Picker

Influence • Inspire • Empower

Exploring the lived experiences of people with Marfan and Loeys-Dietz Syndrome

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.

Delivering high-quality, person centred care requires understanding patients' views and experiences. However, 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

Picker developed the survey in stages to make sure it captured the voices of people affected by MFS and LDS:

- The team conducted a scoping review of recent literature.
- Five in-depth interviews were carried out with people living with, or caring for, someone with MFS or LDS. The aim was to explore current care needs and experiences, and to identify what matters most to people when receiving care.
- A draft questionnaire was developed based on the scoping review and in-depth interviews.
- Five cognitive interviews were conducted to test the draft questionnaire and check that it was clear, accessible and relevant to respondents.

The survey was conducted online and promoted through the Marfan Trust's networks, with paper questionnaires also mailed to 200 Trust members. Eligible participants were UK residents aged 16 or over who either had Marfan syndrome or Loeys-Dietz syndrome, or supported someone with one of these conditions. Data were collected between September and November 2025.

Quantitative data were analysed using descriptive statistics. Free text responses were analysed thematically, and findings were brought together in a final report with recommendations for the Marfan Trust and healthcare services for people with MFS and LDS.

About the survey respondents

395 respondents

Marfan = 84%
Loeys-Dietz = 16%

Respondent type

Person living with MFS or LDS = 76%

Carer = 24%

Gender

Female = 64%

Male = 36%

Age (years)

16-24 = 3%
25-35 = 10%
36-50 = 30%
51-70 = 44%
71+ = 13%

Country

England = 88%
Scotland = 6%
Wales = 4%
N. Ireland= 2%

Ethnicity

White ethnic groups = 92%

Minority ethnic groups = 8%

Findings: Lack of awareness and fragmented care

Lack of knowledge amongst healthcare professionals and poor experiences of care coordination were commonly reported. **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.

26%

said that before their diagnosis their health concerns were not taken seriously by healthcare professionals

56%

did not have a named healthcare professional overseeing their care but wanted this support

67%

'sometimes' or 'often' had to arrange their own care

53%

felt that the different healthcare professionals involved in their care did not work well together.

Findings: Gaps in access to specialist services

- 86% of respondents had received a genetic diagnosis and 81% were *always* or *sometimes* able to access a cardiologist when needed.

Current guidelines recommend that people with MFS or LDS are annually monitored with an echocardiogram (ultrasound of the heart).

- 59% said they have this scan every six months or annually suggesting that some patients may not be receiving the recommended level of monitoring.

While cardiologists were generally accessible, some gaps exist in access to other healthcare specialists:

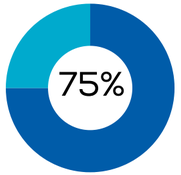
- 69% of those seeking mental health support, did not receive enough help
- 40% reported an unmet need for pain management services
- 34% wanted osteopathy but were unable to access this
- 25% indicated an unmet need for podiatry.

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

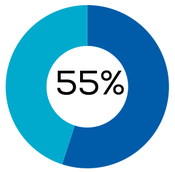
Findings: Gaps in information provision

When first diagnosed, 64% of respondents said the information they were given from healthcare professionals about their condition was either *definitely* or *somewhat* useful.

However, the survey showed a gap in ongoing 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.



did not receive enough information about their condition and ongoing treatment, with many turning to the Marfan Trust website, social media or online peer groups for guidance.



said they did not get enough personalised advice from healthcare professionals about physical activity, despite wanting this support.

Findings: Impact on quality of life

More than half of respondents reported that the following symptoms had affected their daily life in the past 12 months:

- Fatigue (87%)
- Chronic pain (72%)
- Gut problems (58%)
- Headache / migraine (55%)
- Joint hypermobility (52%)

The majority 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, such as excercising, mobility and confidence and self esteem.

46% of respondents reported that their employment had been affected by MFS or LDS. Those experiencing more symptoms were significantly more likely to be on long-term sick leave or retired due to ill health.

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

Turning findings into action

The survey findings have helped the Marfan Trust to better understand the needs of its community and make practical changes to awareness-raising, information provision and patient support.

Raising awareness among healthcare professionals

Survey findings have been shared at a Genetic Alliance meeting and the Annual Aortic Nurses Symposium. Further engagement is planned with groups such as the Royal College of GPs and the British Inherited Cardiac Conditions Society.

Strengthening access to specialist support

The Trust is fundraising to work with Rare Minds, an organisation that provides tailored counselling and mental health support for people affected by rare conditions. A pilot is being developed to test whether this approach meets the needs of the MFS and LDS community and whether it should be considered for longer-term investment.

Improving access to trusted information

QR code cards linking to the Trust's website and leaflets are being produced for distribution in aortopathy and genetic clinics. The Trust has achieved Patient Information Forum quality standards certification and will be added to the TRIP clinical search database. The Trust is collaborating with HCI Digital to develop introductory MFS and LDS videos for use across NHS settings.

Responding to quality of life challenges

The Trust has introduced a dietitian-led webinar on gut health and an Empowered Relief webinar, which teaches practical pain management techniques and is supported by evidence of effectiveness for people with MFS and LDS experiencing pain and fatigue.

Client testimonial

“At the Marfan Trust we receive hundreds of calls and emails to our helpline every year and many highlight issues with the lesser known, invisible symptoms of these rare conditions, symptoms like chronic pain, fatigue, the mental load of living with uncertainty. These survey results have given us an invaluable insight to our member's lived experiences and provided the evidence required to push for change and service development to meet those needs.

The Picker team were great throughout the project; they listened carefully to our priorities and responded well to them. The work was collaborative and very professional, their expertise in the development of the questionnaire was really helpful. All the deadlines we discussed were met and they provided regular feedback and prompt answers to any questions we had”.

Marfan Trust
Supporting Research into Inherited Aortic and Related Disorders

Loeys Dietz

CONNECTING THE UNCONNECTED AND STRENGTHENING LIVES

Picker

Address

Picker Group Suite 6,
Fountain House 1200
Parkway Court John
Smith Drive Oxford
OX4 2JY

Contact details

+44 (0)1865 208100
picker.org
PickerInstituteEurope
pickereurope
@pickereurope.bsky.social

Registered Charity in England and Wales: 1081688
Registered Charity in Scotland: SC045048 Registered
Company Limited by Guarantee: 0390816