

COMPASS

A Publication Dedicated to Research Updates | **SUMMER 2021**

Cure SMA Launches Phase VI of the SMA Industry Collaboration

Established in 2016, the SMA Industry Collaboration (SMA-IC) is a multi-faceted partnership that brings together pharmaceutical companies, Cure SMA, and other nonprofit organizations to share information, ideas, and data. The SMA-IC works together to address scientific, clinical, and regulatory topics critical to advancing drug development in spinal muscular atrophy (SMA). It is currently comprised of our partners at Novartis Gene Therapies, Biogen, Genentech/Roche Pharmaceuticals, Scholar Rock, and SMA Europe.

Through the SMA-IC, we fund research to ensure that effective, safe treatments can progress through clinical trials quickly and gain approval from the U.S. Food and Drug Administration (FDA) and international regulators. Our research also ensures these treatments address the unmet needs of the SMA community, and that the community's priorities and goals are incorporated into the development, review, and approval of therapies. The Collaboration consists of four topic groups, which are discussed throughout. The SMA-IC Joint Steering Committee works closely with Cure SMA to evaluate, prioritize, and fund activities that would advance its goals. Previous projects include:

- Creating interactions with the FDA ([2017 Patient-Focused Drug Development Meeting](#) and [2020 Critical Path Innovation Meeting](#)) and publishing a report, [Voice of the Patient](#).
- Launching an annual [Community Update Survey](#) to better understand the impact of SMA and how things are changing with the advent of therapies.
- Offering [SMArt Moves](#), an education and awareness campaign to reduce the time to diagnosis so babies with SMA can get the most benefit from approved treatments.
- Conducting [clinical trial readiness and education initiatives](#) to increase the number of sites that effectively participate in and conduct SMA clinical trials.

SMA Industry Collaboration Metrics At-a-Glance to Date

Number of Projects Completed Across IC Phases: **74**

Number of Individuals with SMA / Caregivers Surveyed: **2,130**

Number of Healthcare Providers Surveyed: **1,354**

Number of Provider and Researcher Engagements: **33,859**

Number of Manuscripts Published: **7**

Number of Toolkits / Online Resources Published: **17**

Number of In-person / Virtual Events Hosted: **15**

Number of Conference Engagements (e.g., podium presentations, posters, product theaters, booth exhibits): **46**

To learn more about SMA and its effects, please visit www.cureSMA.org, which provides information and resources for patients.



Regulatory Interaction and Outcome Measure Development Topic Group

The Regulatory Topic Group strives to engage the FDA and other regulatory agencies to bring the priorities of the community to the drug development and assessment process. In August 2020, Cure SMA held a successful [Critical Path Innovation Meeting \(CPIM\)](#) with the FDA. In this, key leaders in the Center for Biologics Evaluation and Research (CBER) and Center for Drug Evaluation and Research (CDER) discussed existing priorities in the SMA community—including shifting burden of disease, the identification and development of surrogate biomarkers to assess disease progression, and possible treatment effectiveness and combination therapy trials. A manuscript adapted from the briefing packet and meeting feedback is in the final stages of development.

This year, the Regulatory Topic Group has initiated efforts to develop and improve outcome measures for adults with SMA, in collaboration with the [Pediatric Neuromuscular Clinical Research Network](#) (PNCRN). This is critical as we often hear from the SMA community that developing more sensitive outcomes measures will pick up items more meaningful to individuals with SMA and their families. The group is now working to develop and validate the ATEND, a wheelchair-based motor function scale, as well as the SMA Patient Reported Outcomes (SMA-PRO), a patient reported outcome measure (PROM). The goal of the SMA-PRO project is to broaden the scope of certain measures to better meet the needs of individuals with SMA across ages and phenotypes. Efforts to devise a second PROM—the SMA Caregiver Reported Health Index (SMACR-HI)—are currently underway via a collaboration with the University of Rochester. The SMACR-HI will facilitate tracking of small, but clinically meaningful, changes in disease burden in pediatric populations.

