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Original Article

Spinal muscular atrophy in Kosovo: a national clinical overview

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ABSTRACT

Spinal muscular atrophy (SMA) is a rare, inherited neuromuscular disorder characterized by progressive degeneration of alpha motor neurons, resulting in muscle weakness, loss of motor function, and reduced survival. Over the past decade, the introduction of disease-modifying therapies has significantly altered the natural history of SMA, particularly when treatment is initiated early in the disease course. Nevertheless, in many countries, including Kosovo, newborn screening programs for SMA have not yet been implemented, leading to delayed diagnosis after the onset of clinical symptoms. This study presents the first national clinical overview of genetically confirmed SMA patients from Kosovo, comprising 18 individuals (13 pediatric and 5 adult patients). We analyzed key clinical parameters, including age at symptom onset, age at diagnosis, current age, distribution of SMA types, and the highest motor milestone achieved. Our findings demonstrate substantial diagnostic delays, particularly among pediatric patients with early-onset forms of the disease, which may limit the effectiveness of available therapies. These results highlight the urgent need to implement newborn screening and to improve timely access to treatment in low- and middle-resource settings, in order to optimize clinical outcomes and quality of life for individuals with SMA.

Keywords: Spinal muscular atrophy, Kosovo, Age at diagnosis, Motor milestones, Newborn screening

Introduction

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disorder caused by biallelic mutations or deletions in the *SMN1* gene on chromosome 5q, resulting in reduced survival motor neuron (SMN) protein and progressive loss of anterior horn cells in the spinal cord [1-4]. The clinical spectrum of SMA is broad, traditionally classified according to age at symptom onset and highest motor milestone achieved, ranging from severe infantile-onset forms to milder adult-onset disease [5-7].

Historically, SMA was considered the leading genetic cause of infant mortality. Advances in molecular genetics and the development of disease-modifying therapies such as risdiplan, nusinersen, and onasemnogene abeparvovec have transformed SMA from a uniformly progressive and often fatal disease into a treatable chronic condition [8-10]. Importantly, clinical trials and real-world data have consistently shown that earlier treatment initiation, particularly in the presymptomatic stage, is associated with markedly improved outcomes, including achievement of motor milestones previously thought impossible [11-16].

As a result, many countries have implemented newborn screening (NBS) programs for SMA, enabling diagnosis before symptom onset and early therapeutic intervention [17-21]. Unfortunately, Kosovo currently lacks an SMA newborn screening program. Consequently, all patients included in this study were diagnosed only after the appearance of clinical symptoms, often following prolonged diagnostic journeys [22, 23].

Data from Southeastern Europe regarding SMA remains scarce. National reports are essential not only to describe disease characteristics but also to contextualize global advances within local healthcare systems [24]. This study aims to describe the demographic and clinical characteristics of all known SMA patients in Kosovo, with particular emphasis on age at diagnosis,

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symptom onset, current age, and highest achieved motor milestone.

Materials and Methods

This retrospective observational study includes all genetically confirmed SMA patients currently followed in Kosovo. A total of 18 patients were identified: 13 pediatric and 5 adult patients. Data were extracted from our clinical database and anonymized prior to analysis.

Clinical variables collected included

- Sex
- Current age
- Age at symptom onset
- Age at diagnosis
- Presenting symptoms
- SMA clinical type
- SMN2 copy number
- Highest achieved motor milestone
- Respiratory and feeding involvement (descriptive)
- Current disease-modifying therapy

All data were collected as part of routine clinical care. The study was conducted in accordance with the Declaration of Helsinki. Ethical approval for the study was obtained from the ethics committee, and all data were fully anonymized prior to analysis, with patient identifiers removed.

Results and Discussion

The national SMA cohort comprised 18 genetically confirmed patients, including 13 pediatric patients and 5 adults. There were 11 males and 7 females. All patients were receiving disease-modifying therapy with risdiplam at the time of analysis.

