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Pompe disease in Sweden: a real-world evidence study investigating healthcare resource utilisation and costs

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Introduction and objectives

- Late-onset Pompe disease (LOPD) is a rare disease that has a substantial burden on healthcare systems, resulting in significant healthcare expenditures and resource utilisation[1–3]
  - Up to 2017, a review estimated the average annual cost of supportive therapy per adult patient with LOPD as €22,475[1]
  - Incremental costs of LOPD treatment per quality-adjusted life year (QALY) have been estimated as €1.8–3.2 million.[2,3]
- Effective management and resource allocation require accurate data on disease epidemiology, healthcare resource utilisation (HRU), associated costs, and time to key health outcomes[1]
  - Various studies and guidelines on the economic burden of lysosomal storage diseases, including LOPD, in regional European populations have been conducted, for example, in England,[4] the Netherlands[2] and Switzerland[5]
  - To our knowledge, there have been no such studies in a Swedish population.
- This real-world evidence study aimed to explore disease burden, treatment patterns and HRU associated with Pompe disease in the Swedish population
  - We present HRU and associated costs in patients diagnosed with LOPD.

Conclusions

- The high HRU observed in patients with LOPD in Sweden emphasises the complex management and careful resource allocation required for the disease.
- Variability in time to key health outcomes, such as age at diagnosis and wheelchair use, highlights the heterogeneity of Pompe disease progression and signals increased resource needs.
- Costs associated with LOPD, driven largely by numerous and costly inpatient and outpatient visits, reflect the substantial economic burden on patients and healthcare systems
  - Overall annual inpatient and outpatient costs following enzyme replacement therapy (ERT; at cross-sectional timepoint [CSTP]) were lower than other estimates for LOPD;[1] however, most related publications present data as QALY, precluding direct comparisons
  - Prior diagnoses and high cost of patient care prior to ERT indicate that patients were being investigated by other healthcare professionals before a Pompe disease specialist.
- Study limitations include small sample size, difficulty obtaining patient permission for data use and data masking, which reflect the retrospective nature of the study
  - Furthermore, available data do not allow the assessment of whether treatment costs are higher in patients with greater disease progression, which could be a major factor driving treatment costs.
- These data provide further insight into the substantial HRU, costs, and variability in time to health outcomes associated with LOPD, and underscore the need for early intervention and multidisciplinary healthcare resources to improve outcomes for patients with LOPD.

Methods

How were the data collected?

Data source

Retrospective, observational, secondary disease quality and registry data from the NMiS, NPR, COD and PDR (including deceased patients)

Period of inclusion

The period of inclusion ran from 2005 to 2023. This analysis ran from 1 January 2005 to 31 December 2022 (CTSP)

Patient inclusion (N=22)

5 IOPD | 17 LOPD

Patients diagnosed with Pompe disease during the period of inclusion

COD, Swedish Cause of Death Register; IOPD, infantile-onset Pompe disease; NMiS, Swedish National Registry for Neuromuscular Disorders; NPR, Swedish National Patient Register; PDR, Swedish Prescribed Drug Register.

- The primary objective was to estimate the prevalence and incidence of Pompe disease in Sweden.
- Estimation of HRU, associated costs, and time to key health outcomes were secondary objectives.
- Data masking was used to prevent patient identification
  - The NMiS Steering Committee approved the unmasking of subgroups with ≥3 patients
  - Socialstyrelsen data, including NPR, COD and PDR data, follow the standard masking of data reported in subgroups with <5 patients.
- Data are entered into these registers every time a Swedish citizen visits secondary care, receives a pharmaceutical prescription or dies
  - The data were linked across different national registries (NMiS, NPR, COD and PDR) through unique personal identifiers and extracted for analysis into a single dataset.
- Data from two timepoints were analysed: at diagnosis and a CSTP of 31 December 2022.
- Continuous variables were summarised as median (interquartile range [IQR]), mean (standard deviation [SD]) and number of observations (n).
- HRU included the number of inpatient and outpatient visits and length of inpatient stay for Pompe disease per patient. Overall cost included non-ERT pharmaceutical costs from the PDR.
- For this specific HRU and costs analysis, only adults with diagnosed LOPD (n=17) were included from the overall study cohort.

