

P81 GASTROINTESTINAL ASSESSMENT IN SPINAL MUSCULAR ATROPHY (SMA): The experience of SMA healthcare professionals in France

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Introduction

- The advent of targeted therapies has changed the natural history of spinal muscular atrophy (SMA).
- New phenotypes have emerged with new challenges and goals. However, some classical complications as gastrointestinal symptoms (GIs) are still common, making multidisciplinary approach crucial for proper clinical care and follow-up of SMA patients.

- The aim of our study was to evaluate the current medical approach and remaining unmet needs regarding GIs in French SMA patients.
- In a second stage, we intend to develop national guidelines to ensure a proactive and systematic follow-up for the GIs.

Methods

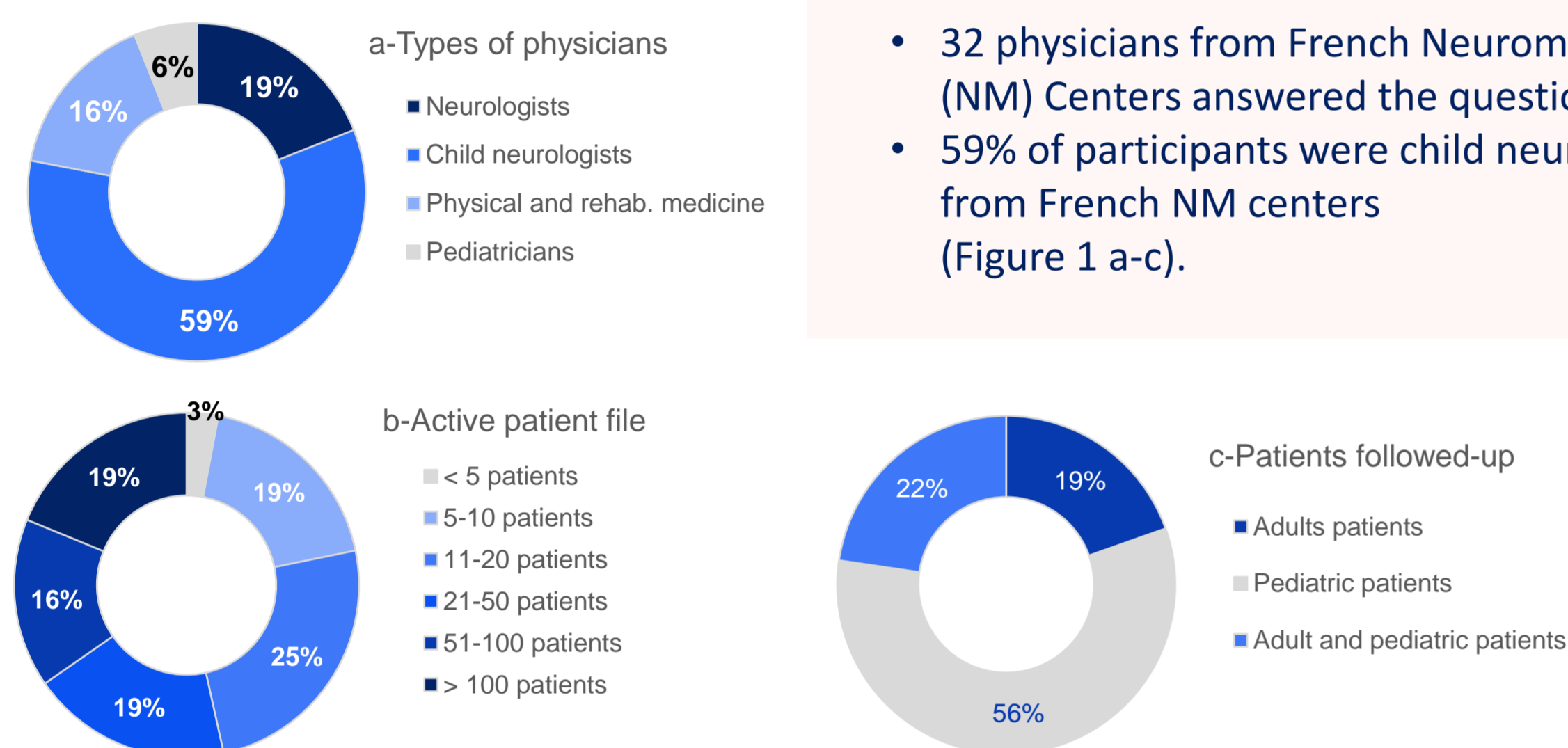
- French multicenter survey collecting data from SMA healthcare professionals (child and adult neurologists and pediatricians).
- Data were collected through an online questionnaire sent via the French neuromuscular network (FILNEMUS, <http://filnemus.fr>).
- This preliminary questionnaire included 9 short questions concerning the observed GIs, ancillary tests and global management in children and adults with genetically confirmed SMA.

