

A Systematic Literature Review on Costs and Healthcare Resource Utilisation in Spinal Muscular Atrophy

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KEY FINDINGS & CONCLUSIONS

- The global SMA burden is substantial, with high costs of patient care and a large impact on caregiver time.
- Non-adherence to risdiplam and nusinersen increased PPPY total cost and HCRU.
- Early identification and treatment may reduce costs and HCRU.

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INTRODUCTION

- Spinal muscular atrophy (SMA) is a rare genetic disorder caused primarily by the homozygous mutation or deletion of the survival motor neuron 1 gene on chromosome 5q.
- SMA causes substantial disability, with an estimated prevalence of approximately 8 per 100,000 births globally and an annual incidence of approximately 10 per 100,000 live births.¹
- Until recently, treatment for SMA primarily focused on supportive care; however, over the past decade, the treatment landscape has evolved significantly with the approval and use of disease-modifying therapies (DMTs) such as nusinersen (administered intrathecally, every 4 months after 4 loading doses over 3 months), risdiplam (administered orally once daily), and onasemnogene abeparvovec (single intravenous infusion; OAV101 IV).

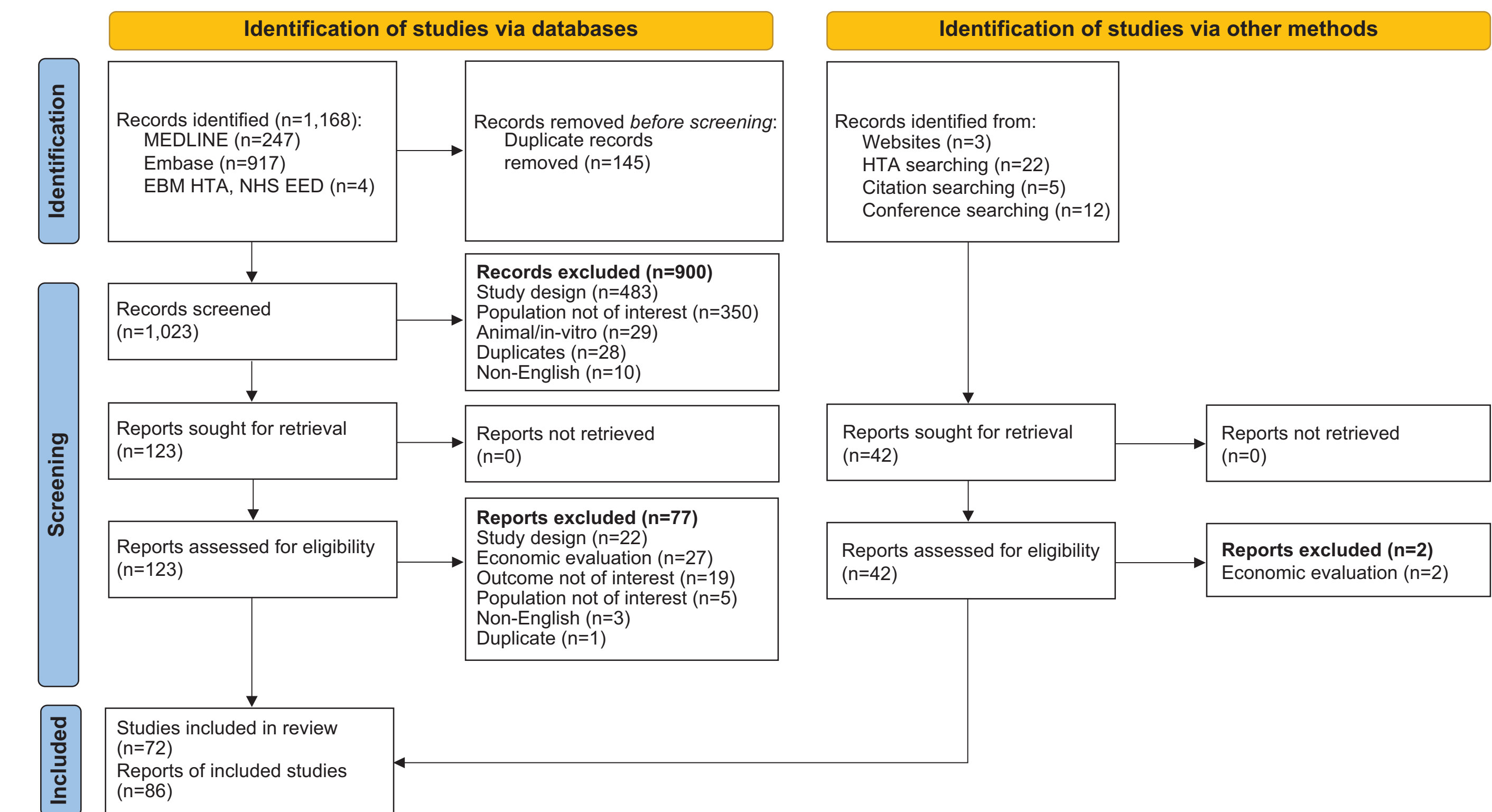
OBJECTIVE

- The objective of this systematic literature review (SLR) was to assess the costs and healthcare resource utilisation (HCRU) associated with SMA.

RESULTS

- Overall, 72 unique studies were included in the review. Of these, 41 studies reported both costs and HCRU, 19 studies reported costs, and 12 studies reported HCRU.
- The study selection process is presented in the PRISMA flowchart (Figure 1).