Pediatric patients ranged from infancy to adolescence, while adult patients represented long-term survivors with childhood- or adult-onset disease. This mixed-age structure reflects both historical survival limitations and the recent therapeutic era **(Table 1)**.

Table 1. Demographic and Clinical Characteristics

Variable	Pediatric (n=13)	Adult (n=5)	Total (n=18)
Male sex, n (%)	7 (54%)	4 (80%)	11 (61%)
Female sex, n (%)	6 (46%)	1 (20%)	7 (39%)
Age at diagnosis (range)	2 months–12 years	30–50 years	—
Median age at diagnosis	24 months	38 years	—

Age at symptom onset

Age at symptom onset varied widely across the cohort and closely reflected SMA clinical subtypes. In pediatric patients, symptoms typically begin in early infancy or early childhood. Common early manifestations included generalized hypotonia, delayed motor development, proximal limb weakness, reduced spontaneous movements, and feeding difficulties in the most severe cases.

In contrast, adult patients reported later-onset symptoms, often during late childhood, adolescence, or adulthood. These individuals frequently described slowly progressive proximal weakness, difficulties with running or climbing stairs, and increased fatigability. In several adult cases, early symptoms were subtle and not initially recognized as pathological.

Age at diagnosis

A marked diagnostic delay was observed across the cohort.

- In pediatric patients (n = 13), age at diagnosis ranged from 2 months to 12 years. When expressed in months, the median age at diagnosis was 24 months, and the mean age at diagnosis was approximately 35 months. Infants with more severe phenotypes were diagnosed earlier, whereas children with intermediate or milder presentations experienced longer diagnostic delays.
- Adult patients (n = 5) were diagnosed between their early 30s and 50s, despite symptom onset often occurring many years earlier.

Overall, age at diagnosis in this cohort was substantially higher than that reported in newborn-screened populations, where diagnosis typically occurs within the first weeks of life [25]. This delay reflects the absence of newborn screening for SMA in Kosovo and limited early access to genetic testing **(Figure 1)**.

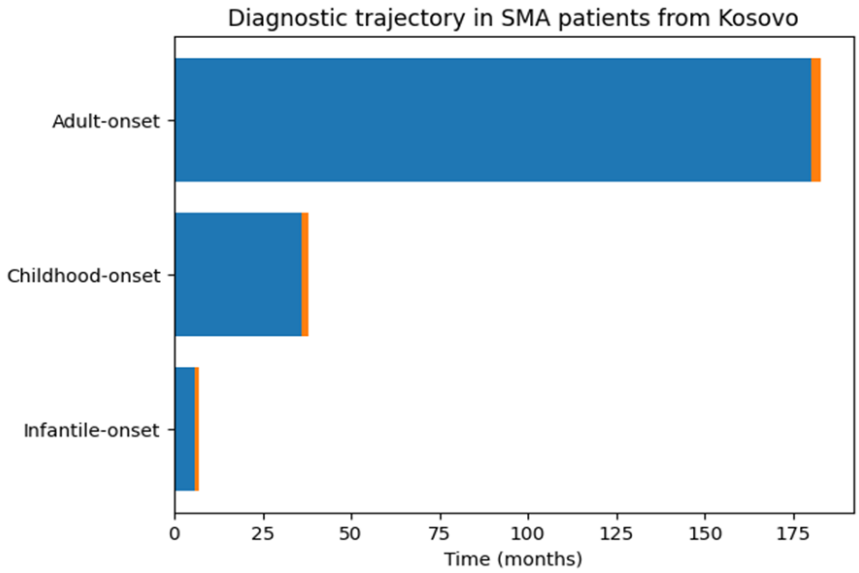

Figure 1. Diagnostic timeline in the Kosovo SMA cohort

SMN2 copy number

SMN2 copy number was available for 18 of the 18 patients (100%).

2 SMN2 copies: 5 patients, all with early-onset and the most severe disease

3 SMN2 copies: 11 patients, including 7 pediatric and 4 adult patients

≥4 SMN2 copies: 2 patients, 1 pediatric and 1 adult, both with later-onset phenotypes

As expected, lower SMN2 copy number was associated with earlier symptom onset and lower motor milestone achievement, whereas higher copy numbers correlated with milder phenotypes. This distribution is consistent with large international cohorts demonstrating an inverse relationship between SMN2 copy number and disease severity **(Table 2)**.

