The impact of Fabry symptoms on patients' quality of life (QoL) and mental health - a qualitative interview study in the UK

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Introduction

Fabry is an incurable, debilitating, and progressive disease. Deficiency in the enzyme alpha-galactosidase A, leads to the accumulation of globotriaosylceramide and its derivatives within lysosomes in a variety of cells throughout the body (1). This buildup in endothelial, cardiac, and nervous tissues causes a progressive loss of organ function and can lead to kidney failure, heart disease and stroke (2). Additionally, patients can experience a wide range of symptoms that afftect their quality of life and mental health.

Neuropsychiatric symptoms, notably depression and cognitive impairment, are prevalent and deficits in social functioning and adaptation, and particular coping styles also contribute to the psychological burden on patients (3).

Aim

Our study aimed to explore the physical and emotional burden of disease and how these affect patients' lives.

Methods

This qualitative study consisted of a screener to check for eligibility, a short questionnaire to collect demographics and a semi-structured in-depth interview.

The sampling method was convenient and purposeful: members of the MPS Society identified to include a representation of the patient population regarding demographics, phenotype and treatment.

- To be eligible, participants had to be: a patient or the parent or caregiver of a person with a confirmed diagnosis of Fabry disease, aged ≥ 18 years, a resident of the UK, able to provide informed consent to participate.

Patients on a clinical trial for a treatment not approved in the UK were excluded. The study was conducted in accordance with the British Healthcare Business Intelligence Association's Legal & Ethical Guidelines for Market Research.

Interviews followed a semi-structure discussion guide and were audio recorded and transcribed. The transcripts were analysed using an inductive thematic approach and NVivo software (4).

Acknowledgements

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References

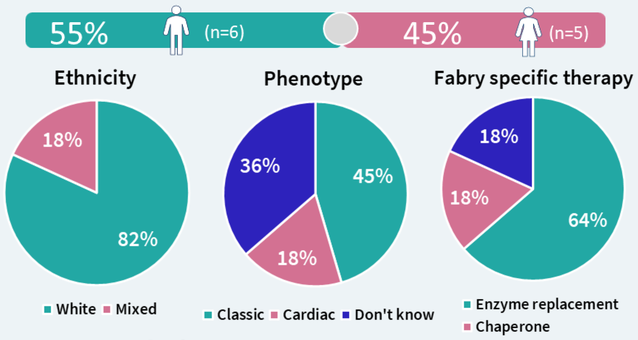
1.Mehta A, Hughes DA. Fabry Disease. In: Adam MP, et al., editors. GeneReviews®. Seattle (WA) 1993–2024. 2.Germain DP. Fabry disease. Orphanet J Rare Dis. 2010; 5:30. 3.Mroczek M, et al. Neuropsychiatric Symptoms and Their Association With Sex, Age, and Enzyme Replacement Therapy in Fabry Disease: A Systematic Review. Front Psychiatry. 2022; 13:829128. 4.Braun V, Clarke V. Reflecting on reflexive thematic analysis. Qualitative Research in Sport, Exercise and Health. 2019; 11(4):589-97.

Participants

- 14 potential participants were contacted and 12 responded
- 11 met the inclusion criteria and completed an interview
- All were resident in England
- Two participants were parents or caregivers
- 9 participants were patients with Fabry

Patient demographics

- Median age 52 years (mean 45.5 ± 21.6, range 12–75)
- Median age at diagnosis 40 years (mean 34.7 ± 22.2, range 2–67)



Burden of disease

The symptom burden of Fabry significantly affected the daily lives and emotional wellbeing of all participants. The most common symptoms were **pain, chronic fatigue, and gastrointestinal (GI) issues**.

Symptoms can disrupt sleep and make physical activity difficult. They affected patients' ability to perform daily tasks, go out of their homes, drive and socialise. They impacted the employment options open to adult patients and children's school attendance, affecting academic performance. Financial burdens were associated with the ability to work and keeping the home warm in winter to avoid increased pain associated with cold temperatures. Patients had to modify their lifestyle, allowing time for rest and to avoid symptom triggers.

Life goals and plans had to be reevaluated in terms of what was possible with Fabry and this could affect family dynamics as the patients' role within the family changed. **Anxiety, stress, frustration** and **depression** were reported in response to physical symptoms, the limits imposed on patients by Fabry disease and the resulting social isolation.

Impact of treatment

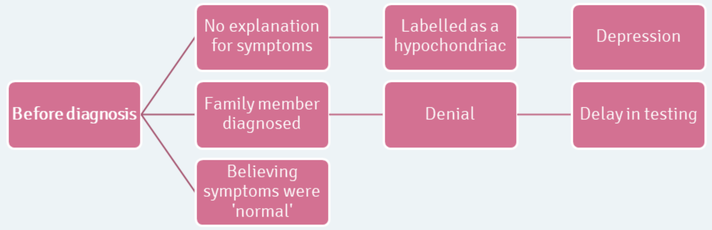
Fabry specific treatment (enzyme replacement or chaperone therapy) improved quality of life by making symptoms more manageable and gave emotional relief to patients by slowing disease progression.

Unmet needs

Patients expressed the need for more treatment options, support for the impacts of Fabry disease on daily life and the importance of mental health services.

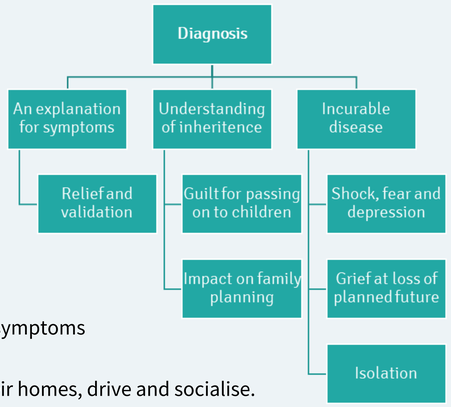
Emotional impacts before diagnosis

Patients may experience symptoms for years before they receive a Fabry diagnosis. Young patients may not recognise symptoms that have always been a part of their experience. Others face difficulties of not being believed or taken seriously when they report symptoms. When a family member is diagnosed, patients may experience denial and delay testing, particularly if they have no symptoms.



Emotional impacts of diagnosis

Most patients with Fabry disease were diagnosed following the diagnosis of a family member, but the majority had experienced Fabry symptoms prior to diagnosis. Emotions were mixed as they learnt about Fabry disease and the implications of their diagnosis.



Conclusions

The findings of this study reveal that Fabry disease symptoms greatly affect the daily lives of patients, including their ability to work, study, and socialize. Participants expressed a range of emotions, which varied depending on their diagnosis, treatment status, and symptoms. While some found relief after receiving a diagnosis, others struggled to come to terms with the implications of having an incurable genetic disease, particulary if they were asymptomatic. Post-diagnosis, the emotional impact of Fabry disease is further influenced by the severity of symptoms and how they affect patients' ability to live their lives as they wish.

By understanding the impact of Fabry disease on patients' daily lives and emotional well-being, healthcare providers can identify areas for improvement in symptom control and psychological care. This will ultimately enhance patients' quality of life.