

LIVED EXPERIENCE: INSIGHTS INTO BARTH SYNDROME

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Introduction:

- Barth syndrome is an ultra-rare, X-linked recessive genetic condition that mostly affects males, occurring in around 1 in every 1 million live male births. It is caused by mutations in the *TAFAZIN* gene, leading to structurally abnormal mitochondria that cannot generate energy efficiently^{1,2}.
- Barth syndrome affects multiple body systems; the most common are cardiac manifestations including dilated cardiomyopathy, congestive heart failure and arrhythmias. In addition to this, people with Barth syndrome often experience muscle weakness, fatigue, neutropenia, and gastrointestinal/feeding problems^{1,3}.
- Until recently, treatment for Barth syndrome was supportive, managed by conventional drugs or heart transplants, where necessary. In 2025, a new treatment for Barth syndrome called elamipretide was approved in the USA. Elamipretide is now being considered for the English market^{4,5,6}.

Aim: To gain insight into the lived experience of people with Barth syndrome

Methods:

- A digital community-facing survey was conducted by Barth Syndrome UK, Barth Syndrome Foundation and Metabolic Support, which ran between 20/07/2026 and 25/08/2026 in the USA and 05/08/2026 and 23/08/2026 in the UK.
- Questions were designed based on the NICE patient organisation submission template and in-house Barth syndrome knowledge.
- The survey was disseminated via social media, newsletters, and targeted outreach.
- Data were analysed using a mixed methods approach using both descriptive statistics and thematic content analysis. EQ-5D-5L data were analysed in line with the guidance set out by the EuroQol Research Foundation and index values were calculated using the UK EQ-5D-5L value set^{7,8}.

Results:

- 63 responses were received and included adults (18+) living with Barth syndrome (n=24, 38.1%), parents/carers of someone living with Barth syndrome (n=35, 55.6%) and parents/carers of a child living with Barth syndrome who had passed away (n=4, 6.4%).
- 27 respondents were from the United Kingdom (23 England, 2 Scotland, 2 Wales), 36 respondents were from the USA.
- 61 affected individuals were male and 2 affected individuals were female.
- 24 respondents had experience with elamipretide (UK, n=3; USA, n=21).



Child with Barth syndrome

Diagnosis and key symptoms

The majority of people reported that individuals started **showing symptoms of Barth syndrome at birth or before (60.3%)**. Only a small proportion of individuals started showing **symptoms after 1 year of age (11.1%)**.

All respondents had experienced at least one cardiovascular symptom, with nearly **94% experiencing cardiomyopathy** and **19% requiring at least 1 heart transplant**.

People living with Barth syndrome have **high levels of fatigue**; **69.3% of children** were classed as **moderately or severely fatigued**, **46.7% of adults** were classed as **moderately or severely fatigued**.

'Fatigue is the symptom of Barth syndrome I feel affects my life the most. I've had to accept long ago that I just wasn't going to be able to do whatever I wanted to do in life' – Adult living with Barth syndrome, UK

The majority of people were **diagnosed with Barth syndrome before they turned six (80.6%)**, with **half** of this group (40.3% of the total) being diagnosed before they turned one.

'He was rushed to A&E for being floppy, grunting & failing to feed. They saw an enlarged heart & rushed us to [...] hospital. He was then diagnosed with DCM. The genetics team later spotted his tafazzin levels and suspected Barth. Genetic Blood tests later confirmed this.' – Parent of a child living with Barth syndrome, UK

Patient experience with elamipretide

38% of respondents had **experience with elamipretide**. Of the 27 respondents from the **UK**, **3** had experience with elamipretide (n=2 via compassionate use and n=1 via clinical trial).

