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#EE415

Economic Perspectives on Niemann–Pick Disease: A Targeted Review of Economic Evidence

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INTRODUCTION

- Niemann–Pick disease (NPD) comprises rare inherited lysosomal storage disorders, including acid sphingomyelinase deficiency (ASMD; types A, A/B, and B) and NPD-C, with distinct pathophysiology and clinical manifestations
- ASMD affects multiple organ systems, impairing physical functioning and quality of life, while imposing financial and productivity burdens on patients and caregivers
- A structured evidence review is needed to characterize NPD's economic burden, healthcare resource use (HCRU), cost drivers, health-state utilities, and evidence gaps

OBJECTIVES

This targeted literature review (TLR) aimed to summarize published economic evidence on NPD, including healthcare costs, HCRU, caregiver burden, utilities, and cost-effectiveness, and identify key evidence gaps to inform future economic research.

METHODS

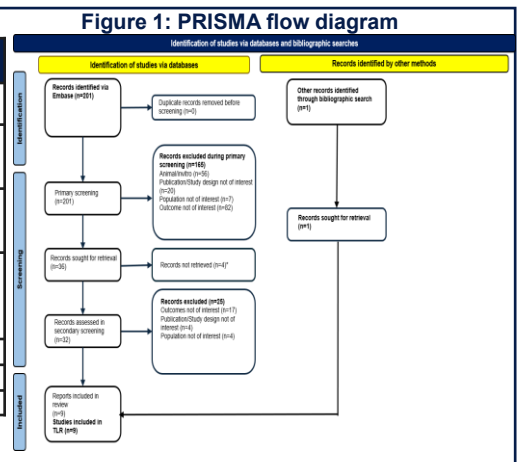
- A TLR was conducted in Embase® from database inception to June 2026 focusing on economic evaluation and HCRU and health-state utilities
- Title/abstract and full-text screening were conducted by a single reviewer, with a random 20% quality check (QC) completed by a senior reviewer. Data were extracted using a standardized grid, with an additional 20% QC. Inclusion criteria are presented in Table 1

Table 1: PICOS for the economic burden TLR

Category	Inclusion criteria
Population	Patients of any age with Niemann–Pick disease
Intervention/ exposure	No intervention limits
Outcomes	HCRU, direct/indirect costs, ICERs, QALYs, and health-state utilities/disutilities
Study design	Observational, economic modeling, cost, resource use, burden-of-illness, and health-state utility studies
Country	No geographic limits
Language	English
Timeline	Database inception to June 2026*

Key: HCRU, healthcare resource utilization; ICER, incremental cost-effectiveness ratio, QALY, quality-adjusted life year; LY, life year; TLR, targeted literature review.
Notes: *Only freely available full-text articles were selected for inclusion, while articles requiring payment or subscription for access were excluded.

Figure 1: PRISMA flow diagram



Key: PRISMA, Preferred Reporting Items for Systematic Reviews and Meta-analyses; TLR, targeted literature review.
Notes: *Only freely available articles were included; all non-retrieved publications required paid/subscription access

RESULTS

Overview of evidence

- Of 201 studies screened, eight (2015–2025) were included, with one additional study identified through bibliographic searching (Figure 1). Designs included retrospective (n=5), prospective (n=2), economic modelling (n=1), and utility (n=1) studies. Settings included the USA (n=3), multinational, the UK, Canada, Brazil, Portugal and England. Outcomes covered HCRU (n=3), costs (n=2), patient/caregiver burden (n=2), cost-effectiveness (n=1), and utilities (n=1)

Healthcare cost and resource utilization

Healthcare cost:

- In Portugal, annual ASMD follow-up costs averaged €923 per patient, rising to €3,668 for cardiovascular complications and €124 for hemorrhagic complications. Annually, 6.1% of patients required hospitalization and 16.3% visited EDs, highlighting complication-related costs[3]
- In England, annual NHS and social care costs (excluding miglustat) averaged £3,800 for adults and £4,200 for children with NPD-C, while miglustat costs were £94,108 and £48,627, respectively, highlighting treatment costs as a major economic burden[4]

Figure 2: Average annual follow-up cost in Portugal[3]

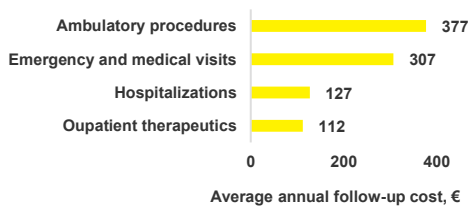
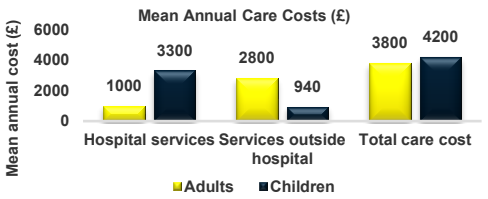


