

20 April 2026

Treatment of Rare Diseases

1. Within your Trust, how many patients currently have a diagnosis for:

Paediatric Service:

- Fabry(-Anderson) disease (ICD10 code E75.21) 9
- Gaucher disease (ICD10 code E75.22) 0
- Pompe disease (ICD10 Code E74.02) <5
- Pompe disease (ICD10 Code E74.02) infantile onset (patients diagnosed before age 1) 0
- Hunter syndrome (MPS II) (ICD10 code E76.1) <5

Adult Service

- Fabry(-Anderson) disease (ICD10 code E75.21) 58
- Gaucher disease (ICD10 code E75.22) <5
- Pompe disease (ICD10 Code E74.02) <5
- Pompe disease (ICD10 Code E74.02) infantile onset 0

2. Of the patients above, how many patients have been newly diagnosed within the past 3 months for:

Paediatric Service:

- Fabry(-Anderson) disease (ICD10 code E75.21) 0
- Gaucher disease (ICD10 code E75.22) 0
- Pompe disease (ICD10 Code E74.02) <5
- Pompe disease (ICD10 Code E74.02) infantile onset (patients diagnosed before age 1) 0
- Hunter syndrome (MPS II) (ICD10 code E76.1) 0

Adult Service

- Fabry(-Anderson) disease (ICD10 code E75.21) 0
- Gaucher disease (ICD10 code E75.22) 0
- Pompe disease (ICD10 Code E74.02) <5
- Pompe disease (ICD10 Code E74.02) infantile onset 0

3. How many patients have been treated in the last 3 months with the following products:

- Replagal (agalsidase alfa) <5
- Fabrazyme (agalsidase beta) 12

20 April 2026

• Galafold (migalastat)	11
• Elfabrio (pegunigalsidase alfa)	<5
• VPRIV (velaglucerase alfa)	0
• Cerezyme (imiglucerase)	<5
• Cerdelga (eliglustat)	0
• Zavesca (miglustat)	<5
• Myozyme (alglucosidase alfa)	0
• Nexviadyme (avalglucosidase alfa)	0
• Pombiliti + Opfolda (cipaglucosidase alfa + miglustat)	0

Use of <5 (less than five): We are unable to provide an exact figure - exempt from release under Section 40(2) of the Freedom of Information Act - Personal Information relating to a third party, as the small numbers involved could make patients personally identifiable. Disclosure would constitute a breach of the principles of the General Data Protection Regulation 2018.

4. **Do you participate in any clinical trials for Fabry(-Anderson) disease? If so, can you please provide the name of each trial along with the number of patients taking part?**

Paediatric Service:

No.

Adult Service:

Adult Service registry participation only with Follow Me registry (#23) and Genzyme Rare Disease Registry ~49.

5. **Do you participate in any clinical trials for Gaucher disease? If so, can you please provide the name of each trial along with the number of patients taking part?**

Paediatric Service:

No.

Adult Service:

No