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# PROMs collection and the UK Spinal Muscular Atrophy Patient Registry

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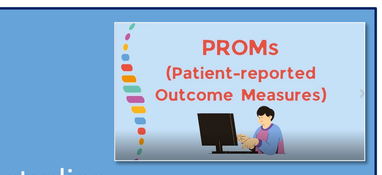
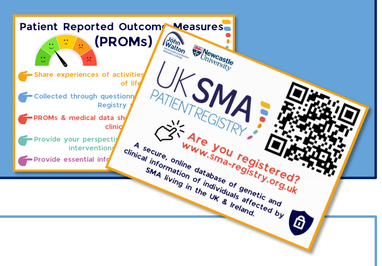
## About the UK Spinal Muscular Atrophy Patient Registry

- Created in 2008
- Collects the TREAT-NMD SMA Expanded Core Dataset
- Collects *patient-reported* data from individuals with SMA living in the UK & Ireland
- Registration is patient-initiated via an online portal (www.sma-registry.org.uk)
- Patients consent online and enter their clinical & genetic data in registry questionnaires
- Part of the TREAT-NMD SMA Global Registries Network, a global network of national SMA registries

## Aims of the registry

- Disseminate SMA-relevant information to participants
- Support the SMA community
- Aid the rapid identification of eligible patients for clinical studies
- Provide a source of information to academics, industry and healthcare professionals, including post-marketing surveillance data




## PROMs

In April 2022, the collection of patient-reported outcome measures (PROMs) was introduced in the registry to supplement clinical and genetic data held therein.

PROMs capture the perspectives of adults and caregivers of young people living with SMA about the impact of their condition and treatment, their quality of life and activities of daily living. Importance of the patient voice is increasingly recognised and valued.

Currently, SMA therapies Nusinersen and Risdiplam are available in the UK via managed access agreements (MAAs). PROMs data from the patient registry will supplement clinical data from SMA REACH UK (children) and Adult SMA REACH databases to inform review of treatment impact by UK regulatory authorities, clinicians, researchers and industry.

## PROMs aims

- To collect PROMs data within +/- 8 weeks of clinic visit from…

**50 adult & 50 paediatric SMA patients on Nusinersen**
**&**
**50 adult & 50 paediatric SMA patients on Risdiplam**

- To align PROMs with SMA REACH clinical data
- To anonymise, analyse and submit data to regulatory authorities for consideration as part of Nusinersen and Risdiplam MAAs.

| | Paediatric | Adult | Total |
|---|---|---|---|
| **Patients** | **199** | **443** | **642** |
| **Genetically confirmed patients** | **143** | **255** | **398 (62.0%)** |

***Table 1.*** *Demographic data (UK only), 29/08/2023*

## Method

Through collaboration with SMA REACH clinics, patients are encouraged to register in the UK SMA Patient Registry and complete questionnaires about their condition and PROMs through a secure online portal. The PROMs collected are…

- ➢Quality of Life - EQ-5D
- ➢SMA Independence Scale (SMAIS)
- ➢Patient Global Impression of Change (PGI)
- ➢Written comments in free-text box

Enabled through patient consent and data sharing agreements, patient-level PROMs are shared with each patient's SMA REACH clinic and with the SMA REACH coordination teams. In clinic, the data will inform patient care. At project coordination level, PROMs will be aligned with clinical data collected by SMA REACH.