Figure 1. PRISMA flowchart showing the study selection process

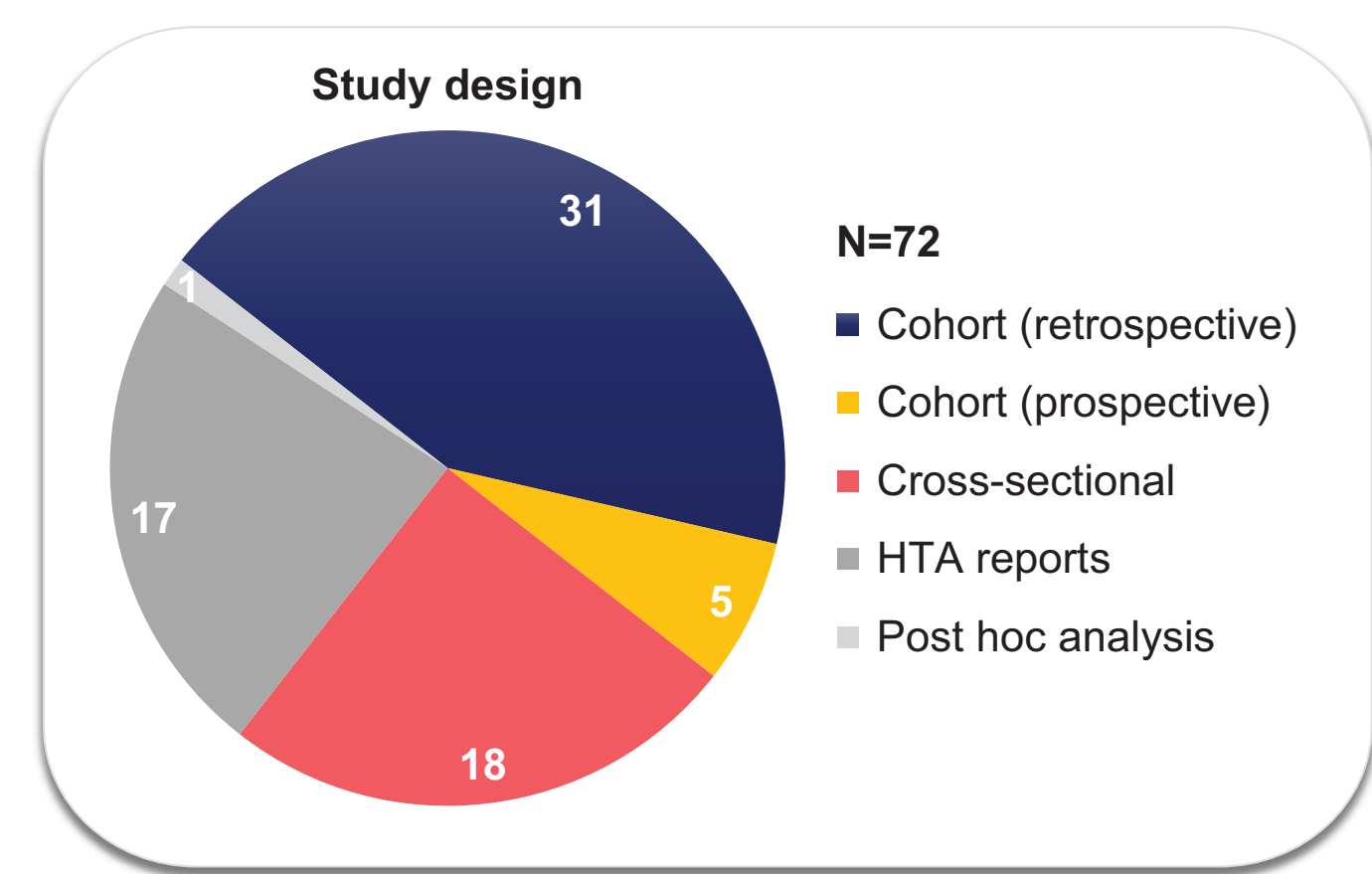


Abbreviations: EBM: Evidence-Based Medicine; Embase: Excerpta Medica Database; HTA: Health Technology Assessment; MEDLINE: Medical Literature Analysis and Retrieval System Online; NHS EED: National Health Service Economic Evaluation Database; PRISMA: Preferred Reporting Items for Systematic Reviews and Meta-Analyses

