

# The specificity of collecting Patient-Reported Outcome Measures by “patient-driven” registries for rare diseases vs “doctor-driven” using the example of the Ukrainian Registry for Spinal Muscular Atrophy

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**Non-governmental patient-driven Foundation “Children with SMA” (CSMA) founded by parents of the children with Spinal Muscular Atrophy, dedicated to improving the quality of life for people living with SMA in Ukraine and supporting of scientific developments.**

Keywords: spinal muscular atrophy, rare disease registries, TREAT-NMD, CSMA

## INTRODUCTION

The Ukrainian SMA Registry refers to the disease-specific geographically defined population and aims to register all SMA cases in the population; using observational study methods to collect uniform data (clinical and other) aiming to evaluate specified outcomes for the population in Ukraine and serves scientific, clinical, and policy purposes. The utility of the Registry is meant to be international rather than internally within Ukraine.

## METHODS

A minimum common dataset (since 2004) and expanded core dataset (since 2018) is collected in collaboration with the TREAT-NMD Network, using patient-driven technology. Involvement of stakeholders such as patients, researchers and clinicians in the design, analysis and governance of the Registry was used to address the complexity and scarcity of knowledge on SMA.

### Patient-Reported Outcome Measures

PROMs can be applied to obtain data from the patient’s perspective. The data can guide decision-making about different clinical inputs, assist with monitoring the outcomes of specific interventions, provide a baseline assessment of the health status, and can offer pharmaceutical companies the chance to quantify the patient perspective on a disease or treatment. They are increasingly being used throughout rare diseases, shaping drug development, regulatory submissions, through discussions with payers, and new models of reimbursement. A number of organizations and industrial players have increased their efforts to develop relevant outcome measures for common disease studies or make recommendations on ways to improve patient relevant outcome measures used in patient-centred outcome research.

## RESULTS

In addition to using the dataset for scientific and social purposes, the dataset was used to conduct a mapping exercise to demonstrate the differences of care worldwide in a TREAT-NMD Network publication. These common and specific datasets enable comparison across Ukraine and internationally. This goal was achieved through easy access to the self-report method to input data. A comparative analysis with the method of entering data by the doctor was carried out. Disadvantages and benefits for patient-driven and “doctor-driven” data entry were assessed.

## CONCLUSION

The amount, frequency, and accessibility of data collection shows the high level of sustainability, and is useful for sharing patient information from “patient-driven” registries to increase the overall patient cohorts when natural history, clinical, patient-centred outcomes and new technologies can be statistically assessed. Attracting Patient Organizations to drive RD registries is more than desirable, but needs financial and legal support from all interested parties and the State as well as pharmaceutical companies. Tools for incentive are valuable.

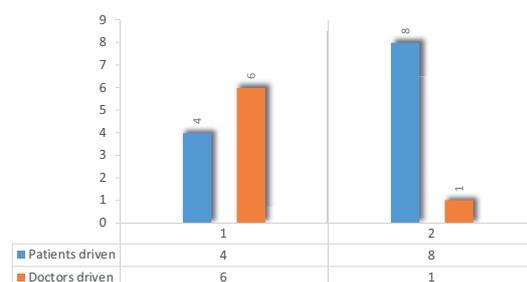
## REMARKS

Since 2004, the Registry registered 429 patients from all regions of Ukraine and neighbouring countries without their own registries. As of February 2020, the registry contains 257 active records. This number does not include 37 patients who relocated from Ukraine, 41 patients who died, 77 citizens of other countries, and 17 patients whose information has not been updated for 12 months or more (loss of contact).

ORIGINAL ITEMS	EXPANDED ITEMS
<ul style="list-style-type: none"> <li>• Personal data, demographics</li> <li>• Wheelchair use</li> <li>• Clinical diagnosis</li> <li>• Genetic test result</li> <li>• Best &amp; current motor function</li> <li>• Feeding function</li> <li>• Scoliosis surgery</li> <li>• Pulmonary function</li> <li>• Family history</li> <li>• SMA type</li> <li>• SMN2 Copies</li> </ul> <p>Module of PROs (under development)</p> <ul style="list-style-type: none"> <li>• Social performance/satisfaction</li> <li>• Fatigue</li> <li>• Activity participation</li> <li>• Emotional health</li> <li>• Pain</li> </ul>	<ul style="list-style-type: none"> <li>• Date&amp;cause of death</li> <li>• Clinical observations incl. contractures</li> <li>• HCP details</li> <li>• IV&amp;NIV use</li> <li>• Airway clearance/secretion mobilisation</li> <li>• FVC results</li> <li>• Medications&amp;disease-modifying therapies</li> <li>• Therapeutic interventions</li> <li>• Allopathic drugs</li> <li>• Hospitalisations&amp;co-morbidities</li> <li>• ≥ 1 validated motor outcome measure</li> <li>• Electrophysiology&amp;biomarkers taken</li> <li>• PRO: Clinical Global Impression of Severity (CGI-S)</li> <li>• Total Global Impression (TGI) according to patient/parent</li> </ul>

	“PATIENT-DRIVEN”	“DOCTOR-DRIVEN”
DISADVANTAGES	<ul style="list-style-type: none"> <li>•The need to quality-check patient-entered data;</li> <li>•the need for a consultant to ensure questions are phrased appropriately in online;</li> <li>•the need for regular missing data reminders;</li> <li>•the need for incentives to update data.</li> </ul>	<ul style="list-style-type: none"> <li>•Limited access to the registry due to the need to schedule a visit;</li> <li>•logistical issues to overcome (e.g. travel arrangements, special equipment required);</li> <li>•difficulties in accurately assessing patients with very severe conditions, can be made worse if patients have to travel;</li> <li>•limited capacity of involved doctors;</li> <li>•the need for financial incentive for a doctor or staff for data entry.</li> </ul>
BENEFITS	<ul style="list-style-type: none"> <li>•24/7 access to personal data;</li> <li>•no need for patient to travel/ attend an appointment;</li> <li>•opportunity to receive an online consultation instead of attending an appointment in person;</li> <li>•free recruitment of new members (no bureaucratic enrolment required);</li> <li>•low knowledge and expertise among healthcare professionals, information is trusted more when disseminated by parents and patient organisations;</li> <li>•the ability of curators to create/update requests at any frequency;</li> <li>•Curator Query Control;</li> <li>•network registry structure with regional curators allows for better management and maintenance of registry.</li> </ul>	<ul style="list-style-type: none"> <li>•Quality of data</li> </ul>

### DISADVANTAGES AND BENEFITS



CSMA is grateful for parents taking part in the Ukrainian Registry.

Partial financial support to implement the expanded TREAT-NMD dataset is provided by TREAT-NMD Network.