Freedom of Information Act 2000 – Request Reference FoI/23/666
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)

Cardiff and Vale University Health Board (the UHB) does not hold this information. 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 cannot ascertain whether the patient still has the condition or if they are definitely still within the Local Health Board area.

The UHB can additionally provide the following with regard to patients who have been coded with specified codes.

Patients coded with specified ICD10 Codes 01/04/22 to date			
Unique patients	E740: 7	E752: 16	E671: 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)

The UHB cannot identify the date of diagnosis, so we are unable to determine if a diagnosis is new.

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

Please note that the UHB only holds information on the number of issues for the generic drug, and not branded drug. This has been reflected in the below figures.

- Replagal (agalsidase alfa): 12
- Fabrazyme (algalsidase beta): 9
- Galafold (migalastat): 15
- Elfabrio (pegunigalsidase alfa): 0
- VPRIV (velaglucerase alfa): 1
- Cerezyme (imiglucerase): 5
- Cerdelga (eliglustat): 5
- Zavesca (miglustat): 0
- Myozyme (alglucosidase alfa): 2
- Nexviazyme (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 (RaILRoAD) – 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.