EPH 243

Epidemiology, Diagnosis and Treatment of Spinal Muscular Atrophy in Türkiye: A Scoping Review

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INTRODUCTION

Spinal Muscular Atrophy (SMA) is a rare neuromuscular disease that has become a priority for healthcare policymakers in Türkiye due to the country's relatively high rate of consanguineous marriages and the strong advocacy of patient organizations. The overall rate of consanguineous marriages is around 25%[1].

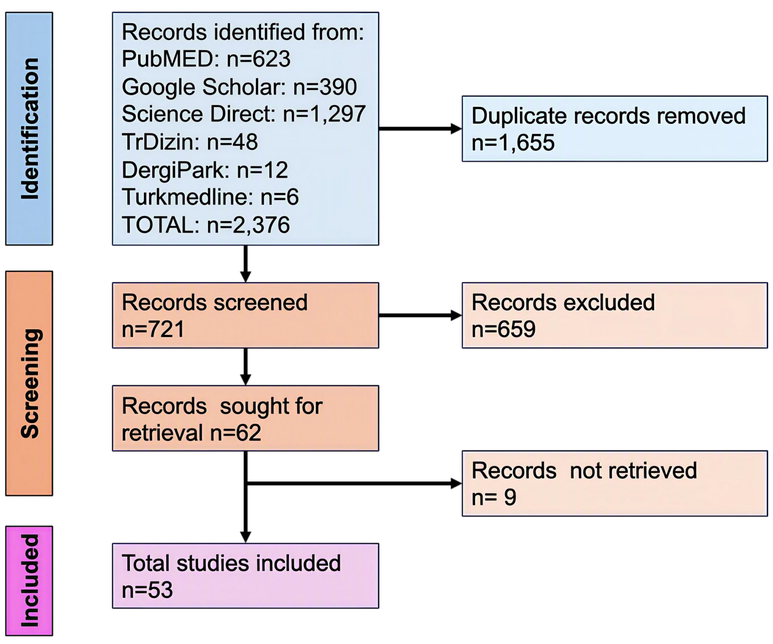
OBJECTIVES

This scoping review aims to provide a comprehensive overview of SMA in Türkiye by examining its epidemiology, patient demographics and clinical characteristics, diagnostic approaches, treatment strategies, and the findings of clinical studies conducted within the country.

METHODS

- A scoping review was conducted to address the research questions following the Preferred Reporting Items for Systematic Reviews and Meta-Analysis 2020 (PRISMA 2020).
- Both English and Turkish databases and websites were searched from January 01, 2000, to December 31,2025.
- As the country's official name changed from 'Turkey' to 'Türkiye' in 2022, an additional search was made using this name from January 01, 2022, to December 31, 2025.
- Studies in languages other than Turkish and English, theoretical papers, commentary pieces and letters, newspaper articles, editorial articles, case studies, conference abstracts and proceedings, and animal studies were excluded.
- Backward and forward research was also conducted to ensure no studies or publications were missed.
- Figure 1 illustrates the PRISMA flow diagram.
- In the initial search, 2,376 records were identified, of which 1,655 were duplicates. Of these, 659 records did not meet the inclusion criteria.
- At the end of this process, 53 articles were analyzed to answer the research questions.

Figure 1: PRISMA Flow Diagram

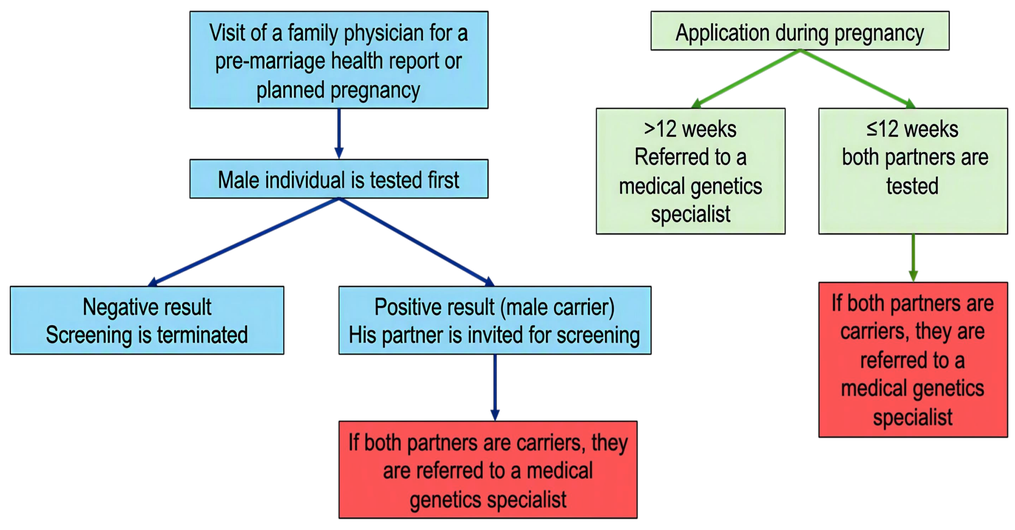


RESULTS

Epidemiology

- In Turkey, although the exact incidence and carrier rates of SMA are not precisely known.
- Considering that approximately 1,100,000 live births occur annually in recent years, the number of new cases per year is estimated to be between 130 and 180 (average: 150)[2].
- Approximately 3000 SMA patients are being monitored in Türkiye, according to data from the Turkish Ministry of Health's Public Health Directorate 2023 SMA Carrier Screening Program[2].
- Turkey launched an SMA Carrier Screening Program in 2021. Figure 2 illustrates the algorithm of the program.
- Studies from different regions and centers have shown a high prevalence of consanguineous marriages among the parents of patients. The prevalence of consanguineous marriages among SMA patients ranged between 25% to 75%[1,4,5].

Figure 2: SMA Carrier Screening Program[3]



- Among the SMA types, SMA Type one is the most frequently observed (50% to 60%)[6,7].
- In a study, the SMN2 gene copy number was one for 2.2% of the patients, two for 68.2%, three for 20.5%, four for 6.8%, and six for 2.2% of the patients[8].

Treatment

- The Social Security Organization (SGK) currently reimburses nusinersen and risdiplam for the treatment of SMA. Nusinersen entered the market in 2019 with the Named Patient Program and was moved to the positive list in 2023. Risdiplam has been reimbursed since 2025.
- Both treatments are reimbursed under the alternative payment route with hidden reimbursement prices.
- Current clinical studies mainly cover the outcomes achieved by nusinersen treatment as it is reimbursed for a longer time.
- Motor achievements have been recorded in the majority of studies involving nusinersen[2,9, 10,11,12, 13].
- Respiratory functions have also improved with nusinersen treatment[9,14,15,16].
- Scoliosis has been identified as a progressive complication associated with nusinersen treatment[17-21].
- Onasemnogene abeparvovec was evaluated concerning telomere length[21]. In the study, SMA patients who had not received gene replacement exhibited significantly shorter telomeres compared to their matched controls ($p = 0.029$), whereas gene therapy-treated patients showed no significant difference from controls ($p = 0.108$). The authors concluded that, while the exact contribution of telomere biology to SMA pathogenesis remains to be clarified, telomere length may represent a biomarker for disease severity and treatment response, as well as a potential therapeutic target.

CONCLUSION

The substantial economic burden of disease management, combined with relatively high prevalence of consanguineous marriages in Türkiye, particularly in certain regions, has made SMA an important public health concern for the country.

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Poster presented at ISPOR Europe 2026 Congress