Belfast Health and Social Care Trust
caring supporting improving together

FOI/31002

18/04/2024

Rare Diseases

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

   Adult Services:

   - **Fabry Disease (ICD10 code E75.21)** 53 (plus 12 children)
   - **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 Service:

   - **Fabry Disease (ICD10 code E75.21)** 61
   - **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): Again we are unable to provide an exact figure - exempt from release under Section 40(2) of the Freedom of Information Act as this could make patients personally identifiable.

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)** <5

Paediatric 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)** <5

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)** <5
- **Fabrazyme (agalsidase beta)** 13
- **Galafold (migalastat)** 14
- **Elfabrio (pegunigalsidase alfa)** 0
- **VPRIV (velaglucerase alfa)** 0
- **Cerezyme (imiglucerase)** 0
- **Cerdelga (eliglustat)** 0
- **Zavesca (miglustat)** 1
- **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 Services:

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 Services:

No.

Paediatric Service:

No