

Date 05/05/2025  
Your Ref  
Our Ref 9958

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Dear

**FREEDOM OF INFORMATION – RARE DISEASES**

I write in response to your request for information in relation to rare diseases.

Question:

1. Within your trust, how many patients currently have a diagnosis for:
  - Fabry(-Anderson) 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)
  - Hunter syndrome (MPS II) (ICD10 code E76.1)
  
2. Of the patients above, how many patients have been newly diagnosed within the past 3 months for:
  - Fabry(-Anderson) 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)
  - Hunter syndrome (MPS II) (ICD10 code E76.1)

Answer:

	Fabry/ Gaucher Disease	Pompe Disease	Pompe Disease (under age 1)	MPS II (Hunter Syndrome)
1	11	5<	0	5<
2	0	0	0	0

1. The number of unique patients who have been admitted to inpatients since April 2008. Lothian patients only.

2. The number of unique patients who have been admitted to inpatients within the last 3 months (1st Jan 2025 - 31st Mar 2025).

The ICD10 code E752 (Other sphingolipidosis) covers both diagnoses of Fabry Disease and Gaucher Disease.

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Chair Professor John Connaghan CBE  
Chief Executive Professor Caroline Hiscox  
Lothian NHS Board is the common name of Lothian Health Board



*The ICD10 code E740 (Glycogen storage disease) covers a diagnoses of Pompe Disease.  
The ICD10 code E761 (Mucopolysaccharidosis, Type II) covers a diagnosis of Hunter Syndrome.*

*There is not a definitive list of all the patients in NHS Lothian with these conditions, as a way of estimating this we have provided the number of inpatients who have been treated in hospital and had one of these conditions recorded on their discharge record. This is not likely to be accurate because not all the patients with these conditions are likely to have had an inpatient stay, the condition may not have been recorded or the patient may have moved out of the area or died since their admission*

Question:

- How many patients have been treated in the last 3 months with the following products:
  - a. Replagal (agalsidase alfa)
  - b. Fabrazyme (agalsidase beta)
  - c. Galafold (migalastat)
  - d. Elfabrio (pegunigalsidase alfa)
  - e. VPRIV (velaglucerase alfa)
  - f. Cerezyme (imiglucerase)
  - g. Cerdelga (eliglustat)
  - h. Zavesca (miglustat)
  - i. Myozyme (alglucosidase alfa)
  - j. Nexviadyme (avalglucosidase alfa)
  - k. AT-GAA (cipaglucosidase alfa/miglustat)

Answer:

a.	5<
b.	5<
c.	0
d.	0
e.	0
f.	5<
g.	5<
h.	0
i.	5<
j.	0
k.	0

Question:

- 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?

Answer:

We have no trials recruiting participants with Fabry disease.

Question:

- 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?

Answer:

We have one (1) trial entitled "RAPSODI, entitled "remote assessment of Parkinsonism supporting ongoing development of interventions in Gaucher's disease". This study is being run by University College London and is a basic science study. The study is split into 2 cohorts: RAPSODI (recruiting participants with Gaucher's disease who may go on to develop Parkinson's disease (dedicated trial website [www.rapsodistudy.com](http://www.rapsodistudy.com)) and PD FRONTLINE (recruiting participants with Parkinson's disease that may carry the gene for Gaucher's disease (dedicated trial website [www.pdf frontline.com](http://www.pdf frontline.com)). In Lothian we are currently only recruiting to the PD FRONTLINE cohort and we have recruited 60 participants.

To protect the identity of the individuals involved any figure of 5 or less has not been shown in the tables above. Since we do not have their consent to release this data from their records, the information is exempt under section 38(1)(b) of the Freedom of Information (Scotland) Act i.e. to provide it would breach the Data Protection Act (2018).

I hope the information provided helps with your request.

If you are unhappy with our response to your request, you do have the right to request us to review it. Your request should be made within 40 working days of receipt of this letter, and we will reply within 20 working days of receipt. If our decision is unchanged following a review and you remain dissatisfied with this, you then have the right to make a formal complaint to the Scottish Information Commissioner within 6 months of receipt of our review response. You can do this by using the Scottish Information Commissioner's Office online appeals service at [www.itspublicknowledge.info/Appeal](http://www.itspublicknowledge.info/Appeal). If you remain dissatisfied with the Commissioner's response you then have the option to appeal to the Court of Session on a point of law.

If you require a review of our decision to be carried out, please write to the FOI Reviewer at the email address at the head of this letter. The review will be undertaken by a Reviewer who was not involved in the original decision-making process.



FOI responses (subject to redaction of personal information) may appear on NHS Lothian's Freedom of Information website at: <https://org.nhsllothian.scot/FOI/Pages/default.aspx>

Yours sincerely

**ALISON MACDONALD**  
**Executive Director, Nursing**  
Cc: Chief Executive