

9 August 2024

Rare Diseases

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

Adult Services:

- Fabry Disease (ICD10 code E75.21) 54
- Gaucher Disease (ICD10 code E75.22) <5
- Pompe Disease (ICD10 Code E74.02) <5
- Pompe Disease (ICD10 Code E74.02) Infantile-onset (Patients Diagnosed before age 1) 0
- MPS II (Hunter Syndrome) (ICD10 code E76.1) <5

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, 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

Paediatric Services:

- Fabry Disease (ICD10 code E75.21) 8
- Gaucher Disease (ICD10 code E75.22) 0
- Pompe Disease (ICD10 Code E74.02) 0
- Pompe Disease (ICD10 Code E74.02) Infantile-onset (Patients Diagnosed before age 1) 0
- MPS II (Hunter Syndrome) (ICD10 code E76.1) <5

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

Adult Services:

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

Paediatric Service:

- Fabry Disease (ICD10 code E75.21) 0
- Gaucher Disease (ICD10 code E75.22) 0
- Pompe Disease (ICD10 Code E74.02) 0
- Pompe Disease (ICD10 Code E74.02) Infantile-onset (patients diagnosed before age 1) 0
- MPS II (Hunter Syndrome) (ICD10 code E76.1) 0

9 August 2024

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

Adult Services:

• Replagal (agalsidase alfa)	<5
• Fabrazyme (algalsidase beta)	13
• Galafold (migalastat)	14
• Elfabrio (pegunigalsidase alfa)	0
• VPRIV (velaglucerase alfa)	<5
• Cerezyme (imiglucerase)	<5
• Cerdelga (eliglustat)	0
• Zavesca (miglustat)	<5 but not for the above indications.
• Myozyme (alglucosidase alfa)	<5
• Nexviazyme (avalglucosidase alfa)	0
• AT-GAA (cipaglucosidase alfa/ miglustat)	0

Paediatric Service:

• Replagal (agalsidase alfa)	0
• Fabrazyme (agalsidase beta)	0
• Galafold (migalastat)	0
• Elfabrio (pegunigalsidase alfa)	0
• VPRIV (velaglucerase alfa)	0
• Cerezyme (imiglucerase)	0
• Cerdelga (eliglustat)	0
• Zavesca (miglustat)	<5 but not for the above indications
• Myozyme (alglucosidase alfa)	0
• Nexviadyme (avalglucosidase alfa)	0
• AT-GAA (cipaglucosidase alfa/miglustat)	0

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

Adult Service:

No – Registries only.

Paediatric Service:

No

9 August 2024

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?

Adult Service:

No

Paediatric Service:

No