

13 December 2024

Rare Diseases

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

Adult Service:

Fabry Disease (ICD10 code E75.21)	56
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

Paediatric Service:

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

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.

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

Adult Service:

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

13 December 2024

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

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

Adult Service:

Replagal (agalsidase alfa)	<5
Fabrazyme (agalsidase beta)	14
Galafold (migalastat)	13
Elfabrio (pegunigalsidase alfa)	0
VPRIV (velaglycerase alfa)	<5
Cerezyme (imiglucerase)	<5
Cerdelga (eliglustat)	0
Zavesca (miglustat)	<5 but not for the above indications
Myozyme (alglucosidase alfa)	<5
Nexviadyme (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 (velaglycerase 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

13 December 2024

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?

Adult Service:

No – Registries only

Paediatric Service:

No

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