Figure 3: Mean annual NHS and publicly funded social care cost by age group in England[4]



Key: NHS, National Health Service; NPC, Niemann–Pick disease

Healthcare resource utilization

Caregiver burden:

- A US survey found that 61% of patients were unaware of Medicaid waivers. Insurance-related coverage gaps negatively affected patients' mental and physical health, while caregivers experienced stress and burnout due to constant vigilance and 24-hour care needs (Fig. 3)[5]
- A US qualitative study found that ASMD affected patients' physical functioning (79.3%), self-esteem (62.1%), emotional well-being, and relationships (55.2% each). Caregivers were primarily affected emotionally (80.0%) and financially (46.7%)[1]

Hospitalization and healthcare visits:

- A Brazilian study reported increased hospitalization over follow-up, with higher rates in ASMD type A/B than type B; no ED or outpatient visits were reported (Fig. 4)[6]
- A US retrospective study found that cognitive, developmental and emotional problems drove office visits (754.5/1,000 PY) and outpatient visits (760.0/1,000 PY), while respiratory disorders led to ED visits (27.5/1,000 PY) and hospitalizations (38.6/1,000 PY) in NPD[7]
- A multinational study reported the highest annual hospital admissions and ED visits in infantile neurovisceral ASMD (2.23 and 0.72), followed by chronic neurovisceral (0.42 and 0) and chronic visceral ASMD (0.41 and 0.07), respectively[8]
- Among adults with chronic visceral ASMD, 51% worked full-time or chose part-time work, 5% worked part-time, 16% were unable to work due to disability, and 27% were not working for unrelated reasons[8]

Figure 4: Patient and caregiver reported life impacts from access burden in the US[5]

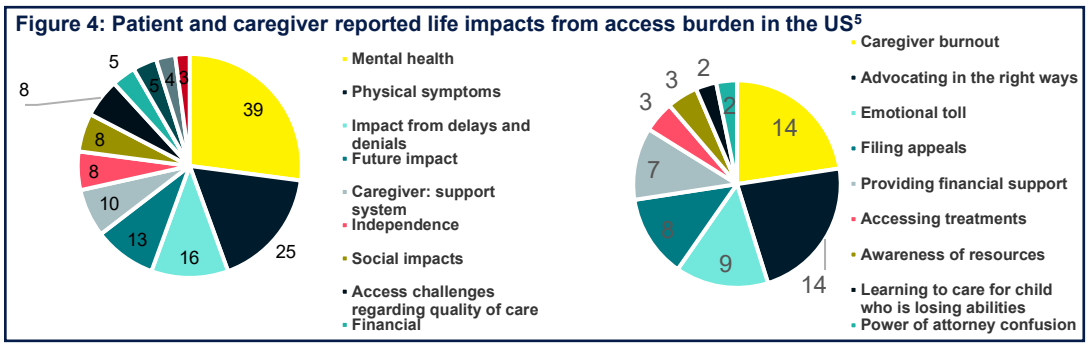
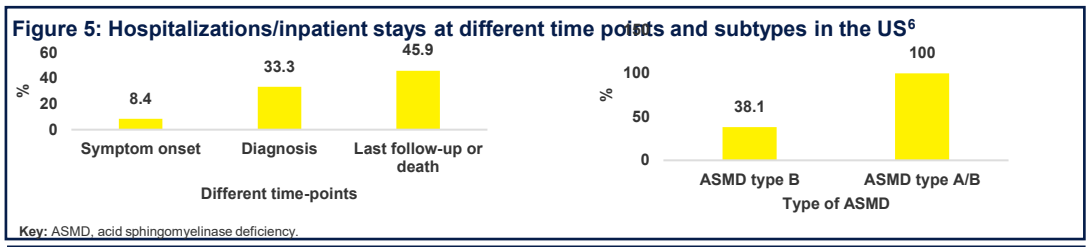
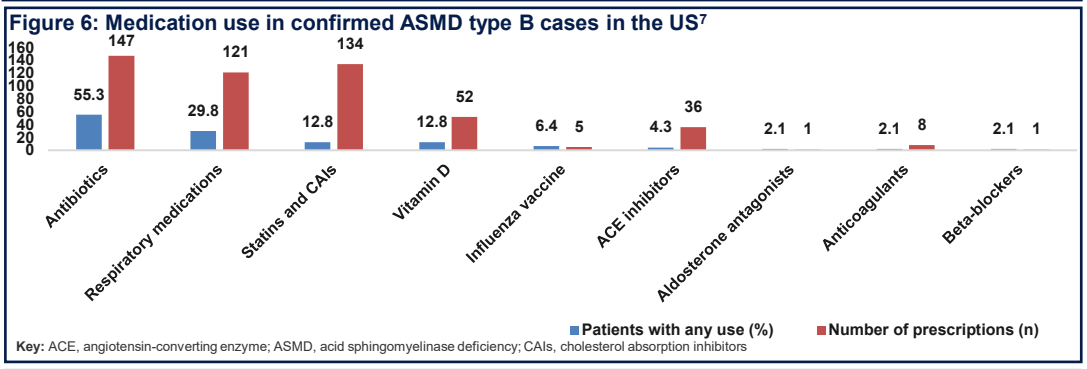