Table 2. Genetic Characteristics and Motor Milestones

Characteristic	Number of patients (%)	children	adults
SMN2 copy number available	18/18 (83%)		
SMN2 copies = 2	6 (40%)	6	0
SMN2 copies = 3	11 (47%)	7	4
SMN2 copies ≥ 4	2 (13%)	1	1
Independent sitting only	6 (33%)		
Standing ± support	4 (22%)		
Independent ambulation	8 (44%)		

Presenting symptoms

Presenting symptoms reflected both age and disease severity **(Table 3)**:

- Infantile-onset patients most commonly presented with hypotonia, delayed head control, reduced limb movements, feeding difficulties, and respiratory involvement.
- Childhood-onset patients typically exhibited delayed motor milestones, proximal muscle weakness, difficulty standing from the floor, and early scoliosis.
- Adult patients frequently reported long-standing weakness, gait abnormalities, muscle fatigability, and gradual functional decline [26, 27].

Notably, several families reported early concerns that were initially attributed to benign developmental variation, contributing to diagnostic delay.

Table 3. Presenting Symptoms by Age at Onset

Symptom	Infantile onset (n=6)	Childhood onset (n=7)	Adult onset (n=5)
Hypotonia	7 (100%)	2 (33%)	0
Delayed motor milestones	7 (100%)	7 (100%)	
Proximal limb weakness	6 (86%)	6 (100%)	5 (100%)
Feeding/swallowing difficulties	4 (57%)	1 (17%)	0
Respiratory involvement	3 (43%)	1 (17%)	0
Gait abnormalities/fatigability		3 (50%)	5 (100%)

Highest achieved motor milestones

The highest achieved motor milestone, before starting the therapy, varied widely and provided a clinically meaningful summary of disease severity.

- Independent sitting only: 6 pediatric patients
- Standing with or without support: 4 pediatric patients
- Independent ambulation: 3 pediatric patients and all 5 adult patients

Importantly, only one patient in this cohort received presymptomatic treatment. The father of the patient was diagnosed with SMA. Two of his newborn twins were tested two

months after birth, both of whom tested positive; one of them was asymptomatic. Compared with newborn-screened populations, where treated infants often achieve independent sitting and, in many cases, walking, motor milestone attainment in this cohort reflects irreversible motor neuron loss before diagnosis and treatment initiation [28, 29].

Treatment with risdiplam

All 18 patients are currently receiving risdiplam as disease-modifying therapy. Treatment initiation occurred after genetic diagnosis was confirmed. Families reported stabilization or subjective improvement in endurance and function, although objective longitudinal analysis was beyond the scope of this study. This first national overview of SMA patients from Kosovo provides an important snapshot of real-world SMA care in a country without newborn screening. The cohort highlights several key themes seen in other late-diagnosis populations but rarely documented in Southeastern Europe.

Diagnostic delay and its consequences

Consistent with historical cohorts, we observed substantial delays between symptom onset and diagnosis, particularly in pediatric patients with early-onset disease. In contrast, countries with established newborn screening programs report diagnosis within the first weeks of life, enabling treatment before irreversible motor neuron loss occurs [25, 28, 30-33].

Comparison with other cohorts

Large international registry and cohort studies show that, in the absence of newborn screening, age at diagnosis of spinal muscular atrophy varies widely by clinical subtype and is frequently delayed. Data from the Cure SMA Registry and European cohorts indicate a mean age at diagnosis of approximately 4–6 months for SMA type I, 15–22 months for SMA type II, and 4–8 years for SMA type III, with longer delays observed in milder phenotypes [11, 16, 27, 34-37]. In contrast, newborn screening programs enable diagnosis within the first 1–2 weeks of life, allowing presymptomatic treatment initiation [17, 28, 38-41]. In this context, the Kosovo cohort, where the median pediatric age at diagnosis was 24 months, reflects a similar pre-screening diagnostic pattern and highlights the importance of newborn screening for an earlier diagnosis.

