SMA in Ukraine: Between Therapeutic Breakthrough and Age-Based Discrimination

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Background.
Spinal muscular atrophy (SMA) remains one of the major challenges in modern neurogenetics. The introduction of disease-modifying therapy (DMT) has substantially improved patient prognosis; however, equitable access to treatment and real-world therapeutic strategies remain unresolved issues. Analysis of data from the "Children with SMA" Patient Registry provides an opportunity to assess actual DMT utilization patterns in Ukraine in 2024–2025.

Methods.
A retrospective analysis of the Registry database was performed. Of 489 genetically confirmed SMA patients, 339 active individuals (data updated within ≤24 months) were included. Variables analyzed included sex distribution, age at symptom onset, SMA clinical type, and structure of pathogenetic therapy (nusinersen, risdiplam, onasemnogene abeparvovec, and combination/switch regimens). Descriptive statistics and cross-tabulation were applied.

Results.
Among 339 patients, 53.7% were male. SMA type 2 was the most prevalent (37.2%), followed by type 3 (26.8%) and type 1 (24.5%). Presymptomatic diagnosis, mainly due to newborn screening, was documented in 4.7% of cases. The peak of symptom onset (28.9%) occurred between 6 months and 2 years of age.

Overall DMT coverage reached 71.4% (n=242). Risdiplam was the leading monotherapy option (48.7%), followed by nusinersen (26.0%). Gene replacement therapy as monotherapy was rare (1.6%). Combination or switch regimens accounted for 23.5% (n=57), predominantly risdiplam-based combinations. Only 10.8% of patients with SMA type 1 remained untreated, whereas 61.9% of individuals with adult-onset disease (≥18 years at manifestation) did not receive DMT, indicating a pronounced age-related disparity.

Conclusions.

The structure of therapy in Ukraine is largely determined by the national reimbursement model rather than purely clinical considerations. Risdiplam is the only state-funded DMT; nusinersen has been accessed mainly through humanitarian programs, and gene replacement therapy is not available domestically. Consequently, treatment choice is constrained by financial mechanisms rather than individualized clinical need. The coexistence of reimbursement-driven therapeutic imbalance and a significant age gap in DMT access—particularly affecting adults—highlights the urgent need to revise eligibility and reimbursement criteria to ensure disease stabilization regardless of age at onset.