Results

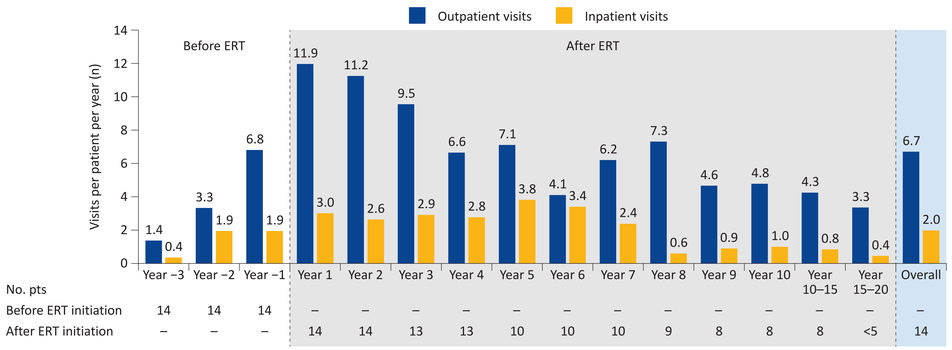
Patients with LOPD showed heterogeneity in age at diagnosis and age at initiation of wheelchair use

	Patient characteristics	
	At diagnosis (n=17)	At CSTP (n=14)
Males, n (%)	11 (64.7)	8 (57.1)
Age, years		
Mean (SD)	43.2 (22.3)	55.7 (16.5)
Median (IQR)	49.9 (23.9, 63.7)	55.4 (47.2, 69.9)
Ambulation status, n (%)		
Ambulatory	17 (100)	10 (71.4)
Non-ambulatory	0	4 (28.6)
Wheelchair use, n (%)		
None	NA	9 (64.3)
Wheelchair used partially/continuously	–	5 (35.7)
Age at initiation of wheelchair use, median (IQR)	–	44.1 (32.6, 60.1)
Age at diagnosis of wheelchair users, median (IQR)	–	28.3 (23.9, 42.4)
Ventilation support/other respiratory management		
Yes, n (%)	≤2 (masked)	6 (42.9)
FVC at first need of ventilation support/other respiratory management, % predicted		
Mean (SD)	–	20.5 (0.7)
Median (IQR)	–	20.5 (20.0, 21.0)
6MWD, metres*		
Mean (SD)	454.0 (149.1)	484.0 (111.7)
Median (IQR)	504.0 (328.0, 546.0)	511.5 (358.0, 532.0)
10MWT, seconds*		
Mean (SD)	8.1 (3.5)	6.6 (2.2)
Median (IQR)	6.5 (6.2, 8.5)	6.9 (5.0, 8.5)
Sitting FVC, % predicted		
Mean (SD)	62.1 (17.1)	50.3 (30.5)
Median (IQR)	67.0 (44.0, 68.0)	57.5 (20.5, 74.5)

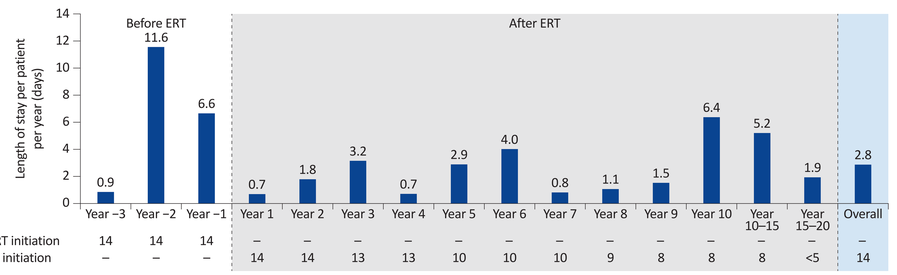
*6MWD and 10MWT data were recorded in five patients at diagnosis and six at CSTP. 6MWD, 6-minute walk distance; 10MWT, 10-metre walk test; FVC, forced vital capacity.

- As of 31 December 2022, the estimated prevalence of diagnosed LOPD in Sweden was 1.3 per 1,000,000 people.
- It was not possible to estimate the incidence of Pompe disease in Sweden due to the difficulty in accurately calculating all incidences since 2005.
- Of 17 patients diagnosed with LOPD, 14 received ERT:
  - There was a median (95% confidence interval) delay of 1.5 (0.48, 16.6) years between LOPD diagnosis and ERT initiation
  - Three patients subsequently discontinued ERT
  - In patients with available dosing data (n=9), all received standard-dose ERT (20 mg/kg biweekly) at treatment initiation.
- Prior to their diagnosis of LOPD, ≥50% of the 17 patients had received prior diagnoses, labelled as symptoms, signs, and abnormal clinical and laboratory findings not classified elsewhere (n=12; 70.6%); factors influencing health status and contact with health services (n=10; 58.8%); endocrine, nutritional and metabolic diseases (n=9; 52.9%); and diseases of the nervous system (n=9; 52.9%; **Supplementary Figure 1**).
- Patients with LOPD (n=14) received a diverse range of concomitant treatments, procedures and pharmacological therapies alongside ERT, including respiratory management (71.4%), psychosocial management (64.3%), and treatments for musculoskeletal and inflammatory conditions (53.7%; **Supplementary Figure 2**).
- Median age at key adverse health outcomes was typically over 50 years in patients with LOPD, with a median age at first ventilation support/respiratory management of 62.8 years (**Supplementary Figure 3**).

