

Freedom of Information Act 2000 – Request Reference FoI/24/213

Rare Diseases

Q1. Within your trust, how many patients currently have a diagnosis for:

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

In completing a search for the information requested, Cardiff and Vale University Health Board (the UHB) has confirmed that it does not centrally collate this data. To retrieve the information requested would require a manual search through individual patient records and the UHB considers that this would exceed the limit set within Regulations for responding to a request. The UHB has therefore relied upon the section 12 exemption of the Freedom of Information Act 2000 ('Exemption where cost of compliance exceeds appropriate limit') and is refusing your request.

The UHB has estimated that to complete the work needed to respond to this request would exceed the time limit as set within Regulations to respond to a Freedom of Information Act request. Under the Act there is an allowance of two and a half days, or 18 hours, to comply with a request and the cost limit set within the Fees Regulations for this amount of work (18 hours) is £450 for the UHB. The Fees Regulations specify that the cost of complying with a request must be calculated at the rate of £25 per hour.

In accordance with a public authority's duty to provide advice and assistance under section 16 of the Freedom of Information Act 2000, the UHB can advise that it can only identify patient records if they have been admitted and coded with a particular condition.

The UHB can additionally provide the following with regard to unique patients who have been coded with specified ICD10 codes from 1 April 2022 to date.

- E740 – 7
- E752 – 18
- E761 – 0

Q2. Of the patients above, how many patients have been newly diagnosed within the past 3 months for:

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

Please see the UHB's response to Q1. In this particular context, the UHB can also confirm that it does not centrally collate information relating to date of diagnosis.

Q3. How many patients have been treated in the last 3 months with the following products:

The UHB cannot provide information on the issues of specific brands; it can only provide a total for the number of issues for the generic drug listed.

- Replagal (agalsidase alfa) – 10
- Fabrazyme (agalsidase beta) – 8
- Galafold (migalastat) – 15
- Elfabrio (pegunigalsidase alfa) – 0
- VPRIV (velaglucerase alfa) – 1
- Cerezyme (imiglucerase) – 5
- Cerdelga (eliglustat) – 5
- Zavesca (miglustat) – 4
- Myozyme (alglucosidase alfa) – 2
- Nexviadyme (avalglucosidase alfa) – 5
- AT-GAA (cipaglucosidase alfa/miglustat) – 0

Q4. 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?

- 7672C The Rare Disease Registry Program (Gaucher, Fabry, Pompe, MPS1) – 0 patients for Fabry.
- 8066 Machine Learning in Fabry Disease Version 1 – no patients recruited, study limited to working with data.
- 8343 Arrhythmia Burden and Risk of Sudden Death in Patients with Fabry Disease: Role of Implantable Loop Recorders (RaLLRoAD) – 3 patients.
- 8382 Intralysosomal Zinc as a Biomarker in Fabry Disease Version 1 – no recruitment.

Q5. 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?

- 7672C The Rare Disease Registry Program (Gaucher, Fabry, Pompe, MPS1) – 0 patients for Gaucher.
- 7759 Remote assessment of Parkinsonism supporting ongoing development of interventions in Gaucher's disease – participant identification centre only – patients referred to participant at other sites.