Included study design characteristics

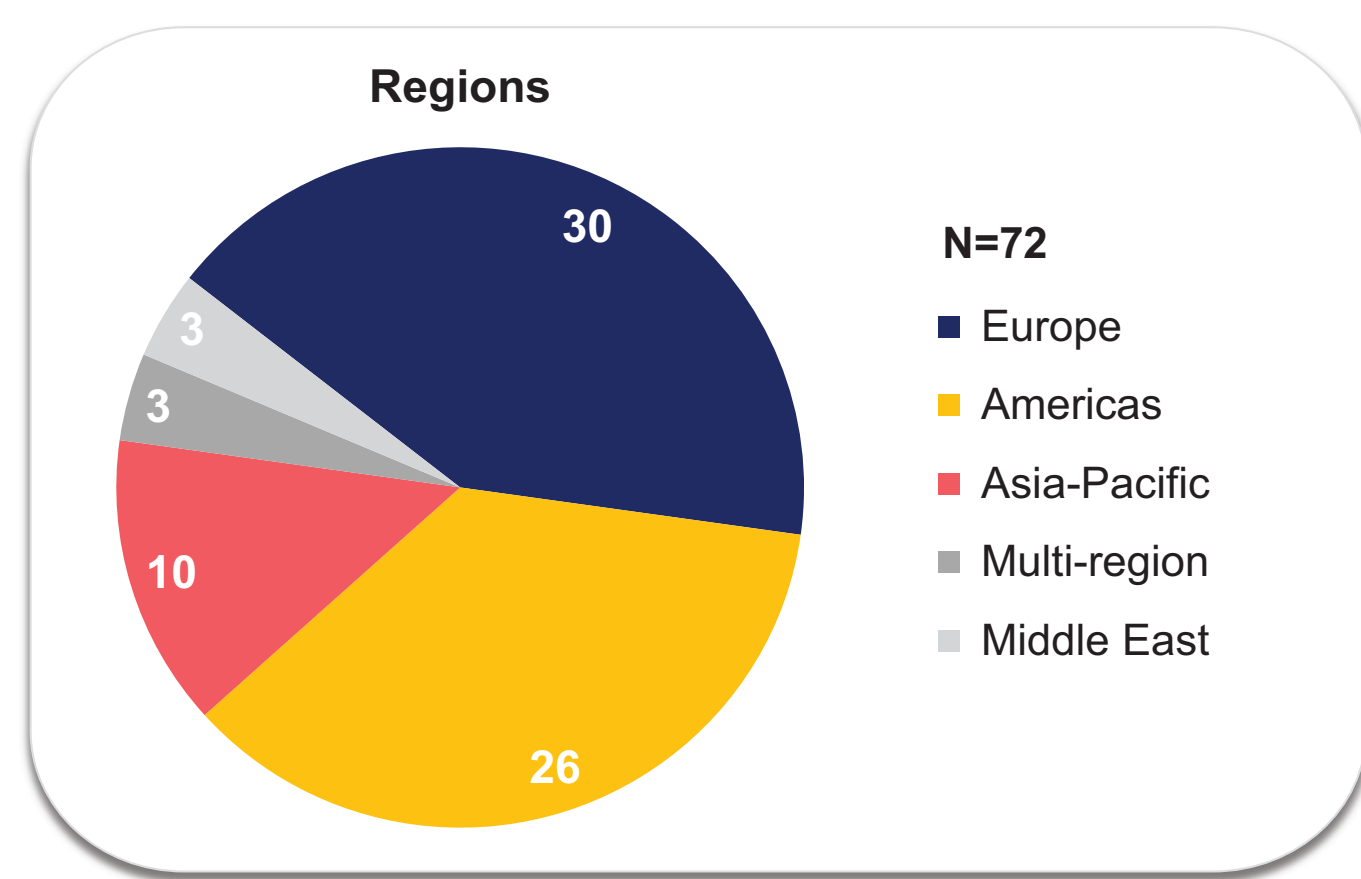
- Most of the included studies were cohort in nature (retrospective cohort: 31, prospective cohort: 5), followed by a cross-sectional study design (n=18) (Figure 2).
- The included studies were conducted globally across various regions (Figure 3).

