Economic Burden of Gaucher Disease: A Systematic Literature Review

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INTRODUCTION

- Gaucher disease (GD) is a rare inherited lysosomal storage disorder caused by deficient activity of the enzyme, glucocerebrosidase, which leads to multi-organ involvement and significant morbidity[1]
- Clinical manifestations of GD such as cytopenia, splenomegaly, hepatomegaly, bone crises, and in some phenotypes, neurological impairment, often necessitate lifelong, high-intensity management[2]
- Management of GD has been revolutionized over the past three decades by enzyme replacement therapy (ERT) and, more recently, substrate reduction therapy (SRT). While these treatments have significantly improved clinical outcomes, they are associated with high and sustained costs, often creating a substantial financial burden

OBJECTIVES

- This systematic literature review aimed to identify comprehensive evidence on the economic burden for patients with GD

METHODS

- Embase® and MEDLINE® were systematically searched following Preferred Reporting Items for Systematic Reviews and Meta-Analyses (PRISMA) guidelines for English language studies reporting data for the economic burden in pediatric and adult patients with GD
- Two independent reviewers screened titles and abstracts, with full-text assessments performed by the same reviewers and reconciled by a third reviewer if needed. The selection process of studies involved evaluating publications retrieved through searches against pre-established population, intervention, comparators, outcome, and study design (PICOS) criteria, as outlined in Table 1
- All costs have been converted to USD using the official exchange rate (exchange-rates.org) for the corresponding years, to ensure year-specific accuracy, consistency, and comparability across different countries

Table 1: Methodology for conducting the economic burden SLR in GD

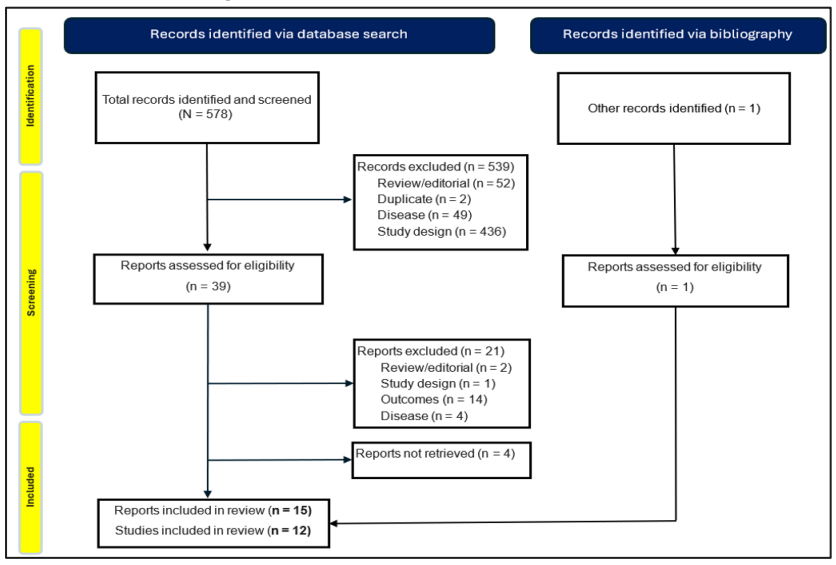
Population	Patients with GD
Intervention and comparator	No restriction
Outcomes	▪ Direct and indirect cost components ▪ Total cost ▪ Healthcare resource costs ▪ Resource use data ▪ Cost and management of treatment-related adverse events ▪ Societal costs, cost of carer, and productivity losses
Study designs	▪ Cost studies ▪ Resource use studies ▪ Economic evaluations reporting costs or resource use ▪ Cost/economic burden studies ▪ Budget impact analysis ▪ Cost−benefit analysis ▪ Cost−consequence analysis ▪ Cost-minimization analysis ▪ Cost−utility analysis ▪ SLR and meta-analysis (for cross-referring only)
Country	No geographical limits on country applied
Language	English
Timeframe	2015−2025

Key: GD, Gaucher disease; SLR, systematic literature review.

RESULTS

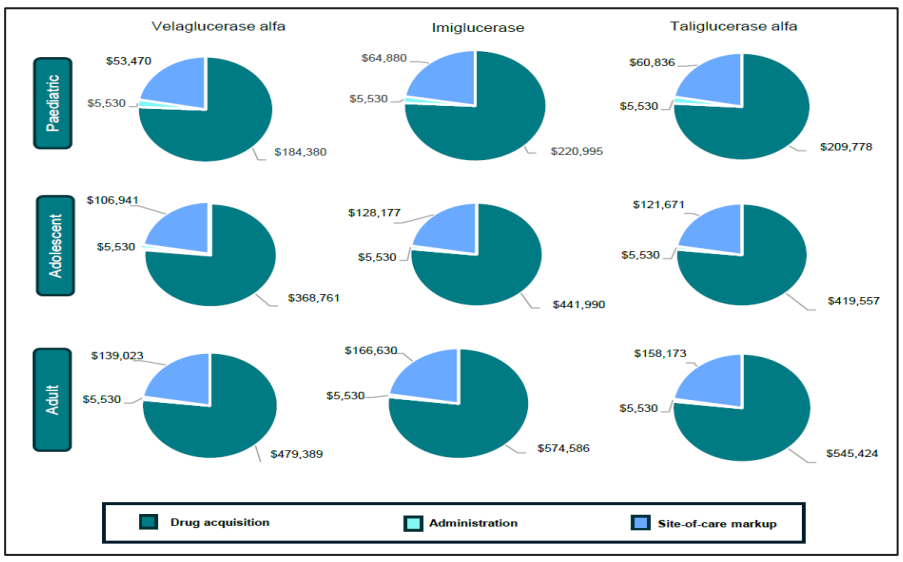
- Twelve unique studies were identified and included; they evaluated the economic burden of GD in nine different countries. Three studies reported data for the US, two for China, and one study each for Greece, Germany, Brazil, Colombia, India, Iran, and Russia (Figure 1)