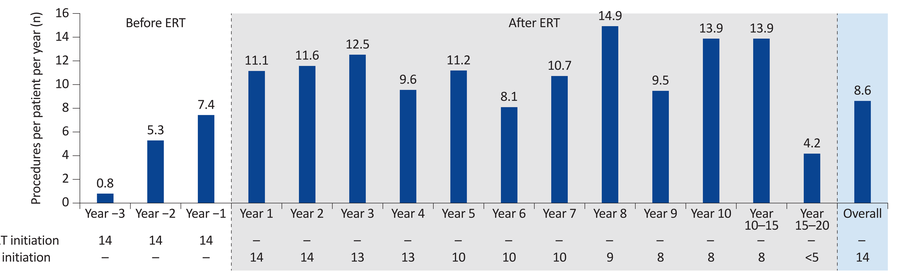
After ERT initiation, the overall annual number of outpatient and inpatient visits were 6.7 and 2.0 visits per patient, respectively



After ERT initiation, the overall annual length of inpatient stay was 2.8 days per patient

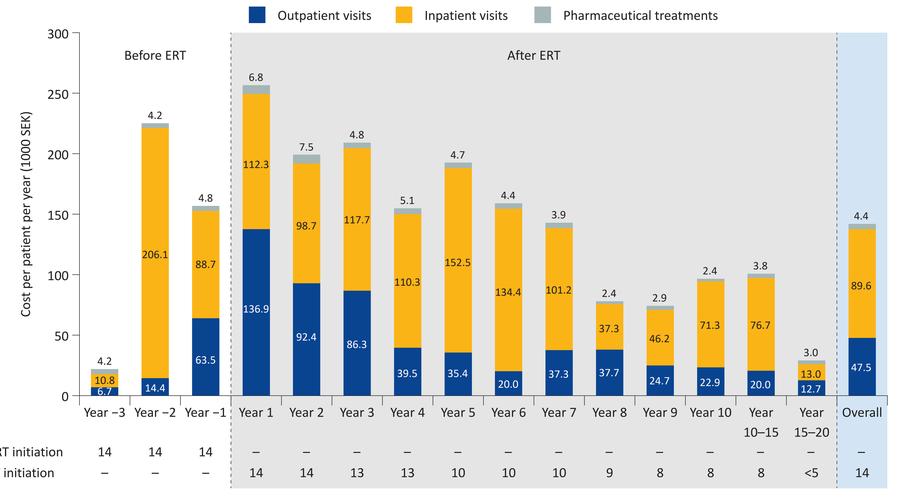


After ERT initiation, the overall annual number of procedures was 8.6 per patient



Data show the sum of inpatient visits, outpatient visits, inpatient stays and medical procedures registered in the NPR divided by the follow-up period. For length of stay, inpatient days were calculated by subtracting the last day of a stay from the first and adding 1 day, eg 1 overnight stay was counted as 2 hospitalisation days, and admission/discharge on the same day was counted as 1 hospitalisation day. Primary care is not included in the analyses as it is not covered by the registers used in the study.

After ERT initiation, overall annual costs of outpatient and inpatient care were 47,500 and 89,600 SEK per patient, respectively



Data show the sum of the cost of inpatient or outpatient visits registered in the NPR divided by the follow-up period. 1 USD is equal to 9.58 SEK as of 25 April 2025. Inpatient and outpatient visits were valued based on the recorded DRG code at the time of the visit and the DRGs corresponding weight published by NBHW. In case of missing DRGs, the most common DRG was imputed to avoid underestimating the costs due to missing values. For pharmaceutical treatments, data show the sum of the total cost (excluding VAT) of pharmaceutical treatment dispensing registered in the PDR divided by the follow-up period and includes costs paid by the patient and the county council. Pharmaceutical treatments include ERT-related (E74.0 Glycogen storage disease ICD-10 coded in NPR) and non-ERT-related (all ICD-10 codes, excluding E74.0 in NPR) treatments. DRG, Diagnosis-related Group; ICD, International Classification of Diseases; NBHW, National Board of Health and Welfare; SEK, Swedish krona; USD, United States dollar; VAT, value-added tax.

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The presenter, Alasdair MacCulloch, is an employee of and holds stocks and shares in Amicus Therapeutics, Inc.

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