[LBS.007] Natural History of Distal Motor Neuron Innervation in Spinal Muscular Atrophy: MUNE and Maximum CMAP Amplitude Studies, and Correlation with SMN 2 Copy Number

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MUNE and maximum CMAP amplitude values were obtained from the hypothenar muscle group in 70 patients with SMA, including 18 SMA 1, 40 SMA 2, and 12 SMA 3 subjects. Prospective longitudinal followup in four subjects identified prenatally or shortly after birth demonstrates a precipitous postnatal loss of distal motor unit innervation. Mean MUNE and CMAP values are substantially different among the three types in agreement with gross motor functional status, with the lowest values in SMA 1, intermediate values in SMA 2, and higher values in SMA 3 subjects. Decreasing MUNE values over time were observed for SMA 1 patients (P=0.001), but not for SMA 2 or 3 subjects, while a decline in CMAP amplitudes was observed in both SMA 1 and 2 subjects (P value < .05). Further analysis showed that SMA 2 subjects with higher CMAP and MUNE values and age less than 30 months are more likely to demonstrate a decrease in these values over time. Cross-sectional data from 40 SMA 2 subjects ranging from 16 months to 49 years of age (age versus MUNE value at first assessment) demonstrates an overall age-dependent decrease in MUNE values across the lifespan in this group. We found a modifying effect of SMN 2 copy number on functional status, and a correlation between SMN 2 copy number and MUNE and CMAP values. These data highlight the dramatic heterogeneity in SMA patients, and support the value of these techniques as outcome measures which may more closely reflect the pathophysiology of the disease process in SMA patients.

Families of SMA

Dr. Swoboda, Dr. Prior, Dr. McNaught, Dr. Wride, and Dr. Bromberg have nothing to disclose.

There is no unlabeled use to disclose.