

**OMTM, Volume 23**

**Supplemental information**

**Implications of circulating neurofilaments**

**for spinal muscular atrophy**

**treatment early in life: A case series**

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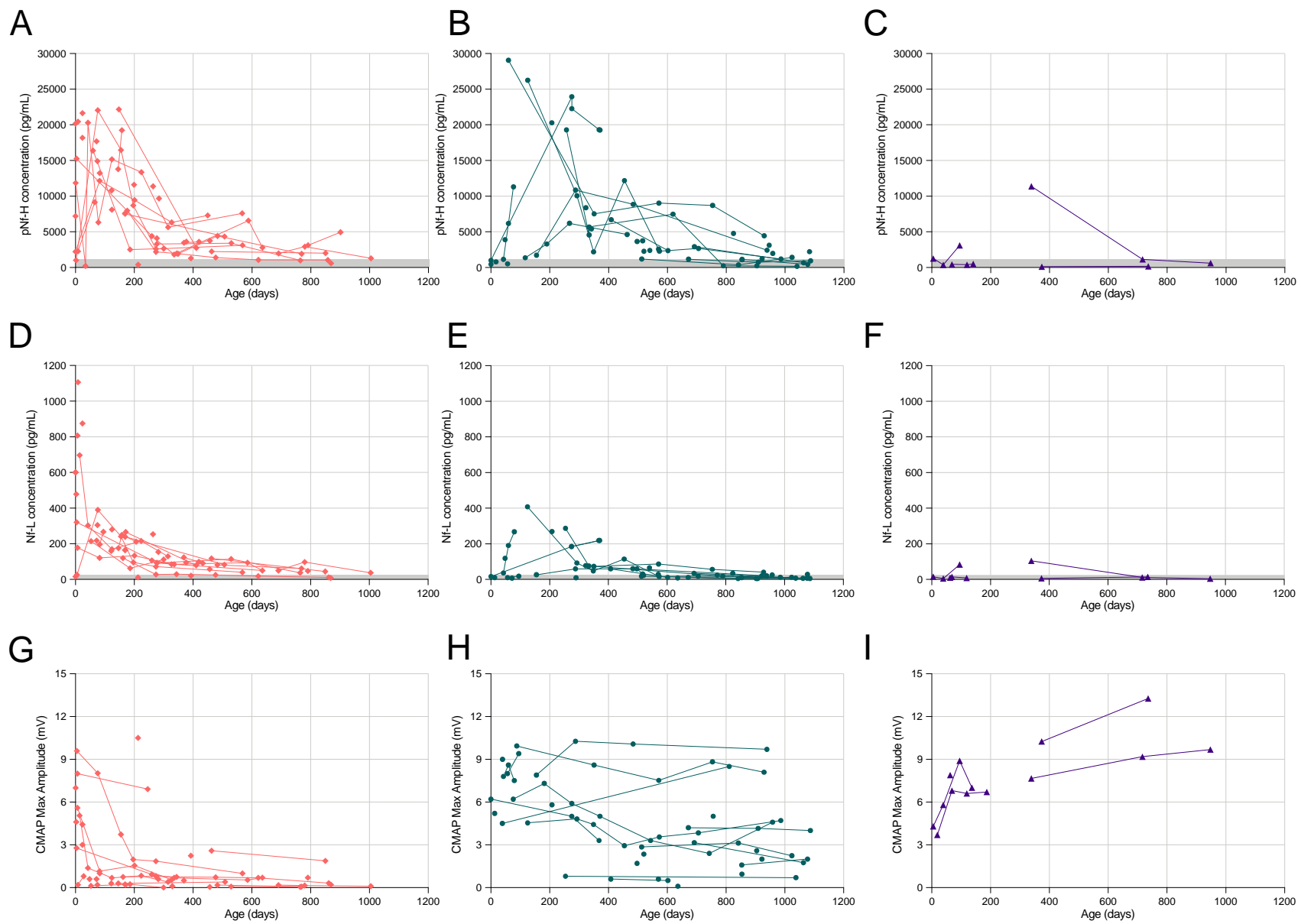
**Table S1.** Source used to determine milestones in each SMA patient receiving nusinersen and/or onasemnogene abeparvovec early in life.

