

## PROMs collection and the UK Spinal Muscular Atrophy Patient Registry

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### About the UK SMA 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](http://www.sma-registry.org.uk))
- Patients consent online and enter their clinical & genetic data in the registry questionnaire
- Part of the TREAT-NMD SMA Global Registries Network, a global network of national SMA registries

The registry aims to:

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

### 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
<b>Patients</b>	<b>196</b>	<b>414</b>	<b>610</b>
<b>Genetically confirmed patients</b>	<b>91</b>	<b>199</b>	<b>290 (47.6%)</b>

### 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 will be 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.

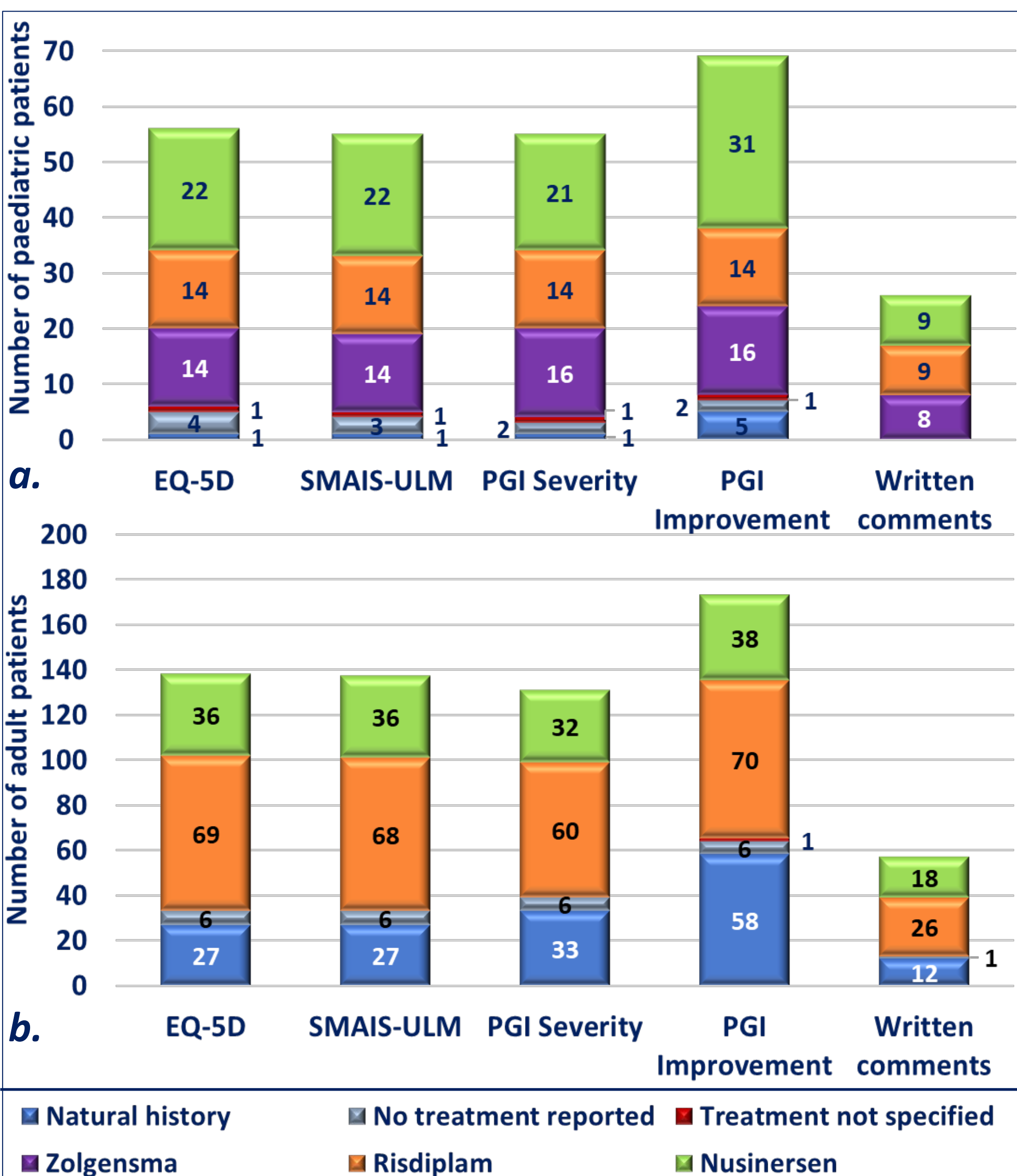


Fig.1. The number of paediatric (a) and adult (b) patients who have reported PROM questionnaires at least once, stratified by treatment.

