two years but said they wanted or needed this support. This suggests a lack of proactive pain management which may contribute to reduced quality of life.

"Local authority has long waiting lists currently on waiting list for pain clinic (2 years) and on waiting list for wheelchair referral."

"I had been in pain for 50 years before I managed to get on a pain management course which was one session a week for 6 weeks many miles from home."

Evidence of gut problems affecting those with MFS or LDS, however, is more limited. A recent Marfan Trust pamphlet highlights some research indicating a link between connective tissue disorders - particularly Ehlers-Danlos syndrome - and gastrointestinal disorders, but emphasises that further studies are needed in populations with MFS and LDS.(22) A survey conducted by the Marfan Foundation in the USA reported that just over a quarter of respondents experienced challenges with gastrointestinal issues.(9) There is evidence that gut problems are more common in those affected by LDS than in those with MFS.(20) This aligns with the survey findings; 71% (n=40) of respondents with LDS reported that gut problems affected their daily life (somewhat or extremely), compared with 56% (n=166) of respondents with MFS (Figure 25). Among those with LDS, 39% (n=23) said they had not been able to access a gastrointestinal (GI) specialist in the past two years but had wanted or needed this (Table 19, Appendix 1).

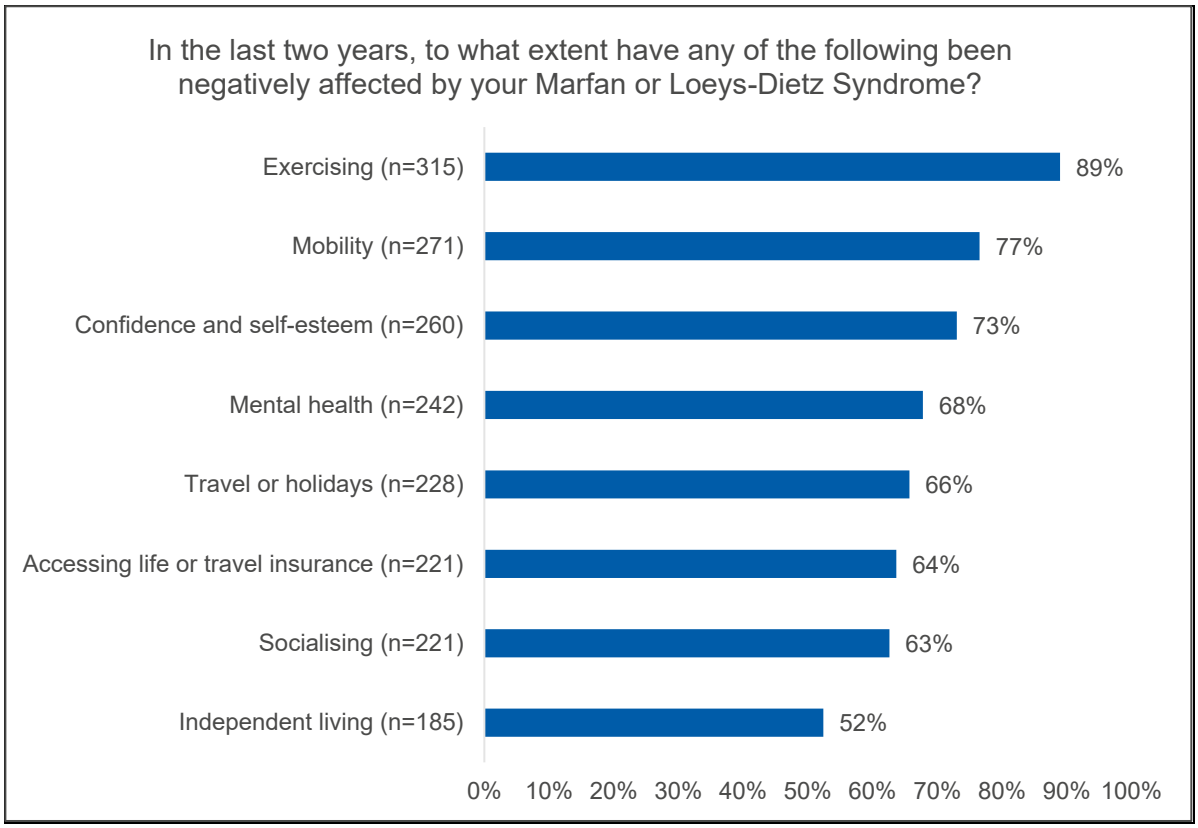
Figure 25 Proportion of respondents reporting gastrointestinal issues, by condition



Impact on quality of life

Figure 26 shows that more than half of respondents reported MFS or LDS had a negative impact - either somewhat or extremely - on various aspects of their lives within the past two years. A large proportion of respondents also reported that MFS or LDS negatively impacted on friendships (48%, n=171), employment (46%, n=154) sexual activity (46%, n=164) and family relationships (44%, n=157).

Figure 26 Areas of life most affected by MFS and LDS



"I am unable to undertake regular employment because of the joint and muscle pain and the fatigue from the Marfans."

"Been waiting for my spinal surgery from before COVID. Been pushed from pillar to post and the surgeon retires. My fusion has collapsed and my curvature continues to get worse. I am in pain and can't walk far distances. No one understands the impact this has on my mental health."