Figure 2. Included study characteristics



The majority of the included studies were cohort in nature

Figure 3. Geographic distribution of included studies



The majority of the studies were conducted in Europe, followed by the Americas

Costs and HCRU among SMA vs. non-SMA cohort

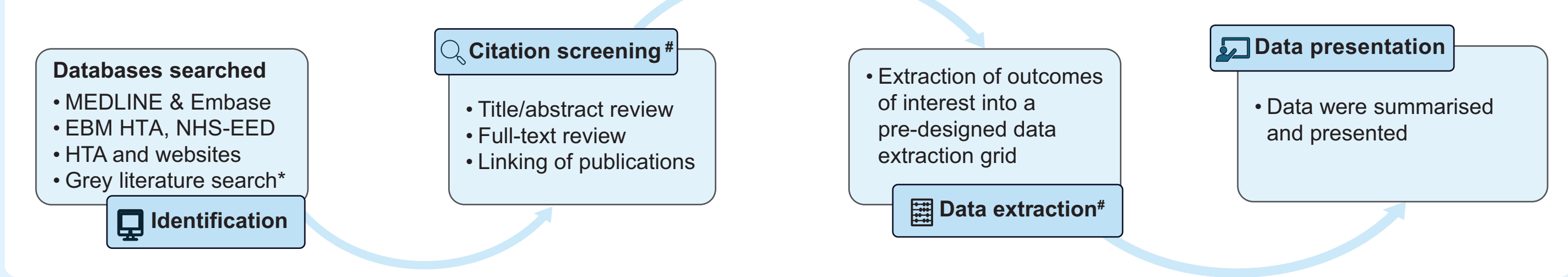
- Three retrospective cohort studies from the United States (US), two using claims databases and one using military healthcare system data, compared the costs and HCRU among patients with SMA vs. non-SMA matched controls.²⁻⁴ Additionally, one retrospective cohort study from Sweden using national patient registry data compared costs and HCRU among patients with SMA vs. a non-SMA reference cohort.⁵
- The mean per patient per year (PPPY) total direct medical cost (excluding DMT cost) for patients with SMA was significantly higher than that for non-SMA matched (without SMA diagnosis and matched based on age and gender) controls (US\$47,862 vs. US\$1,861, respectively; $p < 0.001$).⁴
- Direct costs of SMA are significant, often >50-fold higher than for matched controls without SMA, matched based on age, gender, and region.³
- Patients with SMA had higher hospitalisation rates and length of hospital stay compared with controls without SMA matched for age, gender, and region.³
- Annual HCRU was higher in patients with SMA compared with reference cohorts (matched non-SMA).⁵

METHODS

Studies meeting the following criteria were eligible for inclusion in the review:

- Studies including patients with any type of SMA diagnosis with no restriction on interventions or comparators, reporting original cost/HCRU outcomes: total cost, direct/indirect cost, and cost drivers.
- Studies published between 1 January 2014 and 14 February 2025.
- The search date restriction was set to capture time before the first SMA drug approval.

