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ABSTRACT PREVIEW

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UNEXPECTED OUTCOMES IN A CHILD WITH SMA TYPE 1 (PRE-SPINRAZA TREATMENT)

Proposal Category: Poster - Case Study Report or Series

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Background & Purpose

Spinal muscular atrophy is an autosomal recessive genetic disorder. Type I (Werdnig-Hoffman, acute) is the most severe form of SMA. Onset is noted at 0-3 months due to hypotonia and severe weakness, feeding difficulties and/or need for respiratory support. Type I SMA has historically been characterized as fatal, with lifespan reported as 1 or 2 years. Children with SMA have a reduced number of anterior horn cells in the spinal cord, and there is progressive deterioration of the ones that are present, which leads to progressive loss of strength and function. The genetic defect in SMA is the survival motor neuron (SMN) gene on chromosome 5q11.2-13. SMN codes for SMN protein, which maintains the anterior horn cells. In the absence of adequate SMN protein, the anterior horn cells die.

Physical therapy intervention focused on 24–hour positioning programs and maintenance of upright positioning is crucial for children with Type I SMA for maintenance of optimal spinal alignment, prevention of contractures and optimization of function.

Case Description

Patient characteristics:
3 year old boy with SMA type 1
6/2014: Formally diagnosed at 7 months, significant weakness noted earlier
0 copies SMN 1, 3 copies of SMN 2 (mild type 1 SMA)
5/2015: initial PT equipment evaluation at Duke. Goals to progress towards upright activities and improved functional strength.
5/2015: functional mobility (19 months old): rolled supine to either side with mod-min A. Required max A to be supported in sitting
12/16: 2 week PICU admission due to acute respiratory failure
7/14/2017: first Spinraza dose

Outcomes

Functional skills attained prior to Spinraza initiation: independent rolling, independent head control, independent sitting (hands free), standing with KAFO's at a posterior support independently, independent reaching and manipulation of toys, independent propulsion of manual wheelchair (Panthera Micro), independent maintenance of prone on extended elbows with head erect once placed, independent step taking with/forward propulsion of Kaye body weight support walker.
Improved CHOP-INTEND scores over time:
11/30/2016: 53/64
4/26/2017: 48/64 (after hospitalization/PICU stay 12/2016-1/2017 due to acute respiratory failure
7/12/2017: 60/64

Discussion

What we know about the natural history of all types of SMA is likely to be changing. Disease modifying treatment with an antisense oligonucleotide (Spinraza) is now available for Spinal Muscular Atrophy (SMA), and gene therapy for SMA is also currently in clinical trials with exciting preliminary results reported. If Spinraza or gene therapy is administered in the newborn period, before cell death has occurred, the potential exists for children with SMA type 1 to sit, stand and/or walk. Physical therapy intervention has never been more critical for these children as we now will be focused on prevention of primary and secondary musculoskeletal impairments and improving function and participation more aggressively than ever before due to increased capacity for change. This patient demonstrated unexpected improvement before Spinraza and has continued to gain strength post-Spinraza administration.