Respondents who reported a higher number of symptoms affecting daily life were much more likely to say that MFS or LDS had a negative impact on their quality of life. For example, among those experiencing ten or more symptoms, 82% (n=31) reported that their mobility was *extremely* negatively affected, compared with just 9% (n=12) of those reporting between one and five symptoms (Table 20, Appendix 1).

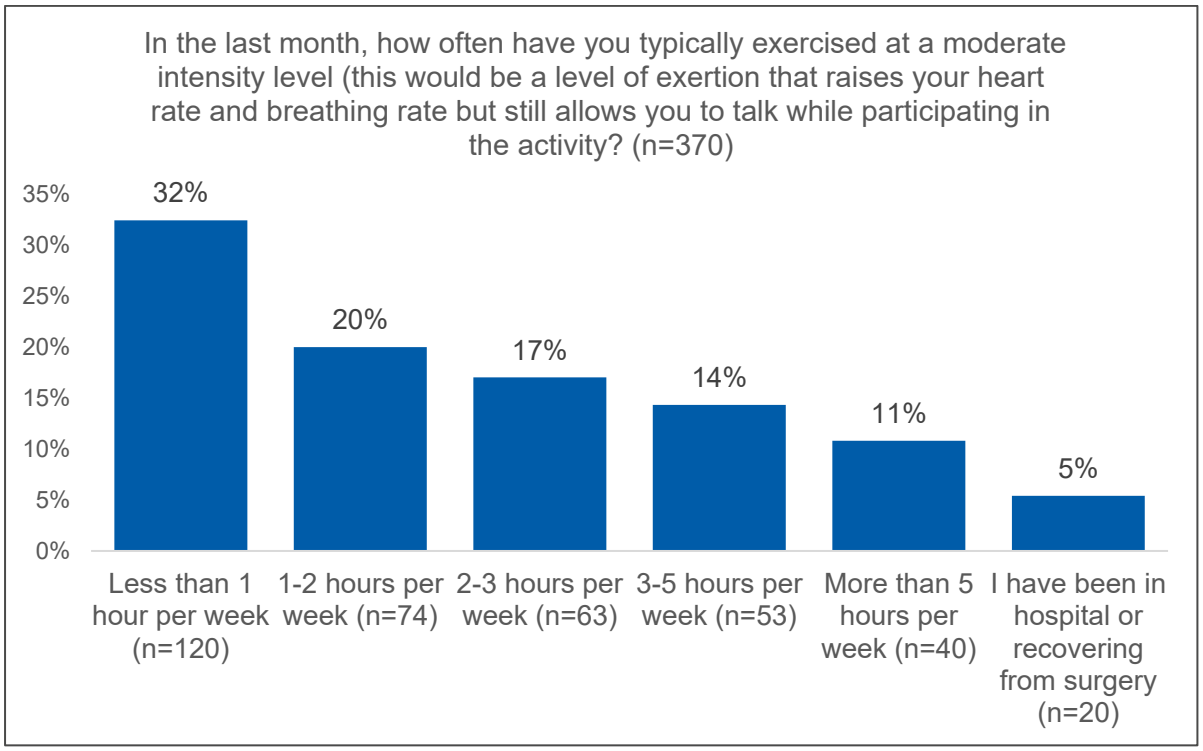
Exercise

Previous research has shown that people with MFS or LDS often view exercise as a dilemma between balancing health benefits against the risk of harm.(11) Guidance on exercise for people with MFS/LDS has evolved over time; whilst it was initially discouraged

due to the risk of exacerbating the condition, more recent recommendations suggest that low-intensity, low-impact exercises can be beneficial.(1)

The survey found that almost one third of respondents (32%, n=120) typically exercise for less than one hour a week (Figure 27), suggesting that concerns about physical activity remain common among people with MFS and LDS.

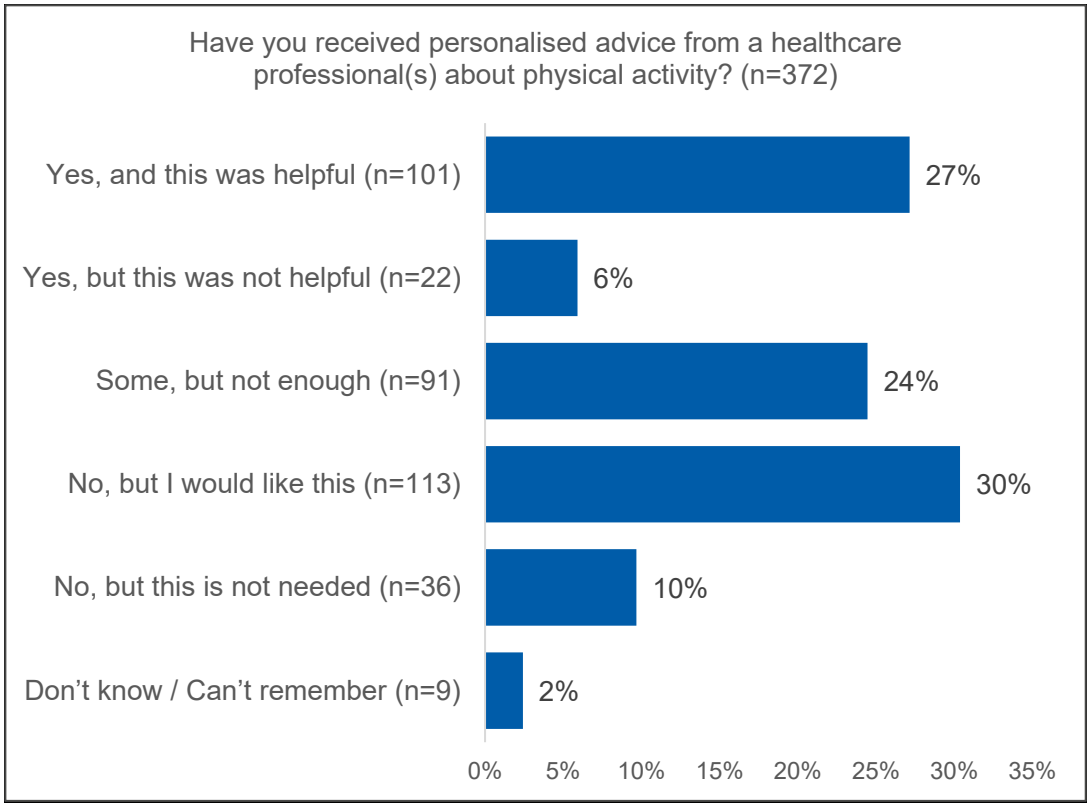
Figure 27: Frequency respondents typically exercised in the last month



