with non-mandatory data items. Data collected through the dataset includes genetic diagnosis, clinical observations, scoliosis, motor function, ambulation, nutrition, pulmonary function, disease-modifying therapies, medication and rehabilitation, hospitalisations and comorbiditis. The dataset dictionary is publicly available and data harmonization has been undertaken with key stakeholders in the wider SMA community. One year on, this poster will present an updated overview on the progress of the Dataset implementation, collected through online surveys and virtual interviews with curators across the network. Additionally, we will present data from an enquiry into the TREAT-NMD global registry network for SMA, to demonstrate the impact of the expanded dataset and illustrate its potential use in PMS.

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Evaluating perceived fatigue within an adult SMA population

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Patients with SMA often express an overwhelming sense of tiredness where they become exhausted with activity and have reduced endurance. Starting or sustaining voluntary activities can be difficult. They often suffer from a marked reduction in pulmonary capacity which can intensify fatigue. However, very few studies have measured and/or evaluated perceived fatigue within an SMA population. To quantify the degree of fatigue experienced among adults with SMA, Cure SMA conducted an online survey containing various patient-reported fatigue instruments and quality of life scales. 253 adults completed the online survey between December 9th-December 31st, 2020. Survey participants were randomized to receive three of the following five fatigue instruments: the modified fatigue impact scale, the multidimensional fatigue inventory, the fatigue severity scale, the PedsQL multidimensional fatigue scale, and the SMA-HI. All survey participants completed two quality of life scales, the SF-12 and the Health Utilities Index (HUI). The mean (sd) age of the survey participants was 38.2 (13.6) years. 5.9%, 44.7%, and 44.3% of survey participants self-reported to have SMA type I, II, and III, respectively. The summary subscale scores of each fatigue and quality of life instrument will be presented by SMA type, *SMN2* copy number, and motor function. If available, the results will be compared to the general or "healthy" population for reference. The results from this survey will highlight the perceived fatigue the SMA population experiences. We also hope to demonstrate this in comparison to the general population. Additionally, the various instruments will provide a baseline measure for future clinical trials.

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Longitudinal follow-up of adult patients with spinal muscular atrophy undergoing nusinersen treatment using an innovative magneto-inertial device

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Nusinersen was approved as the first medical treatment for all patients with spinal muscular atrophy (SMA). A recent study in a large cohort of adult patients treated with nusinersen demonstrated significant improvements in the Hammersmith functional motor scale expanded (HFMSE). However, clinically meaningful improvements were seen in only 40% of patients at 14 months despite a positive patients' experience. The main issue in open label study remains the risk of placebo effect, which could be largely compensated by the use of continuous and passive measures. We have demonstrated in a longitudinal natural history that a significant drop-in motor activity could be measured over a period of 1 year in SMA using magneto-inertial technology. We aimed to assess the efficacy of nusinersen in adults with spinal muscular atrophy with standard outcomes and by continuous measure issued from a wearable device. In order to achieve this, 13 (6 ambulant, 7 non-ambulant) patients with SMA type 2 and 3 aged from 23 to 59 years, were assessed before treatment and every 4 months for 14 months after the initiation. Evaluations included the HFMSE, the motor function measure, the revised upper limb module, dynamometric measures, the 6-minute walk, 4-stair climb, rise from floor, 10-metre walk tests. Around each visit, patients wore a magneto-inertial sensor at the ankle and at the wrist. Baseline HFMSE was 30.27± 23.04. HFMSE scores change over 14 months period was 1 ± 3.19. In non-ambulant patients, no decline was observed in the different variables computed (power at the upper limb level, vertical component of the movement), which contrasts with natural history data in which a drop around 5% was observed. In ambulant patients, stride length, stride velocity and maximal distance walked remain stable over 14 months.

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Assessment of current best-practices to triage and expedite incoming referrals for the evaluation of spinal muscular atrophy

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Spinal muscular atrophy (SMA) is an autosomal recessive neuromuscular disease characterized by progressive muscle weakness and atrophy. Clinical trial data suggests early treatment is critical to modifying disease progression and is expected to alter phenotype. Thus, early diagnosis is crucial. The purpose of the study was to evaluate appointment wait time for initial referral, and current best practices to expedite and triage referrals across neurology and SMA care centers in the US, when SMA is suspected. Cure SMA distributed the survey from July 9, 2020 through August 31, 2020 via Medscape to neurologists, child neurologists, and neuromuscular specialists; 279 responses were obtained. Cure SMA also distributed the survey to providers affiliated with SMA care centers yielding 26 additional completers. 43% of the general cohort and 85% of the SMA care centers reported average wait time of 0 to 2 weeks for referrals evaluating 'suspected SMA'. Additionally, 39% of the general cohort and 62% of the SMA care centers reported average wait time of 0 to 2 weeks for referrals evaluating 'hypotonia & motor delays' in infants. 85% of the general cohort and 100% of SMA care centers triaged incoming referrals. When evaluating triage methods, SMA care centers utilized 'centralized call center staff' (46%) and 'nurse coordinators' (38%) to prioritize appointments if key emergency words were included within the referral. 58% of physicians at SMA care centers and 44% within the general cohort reviewed incoming referrals and expedited urgent cases. Additional analysis revealed that amongst the general cohort, there was a 98.2% chance that infants with 'hypotonia and motor delay' would experience a 0 to 2 week *vs* 1 to 2 month wait time when the respondent indicated that at their site, the physician reviews referrals in advance and identifies any patients that may be urgent. Appointment wait time when SMA was suspected was significantly less at SMA care centers compared to the general cohort (p= 0.004). Guidelines supporting the triage of referrals specific to hypotonia and motor delays may relieve wait time and support early diagnosis and treatment of SMA.

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"Registry - SMA France: one year after launching the National French registry on SMA

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