<i>Subject ID</i>	<i>Elicited (E) or Independent Rolling</i>	<i>Sitting Independently</i>	<i>Standing Supported</i>	<i>Walking Independently</i>
<i>Nusinersen</i>				
507	HINE-2 (3)	HINE-2 (3)	HINE-2 (2)	-
556	-	-	-	-
618	Parent reported no video	Parent reported no video	-	-
630	Parent reported no video	-	-	-
508	CHOP-INTEND items 6/7 (4) (E)	HINE-2 (3)	HINE-2 (2)	HINE-2 (3)
579	HFMSE items 8/9 (2)	HFMSE item 1 (2)	Parent reported no video	HFMSE item 20 (2)
615	CHOP-INTEND items 6/7 (3) (E)	WHO-MM 10s	WHO-MM	WHO-MM
634	Parent reported no video	Parent reported no video	Parent reported with video	-
829	HINE-2 (3) (E)	HINE-2 (3)	-	-
<i>Onasemnogene abeparvovec</i>				
669	BSID-III item 25	WHO-MM 10s	WHO-MM	BSID-III item 43
813	CHOP-INTEND items 6/7 (3) (E)	BSID-III item 26 30s	BSID-III item 33	BSID-III item 43
825	CHOP-INTEND item 7 (4) (E)	HINE-2 (3)	WHO-MM	WHO-MM
849	CHOP-INTEND items 6/7 (3) (E)	WHO-MM 10s	HINE-2 (2), WHO-MM, BSID-III item 33	Parent reported with video
907	CHOP-INTEND items 6/7 (4) (E)	BSID-III item 26 30s	WHO-MM and BSID-III item 33	WHO-MM
818	CHOP-INTEND items 6/7 (3) (E)	WHO-MM 10s	WHO-MM	Parent reported with video
851	CHOP-INTEND item 7 (3) (E)	WHO-MM 10s	WHO-MM	WHO-MM
<i>Co-therapy</i>				
571	CHOP-INTEND items 6/7 (3) (E)	Parent reported with video	HINE-2 (2)	-
809	CHOP-INTEND items 6/7 (3) (E)	HINE-2 (3)	BSID-III item 33	-
832	CHOP-INTEND items 6/7 (3) (E)	Parent reported no video	Parent reported with video	-
846	CHOP-INTEND items 6/7 (3) (E)	Parent reported no video	-	-
913	CHOP-INTEND items 6/7 (3) (E)	HINE-2 (3)	HINE-2 (2), WHO-MM, BSID-III item 33	-
927	CHOP-INTEND items 6/7 (3) (E)	WHO-MM 10s	WHO-MM	-

**Table S2.** Demographic and clinical characteristics of SMA patients and healthy controls.

	<b>Controls</b> (n = 10)	<b>Natural History</b> (n = 47)	<b>Nusinersen</b> (n = 9)	<b>Onasemnogene abeparvovec</b> (n = 7)	<b>Co-Therapy</b> (n = 6)
<b>Age (days)</b>	57.4 ± 102.1 (0-360)	874.5 ± 879.7 (3-3791)	458.4 ± 329.8 (28-1082)	321.7 ± 216.7 (39-928)	389.5 ± 305.8 (56-1056)
<b>Sex, no. (%)</b>					
Male	4 (40.0)	28 (59.6)	4 (44.4)	5 (71.4)	3 (50.0)
Female	6 (60.0)	19 (40.4)	5 (55.6)	2 (28.6)	3 (50.0)
<b>Race, no. (%)</b>					
White	9 (90.0)	43 (91.5)	8 (88.9)	5 (71.4)	6 (100.0)
Asian	1 (10.0)	3 (6.4)	–	1 (14.3)	–
Black	–	–	1 (11.1)	–	–
Unknown	–	1 (2.1)	–	1 (14.3)	–
<b>Ethnicity, no. (%)</b>					
Hispanic or Latino	1 (10.0)	8 (17.0)	6 (66.7)	4 (57.1)	5 (83.3)
Not Hispanic/Latino	9 (90.0)	39 (83.0)	3 (33.3)	3 (42.9)	1 (16.7)
<b>SMN2, no. (%)</b>					
2	–	24 (42.1)	4 (44.4)	1 (14.3)	6 (100.0)
3	–	18 (31.6)	5 (55.6)	4 (57.1)	–
4+	–	3 (5.3)	–	2 (28.6)	–
<b>SMA Type, no. (%)</b>					
Type 1	–	25 (53.2)	2 (22.2)	–	4 (66.7)
Type 2	–	18 (34.0)	4 (44.4)	–	1 (16.7)
Type 3	–	3 (6.4)	1 (11.1)	–	–
Presymptomatic	–	3 (6.4)	2 (22.2)	7 (100.0)	1 (16.7)
<b>pNf-H (pg/mL)</b>	498 ± 261 (177-1092)	6092 ± 6710 (123-29060)	2797 ± 3864 (208-19431)	8127 ± 6320 (1367-29788)	3787 ± 306 (597-16940)
<b>Nf-L (pg/mL)</b>	10 ± 5 (3–19)	112 ± 198 (4-1826)	45 ± 57 (7-267)	121 ± 97 (22-433)	56 ± 49 (8-178)
<b>CMAP Amplitude (mv)</b>	12.0 ± 4.0 (9.1-14.8)	3.2 ± 3.2 (0-13.3)	5.7 ± 3.6 (0.4-12.1)	7.1 ± 3.0 (0.4-12.1)	1.6 ± 0.8 (0.5-3.3)

Data is presented as mean ± SD (range) or number of subjects (%)



**Figure S1.** Natural history data in SMA patients with 2, 3, and 4 *SMN2* copies.