Coming up in 2021, this Topic Group will also conduct a two-pronged consensus-building activity on outcomes for adults and older patients. We seek to formulate a working group of clinical experts to participate in a survey that will determine the most clinically meaningful outcomes for the SMA population and identify critical gaps in currently available measures. The Topic Group will subsequently capture the patient perspective via a survey patterned after the options considered by these clinical experts.

Education and Engagement Topic Group

The primary objective of Education and Engagement Topic Group is the reduction of diagnostic delays in symptomatic patients through education and awareness of parents, caregivers, healthcare professionals, and the public. Despite cancellation of in-person events, Cure SMA hosted virtual product theater and booth exhibition events at various professional conferences, engaging healthcare providers on the urgency for early diagnosis of SMA and administration of FDA-approved treatments. The Topic Group is also developing a telehealth webinar series looking at opportunities to optimize post-pandemic telehealth utilization and share guidance regarding the remote identification of suspected SMA in older children. Additionally, Education Topic Group members published the manuscript "[Awareness Screening and Referral Patterns Among Pediatricians in the United States Related to Early Clinical Features of SMA](#)," in *BMC Pediatrics*.

We are also working to engage individuals with SMA and healthcare providers regarding the unmet educational needs of the community. In Summer 2021, the Topic Group will survey adults with SMA aged 18 to 29 years, to evaluate needs upon transition from pediatric to adult care. The tool will assess the process to devise and implement transitional plans and explore the challenges these adults experience when engaging adult healthcare providers. Members will also evaluate the unmet needs of providers at known treatment sites via the distribution of a survey exploring challenges with treatment plan implementation and care coordination, as well as needed patient and provider educational resources. Eligible adults are encouraged to participate at <https://surveys.curesma.org/s3/Transition-to-Adult-Care-Survey>.



Clinical Trials Topic Group

The Clinical Trials Topic Group seeks to optimize readiness for SMA clinical trials at sites throughout the U.S. and Europe to better meet the needs of trial sponsors and the SMA community. This year, the group expanded its work on resources for physical therapists, recognizing their critical role in serving the SMA community (see Figure 1 below). Related activities include the development of a new competency-based training program for physical therapists, called [StepinSMA](#), and publication of an updated toolkit on outcome measures used in SMA. Considering the changes in care and research approaches spurred by the pandemic, the group has also taken on new research projects to assess community perspectives on telemedicine and to determine the impact of COVID-19 on clinical trial operations. Additionally, the group published a manuscript, titled “[I Have SMA, SMA Doesn’t Have Me](#),” in the *Orphanet Journal of Rare Disease*.

Lastly, in collaboration with SMA Europe, this Topic Group has continued engagement with clinical trial centers in Europe. During Q1 2021, SMA Europe launched Infopack 1, entitled “SMA- Pathology, Diagnosis, Clinical Presentation, Therapeutic Strategies, and Treatments,” to member countries. Efforts to translate materials into additional languages are underway. Infopack 2 (“SMA Standard of Care”) and Infopack 3 (“Experiences of Clinical Trials”) are in development.



Figure 1: Ongoing PT-related activities across the SMA-IC

Patient Reported Data Topic Group

The goal of this Topic Group is to collect patient-reported data to better understand SMA, the evolving phenotypes as new therapies are approved, and the continued burden of SMA. This year, the Topic Group launched the 5th Annual Cure SMA Community Update Survey, designed to capture self- and caregiver-reported data on the characteristics, experiences, and outcomes of individuals with SMA. Findings will be shared with federal and state policymakers, insurers, and other key officials to support the ongoing care and program needs of the SMA community. Additionally, the Topic Group is utilizing data from the Cure SMA membership database and the annual Community Update Survey to evaluate disparities in treatment access. A caregiver survey will also be launched in Fall 2021, to better understand the experiences, quality of life, and unmet needs of individuals caring for someone with SMA.