Methodology: Overview of the SLR process



*Includes manual searches (key conference proceedings and bibliographic searches)

*Literature screening was performed by two independent reviewers, while data extraction was performed by one reviewer and data accuracy was ensured by another reviewer

Any conflicts in both the steps were resolved by discussion or by a third independent reviewer

Abbreviations: EBM: Evidence-Based Medicine; Embase: Excerpta Medica Database; HTA: Health Technology Assessment; MEDLINE: Medical Literature Analysis and Retrieval System Online; NHS EED: NHS EED: National Health Service Economic Evaluation Database; SLR: Systematic literature review

Impact of SMA on overall costs and HCRU

- Patients with SMA type 1 incurred the highest total costs (excluding DMT cost) PPPY (€107,807) compared with those with SMA type 2 (€90,267) and SMA type 3 (€52,440).⁶
- The mean direct medical and non-medical costs, excluding DMT costs, PPPY for SMA types 1–3 are presented in Figures 4 and 5.⁶⁻⁸

Figure 4. Direct medical costs

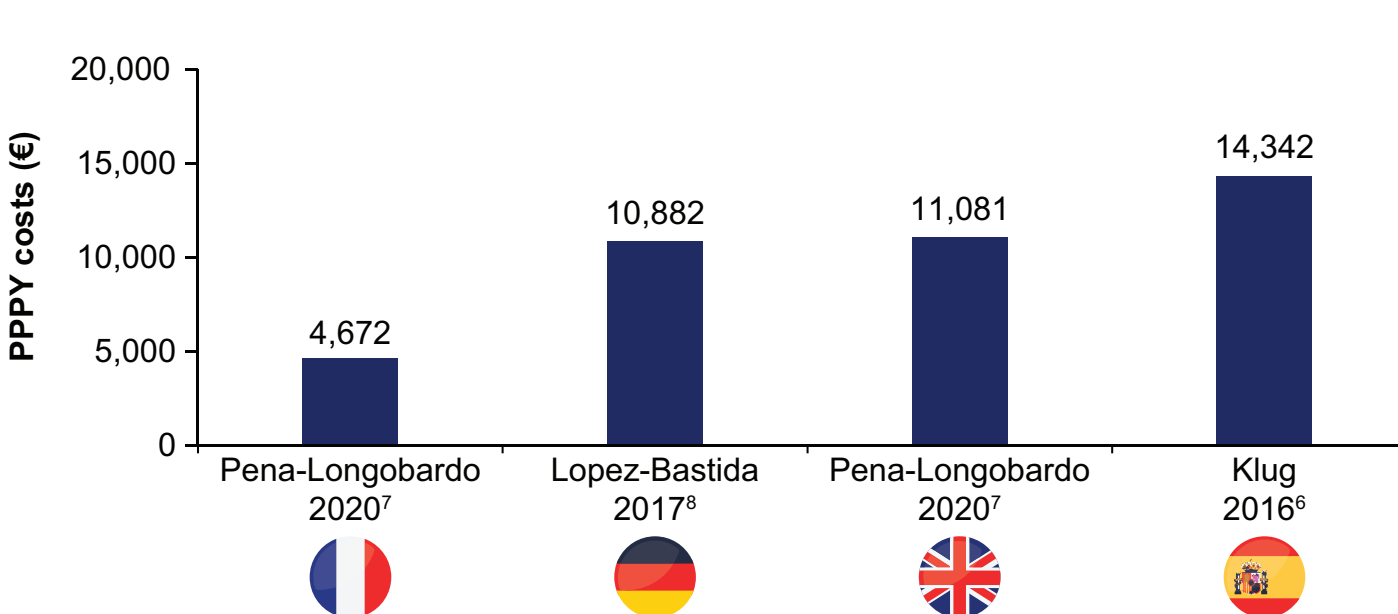
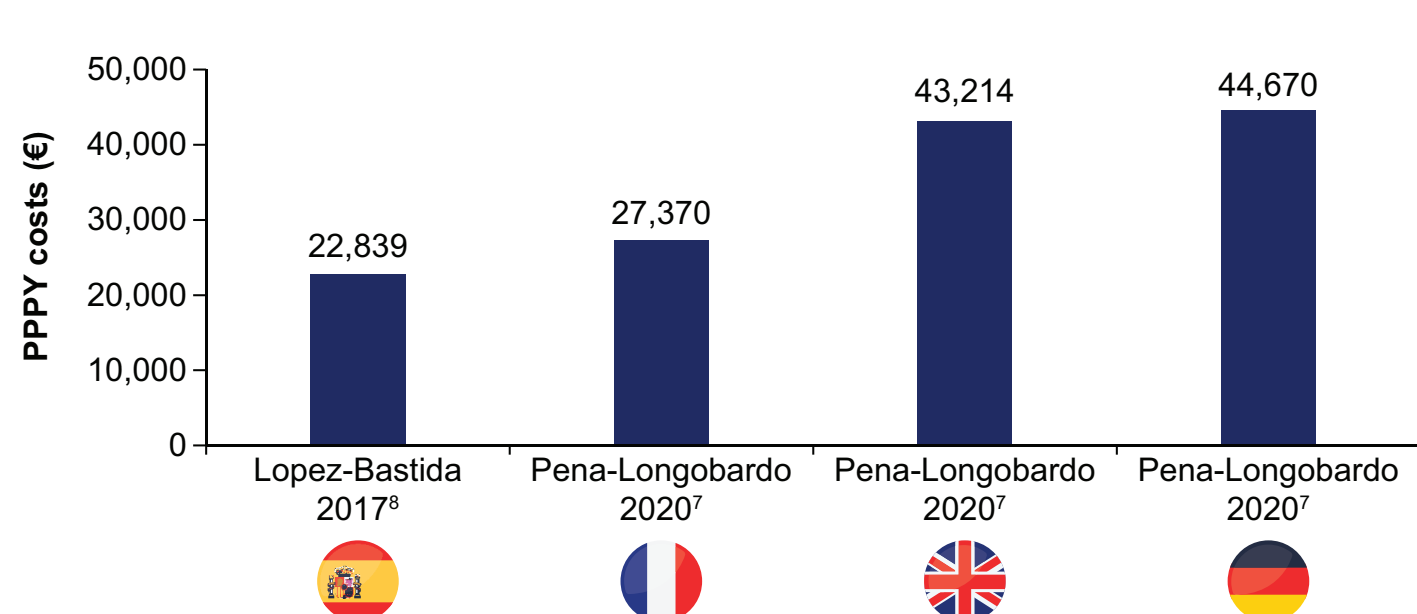