Clinical guidelines for the management of MFS recommend physical activity is individualised based on aortic diameter, family history of aortic dissection and pre-existing fitness.(1)

Respondents were asked if they had received personalised advice from healthcare professional(s) about physical activity. While 27% (n=101) said they had received helpful advice, more than half (54%, n=204) reported either not getting enough advice or none at all, despite wanting this support (Figure 28). This reinforces the earlier point that exercise is often viewed as a dilemma for people with Marfan and Loeys-Dietz syndromes - balancing potential health benefits against the risk of harm. Without clear, tailored advice, some people may avoid exercise altogether. This finding supports the argument that people with MFS and LDS need help to minimise fear and anxiety associated with exercise.(6,18,19) Addressing this gap by providing specialist-led, evidence-based guidance could help patients maintain safe levels of physical activity, and improve overall quality of life.

Figure 28: Proportion of respondents receiving personalised advice on physical activity



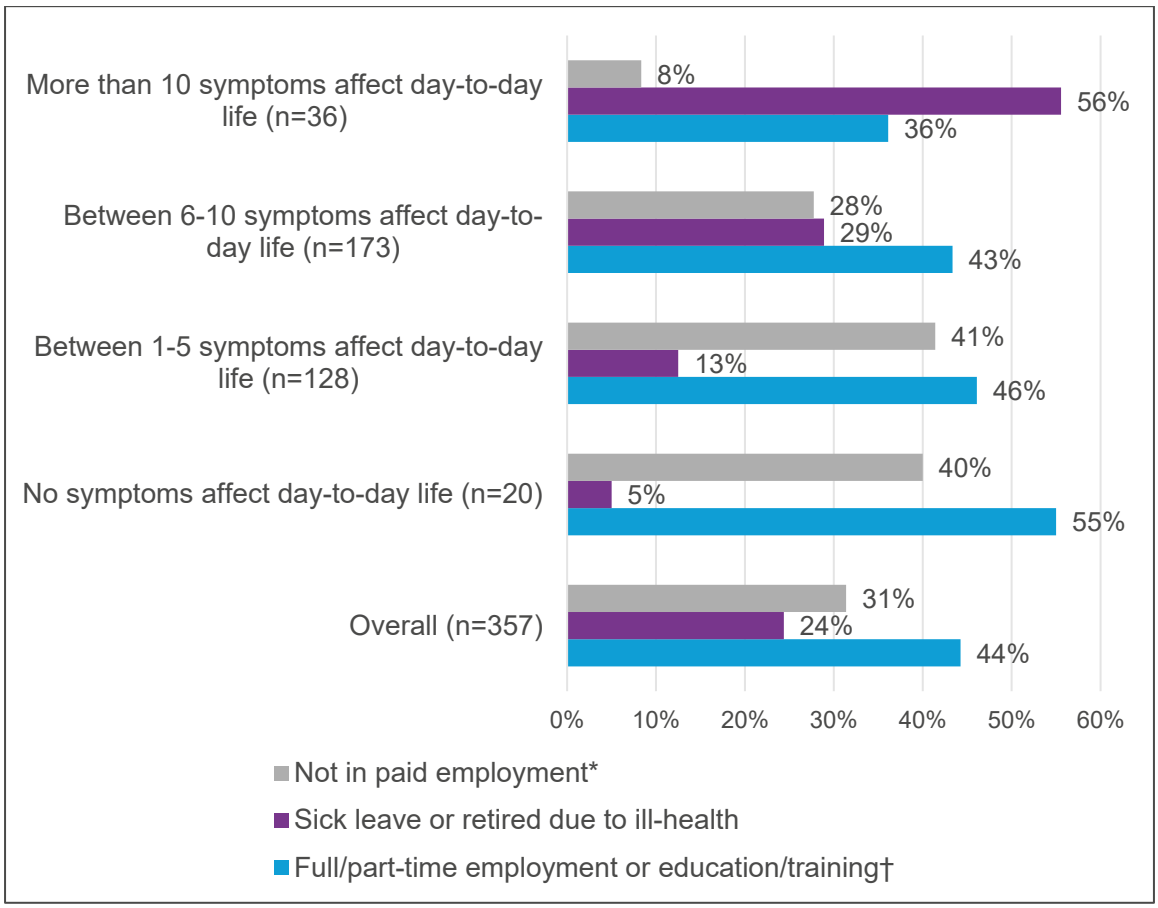
Employment and finances

As noted above, 46% (n=154) of respondents reported that their employment had been *somewhat* or *extremely* affected by MFS or LDS. Those experiencing a higher number of symptoms affecting daily life were more likely to report a negative impact on employment. Among respondents with ten or more symptoms, 62% (n=21) said their employment was *extremely* affected, compared with just 9% (n=11) of those reporting between one and five symptoms (Table 21, Appendix 1). Unsurprisingly, this pattern extended to work status: respondents with more symptoms were more likely to be on long-term sick leave or unable to work due to MFS or LDS (Figure 29). Among those with ten or more symptoms, 56% (n=20) were on sick leave or had retired due to ill-health, compared with 13% (n=16) of those reporting between one and five symptoms. It is perhaps worth noting that of those not in paid employment, 60% (n=67) reported they were retired which aligns with the finding that more than half of the respondents were aged 51 years and over.

"My employer (name) were awful, failed to recognise LDS as an illness, they closed my contract due to ill health after 30 years which left me devastated. They would not take me back as I was a risk and unable to do my job as a floor manager physically - no other adaptations offered they were scared I would have another heart attack. I have fought them for two years and finally won my legal appeal. Now on full pension and retired partner status. This has cost me dearly financially and mentally. I nearly lost my house, sold my car to pay the bills etc."

"I have been off work with depression 2.5 months and have received fit note after fit note without discussion/referral/support."

Figure 29: Respondent work status by number of symptoms experienced



* Includes those who selected 'maternity/paternity leave', 'carer', 'seeking work', 'retired' or 'other'
† Includes those who selected 'self-employed' and 'freelance'

Most respondents reported that benefits or financial support related to their MFS or LDS were either not needed or not applicable (Table 22, Appendix 1). The most commonly received benefit was Personal Independence Payment (PIP), reported by 30% (n=101) of respondents. However, at least one in ten respondents were unaware of each benefit listed, indicating a gap in knowledge about available financial support that people may be eligible for. This lack of awareness suggests that some individuals may miss out on assistance they are entitled to, highlighting the need for clearer information and guidance on benefits for people living with MFS and LDS. A few respondents commented that obtaining benefits can be challenging:

