Scoliosis development in spinal muscular atrophy under disease modifying therapies. How is changed the orthopedic approach.

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INTRODUCTION:

Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease that affects motor neurons and leads to hypotonia and muscle weakness. SMA causes systemic involvement and requires a multidisciplinary approach. Orthopedic management focuses primarily on the treatment of scoliosis, one of the most serious complications contributing to worsening lung function. The early identification of severe scoliosis progression and the implementation of preventive treatment are a primary objective in current therapeutic approaches. However, in most patients' spinal surgery remains the treatment of choice. The study intends to investigate the natural progression of scoliosis through a retrospective analysis of a single institute experience.

METHODS:

A retrospective analysis of longitudinal data from 43 patients treated in Hub Center was conducted. The inclusion criteria comprised patients with a genetically confirmed diagnosis of SMA, as evidenced by non-expression of SMN1, and scoliosis confirmed by X-ray. Linear regression analyses are used for studying the progression of scoliosis in relation to age. The study population was divided into subgroups to investigate scoliosis progression in relation to prognostic factors such as type of SMA, motor function, treatment with DMTs and HFMSE scores. For each subgroup, a trend of natural scoliosis progression was obtained.

RESULTS: 34 patients met the inclusion criteria. The most severe scoliosis progression was observed in patients with SMA type 1, with an average annual Cobb angle progression of 7.928°, and in non-sitter patients, who exhibited an average annual progression of 6.792°. Severe scoliosis developed early in SMA type 1 patients, with 78% of these patients reaching a Cobb angle greater than 40° by age 6. In walker patients' regression analysis showed a non-significant association between Cobb's Angle and age (p-value = 0.329) and scoliosis is maintained below values of 12°. Motor function, measured using the HFMSE scale, was inversely correlated with the Cobb angle ($p < 0.05$).

DISCUSSION AND CONCLUSION:

Compromised motor function and loss of ambulation are predictors of severe scoliosis progression. the effects of SMA therapies do not prevent scoliosis development. The improved prognosis may lead to a growing cohort of SMA type 1 and 2 patients with early onset scoliosis who require early growth-friendly surgical interventions. Scoliosis monitoring should begin in the first years of age for SMA type 1 patients. Continuous follow-up is essential to detect early severe scoliosis progression.