

The impact of COVID-19 on Fabry patients receiving enzyme replacement therapy

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Introduction

Fabry is a rare, life-limiting disease characterised by deficiency of the enzyme α -galactosidase A. Enzyme replacement therapy (ERT) is one of the available treatment options for patients in the UK, administered via bi-weekly infusions, usually in the patients' own home. Breaks in treatment are rare and therefore information on the impact of treatment withdrawal on patients is sparse.

During the first wave of the COVID-19 pandemic in the UK, some Fabry patients who usually receive ERT were placed on a shielded patients list and were not able to access treatment as a result.¹

Aim

The aim of the study was to determine **the impact of treatment breaks on the patients' experience of Fabry symptoms.**

Methods

Data was collected from respondents over three research activities:

- Treatment break online symptom diary, April 2020
- Effects of treatment break online survey, December 2020
- Semi-structured follow up telephone interviews, June 2021

Respondents

- Data was collected from nine respondents overall: symptom diary (n=9), treatment break survey (n=8) and semi-structured telephone interviews (n=4)
- All respondents were female
- Median age, 57 years (range, 30–74 years)
- ERT treatment was with agalsidase alfa (78%) or agalsidase beta (22%)

Conclusions

The findings suggest that even short term pauses in ERT treatment can have a negative impact on pre-existing symptoms of Fabry disease, particularly fatigue and lack of energy, and may lead to the occurrence of new symptoms. While some symptoms improved when treatment was re-started, some respondents reported a continued impact of their treatment break after re-starting ERT.

References

1. GOV.UK. Coronavirus (COVID-19), Health and wellbeing during coronavirus 2021 [updated 2021 Feb 25; cited 2021 Mar 12]. Available from: <https://www.gov.uk/government/publications/guidance-on-shielding-and-protecting-extremely-vulnerable-persons-from-covid-19>

Results

Treatment breaks

- For the majority of respondents (56%), ERT stopped in March 2020
- The median length of treatment break was 3 months (range 1–7 months)

Pre-existing symptoms

- 89% of respondents reported that they usually experienced some symptoms associated with their Fabry disease

67% of respondents reported a worsening to some of their pre-existing Fabry symptoms during their time off treatment

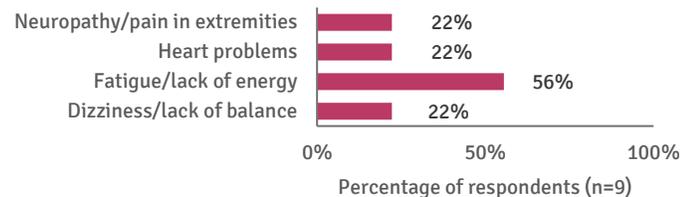


Figure 1. Most reported pre-existing symptoms that worsened during the treatment break

Heart symptoms experienced during the treatment break included a worsening of palpitations necessitating the fitting of a heart loop monitor and an episode of pericarditis that resulted in hospitalisation.

- Both respondents were still affected by an increase in palpitations after re-starting ERT.

Table 1. Worsening pre-existing symptoms, respondent comments

Symptom	Respondent comments
Fatigue/lack of energy	"... I can usually get myself out of bed and get myself dressed. But when I was on the treatment break, just to give you an example, last year I spent probably from April to November, most of that time was either in bed at home, or in bed in hospital"
Heart problems	"I got a really bad episode of pericarditis, the worst that I've ever had. And it just took hold of me, and I was ill for months with it, and it only started to really improve when I started back on the ERT"

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New symptoms

44% of respondents reported a new symptom that first occurred during their time off treatment

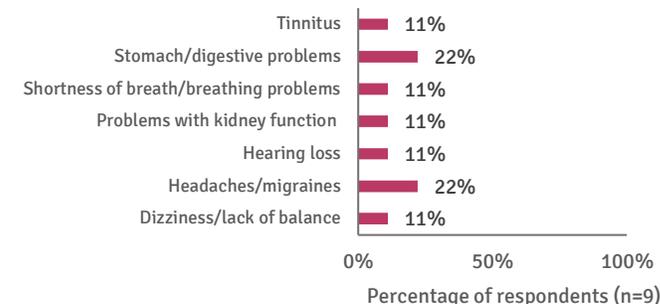


Figure 2. Reported new symptoms first experienced during the treatment break

Table 2. New symptoms, respondent comments

Symptom	Respondent comments
Stomach/digestive problems	"Never had digestive or stomach issues until the treatment break. Now have problems eating and have lost several kilograms in weight", "Frequent heartburn, colic"

Use of additional medication

56% of respondents took additional medication during their treatment break

- 44% used additional medications to treat headaches/migraines and pain, medications used included paracetamol, aspirin, codeine and tramadol
- **22% of respondents reported the use of morphine to treat pain**
- 22% of respondents increased their use of common over the counter remedies for stomach/digestive problems, such as peppermint oil

Returning to treatment

Respondents who took part in telephone interviews (n=4) had all resumed ERT treatment for a median of 10 months (range, 9–12 months). This had a varied effect on their symptoms:

- One respondent felt their symptoms had returned to the same levels as before the treatment break
- For the remaining three respondents some symptoms, such as headaches/migraines, had returned to their usual state, but other symptoms had remained worse than before the treatment break
- Symptoms that remained worse included neuropathy/pain in extremities, stomach/digestive problems and heart problems