These differences are not due to biological variation but rather to systemic factors, most notably the lack of newborn screening and delayed results of genetic tests.

Human impact

Beyond numbers, each delayed diagnosis represents a family navigating uncertainty, functional loss, and missed opportunities. Parents also described noticing early signs but facing prolonged referral pathways. Adult patients frequently normalized their weakness for years before receiving a definitive diagnosis.

Implications for policy

Our findings strongly support the urgent implementation of newborn screening for SMA in Kosovo. With disease-modifying therapies already available, screening represents the single most impactful intervention to improve long-term outcomes.

Limitations

This study is limited by its small sample size and retrospective design. Functional outcomes were described qualitatively rather than through standardized scales. Nevertheless, given the rarity of SMA and the national scope of the cohort, these data provide valuable insight.

Conclusion

SMA patients in Kosovo are currently diagnosed only after symptom onset, resulting in delayed treatment and limited motor outcomes compared with screened populations. Despite these challenges, access to risdiplam has enabled stabilization and ongoing care for both pediatric and adult patients.

The introduction of newborn screening for SMA in Kosovo is urgently needed to align national practice with international standards and to ensure that future generations of children with SMA have the opportunity for optimal outcomes.

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References

1. Lefebvre S, Burglen L, Reboullet S, Clermont O, Burlet P, Viollet L. Identification and characterization of a spinal muscular atrophy-determining gene. Cell. 1995;80(1):155-65.
2. Melki J, Abdelhak S, Sheth P, Bachelot MF, Burlet P, Marcadet A. Gene for chronic proximal spinal muscular atrophies maps to chromosome 5q. Nature. 1990;344:767-8.
3. Grant O, Wallace E. The influence of diversity-focused leadership on employee advocacy in selected Indian Fortune companies: The mediating roles of symmetrical internal communication and work engagement. Ann Organ Cult Leadersh Extern Engagem J. 2024;5:159-73. doi:10.51847/X2YHdX2Qz7

