

Patient experience with Fabry disease and its management in the real world: a survey of 280 patients with Fabry disease

Supplemental information to Berry L et al. *Orphanet Journal of Rare Diseases*. 2024.

This is a summary of an article about the patient experience with Fabry disease, including symptoms and treatment, which was published in a medical journal called *Orphanet Journal of Rare Diseases* in 2024.

How to say...

Agalsidase alfa: "AY-gal-suh-days AL-fuh"

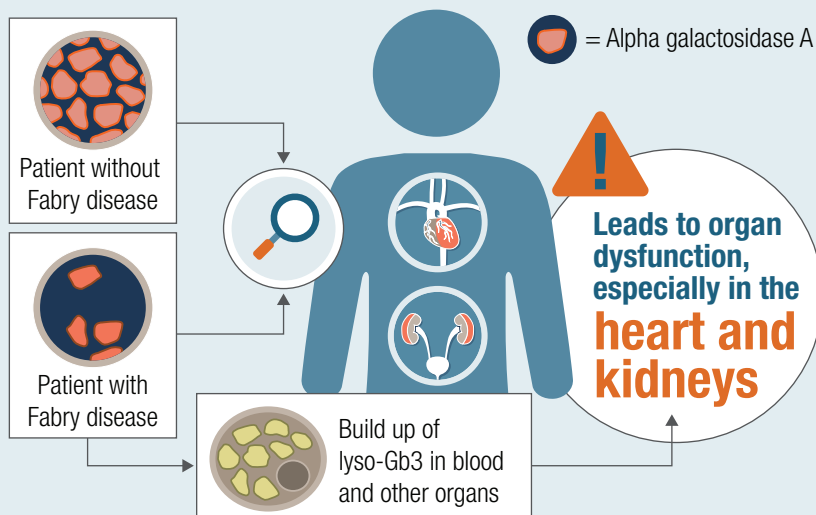
Agalsidase beta: "AY-gal-suh-days bay-tuh"

Alpha galactosidase: "al-fuh guh-lak-tow-suh-days"

Fabry: "FAB-ree"

What is Fabry disease?

Fabry disease is a rare, inherited disorder in which patients have a missing, decreased, or nonworking enzyme in their cells. In healthy cells, enzymes are molecules that speed up chemical reactions in the body, such as reactions that create or break down other substances. Patients with Fabry disease have little or no alpha galactosidase A enzyme in their body. As a result, their cells cannot break down large molecules, such as lyso-Gb3. This leads to a build-up of these large molecules in the blood and organs throughout the body. Over time, this causes different symptoms in patients with Fabry disease, such as nerve pain, stomach pain, diarrhoea, fatigue, decreased sweating, hearing loss, and eye problems. Symptoms may get worse over time, and organs such as the heart and kidneys may not function properly. **Fabry disease affects both men and women and is a lifelong disorder.**



What treatments are available for patients with Fabry disease?

Currently, there are 2 kinds of treatments available for patients with Fabry disease called **enzyme replacement therapy** and **chaperone therapy**. Enzyme replacement therapy replaces the missing enzyme in patients with Fabry disease so their cells can break down lyso-Gb3. At the time of this study, there were 2 medications available for enzyme replacement therapy for patients with Fabry disease: agalsidase alfa and agalsidase beta. A third medication for enzyme replacement therapy, pegunigalsidase alfa-ixwj, was approved in Europe and the United States after this study was completed. All 3 medications are injected directly into the bloodstream by a slow infusion given every 2 weeks.

The other type of treatment, chaperone therapy, works by binding to the nonworking enzyme to restore some of the normal activity. The medication used for chaperone therapy is called migalastat and is taken by mouth every other day. Migalastat can only be used by patients with certain gene changes causing their Fabry disease.



Why did researchers conduct this survey?

There are not many studies that have surveyed patients directly about their disease and treatment experience.

This study was conducted to find out how patients with Fabry disease feel about their disease severity, symptoms, monitoring, and treatment.

How was this survey conducted?

Researchers created a 30-minute online survey that had 33 questions. The questions were based on input from healthcare providers who treat patients with genetic diseases and from Fabry disease patient advocacy groups. The survey was completed by patients with Fabry disease who were 18 years or older and could understand English.

Who completed this survey?



280 patients with Fabry disease in the **US** and **Canada** completed the survey



Treatment history

Currently on any type of treatment:

84% (234 of 280 patients; women: 78% [149 of 191]; men: 95% [84 of 88]; nonconforming: 100% [1 of 1])

Currently on enzyme replacement therapy:

74% (208 patients)



Sex

Women:

68% (191 patients)

Men:

31% (88 patients)

Nonconforming:

<1% (1 patient)



Age

Range: 18 to 77 years

Average: 47 years



Work status

Full or part time: 54% (152 of 280 patients;

women: 50% [95 of 191]; men: 65% [57 of 88])

Retired with or without disability: 29% (82 patients)

Student: 5% (15 patients)

1 What type of doctor primarily managed each patient's Fabry disease?

Doctor Type	Percentage
Specialist in genetic diseases	46%
Kidney specialist	23%
Family doctor	16%
Heart specialist	7%
Other	4%
Brain specialist	3%
No one	2%

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