Figure 5: Hospitalizations/inpatient stays at different time points and subtypes in the US[6]



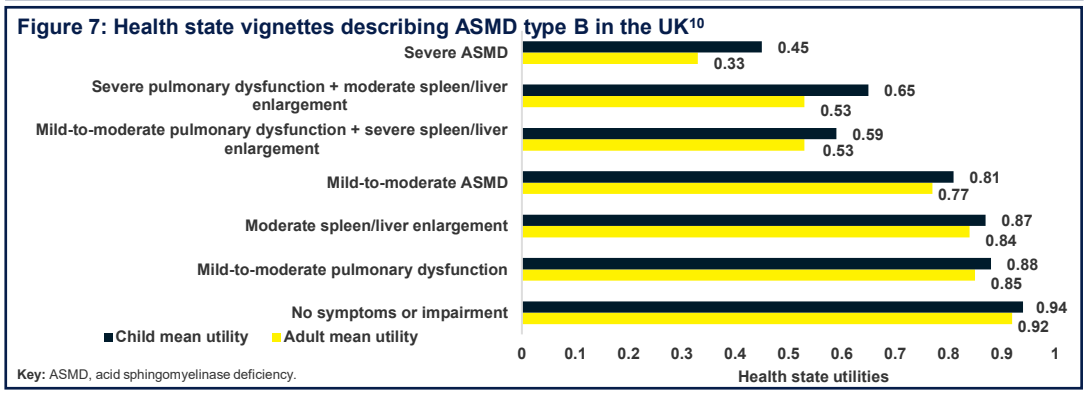
Key: ASMD, acid sphingomyelinase deficiency.

Figure 6: Medication use in confirmed ASMD type B cases in the US[7]



Key: ACE, angiotensin-converting enzyme; ASMD, acid sphingomyelinase deficiency; CAIs, cholesterol absorption inhibitors

Figure 7: Health state vignettes describing ASMD type B in the UK[10]



Key: ASMD, acid sphingomyelinase deficiency.

Economic evaluation review:

A Canadian decision-analytic model found that plasma oxysterol screening in intellectually disabled children and adolescents with NPD increased QALYs by 0.0022 and reduced costs by CAD 3,642 per patient, making it a dominant strategy versus current practice.[9]

Health states utility review:

- A UK vignette time trade-off (TTO) study generated 14 utility values for adult and pediatric ASMD type B, showing higher utilities in children than adults (differences: 0.02–0.13) and declining utilities with increasing disease severity (0.33 in adults; 0.45 in children) (Fig. 5)[10]
- The Canadian cost–utility model used utilities of 0.64 for untreated NPD-C, 0.73 for miglustat-treated NPD-C, and 0.60 for intellectual disability to estimate incremental QALYs from screening[9]
- A UK HTA found that female sex and older age were associated with lower EQ-5D utility, while longer miglustat treatment was associated with higher EQ-5D utility. No significant association was observed between SF-6D utility and treatment duration[4]

DISCUSSION

- Economic burden: NPD incurs substantial healthcare and treatment costs, driven by miglustat in the UK and cardiovascular complications in ASMD
- HCRU: Hospitalization varied by phenotype, peaking in infantile neurovisceral ASMD; respiratory disorders were major drivers of healthcare use
- Patient and caregiver burden: ASMD impaired physical and psychosocial well-being, while caregivers faced emotional and financial impacts, compounded by insurance barriers and work limitations
- Utility evidence: Plasma oxysterol screening was cost-saving and more effective than current practice; utilities declined with ASMD severity but varied by method and population

LIMITATIONS

- Limited evidence base (n=9 studies)
- Exclusion of non-English and subscription-based articles may have reduced coverage
- Heterogeneity in populations, subtypes, methods, outcomes, and currencies limited comparisons
- Limited geographic coverage, with some findings based on small or phenotype-specific samples
- Few economic evaluations; utility estimates varied by methodology

CONCLUSION

NPD, including ASMD and NPD-C, is associated with substantial healthcare costs, resource use, and patient and caregiver burden. HRQoL declines with increasing ASMD severity, while limited evidence suggests cost savings from plasma oxysterol screening. Further research is needed to address evidence gaps, long-term burden and the cost-effectiveness of emerging treatments and supportive care.

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