Take-home messages

- Digestive symptoms resurface in the new phenotypes of SMA treated patients.
- Our questionnaire is an easy to apply tool that has allowed to highlight the practices of physicians in charge of SMA patients and the gastrointestinal (GI) needs in France.
- Preliminary results have shown that although most of the symptoms were monitored their follow-up was not always optimal, confirming the need for consensus on standards of care and systematic GI, nutritional and endocrine-metabolic monitoring in patients with SMA.
- This first survey has some limitations, as it only assessed digestive symptoms. Growth charts, nutritional and hydration status as well as metabolic complications will be included in future versions of the questionnaire.
- To improve the current situation and to ensure a proper clinical care for SMA patients, our study group will design a user-friendly GI symptoms (GIs) questionnaire, which can be easily conducted during multidisciplinary follow-ups and distributed within the SMA medical community.
- Systematic assessment of GIs and appropriate treatment could prevent serious complications, improve patients' quality of life and potentially reduce the burden on caregivers.

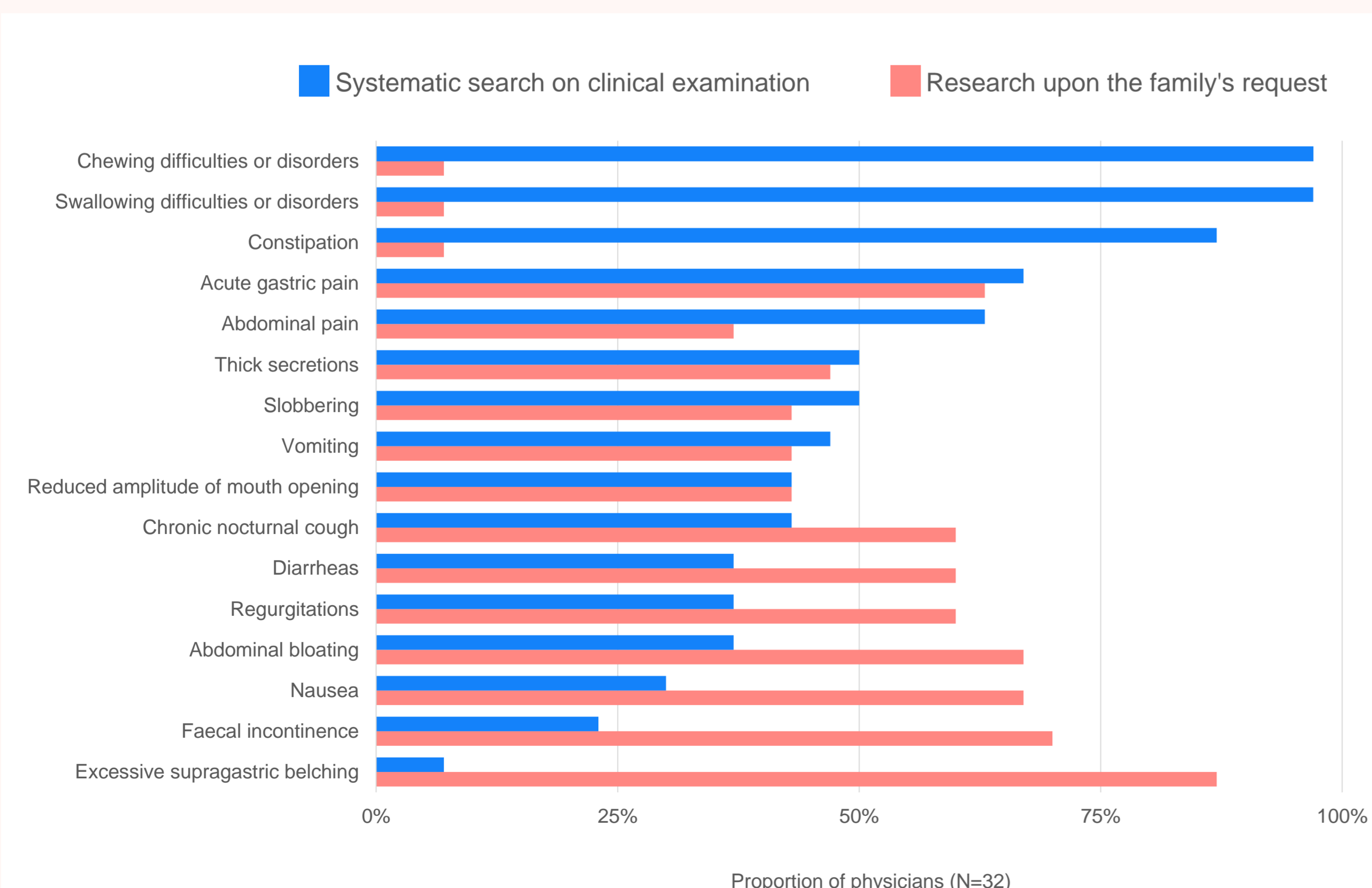
Results

Physicians population and their patients



Symptoms documented

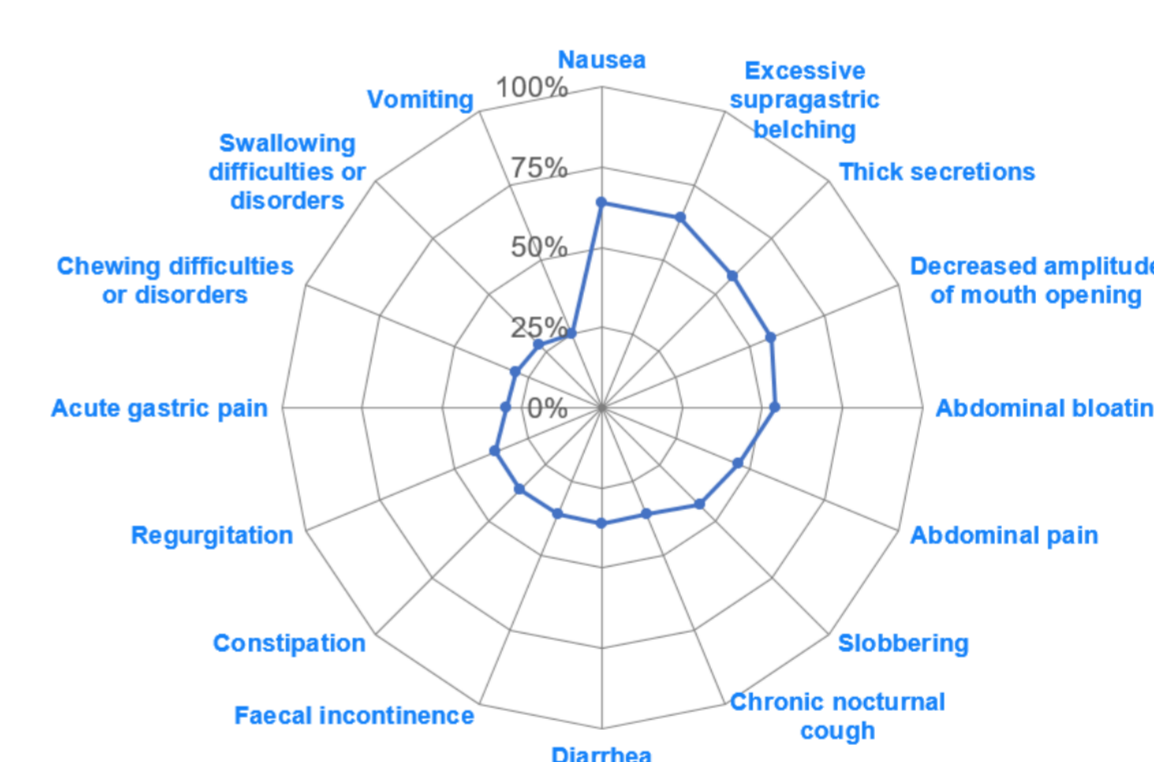
- The most common documented GIs were constipation and swallowing dysfunction (97% each), chewing difficulty and abdominal pain (90% each).
- A majority of the experts affirmed to **systematically** investigate swallowing (97%) and chewing disorders (87%), as well as gastric or abdominal complications during their routing examination (Figure 2).
- While physicians primarily focused on common GIs such as swallowing difficulty and constipation, recurrent symptoms like excessive belching, nausea, diarrhea and regurgitation were not consistently investigated or explored by healthcare professionals. Instead, they were often addressed in **response to a specific complaint or a request from the patient's family** (Figure 2).
- This questionnaire has shown that some important symptoms as limitation of mouth opening (danger of difficult intubation), presence of abdominal bloating and/or fecal incontinence were not systematically explored during follow-up visits (Figure 2).



