SCOLIOSIS IN SMA PATIENTS TREATED WITH DISEASE-MODIFYING DRUGS: A PRELIMINAR STUDY

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BACKGROUND

Disease-modifying drugs have improved survival and motor function in Spinal Muscular Atrophy (SMA) patients. However, few studies have investigated the evolution of scoliosis in treated SMA patients. This study aimed to explore the possible correlation between treatment and scoliosis evolution.

METHODS

Four SMA patients treated with disease-modifying drugs (3 with SMA type 1, 1 with SMA type 3) were enrolled. Clinical history and evaluations—including Cobb angle and motor scores (HFMSE, CHOP-INTEND, RULM)—were retrospectively collected when available, within a time window ranging from 5 to 1 year before T0 and compared to data at T0.

RESULTS

The mean Cobb angle from the first available spine X-ray was 20.25°. One patient did not meet the criteria for scoliosis (Cobb angle of 7°), while the other three presented with mild (n=1, Cobb angle of 12°) or moderate (n=2, Cobb angles of 30° and 32°, respectively) scoliosis. The highest values were observed in the three patients with SMA type 1.

RETROSPECTIVE DATA

The average time from therapy initiation to the first available Cobb angle measurement was 2.75 years. The first available retrospective motor scale scores were as follows: S.1 — CHOP INTEND: 24; S.2 — HFMSE: 17, RULM: 20; S.3 — HFMSE: 4, RULM: 10, CHOP INTEND: 41; S.4 — HFMSE: 44, RULM: 36.

T0 DATA

At our first evaluation (T0), the mean Cobb angle was 38.25°. An increase in Cobb angle was observed compared to retrospective data, despite patients already being on treatment.

The Cobb angles at T0 were as follows: S.2 (SMA1) increased from 30° to 60°; S.3 (SMA1) from 32° to 60°; S.4 (SMA3) from 7° to 18°; and S.1 (SMA1) from 12° to 15°.

Motor scale scores at T0 were S.1 — CHOP INTEND: 44; S.2 — HFMSE: 11, RULM: 20; S.3 — HFMSE: 2, RULM: 9, CHOP INTEND: 31; S.4 — HFMSE: 44, RULM: 37.

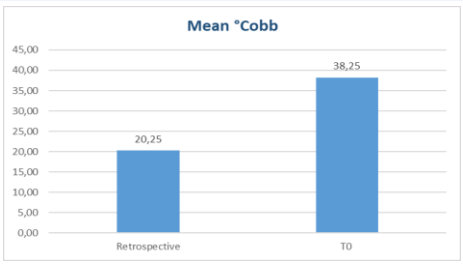

Fig 1.0 Mean °Cobb: retrospective –baseline.

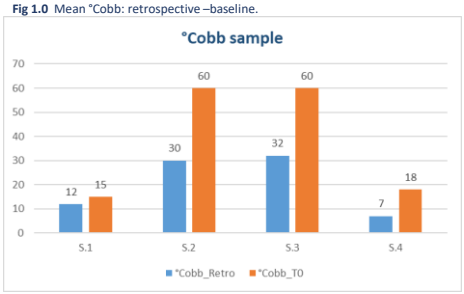

Fig 2.0 °Cobb sample: retrospective –baseline.

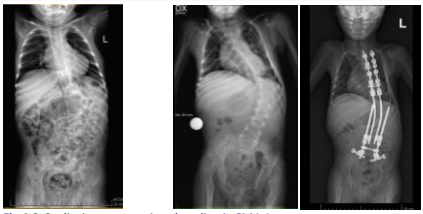

Fig 4.0 Scoliosis retrospective –baseline in SMA 1.

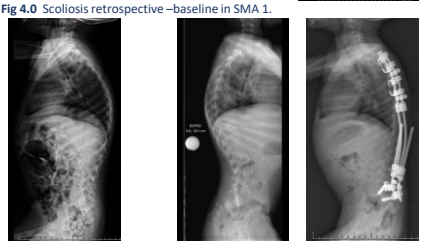

Fig 5.0 Kyphosis: retrospective –baseline in SMA 1.

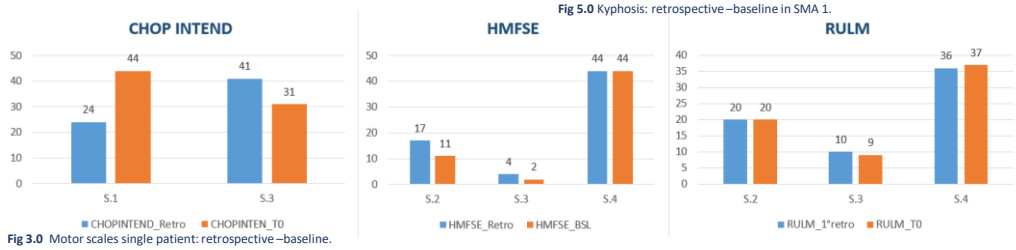

Fig 3.0 Motor scales single patient: retrospective –baseline.

CONCLUSION

Progression of scoliosis seems to be an expected feature even in treated patients. Changes in scoliosis seem to correlate with motor function and its progression over time, particularly in SMA1 patients.