"My condition has worsened since I was 18. I am now unable to work due to extreme fatigue and joint pain. I have been fighting for PIP for two years and just been awarded middle grade on appeal."

Summary and recommendations

This survey provides evidence on the experiences of people living with Marfan syndrome (MFS) and Loeys-Dietz syndrome (LDS) in the UK. It highlights significant gaps in diagnosis, awareness, care coordination, and support for individuals with MFS and LDS which all align with key priorities within the 2021 UK Rare Diseases Framework.(23) While cardiology services are generally accessible, many respondents reported delays in diagnosis, fragmented care pathways, and unmet needs in areas such as pain management, mental health support, and gastrointestinal care. Quality of life is significantly impacted, with fatigue, chronic pain, and mobility challenges affecting daily living, employment, and mental wellbeing.

Diagnosis and genetic testing

Many respondents experienced delays and dismissive attitudes from healthcare professionals before diagnosis. LDS respondents were more likely to report that their concerns were not taken seriously and faced longer diagnostic timelines.

While most respondents had a genetic diagnosis, one in five said family members were not offered testing.

Access to Care

Cardiologists were generally accessible, but gaps were evident in access to other healthcare professionals, such as pain specialists, osteopaths, and podiatrists. Comments at the end of the survey indicated some respondents had waited a long time to receive appointments with some specialists.

Among those who wanted **mental health support**, more than two-thirds said they did not receive enough, and some reported it was inappropriate for their needs.

Treatment and monitoring

Most respondents reported taking heart medication. While over half received an echocardiogram at least once a year, a notable proportion were not monitored as frequently as recommended by clinical guidelines.

Care Coordination

Only one-third of respondents reported having a named healthcare professional responsible for overseeing their care, while more than half said they would like this support. Comments provided by respondents highlighted concerns about fragmented care and a strong desire for a single point of contact to coordinate services.

Two-thirds of respondents indicated they had to arrange their own care, and over half felt that healthcare services did not work well together.

Care from healthcare professionals

Only around one in five respondents felt healthcare professionals fully considered their needs and preferences, and a similar proportion reported consistently having confidence and trust in those involved in their care.

Fewer than one in four respondents said they had received enough information about their condition from healthcare professionals. Many looked for additional information and support from the Marfan Trust website and social media.

Despite a desire for personalised advice on physical activity, more than half reported not receiving this support.

Quality of Life

Fatigue, chronic pain, gastrointestinal issues, headache/migraine and joint hypermobility were widely reported. More than one third felt healthcare professionals did not do everything they could to control their pain.

Two-thirds or more of respondents reported MFS or LDS had a negative impact on exercise, mobility, confidence and self-esteem, mental health, and travel and holidays. Respondents with more symptoms experienced greater challenges across all areas.

Nearly half reported employment was affected, and those with more symptoms were significantly more likely to be on long-term sick leave or retired due to ill health. Awareness of some benefits was low, and some faced difficulties accessing financial support.