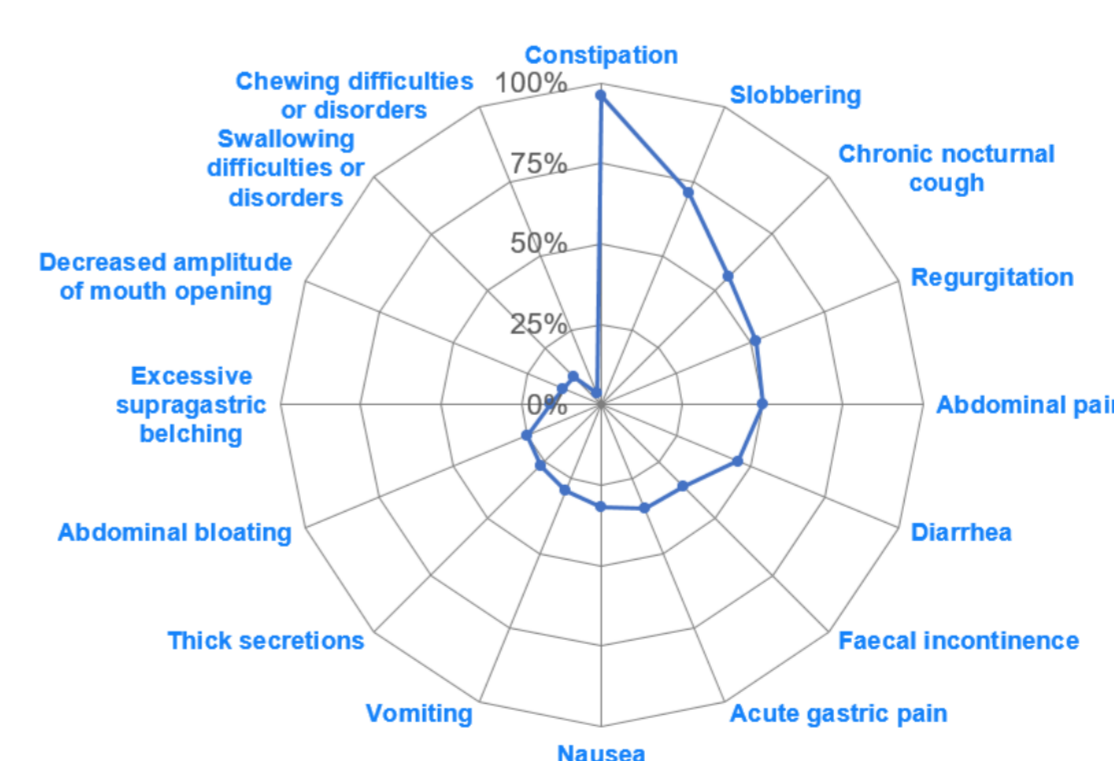
Clinical management of symptoms

- Figures 3-8** show the practices of the SMA healthcare experts during their routine follow-up concerning the SMA GIs.
- We observe that the attitude of the experts (monitoring until the next visit [Fig. 3], requesting complementary examinations [Fig. 4] or initiating treatment [Fig. 5]) varies widely depending on the symptom.
- Interestingly, more than 50% of physicians preferred waiting until next visit in case of excessive gastric belching and abdominal bloating (Fig. 3), which are specific gastrointestinal symptoms that require advice from an expert gastroenterologist, with high probability of resorting to complementary tests.
- The same attitude was observed in case of decreased amplitude of mouth opening, even if treating it early is primordial (infections, risk for intubation...).
- As expected, most of physicians seemed to care about vomiting, which can be a life-threatening event in SMA with the risk of inhalation, dehydration and hypoglycemia. More than 75% asked for an expert gastroenterologist advice (Fig. 6).