4. Kunie K, Kawakami N, Shimazu A, Yonekura Y, Miyamoto Y. Examining the impact of managerial communication on the link between nurses' job performance and psychological empowerment. Ann Organ Cult Leadersh Extern Engagem J. 2025;6:1-7. doi:10.51847/SF5ZX3J4OT
5. Mercuri E, Sumner CJ, Muntoni F, Darras BT, Finkel RS. Spinal muscular atrophy. Nat Rev Dis Primers. 2022;8:52.
6. Osluf ASH, Shoukeer M, Almarzoog NA. Case report on persistent fetal vasculature accompanied by congenital hydrocephalus. Asian J Curr Res Clin Cancer. 2024;4(1):25-30. doi:10.51847/0gjOEudJNr
7. Morgan AL, Foster DK, Collins IJ. Disparities in HER2-targeted therapy adoption and survival impact in metastatic HR−/HER2+ breast cancer: NCDB cohort study. Asian J Curr Res Clin Cancer. 2025;5(2):1-11. doi:10.51847/AZI4JURGlQ
8. Novak TJ, Dvorak PM. A spatiotemporal neural network framework for EEG-based emotion recognition in depression assessment. J Med Sci Interdiscip Res. 2025;5(2):24-38. doi:10.51847/A2pBOYHJW1
9. Solmell O, Sterner PD, Berg S. MRI of chronic low back pain: Correlation between pain, disability, and disc herniation. J Med Sci Interdiscip Res. 2024;4(1):22-7. doi:10.51847/hTOnlU7PdK
10. Schneider TL, Krüger BE. Breast cancer-specific mortality in stage IV patients with small tumors: Insights from a population-based cohort. Arch Int J Cancer Allied Sci. 2025;5(2):1-12. doi:10.51847/b9vFcweAVg
11. Nishio H, Niba ETE, Saito T, Okamoto K, Takeshima Y, Awano H. Spinal muscular atrophy: the past, present, and future of diagnosis and treatment. Int J Mol Sci. 2023;24(15):11939.
12. Lindstrom H, Jansson S, Lundgren P. Hospital pharmacists' knowledge, attitudes, and practices toward clinically significant drug interactions: A multi-center regional survey in Indonesia. Ann Pharm Pract Pharmacother. 2025;5:13-22. doi:10.51847/AtEgvCNECd
13. Csep AN, Voiţă-Mekereş F, Tudoran C, Manole F. Understanding and managing polypharmacy in the aging population. Ann Pharm Pract Pharmacother. 2024;4:17-23. doi:10.51847/VdKr0egSln
14. Miciak M, Jurkiewicz K. Recent advances in the diagnostics and management of medullary thyroid carcinoma: Emphasis on biomarkers and thyroidectomy in neuroendocrine neoplasms. Arch Int J Cancer Allied Sci. 2024;4(1):17-23. doi:10.51847/ar1ylTQfNa
15. Rani N, Gehrke P. Promoting intercultural competence in German medical students via innovative medical ethics education focused on Muslim patients – A pilot study. Asian J Ethics Health Med. 2025;5:1-12. doi:10.51847/0foncaeXr1
16. Oskoui M, Levy G, Garland CJ, Gray JM, Ohagen J, De Vivo DC. The changing natural history of spinal muscular atrophy type 1. Neurology. 2007;69(20):1931-6.
17. Glascock J, Sampson J, Connolly AM. Treatment algorithm for spinal muscular atrophy in the era of newborn screening. Neurology. 2024;102(6):e623-34.
18. Anunziata OA, Cussa J. Development and assessment of cyclophosphamide-loaded microspheres for enhanced topical drug delivery. Pharm Sci Drug Des. 2024;4:35-42. doi:10.51847/mrkjejeAVc
19. Clark A, Foster H. Network pharmacology integration and experimental verification to elucidate the molecular mechanisms of triptolide in treating membranous nephropathy. Pharm Sci Drug Des. 2025;5:33-47. doi:10.51847/X9UVmVSJ4E
20. Iriti A, Lupo M, Khazaal E. Perspectives and apprehensions of healthy individuals toward post-mortem brain donation: A qualitative study across Italy. Asian J Ethics Health Med. 2024;4:68-80. doi:10.51847/p7nqk1jS4l
21. Alnabulsi M, Ali EAA, Alsharif MH, Filfilan NF, Fadda SH. Medical students' perceptions, self-confidence, and willingness to handle in-flight medical emergencies: A cross-sectional study. Bull Pioneer Res Med Clin Sci. 2025;5(2):63-74. doi:10.51847/EQuNo67MNf
22. Jaafar NH, Rahman IA, Ter KZ, Ahmad B. The impact of non-classroom teaching on musculoskeletal pain in university students amid the COVID-19 pandemic. Bull Pioneer Res Med Clin Sci. 2024;4(1):50-7. doi:10.51847/UZ9DyvWUrn
23. Shen F, Bao L. Studying the effects of music on the time to gain independent oral feeding in premature infants. J Integr Nurs Palliat Care. 2025;6:1-6. doi:10.51847/xBTC4CiH10
24. Uneno Y, Morita T, Watanabe Y, Okamoto S, Kawashima N, Muto M. Supportive care requirements of elderly patients with cancer refer to Seirei Mikatahara General Hospital in 2023. J Integr Nurs Palliat Care. 2024;5:42-7. doi:10.51847/lmadKZ2u1J
25. Dangouloff T, Servais L. Newborn screening for spinal muscular atrophy: from trials to real-world implementation. Lancet Neurol. 2024;23(1):12-4.
26. Dubowitz V. Chaos in the classification of the spinal muscular atrophies of childhood. Neuromuscul Disord. 1991;1(2):77-80.
27. Mercuri E, Finkel RS, Muntoni F, Wirth B, Montes J, Main M. Diagnosis and management of spinal muscular atrophy: Part 1. Neuromuscul Disord. 2018;28(2):103-15.
28. Vill K, Kolbel H, Schwartz O. One-year outcomes of presymptomatically treated infants with spinal muscular atrophy identified by newborn screening. JAMA Neurol. 2024;81(2):142-51.
29. Peeters K, Boemer F, Deconinck N. Real-world effectiveness and safety of risdiplam in children and adults with spinal muscular atrophy. Neuromuscul Disord. 2024;34(3):235-44.
30. Ganea M, Horvath T, Nagy C, Morna AA, Pasc P, Szilagyi A, et al. Rapid method for microencapsulation of Magnolia officinalis oil and its medical applications. Spec J