Recommendations

Based on these findings, the following actions are recommended:

For the NHS

1. **Reduce diagnostic delays and improve awareness**
    - Provide targeted training for GPs and non-specialist clinicians to recognise MFS and LDS symptoms.
    - Develop clear referral pathways and guidance for suspected cases.
    - Enhance system-wide education and awareness of rare conditions by integrating dedicated training modules into medical curricula and ongoing professional development, helping clinicians to better identify, understand and support individuals with MFS/LDS and other rare disorders.
2. **Improve genetic testing provision**
    - Ensure timely genetic testing for patients and offer cascade testing for family members as standard practice.

3. **Strengthen care coordination**
    - Introduce named care coordinators or specialist nurses to oversee multi-disciplinary care and act as a single point of contact.
    - Encourage multi-disciplinary team (MDT) meetings for complex cases.
4. **Expand access to specialist services**
    - Address gaps in pain management, physiotherapy, mental health support, and gastrointestinal care.
    - Offer tailored psychological support for patients and families, recognising the emotional impact of living with a rare condition.
5. **Address quality of life needs**
    - Provide proactive pain and fatigue management strategies.
    - Offer personalised advice on safe physical activity to reduce fear and inactivity.
6. **Support employment and financial wellbeing**
    - Improve signposting to benefits and financial support.
    - Work with employers to raise awareness and encourage reasonable adjustments for people with MFS and LDS.

For the Marfan Trust

1. **Raise awareness and education**
    - Deliver awareness campaigns for healthcare professionals and the public to improve understanding of MFS and LDS.
    - Share best practice guidance and patient stories to highlight the importance of early diagnosis and holistic care.
2. **Enhance information provision**
    - Continue developing comprehensive, accessible resources for patients and carers, including guidance on exercise and symptom management.
3. **Improve signposting and advocacy**
    - Strengthen links with NHS services to ensure patients are directed to reliable sources of information.
    - Advocate for improved care pathways and equitable access to specialist services.

4. **Support mental health and wellbeing**
    - Provide resources and signposting for psychological support tailored to rare conditions.
    - Explore partnerships with counselling services to offer condition-specific support.
5. **Assist with practical and financial support**
    - Offer clear guidance on benefits and entitlements.
    - Provide tools and advice for navigating employment challenges and workplace adjustments.

Limitations of the study

Some pragmatic choices were made in designing the survey methodology due to budget and time constraints, which introduced some limitations. The recruitment approach relied on self-selection, meaning the sample is unlikely to represent the entire UK population of people with MFS and LDS. Those more engaged with the Marfan Trust, and/or those diagnosed more than ten years ago, may have been disproportionately represented. To achieve a more representative sample, a standardised approach using patient lists from healthcare providers would be preferable, though this may not be feasible for a charity to implement alone.

Responses from people living in Northern Ireland, Scotland and Wales were relatively few compared to England, which limits the ability to generalise findings across the UK. A full assessment of representativeness would require comparison of the respondent profile with the overall UK population of individuals diagnosed with MFS and LDS.

Offering the survey in additional formats (such as other languages, large print, or Braille), may have improved accessibility and diversity, but is unlikely to have significantly increased number of responses.

Interpretation of the findings on time to diagnosis should be approached with caution, as almost one third of respondents were unsure or unable to recall the interval between first raising symptoms and receiving a diagnosis. This may reflect the long period since diagnosis for some participants. Although the question was not restricted to those diagnosed within the past year - on the assumption that most would recall the approximate timeframe - future surveys may wish to apply a 12-month cut-off to reduce potential recall bias. Additional uncertainty may have arisen due to differing interpretations of the time period to report, influenced by the varied nature of symptoms and by cases diagnosed following a medical emergency. The unexpectedly high proportion reporting diagnosis within three months, regardless of time since diagnosis, further suggests differing interpretations. The findings are also complicated by the fact that more than one third of respondents did not seek medical attention prior to diagnosis, resulting in fundamentally different diagnostic pathways. Future iterations of the survey should further test this question and consider including supplementary items to better capture the diversity of diagnostic experiences and/or conduct qualitative interviews to explore this.

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Appendix 1: Frequency Tables

Please note: these tables are referenced in the report and do not represent frequency tables for every survey question and subgroup analyses.

Table 1: How long ago were you first diagnosed with Marfan or Loeys-Dietz Syndrome? (this could be a clinical diagnosis OR a genetic diagnosis)

	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Under 1 year ago	20	5%	13	4%	7	11%
1-2 years ago	24	6%	9	3%	15	23%
2-5 years ago	42	11%	27	8%	15	23%
5-10 years ago	51	13%	40	12%	11	17%
More than 10 years ago	256	65%	240	73%	16	25%
Don't know / Can't remember	2	1%	2	1%	0	0%
Total Responses	**395**	**100%**	**331**	**100%**	**64**	**100%**

Table 2: Reason for seeking medical attention before diagnosis

	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
When symptoms first appeared	32	8%	26	8%	6	9%
When symptoms got worse	8	2%	8	2%	0	0%
Family history of Marfan or Loeys-Dietz Syndrome / a family member was diagnosed with Marfan or Loeys-Dietz Syndrome	122	31%	90	28%	32	50%
A friend or family member was worried about symptoms	12	3%	12	4%	0	0%
I was advised by a healthcare professional to have genetic testing	33	8%	28	9%	5	8%
I didn't, my diagnosis followed a check-up or medical attention for another condition	76	19%	70	21%	6	9%
I didn't, medical attention was sought on my behalf (e.g. medical emergency)	25	6%	21	6%	4	6%
My diagnosis followed a medical emergency in another family member	37	9%	29	9%	8	13%
Other	36	9%	33	10%	3	5%
Don't know / Can't remember	9	2%	9	3%	0	0%
Total Responses	**390**	**100%**	**326**	**100%**	**64**	**100%**