Figure 5. Direct non-medical costs



- When DMT costs were taken into account, a study from Germany reported total cost of illness (COI) at €206,856, with direct medical cost at €166,242 (80.5% of the total COI), with DMT cost accounting for the majority of direct medical cost 66.3% of the total COI (€137,118).⁹
- DMT cost, hospitalisation, and medical visits were the major direct medical cost drivers, while caregiver costs were the major direct non-medical cost driver.
- Mean PPPY indirect costs varied from €6,856 to €15,845 across Europe.^{6,9} Productivity loss was the major indirect cost driver.

Impact of non-adherence on costs and HCRU

- Patients with SMA types 1, 2, and 3 who were non-adherent to nusinersen treatment had greater HCRU days PPPY compared with those adherent to their dosing schedule (Table 1).¹⁰
- Compared with adherent patients, those non-adherent to risdiplam had higher median total healthcare costs by US\$335,049 for SMA type 2, US\$41,204 for SMA type 3, and US\$12,223 for SMA type 4.¹¹

Table 1. Summary of cost and HCRU studies: adherent to DMTs vs. non-adherent to DMTs

Study author, year	Items	SMA type 1		SMA type 2		SMA type 3	
		Adherent	Non-adherent	Adherent	Non-adherent	Adherent	Non-adherent
Gauthier-Loiselle, 2021 ¹⁰	HCRU days (PPPY); Nusinersen	42.2 days	48.4 days	45.2 days	109.6 days	54.8 days	71.7 days
Gauthier-Loiselle, 2021 ¹⁰	Mean total healthcare cost (PPPY)*: Nusinersen	US\$59,244	US\$85,042	US\$51,937	US\$146,830	US\$74,647	US\$105,657
Patel, 2024 ¹¹	Mean total healthcare cost (PPPY)*: Risdiplam	NR	NR	US\$49,062	US\$378,126	US\$34,249	US\$64,076