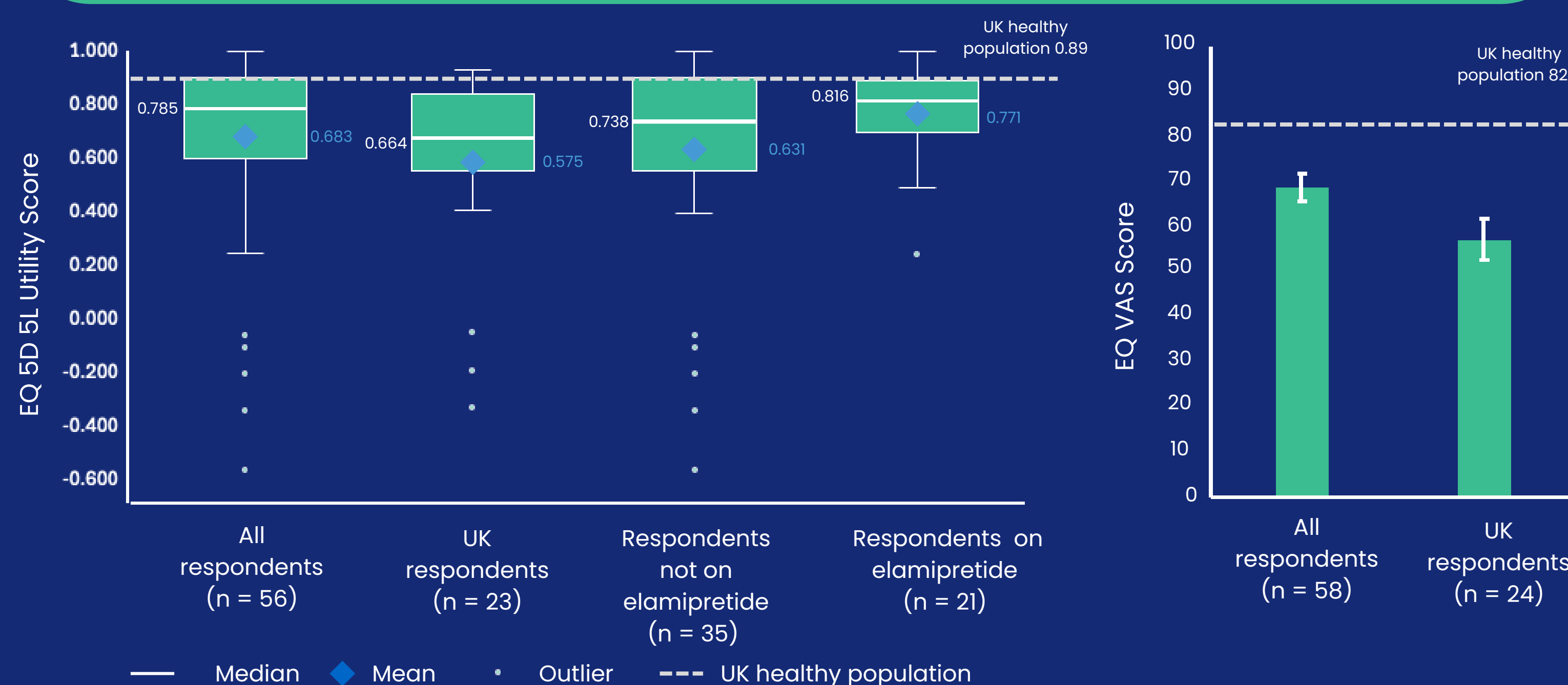
79.1% of respondents reported a **benefit to taking elamipretide**. The most common self-reported benefits were **increased stamina** (n=9; 37.5%), **increased strength** (n=7, 29.1%) and **more energy** (n=5, 20.8%).

54.1% of people reported an **injection site reaction** as a side effect to taking elamipretide and **4.2%** of people reported **injection site scarring** as a side effect of taking elamipretide. **45.8%** of people reported **no side effects** to taking elamipretide.

81.8% (n=18/22) of people said that elamipretide had **met their expectations**. **95.8%** (n=23/24) of people said elamipretide was **easy to administer**. **95.8%** (n=23/24) of people said they would **recommend elamipretide**.

'I'm not sure I would still be here without this medication. Before I was taking this medication I was progressively getting worse. I didn't want to go out with friends and didn't want to live a life where I couldn't perform everyday activities. This medication allows me to do that and so much more.'
– Adult living with Barth syndrome, USA

EQ-5D-5L data confirmed reduced quality of life compared with general population norms



The **average EQ-5D-5L score from UK participants was lower than the UK average** for healthy adults (0.89, a higher score indicates better quality of life). The average EQ-5D-5L score from participants on elamipretide was lower than the UK average for healthy adults but higher than participants who do not receive elamipretide.

Conclusions:

This survey captures perspectives from a significant portion of the UK and USA Barth syndrome population, offering rare and valuable insights into daily life with this ultra-rare condition. The data shown here show that Barth syndrome greatly impacts the quality of life of affected individuals due to high fatigue, cardiac impairment, caregiver stress (data not shown) and healthcare resource utilisation.