Table 3: Extent to Which Healthcare Professionals Took Health Concerns Seriously, by Time to Diagnosis

	Overall		Less than 6 months		6-12 months		1-2 years		More than 2 years	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Yes, definitely	47	12%	24	20%	2	4%	5	15%	5	8%
Yes, to some extent	69	18%	17	14%	14	29%	3	9%	17	27%
No	100	26%	30	24%	12	25%	15	45%	28	44%
I did not seek medical attention before my diagnosis	136	35%	50	41%	20	42%	9	27%	11	17%
Don't know / Can't remember	36	9%	2	2%	0	0%	1	3%	3	5%
Total Responses	**388**	**100%**	**123**	**100%**	**48**	**100%**	**33**	**100%**	**64**	**100%**

Table 4: Time to diagnosis for those diagnosed within the last 12 months

	Overall		Under 1 year ago	
Option	**Count**	**Percent**	**Count**	**Percent**
Less than 3 months	62	16%	5	26%
3-6 months	62	16%	3	16%
6-12 months	48	12%	2	11%
1-2 years	33	9%	3	16%
2-5 years	24	6%	1	5%
More than 5 years	40	10%	4	21%
Don't know / Can't remember	117	30%	1	5%
Total Responses	**386**	**100%**	**19**	**100%**

Table 5: Have you had a genetic diagnosis of Marfan Syndrome or Loeys-Dietz Syndrome?

	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Yes	328	86%	268	84%	60	95%
No, but I would like a genetic diagnosis	28	7%	27	8%	1	2%
No, but I did not want a genetic diagnosis	21	5%	21	7%	0	0%
I am waiting for genetic testing	3	1%	3	1%	0	0%
I am waiting for the results of genetic testing	3	1%	1	0%	2	3%
Total Responses	**383**	**100%**	**320**	**100%**	**63**	**100%**

Table 6 Time waited for genetic testing from referral, by time to diagnosis

	Overall		Less than 6 months		6-12 months		1-2 years		More than 2 years	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Less than 3 months	90	27%	53	50%	5	10%	6	22%	10	19%
3-6 months	81	24%	34	32%	20	42%	4	15%	9	17%
6-12 months	46	14%	3	3%	17	35%	9	33%	10	19%
1-2 years	9	3%	1	1%	0	0%	5	19%	2	4%
More than 2 years	20	6%	2	2%	1	2%	1	4%	11	21%
Don’t know / Can’t remember	89	27%	14	13%	5	10%	2	7%	11	21%
Total Responses	**335**	**100%**	**107**	**100%**	**48**	**100%**	**27**	**100%**	**53**	**100%**

Table 7 In the last two years, have you been able to access a Cardiologist when you needed to?

	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
Option	Count	Percent	Count	Percent	Count	Percent
Yes, always	190	51%	165	53%	25	40%
Yes, sometimes	111	30%	88	28%	23	37%
No, but I wanted or needed this	46	12%	35	11%	11	17%
I did not want or need this	23	6%	20	6%	3	5%
I accessed this privately	4	1%	3	1%	1	2%
Don't know / Can't remember	0	0%	0	0%	0	0%
Total Responses	**374**	**100%**	**311**	**100%**	**63**	**100%**

Table 8: Respondents who accessed NHS healthcare professionals privately in the last two years

Healthcare professional	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
	Count	Percent	Count	Percent	Count	Percent
Cardiologist	4	1%	3	1%	1	2%
Ear Nose Throat (ENT) specialist	8	2%	8	3%	0	0%
Gastrointestinal (GI) specialist	8	2%	7	2%	1	2%
Genetic specialist	1	0%	0	0%	1	2%
Mental health professional	13	4%	11	4%	2	3%
Neurologist	5	1%	4	1%	1	2%
Occupational therapist	3	1%	2	1%	1	2%
Ophthalmic (eye) specialist	15	4%	13	4%	2	3%
Orthotic services (e.g. prescription insoles, braces, splints, footwear)	14	4%	13	4%	1	2%
Osteopathy (physical manipulation, stretching and massaging muscles and joints)	**37**	**10%**	**37**	**12%**	**0**	**0%**
Pain specialist	6	2%	6	2%	0	0%
Physiotherapist	**25**	**7%**	**24**	**8%**	**1**	**2%**
Podiatrist (specialises in foot, ankle and leg healthcare)	**34**	**9%**	**32**	**11%**	**2**	**3%**
Rheumatology/Orthopaedic surgeon	3	1%	1	0%	2	3%

Table 9: Do you need to travel to more than one hospital/clinic to get the care you need for your Marfan or Loeys-Dietz Syndrome?