Pharmacogn Phytochem Biotechnol. 2024;4:29-38. doi:10.51847/UllqQHbfeC

31. Raza S, Khan A, Mehmood F, Farooq U. Nationwide implementation of essential pharmacogenomic testing in the Netherlands: A decision-analytic model of lives saved and cost-effectiveness. Spec J Pharmacogn Phytochem Biotechnol. 2025;5:39-49. doi:10.51847/PUWEymkYkk
32. Ming S, Lei Z, Jie W. Peripheral neuropathy in diabetes patients at Jimma University Medical Center: Magnitude and contributing factors. Interdiscip Res Med Sci Spec. 2025;5(2):1-9. doi:10.51847/2aT3p1KejS
33. Ribeiro A, Martins S, Fonseca T. Progress and gaps in national medicines policy implementation in SADC member states: A comprehensive desktop review. Interdiscip Res Med Sci Spec. 2024;4(1):42-56. doi:10.51847/0eVBxAI8y0
34. Arnold WD, Kassar D, Kissel JT. Spinal muscular atrophy: diagnosis and management in a new therapeutic era. Muscle Nerve. 2015;51(2):157-67.
35. Cuenca-Martínez F, Herranz-Gómez A, Madroñero-Miguel B, Reina-Varona Á, Touche RL, Angulo-Díaz-Parreño S, et al. A systematic review of the literature on the connection between cervical spine abnormalities and internal disorders of the temporomandibular joint. J Curr Res Oral Surg. 2025;5:1-10. doi:10.51847/e4CoCM6iSZ
36. Mickevičius I, Astramskaitė E, Janužis G. A systematic review of the implant success rate following immediate implant placement in infected sockets. J Curr Res Oral Surg. 2024;4:20-31. doi:10.51847/PcPJL1v1XF
37. Jabin A, Guthrie A. Understanding treatment gaps in type 2 diabetes: A qualitative study on why patients stop and restart care. Int J Soc Psychol Asp Healthc. 2025;5:24-34. doi:10.51847/K4r85uzgEQ
38. Angilletta I, Ferrante R, Giansante R, Lombardi L, Babore A, Dell'Elice A. Spinal muscular atrophy: An evolving scenario through new perspectives in diagnosis and advances in therapies. Int J Mol Sci. 2023;24(19):14873.
39. Hsiao FH, Chen PL, Ho CC, Ho RTH, Lai YM, Wu JL. Exploring the impact of cognitive-behavioral therapy on anxiety disorders in children and adolescents. Int J Soc Psychol Asp Healthc. 2024;4:26-31. doi:10.51847/jcgvRFfQPM
40. Wong Y, Lin S, Cheng H, Hsieh T, Hsiue T, Chung H, et al. Understanding the impact of medical humanities on internship training and performance. Ann Pharm Educ Saf Public Health Advocacy. 2025;5:12-21. doi:10.51847/Z1fogzPksy
41. Alhossan A, Al Aloola N, Basoodan M, Alkathiri M, Alshahrani R, Mansy W, et al. Assessment of community pharmacy services and preparedness in Saudi Arabia during the COVID-19 pandemic: A cross-sectional study. Ann Pharm Educ Saf Public Health Advocacy. 2024;4:43-9. doi:10.51847/C52qAb0bZW