Acknowledgements:

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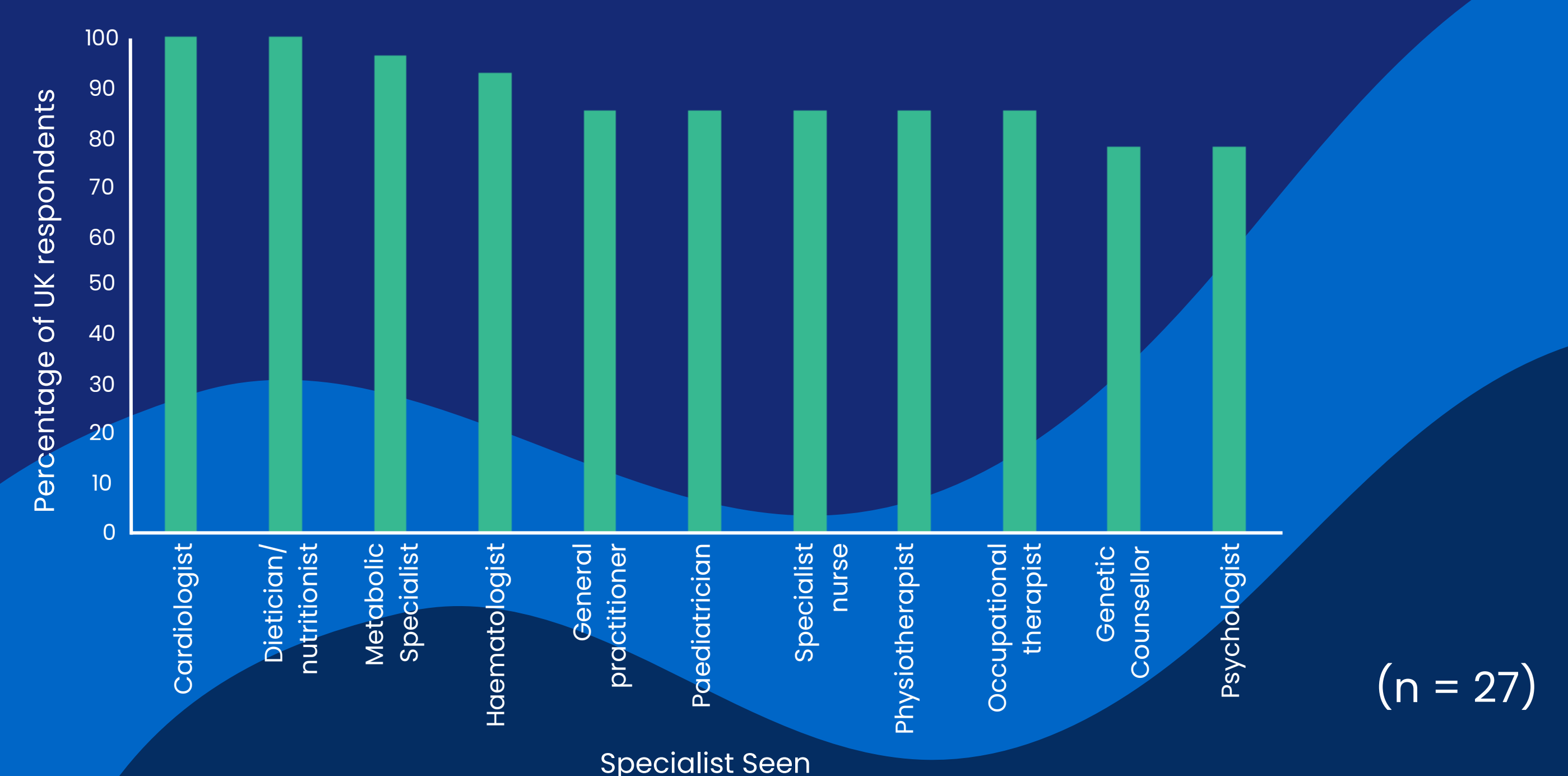
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Healthcare resource use is high in people living with Barth syndrome

On average people with Barth Syndrome in the UK had **12.9 outpatient appointments in the last year** (n=25, median: 10, range 5 to 50)

On average people with Barth Syndrome in the UK **see 11.4 different specialists** (n=27, median: 11, range 5 to 17)



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