Figure 1: PRISMA flow diagram



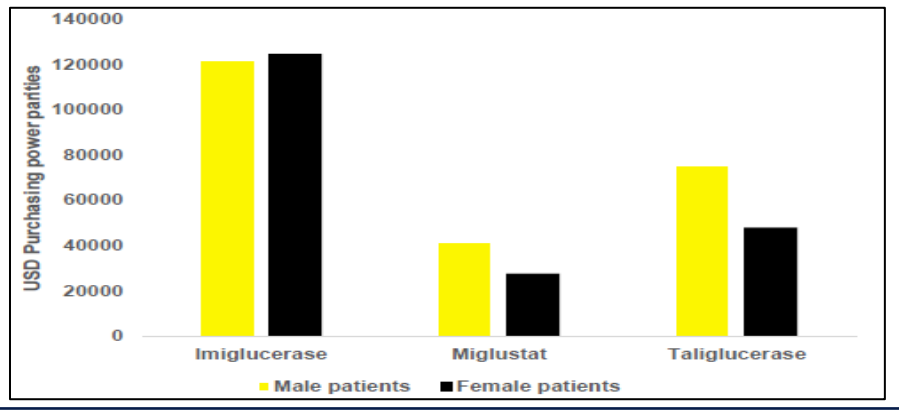
- In the US, during 2021, total annual direct healthcare costs per patient ranged from $243,281–$276,144 in the pediatric population, and from $623,942–$709,127 in the adult population. Among available therapeutic options, treatment with taliglucerase alfa was associated with the greatest financial burden (Figure 2)[6,12]
- In Germany, from 2019 to 2021, the mean annual treatment cost was $528,070 (€471,449) per patient, of which 99%, i.e., $522,049 (€466,074) was attributable to infusion-related expenses. The average cost per infusion per patient was $21,584 (€19,270)[2]
- In China, in 2018, the annual direct cost of treatment was $46,790.60 per patient with a pharmaceutical therapy cost of $29,907.60. The mean costs for inpatient and outpatient medical care were $7,451.20 and $1,838.10, respectively[7,8]
- In Colombia, in 2023, the average annual cost of treatment per patient was $125,649 and the annual economic burden for overall diagnosed patients was $31,968,502[4]
- In Iran, in 2017, the average annual treatment cost per patient was $19,763, while the total medical care expenditure was $3,399,258 for the entire GD population[10]

Figure 2: Total annual direct healthcare costs per patient with GD in the US



- In Brazil, between 2000–2015, a cohort of patients with GD incurred a mean annual overall medical care cost (adjusted according to the purchasing power parity index of The World Bank 2024) of $120,880.14 for female patients, and of $120,264.41 for male patients according to a retrospective analysis (Figure 3)[3]
- In India, in 2020, the total cost for 30 patients in a single study was $3,440,199.20 (₹254,574,743). Among these, 14 patients treated with ERT or SRT incurred a total cost of $3,418,476.74 (₹252,967,279), while 16 treatment-naïve patients incurred a cost of $21,825.19 (₹1,615,064) across the population[5]
- The cost of a 1-year treatment course per patient of taliglucerase alfa, velaglucerase alpha, and imiglucerase was $194,295, $388,825, and $392,080 (12,196,800.0 RUB, 24,408,346.5 RUB, and 24,612,737.0 RUB), respectively, in Russia calculated from the maximum manufacturer's prices indicated in the list of vital and essential medicines of March 1, 2018[11]
- A cross-sectional study conducted between December 2022 and March 2023 in Greece reported the average annual cost for 174 patients with GD as $25,808,939 (€23,850,789) with a per-patient cost of $148,327 (€137,074)[13]

Figure 3: Mean annual overall medical costs in Brazil for patients with GD



- Resource utilization data in China showed an average of 8.8 outpatient and 4.0 inpatient visits per year in 2018, while a US study found that only 0.3% of patients required hospitalization in 2016[7,9]

CONCLUSIONS

- GD has a substantial and persistent economic burden on healthcare systems worldwide. As a rare disorder requiring life-long treatment, the costs are driven predominantly by ERT and, in many settings, by infusion-related expenses
- Direct medical costs per patient are consistently high across regions, often exceeding those of many other chronic conditions, and remain relatively stable over time due to limited opportunities for cost reduction in the therapeutic approach
- Our findings emphasize the global need for cost-effective management strategies and further research to reduce the financial impact on healthcare systems and patients

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