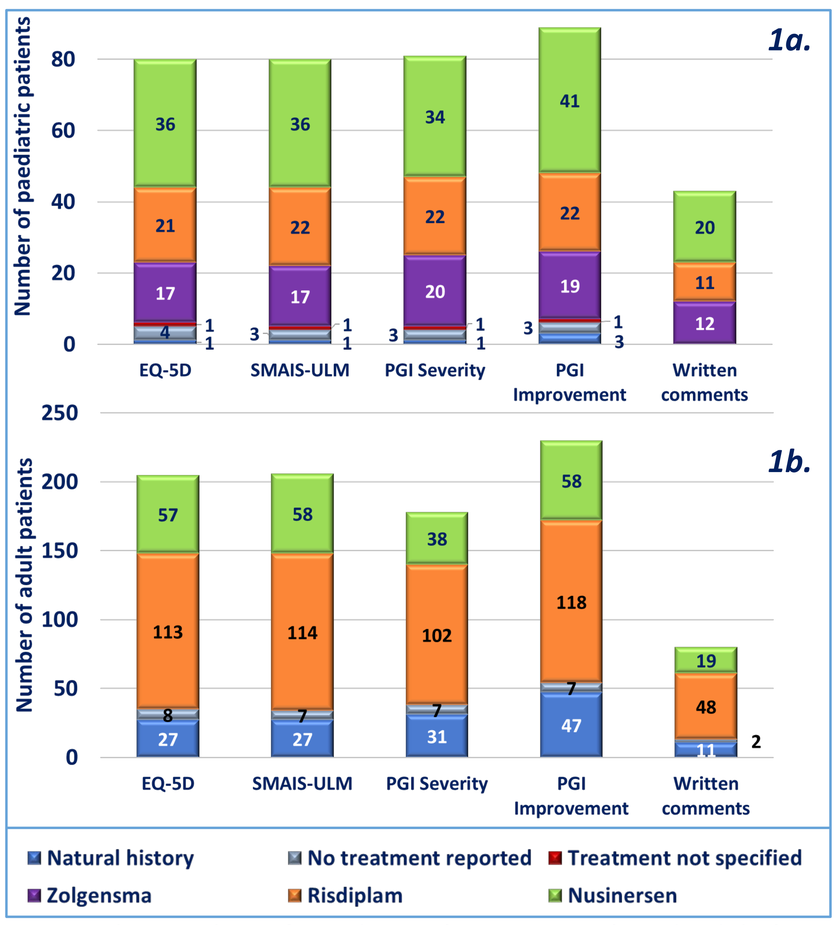



***Fig.1.*** *The number of paediatric (<16 years, a) and adult (≥16 years, b) patients who have reported PROM questionnaires at least once, stratified by treatment.*

## Results

The registry has 642 UK participants, 398 (62%) of whom have reported their genetic diagnosis of SMA (**Table 1**). PROMs have been completed by 230 adults and by the caregivers of 89 paediatric patients (**Fig.1**). To date, in the registry 144 adults and 46 children report treatment with Risdiplam, and 73 adults and 68 children report treatment with Nusinersen. The percentage of reported PROMs of each group is presented in **Fig.2**. Currently, the SMA REACH reference numbers of 208 adult and 136 child participants have been reported in the registry. Knowledge of a patient's SMA REACH reference number is essential to enable registry PROMs to be aligned with SMA REACH clinical data. Patients are encouraged to report their PROMs every six months, resulting in a steady accumulation of longitudinal PROMs data (**Fig.3**).

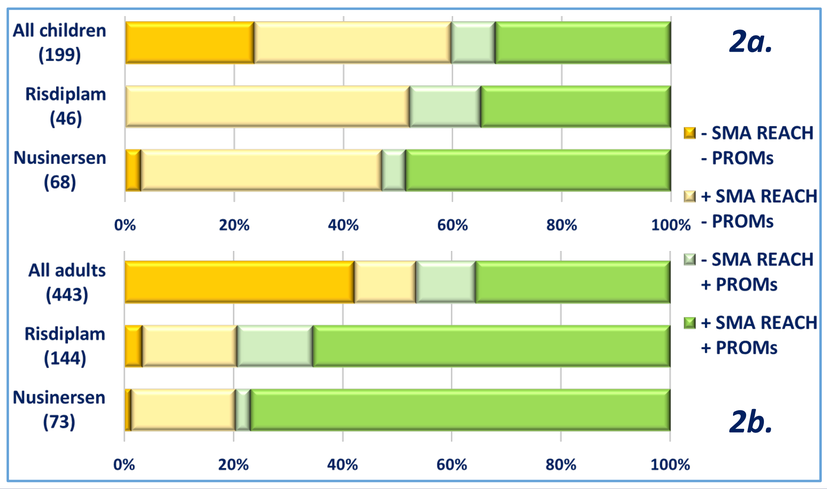



***Fig.2.*** *Percentage of paediatric (a) and adult (b) patients who have reported PROMs and for whom their SMA REACH reference number (SMA REACH) is known by the registry (total patient number per group is presented in brackets)*