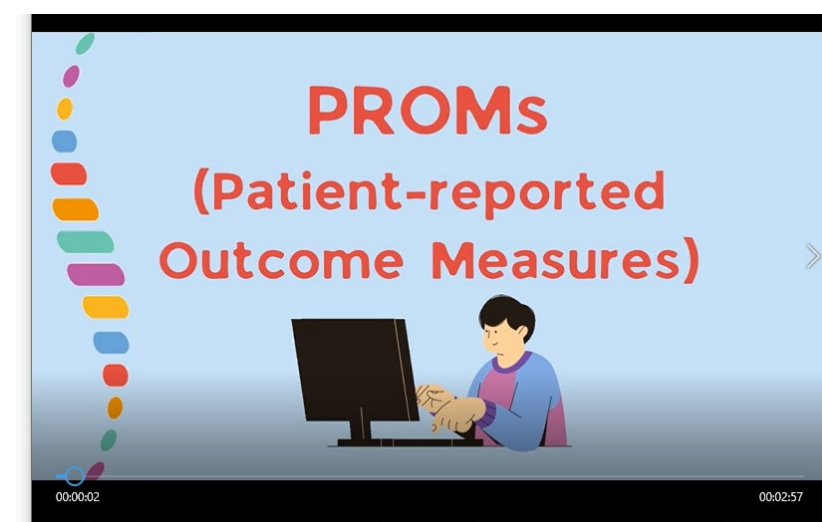


Table 1. Demographic data (UK only), 21/03/2023

### Results

The registry has 610 UK participants, 290 (48%) of whom have reported their genetic diagnosis of SMA (Table 1). PROMs have been completed by 137 adults and by the caregivers of 58 paediatric patients (Fig.1). To date, in the registry 93 adults and 22 children report treatment with Risdiplam, and 50 adults and 44 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 83 adult and 40 child participants have been reported in the registry. The number of patients with an SMA REACH reference number who have i. reported PROMs and ii. reported PROMs in a +/- eight week window of their clinic visit are reported in Table 2.

### 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.

	Paediatric		Adult	
	Risdiplam	Nusinersen	Risdiplam	Nusinersen
<b>Patients in registry (self-reported treatment)</b>	22	44	93	50
<b>Patients with SN reported in registry</b>	11	24	47	32
<b>SN patients who have reported PROMs</b>	10	10	32	23
<b>SN patients with +/- 8 week in-window PROMs</b>	7	8	25	21
<b>Total number of +/- 8 week in-window PROMs</b>	8	8	37	31

Table 2. PROMs reported by Risdiplam and Nusinersen patients (SN: SMA REACH reference number)

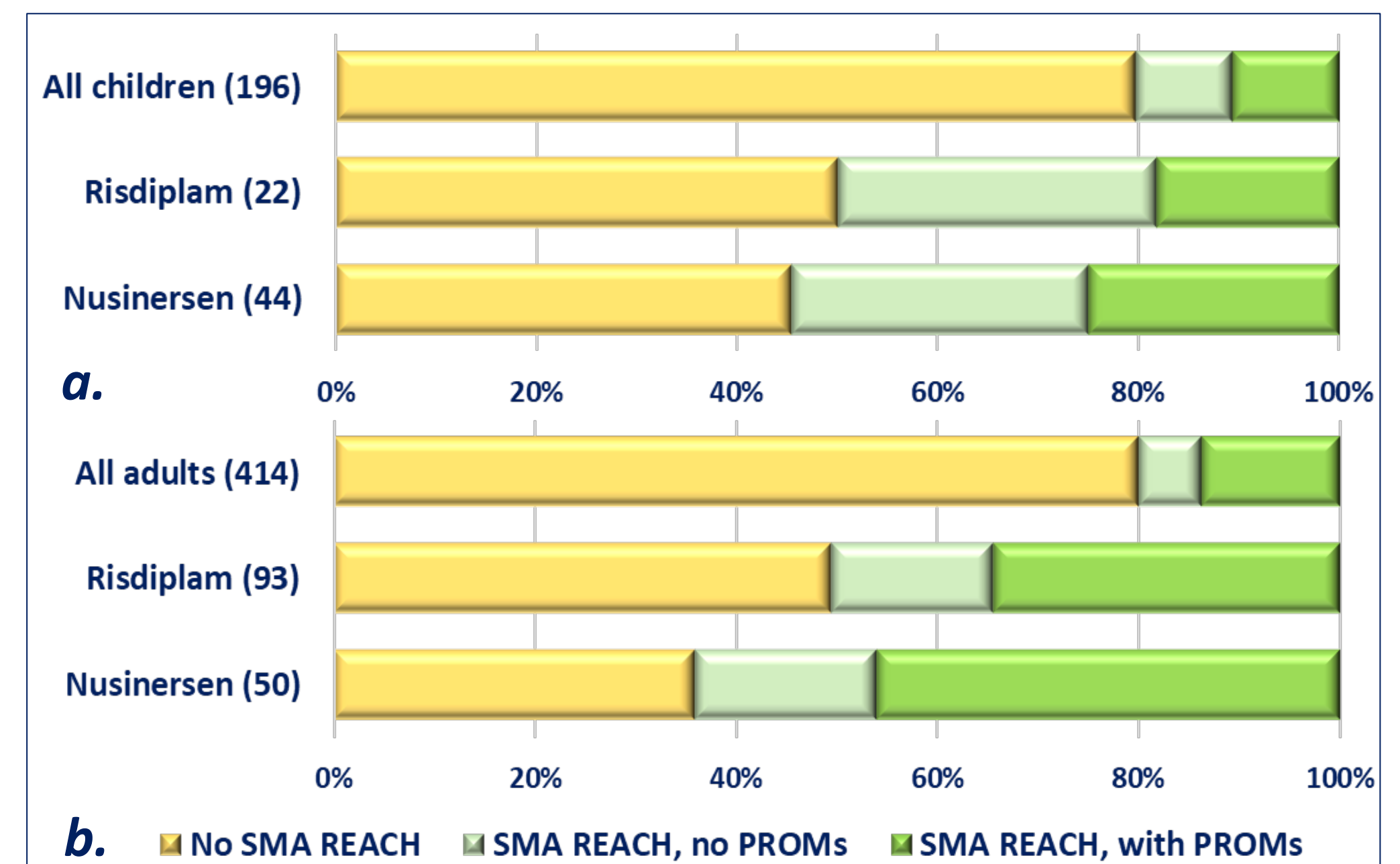


Fig.2. Percentage of paediatric (a) and adult (b) patients who have reported PROMs and for whom their SMA REACH reference number (SMA REACH) is known by the registry (total patient number per group is presented in brackets)



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