	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
Option	Count	Percent	Count	Percent	Count	Percent
Yes, and this bothers me	92	25%	76	25%	16	25%
Yes, but I do not mind	194	52%	161	52%	33	52%
No	84	23%	70	23%	14	22%
Total Responses	**370**	**100%**	**307**	**100%**	**63**	**100%**

Table 10: Travel to Multiple Hospitals or Clinics for Care, by Number of Symptoms Experienced

	Overall		No symptoms affect day-to-day life		Between 1-5 symptoms affect day-to-day life		Between 6-10 symptoms affect day-to-day life		More than 10 symptoms affect day-to-day life	
Option	Count	Percent	Count	Percent	Count	Percent	Count	Percent	Count	Percent
Yes, and this bothers me	92	25%	6	20%	20	16%	46	26%	20	53%
Yes, but I do not mind	194	52%	11	37%	66	52%	100	57%	17	45%
No	84	23%	13	43%	41	32%	29	17%	1	3%
Total Responses	**370**	**100%**	**30**	**100%**	**127**	**100%**	**175**	**100%**	**38**	**100%**

Table 11: Do you regularly see a cardiologist for your Marfan or Loeys-Dietz Syndrome? (by "cardiologist", we mean a doctor who specialises in heart and blood vessel diseases)

	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Yes, at least once a year	229	61%	185	59%	44	70%
Yes, at least once every 2 years	106	28%	95	30%	11	17%
No	38	10%	31	10%	7	11%
I'm waiting to attend my first appointment	4	1%	3	1%	1	2%
Don't know / Can't remember	0	0%	0	0%	0	0%
Total Responses	**377**	**100%**	**314**	**100%**	**63**	**100%**

Table 12 Respondents taking regular heart medication, by time since diagnosis

	Overall		Under 1 year ago		1-5 years		5-10 years		More than 10 years	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Yes, Beta Blocker (e.g. Bisoprolol, Atenolol)	220	59%	5	31%	32	52%	28	57%	154	62%
Yes, Angiotensin Receptor Blocker (ARB e.g. Irbesartan, Losartan)	179	48%	5	31%	34	56%	21	43%	119	48%
Yes, other heart medication(s)	111	30%	2	13%	12	20%	9	18%	87	35%
No, I haven't been offered them	48	13%	7	44%	11	18%	8	16%	22	9%
No, I stopped them due to side effects	13	3%	0	0%	1	2%	1	2%	11	4%
Don't know	3	1%	0	0%	1	2%	1	2%	1	0%
Total Responses	**375**	**100%**	**16**	**100%**	**61**	**100%**	**49**	**100%**	**248**	**100%**

Table 13: In the last two years, have you been given enough support for your mental health and wellbeing, such as counselling or psychotherapy? This may be support provided by the NHS or a local authority (such as school special educational needs and disabilities (SEND) provisions)

	Overall	
Option	**Count**	**Percent**
Yes, and it was helpful/appropriate	34	9%
Yes, but it was not helpful/appropriate	30	8%
No, I have not had any support but wanted this	143	38%
I did not want or need this	170	45%
Total Responses	**377**	**100%**

Table 14: Support for mental health and wellbeing among respondents who wanted or needed It

	Overall	
Option	**Count**	**Percent**
Yes, and it was helpful/appropriate	34	16%
Yes, but it was not helpful/appropriate	30	14%
No, but wanted this	143	69%
Total Responses	207	100%

Table 15 Access to mental health support, by region

		Given support and it was helpful	Given support but it was not helpful	Not given support, but wanted this	Total Responses
Overall	**Count**	34	30	143	**207**
	Percent	16%	14%	69%	**100%**
North East (England)	**Count**	4	2	2	**8**
	Percent	50%	25%	25%	**100%**
North West (England)	**Count**	8	3	11	**22**
	Percent	36%	14%	50%	**100%**
Yorkshire and The Humber	**Count**	1	1	9	**11**
	Percent	9%	9%	82%	**100%**
East Midlands (England)	**Count**	3	2	9	**14**
	Percent	21%	14%	64%	**100%**
West Midlands (England)	**Count**	1	0	13	**14**
	Percent	7%	0%	93%	**100%**
East of England	**Count**	1	3	12	**16**
	Percent	6%	19%	75%	**100%**
London	**Count**	0	3	8	**11**
	Percent	0%	27%	73%	**100%**
South East (England)	**Count**	6	7	32	**45**
	Percent	13%	16%	71%	**100%**
South West (England)	**Count**	2	7	28	**37**
	Percent	5%	19%	76%	**100%**
Scotland	**Count**	2	1	8	**11**
	Percent	18%	9%	73%	**100%**
Wales	**Count**	4	0	7	**11**
	Percent	36%	0%	64%	**100%**
Northern Ireland	**Count**	2	1	4	**7**
	Percent	29%	14%	57%	**100%**

Table 16 Which of the following types of surgery, if any, have you had because of your Marfan or Loeys-Dietz Syndrome? Please select all that apply

	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
Option	Count	Percent	Count	Percent	Count	Percent
Aorta surgery	159	43%	136	44%	23	37%
Back surgery	39	10%	36	12%	3	5%
Bowel surgery	14	4%	12	4%	2	3%
Dental surgery	56	15%	51	16%	5	8%
ENT (Ear, Nose, Throat) surgery	43	12%	38	12%	5	8%
Eye surgery	93	25%	89	29%	4	6%
Growth restriction surgery	7	2%	6	2%	1	2%
Heart valve surgery	101	27%	92	30%	9	14%
Lung surgery	19	5%	18	6%	1	2%
Orthopaedic surgery	53	14%	45	15%	8	13%
Pectus surgery	15	4%	14	5%	1	2%
Other (please specify)	68	18%	50	16%	18	29%
I have not had any surgery because of my Marfan or Loeys-Dietz	80	21%	59	19%	21	33%
Total Responses	**373**	**100%**	**310**	**100%**	**63**	**100%**

Table 17 When you were first diagnosed with Marfan or Loeys-Dietz Syndrome, was the information you were given from healthcare professionals about your condition useful?

