Comparison of Adult vs. Pediatric Patients with Spinal Muscular Atrophy: An Axon Registry® Study

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Objective

To characterize patients with spinal muscular atrophy (SMA) in the American Academy of Neurology Axon Registry®.

Background

SMA was first described in the 19th century, however the genetic mutation was not discovered until the 1990's. The first treatment was not approved until 2016, with newborn screening not recommended until 2018. While these discoveries and protocols fundamentally changed the approach to care for children born with SMA, treatment and care for the adult population remain uncertain. The Axon Registry captures real-world data (RWD) on this unique adult population for whom clinical care may be different from children.

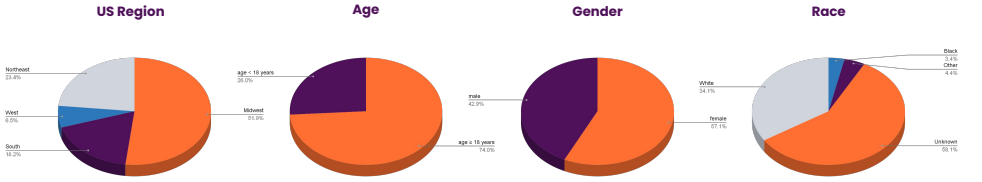
Methods

- 296 de-identified patients with SMA were manually selected in the Axon Registry if they had SMA in the opinion of the treating provider.
- SMA features (type, genetic copy number) were manually abstracted from EHR data contained in the Axon Registry.
- Demographic, care location and treatments were extracted from structured EHR data contained in the Axon Registry.
- De-identified Adult SMA patients(Age ≥ 18 at time of registry entry) were compared to de-identified pediatric SMA patients.

Conclusions

The Axon Registry contains RWD about diagnosis and treatment for adult patients with SMA who are not receiving care in academic practices. Though recent diagnostic and treatment advances focus on care of pediatric SMA patients, there is an opportunity to better understand the factors driving diagnosis and treatment in adults.

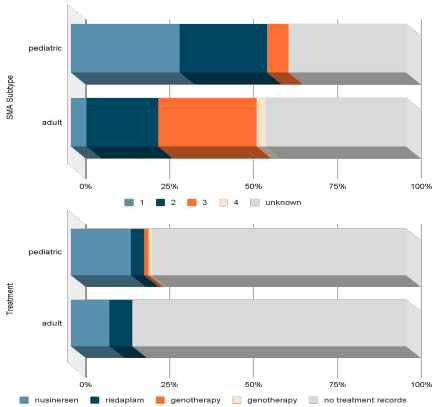
Results

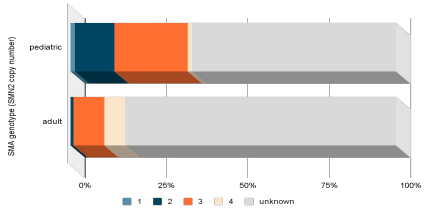


Academic practices:
Adults 22%, pediatrics 12% (p=0/0009)

Higher proportion adults than published registries (Belter et al).

Ethnicity:
60% not hispanic, 4% hispanic, 36% unknown





Compared with children, adults

- had less severe SMA type
- were similarly likely to have type documented
- were less likely to have genotype documented (p<0.0005)
- were similarly likely to receive disease targeting treatment (p=0.4).

Rad N, Cai H, Weiss MD. Management of Spinal Muscular Atrophy in the Adult Population. Muscle Nerve. 2022 May;65(5):498-507. doi: 10.1002/mus.27519. Epub 2022 Feb 26.

Belter L, Jarecki J, Reyna SP, Cruz R, Jones CC, Schroth M, O'Toole CM, O'Brien S, Hall SA, Johnson NB, Paradis AD. The Cure SMA Membership Surveys: Highlights of Key Demographic and Clinical Characteristics of Individuals with Spinal Muscular Atrophy. J Neuromuscul Dis. 2021;8(1):109-123. doi: 10.3233/JND-200563. PMID: 33104036; PMCID: PMC7902958.

Baca C.M., et al., Axon Registry® data validation: Accuracy assessment of data extraction and measure specification. Neurology. 2019. 92(18): p. 847-858

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The Axon Registry® is an initiative of the American Academy of Neurology ("AAN"). There are limitations of the Axon Registry data (Baca, 2019). The views expressed in this abstract represent those of the authors and do not necessarily represent the official views of the AAN

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