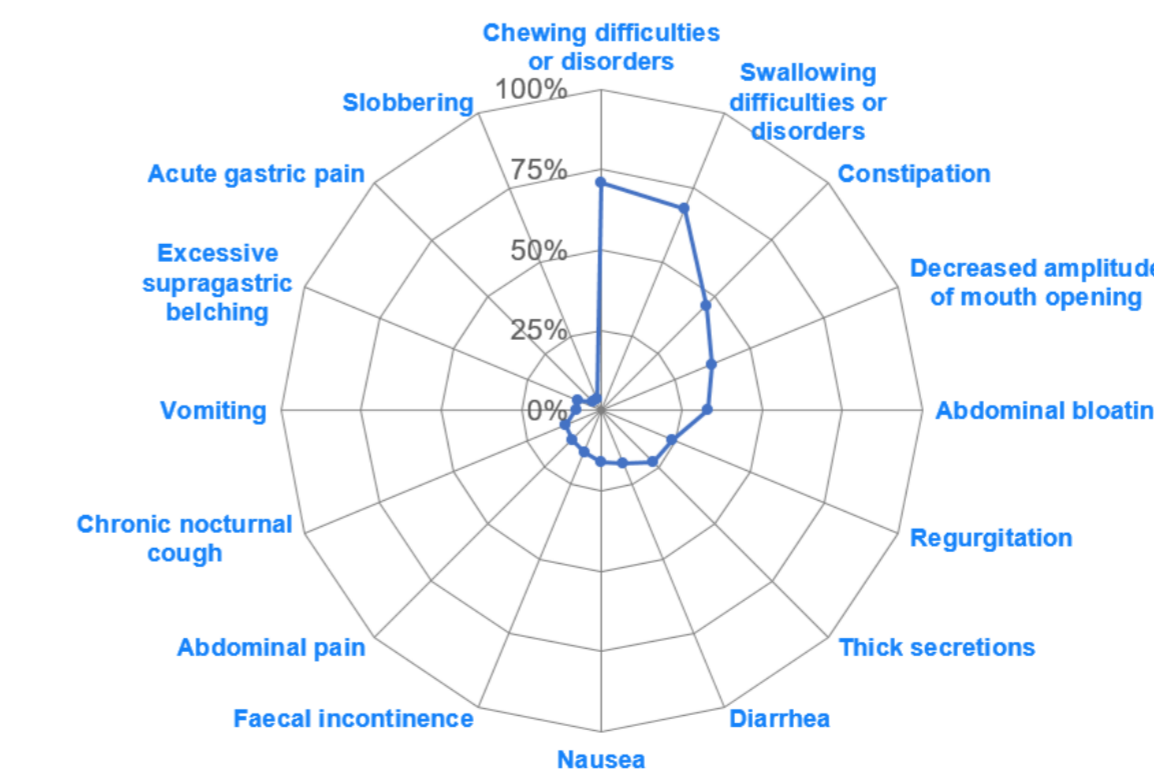
Monitoring until next visit



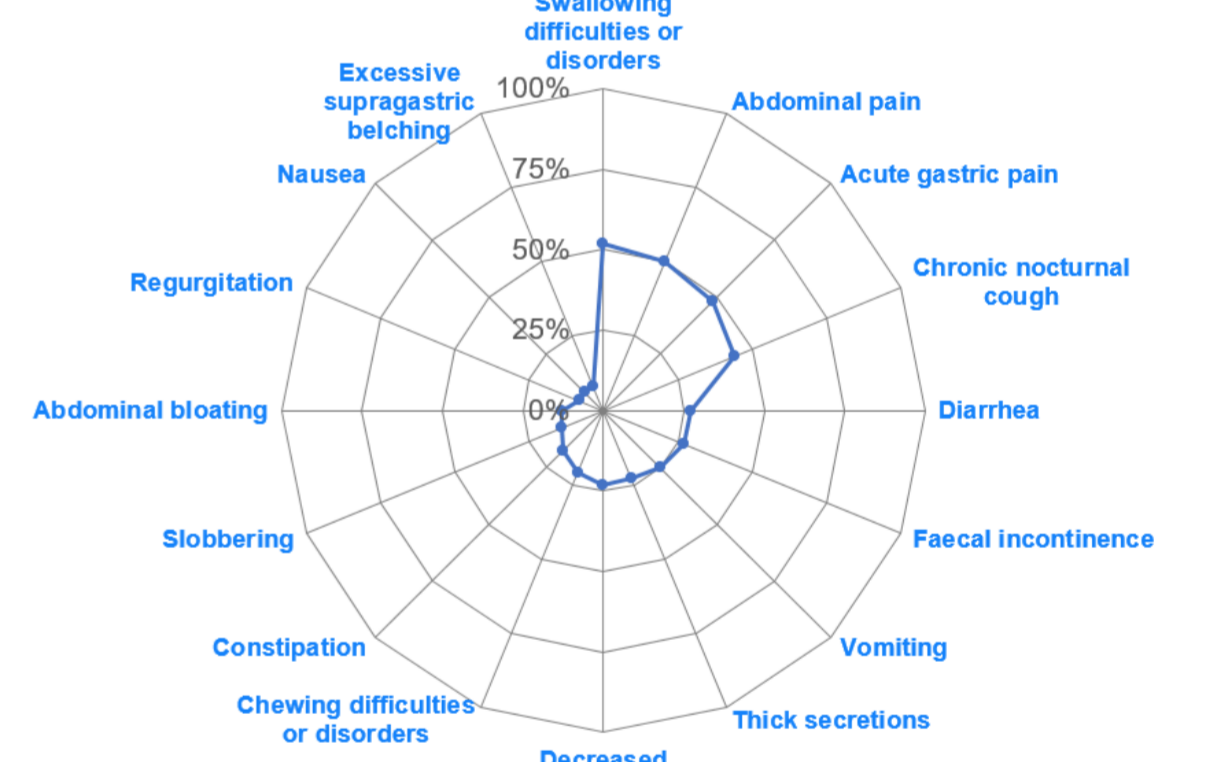
Drug therapy