The findings from the 2019 Community Update Survey were published within *Orphanet Journal of Rare Diseases* in a manuscript entitled, “[SMA: A baseline dataset from the Cure SMA Community Update Survey](#).” The manuscript presents data captured via various PROMs, including the Health Utilities Index (HUI), the Work Productivity and Activity Impairment questionnaire (WPAI), and the Patient Reported Outcome Measurement Information System (PROMIS) Fatigue questionnaire. Additionally, in December 2020, the Topic Group launched a survey measuring perceived fatigue using PROMs instruments validated in diverse populations (see Figure 2 on next page) to compare how each instrument performs with adults with SMA. Analysis revealed fatigue levels did not differ by self-reported SMN2 copy number. Figure 2 summarizes the measures that we assessed. A manuscript summarizing key findings is currently in progress. The testing and selection of PROMs that can measure important and meaningful change in SMA is important to regulators, payers, and drug developers. Thus, during Q3 and Q4 of 2021, Cure SMA will also formulate a working group to develop a consensus on recommended PROMs for inclusion within future surveys, studies, and clinical trials, including for those tested in our annual survey.

Lastly, in collaboration with SMA Europe, this Topic Group seeks to evaluate the experiences of individuals with SMA in over 20 European countries with the distribution of The Daily Life Survey. This survey will be distributed to more than 1,500 patients and caregivers, assessing physical therapy, treatment expectations, and nutrition. The measure will also evaluate changes in patients’ feelings, expectations, and physical standing over time to capture the complexity of daily life with SMA.

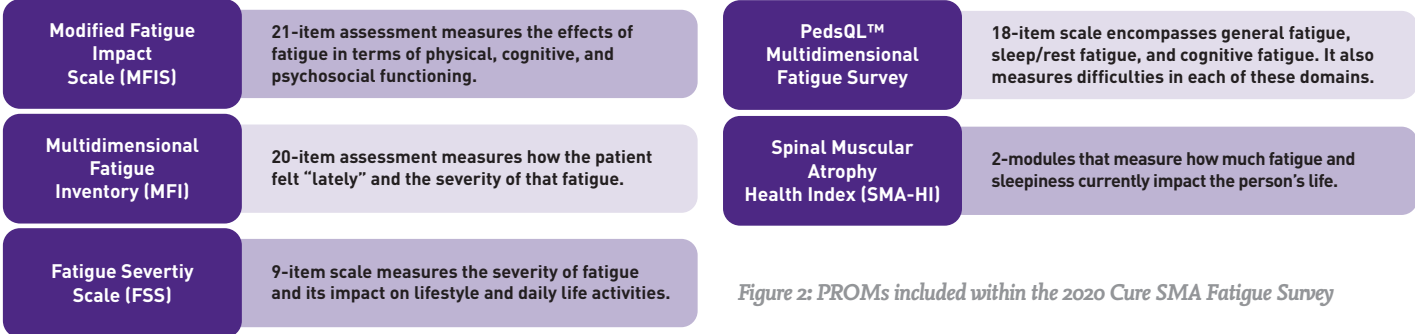


Figure 2: PROMs included within the 2020 Cure SMA Fatigue Survey

Conclusion

Thanks to the dedication of our community and the ingenuity of our researchers, we now have three approved disease-modifying treatments that target the underlying genetics of SMA and are expected to alter the phenotype. However, many unmet needs remain for the SMA community. As the experience of SMA continues to change, the priorities of the SMA-IC will also evolve. The SMA-IC provides a unique opportunity for Cure SMA to collaborate with academics, industry, government, and individuals with SMA and their families to actively identify and

advance goals important to the SMA community. Cure SMA appreciates the invaluable funding support for the SMA-IC, provided by our partners at Novartis Gene Therapies, Biogen, Genentech/Roche Pharmaceuticals, and Scholar Rock.

For additional information, please visit <https://www.curesma.org/sma-industry-collaboration/>.

Non-Profit Org.
U.S. Postage
PAID
Chicago, IL
Permit No. 1705

Cure SMA
925 Busse Road
Elk Grove Village, IL 60007
1.800.886.1762
Return Service Requested