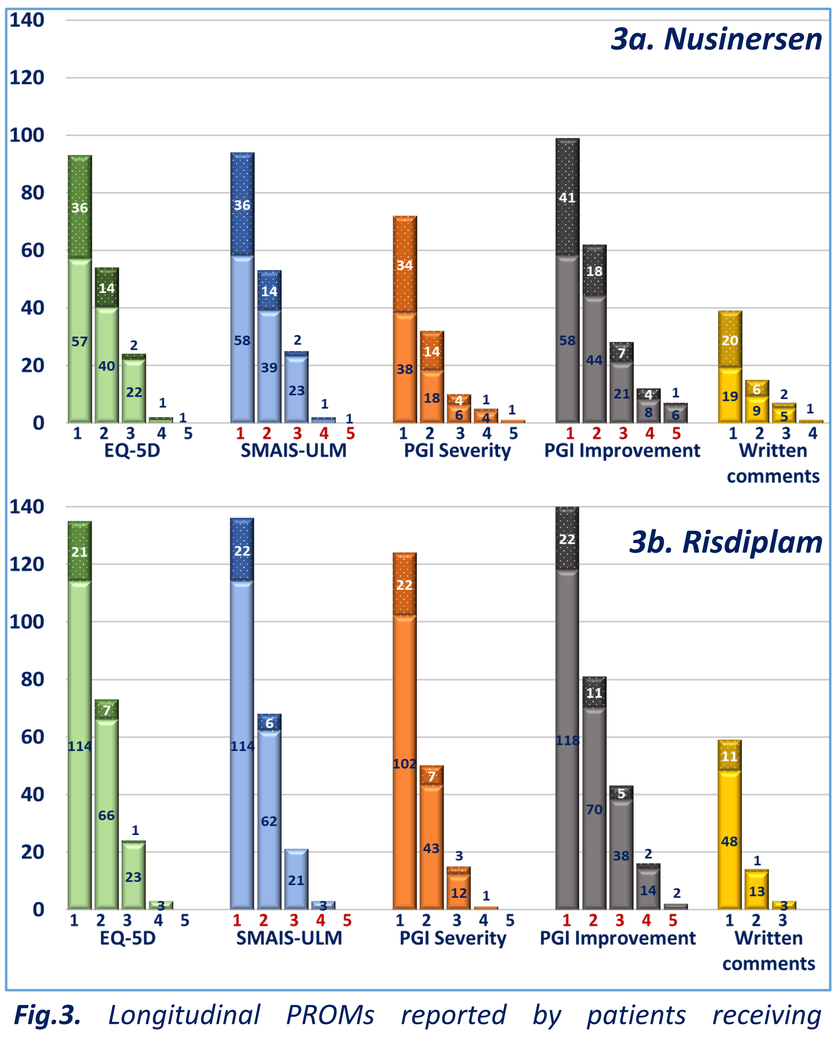



***Fig.3.*** *Longitudinal PROMs reported by patients receiving Nusinersen (a) and Risdiplam (b). The number of patients is presented against the number of reports of each PROM, at intervals of approximately six months. (Solid colour bars – adult data; Spotted colour bars – paediatric data)*

## Conclusions

Expansion of the UK SMA Patient Registry to collect PROMs supports UK SMA data collection and supplements SMA REACH clinical data, thereby assisting in therapy evaluation by regulatory authorities. Analysis presented herein identifies that there are a number of registry participants receiving treatment who need to be further targeted and encouraged to report PROMs. The accumulation of PROMs is positive and steady but needs continued effort from the registry, healthcare professionals and patient organisations to reach out to patients, emphasise the importance of PROMs and their impact on future decisions made about SMA treatments within the UK.

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