	Overall		Less than 6 months		6-12 months		1-2 years		More than 2 years	
Option	Count	Percent	Count	Percent	Count	Percent	Count	Percent	Count	Percent
Yes, definitely	105	27%	51	41%	13	28%	9	28%	14	22%
Yes, to some extent	144	37%	40	32%	25	53%	14	44%	20	32%
No, the information was not useful	24	6%	6	5%	2	4%	4	13%	8	13%
I did not receive any information when I was diagnosed	70	18%	18	15%	6	13%	4	13%	18	29%
Don't know / Can't remember	43	11%	9	7%	1	2%	1	3%	3	5%
Total Responses	**386**	**100%**	**124**	**100%**	**47**	**100%**	**32**	**100%**	**63**	**100%**

Table 18 Information from healthcare professionals about condition and ongoing treatment, by number of symptoms experienced

	Overall		No symptoms affect day-to-day life		Between 1-5 symptoms affect day-to-day life		Between 6-10 symptoms affect day-to-day life		More than 10 symptoms affect day-to-day life	
Option	Count	Percent	Count	Percent	Count	Percent	Count	Percent	Count	Percent
Yes, enough information	85	23%	13	41%	37	29%	29	17%	6	16%
Some, but not enough information	157	42%	8	25%	56	44%	73	42%	20	53%
Little or no information	127	34%	10	31%	35	27%	70	40%	12	32%
Don't know / Can't remember	3	1%	1	3%	0	0%	2	1%	0	0%
Total Responses	**372**	**100%**	**32**	**100%**	**128**	**100%**	**174**	**100%**	**38**	**100%**

Table 19 Do you think healthcare professionals do everything they can to help control your pain?

	Overall	
Option	**Count**	**Percent**
Yes, always	20	8%
Yes, sometimes	129	51%
No, never	87	35%
I am not in any pain due to Marfan or Loeys-Dietz Syndrome	15	6%
Total Responses	**251**	**100%**

Table 20: The proportion of respondents able to access a Gastrointestinal (GI) specialist when they needed to, by condition.

	Overall		Marfan Syndrome		Loeys-Dietz Syndrome	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Yes, always	18	5%	17	6%	1	2%
Yes, sometimes	45	12%	37	12%	8	14%
No, but I wanted or needed this	75	21%	52	17%	23	39%
I did not want or need this	211	58%	187	62%	24	41%
I accessed this privately	8	2%	7	2%	1	2%
Don't know / Can't remember	6	2%	4	1%	2	3%
Total Responses	**363**	**100%**	**304**	**100%**	**59**	**100%**

Table 21 Impact of MFS or LDS on mobility, by number of symptoms experienced

	Overall		No symptoms affect day-to-day life		Between 1-5 symptoms affect day-to-day life		Between 6-10 symptoms affect day-to-day life		More than 10 symptoms affect day-to-day life	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Not affected at all / Not applicable	83	23%	15	83%	48	38%	19	11%	1	3%
Somewhat affected	154	44%	2	11%	68	53%	78	46%	6	16%
Extremely affected	117	33%	1	6%	12	9%	73	43%	31	82%
Total Responses	**354**	**100%**	**18**	**100%**	**128**	**100%**	**170**	**100%**	**38**	**100%**

Table 22 Impact of MFS or LDS on employment, by number of symptoms experienced

	Overall		No symptoms affect day-to-day life		Between 1-5 symptoms affect day-to-day life		Between 6-10 symptoms affect day-to-day life		More than 10 symptoms affect day-to-day life	
Option	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**	**Count**	**Percent**
Not affected at all / Not applicable	184	54%	18	95%	90	74%	68	42%	8	24%
Somewhat affected	63	19%	0	0%	21	17%	37	23%	5	15%
Extremely affected	91	27%	1	5%	11	9%	58	36%	21	62%
Total Responses	**338**	**100%**	**19**	**100%**	**122**	**100%**	**163**	**100%**	**34**	**100%**

Table 23: In the last two years, have you needed any of the following benefits and/or financial support as a result of your Marfan or Loeys-Dietz Syndrome?

	Employment and Support Allowance		Universal Credit		Carers Allowance		Personal Independence Payment (PIP)		Disability Living Allowance (DLA)		Statutory Sick Pay (SSP)		Pension Age Disability Payment/Attendance Allowance		Local authority or council funding for care needs	
Option	**N**	**%**	**N**	**%**	**N**	**%**	**N**	**%**	**N**	**%**	**N**	**%**	**N**	**%**	**N**	**%**
Yes, and I have received this	39	12%	50	15%	25	8%	101	30%	37	11%	43	13%	11	3%	27	8%
Yes, but I have not received this	13	4%	7	2%	8	2%	24	7%	20	6%	9	3%	3	1%	14	4%
No, but I don't need this / Not applicable	215	65%	238	71%	250	76%	172	50%	210	63%	248	75%	271	82%	229	69%
I did not know this was available	61	18%	38	11%	42	13%	40	12%	58	17%	22	7%	38	12%	56	17%
Don't know / Can't remember	5	2%	3	1%	5	2%	4	1%	8	2%	9	3%	7	2%	6	2%
Total Responses	**333**	**100%**	**336**	**100%**	**330**	**100%**	**341**	**100%**	**333**	**100%**	**331**	**100%**	**330**	**100%**	**332**	**100%**

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