

# EFFECTIVENESS OF NUSINERSEN IN PAEDIATRIC PATIENTS SMA1 AND SMA2

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## Introduction

Nusinersen is an innovative drug given by intrathecal injection, used to treat 5q-spinal muscular atrophy (SMA), a severe neuromuscular disorder due to a defect in the survival motor neuron1 (SMN1) gene. This antisense oligonucleotide drug modifies RNA splicing of SMN2 gene, thus increasing the production of full length SMN protein.

The first dose, given as soon as possible after the diagnosis, should be followed by 3 more doses after 2,4, and 9 (L1,L2,L3,L4) weeks and one dose every 4 months (M1, M2,M3...) thereafter.

This study aims to describe the efficacy of Nusinersen in terms of improvements in motor function in pediatric patients with SMA1 and SMA2.

## Methodology

From Feb-2018 we collected data from 8 patients, 3 with SMA 1 and 5 with SMA 2, using specific neuromuscular functional tests: CHOP-INTEND, HINE and HFMSE

## Results

Results are expressed in points of increase (p) of motor function scores from baseline (or from the first score recorded in our Center\*) to the score obtained at the time of last injection for each patient.

### SMA1 patients:

2 months old at the time of first injection(TFI):

CHOP-INTEND 8/64 to +38p(M2);

HINE 0/26 to +5p(M2).

3.3 years old TFI\*:

CHOP-INTEND 18/64(M2) to +16p(M6);

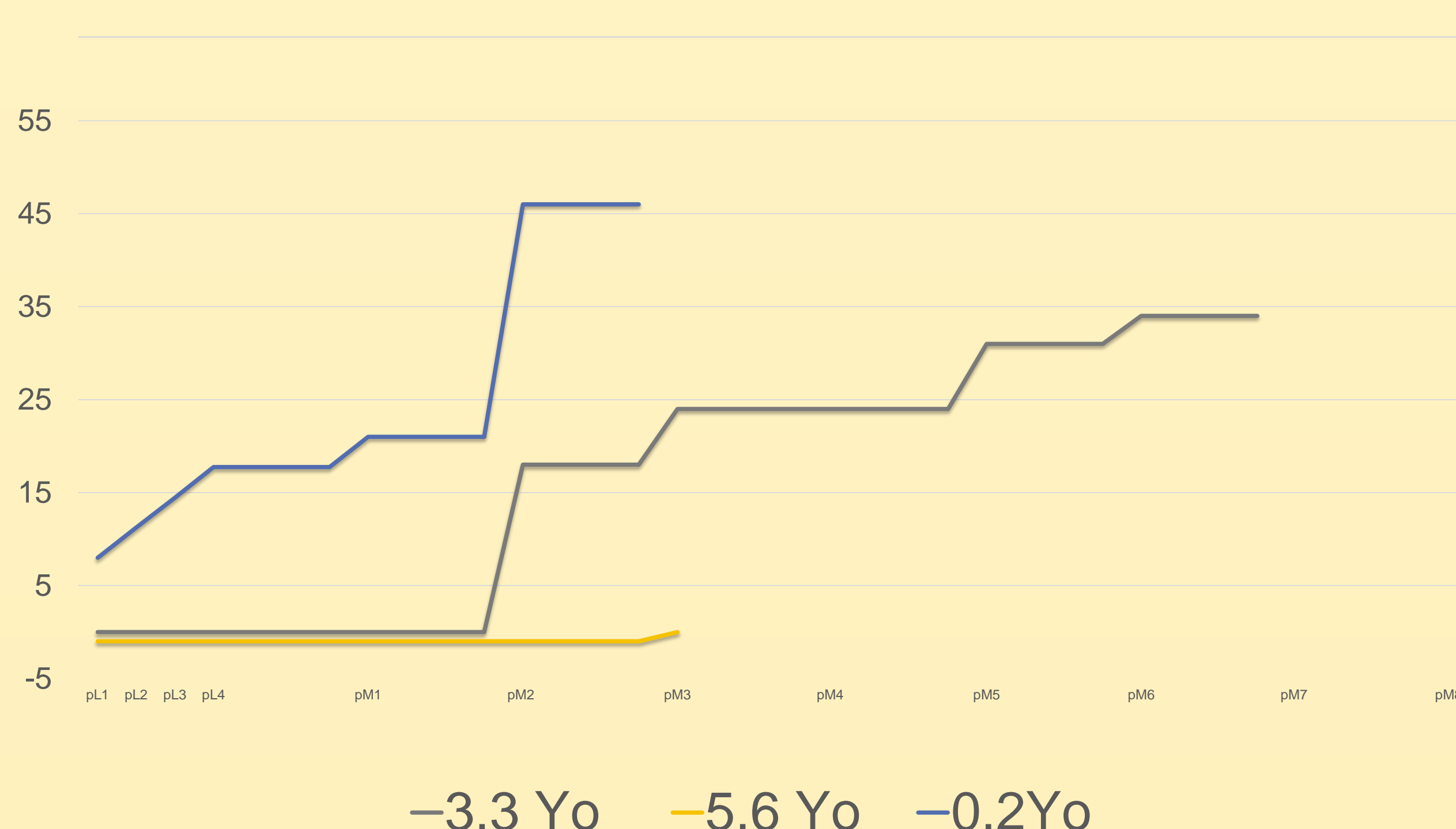
HINE 2/26, stable at M6.