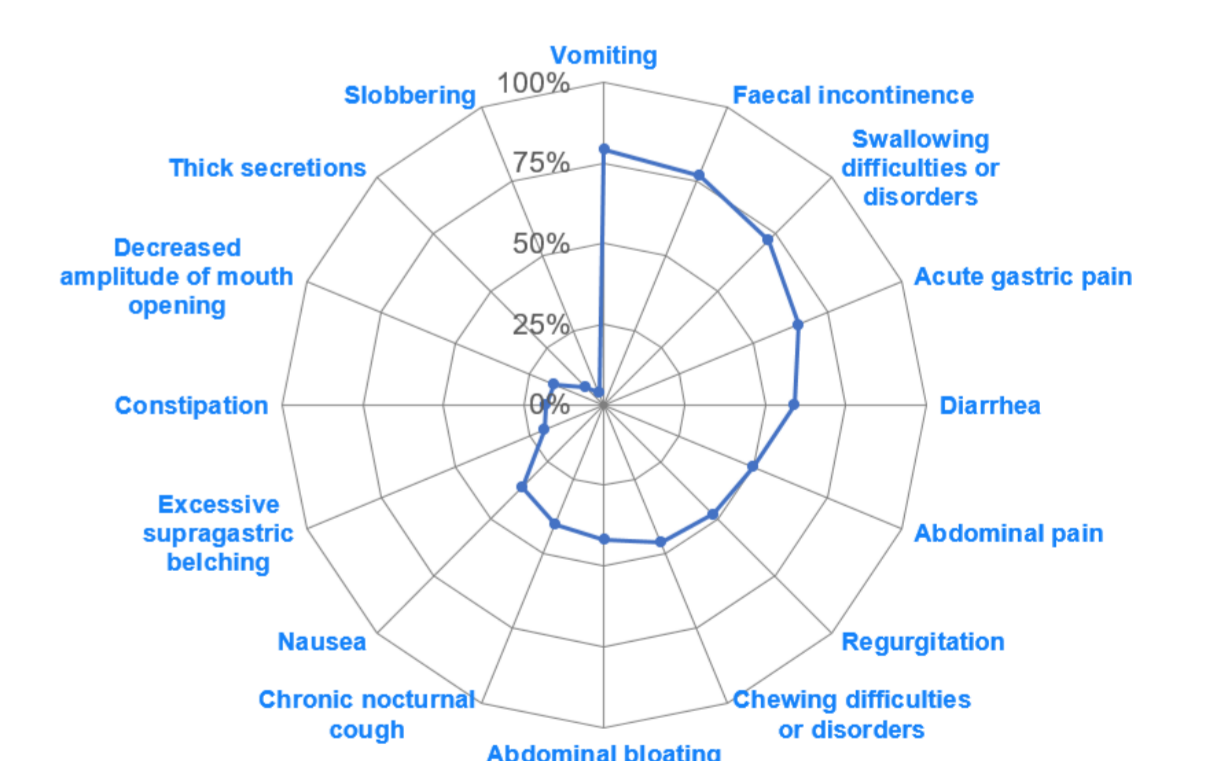
Dietary advice



Complementary exam



Request for GI advice



Referral to the treating physician