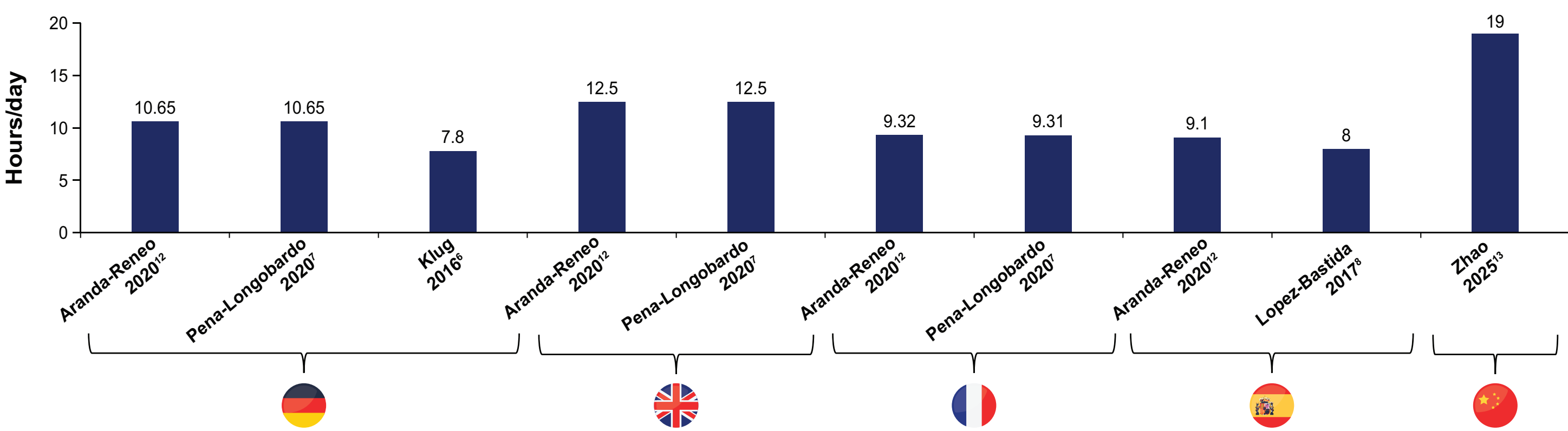
Abbreviations: HCRU: Healthcare resource utilisation; NR: Not reported; PPPY: Per patient per year; SMA: Spinal muscular atrophy

*Excluding costs related to nusinersen and OAV101 IV; *Excluding costs related to DMTs: Nusinersen, OAV101 IV, and risdiplam

Impact of SMA on caregivers

- Across the European countries, including the United Kingdom (UK), caregivers spent an average of 7.8–12.5 hours/day in caregiving for patients with SMA (Figure 6).^{6,7,12}
- The mean patient care time was highest for SMA type 1 (62.8 hours/week in Brazil, 79.4 hours/week in Taiwan),^{14,15} followed by SMA type 2 (79.7 hours/week in Argentina).¹⁶
- High patient care time negatively impacted employment, which led to either job change, reduced working hours, or stopping work entirely.¹⁴⁻¹⁶

Figure 6. Mean caregiving time spent by caregivers in caring for patients with SMA types 1–3



Costs and HCRU among presymptomatic SMA vs. symptomatic SMA

- Presymptomatic treatment resulted in lower direct costs vs. no or symptomatic treatment.^{17,18}
- Total costs were lower for treated patients who were identified by early testing than for patients identified and treated symptomatically (median cost PPPY: €294,463 vs. €311,002; $p = 0.006$).¹⁷
- In Turkey, annual direct medical costs (excluding DMT costs) were lower (US\$1,347) in presymptomatic DMT-treated patients vs. non-DMT-treated patients with SMA type 1 (US\$4,919).¹⁸

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DISCLOSURES

Salman Hussain, Michel Kroes, and Grace McCarthy are employees of Novartis.



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