5.6 years old TFI\*:

CHOP-INTEND 1/64 (M2) to -1p(M4);

HINE 0/26 (M2) to +1p (M4), then suspended for absence of efficacy.

### CHOP-INTEND SMA1



## Results

### SMA2 patients:

1.2 years old TFI:CHOP-INTEND 59/64 to -1p (M1).

3.4 years old TFI:CHOP-INTEND 41/64 to +8p (M1), +14p (M3);

HFMSE 8/66 to stable at M1 +4p (M3).

4.6 years old TFI:CHOP-INTEND 55/64 to +6p (M1) +7 (M2);

HFMSE 22/66 to, +3p (M1) +3p (M3).

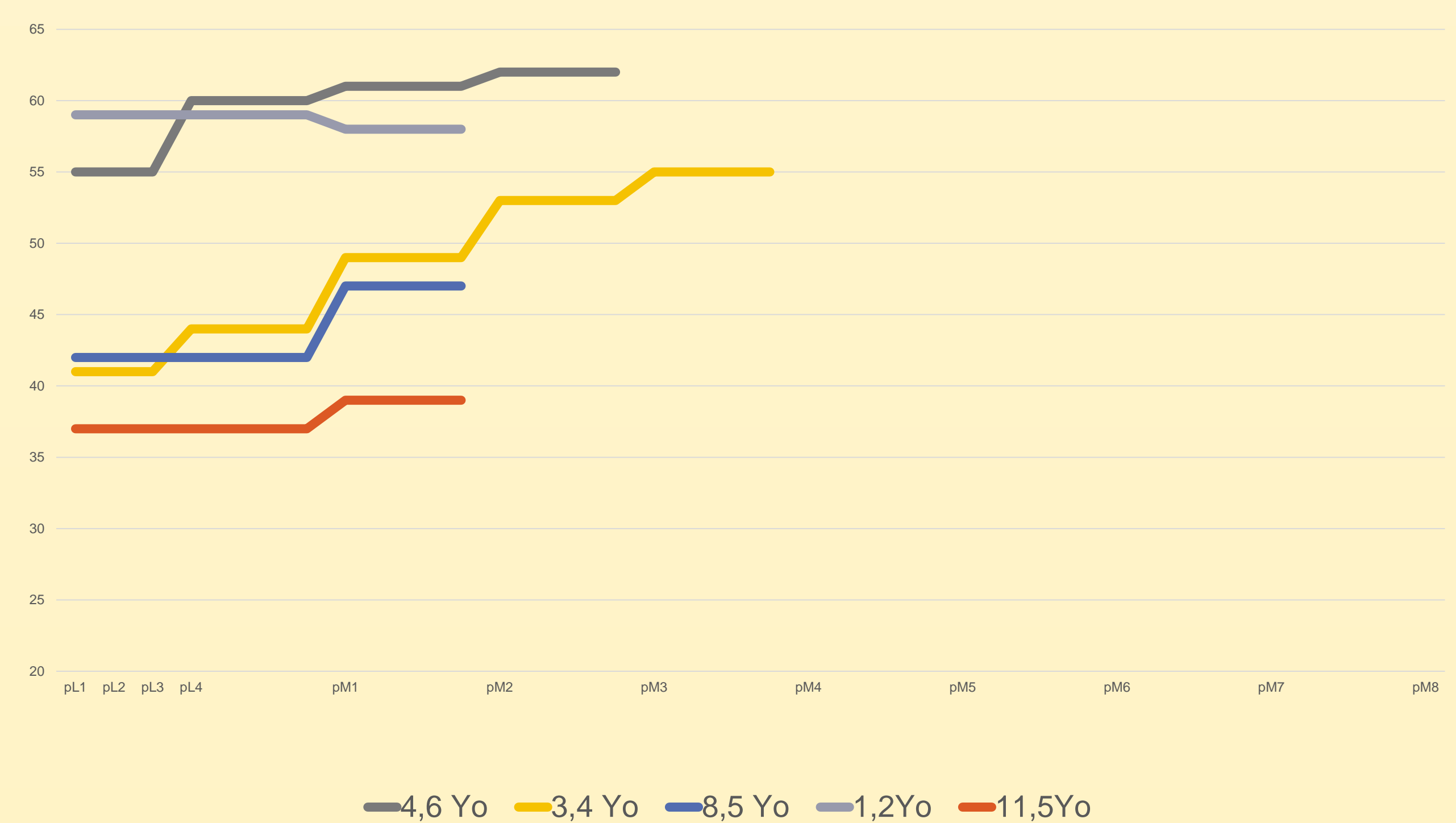
8.5 years old TFI:CHOP-INTEND 42/64 to +5p (M1);

HFMSE 17/66 to +10p (M1).

11.5 years old TFI:CHOP-INTEND 37/64 to +2p (M1);

HFMSE 8/66 **stable** at M1.

### CHOP-INTEND SMA 2



## Conclusion

Our results show an average increase of 4 points for CHOP-INTEND and 3.75 points for HFMSE in SMA2 patients, after 6-months(M1) of treatment.

For SMA1 patients it wasn't possible to evaluate the average trend for CHOP-INTEND and HINE scores after 6-months of treatment, because two patients started Nusinersen in other Hospitals (motor scores at L1-M1 not available).

Longer follow-up and data from other parameters such as swallowing and respiratory function are important to better understand the overall efficacy of Nusinersen.

