Evolving phenotypes and ventilation needs in SMA in the post-treatment era: Results from a European community survey

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About

- Spinal muscular atrophy (SMA) is characterised by significant phenotypic variability.
- This heterogeneity has increased with the advent of disease-modifying treatments (DMTs), earlier diagnosis, and improved standards of care.
- Disease trajectory and phenotypes no longer fit the traditional type-based classifications in SMA.
- Evolving phenotypes have been described in the motor domain, but respiratory function, and ventilation needs in particular, have received less attention despite being used as a criterion for DMT reimbursement.

Goal

Explore respiratory phenotypes in SMA in the era of DMTs.

Method

- **EUPESMA 2025** (European Patient Experience Survey in SMA) is part of a pan-European biennial survey series designed to capture the treatment experiences of people living with SMA and their families.
- **Patient-led, participatory, mixed-methods design,** including focus groups with adults and carers of children living with SMA and systematic community input in all phases of research design.
- Survey translated to 18 languages and distributed through 30 SMA Europe member patient organisations.
- Data collected in Qualtrics and analysed in SPSS and R.
- Patient characteristics and ventilator and tracheostomy use were assessed. Change in response to treatment was measured with a categorical scale indicating the direction of change (improvement, no change, worsening).

Results

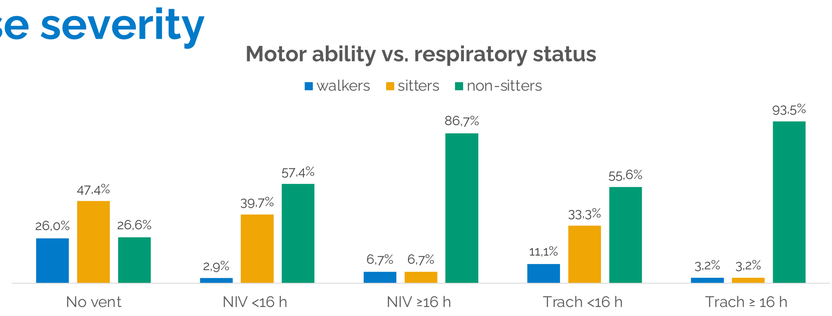
826 individuals

Respondents: people living with SMA (60.8%), legal guardians (37.5%), others (1.7%).

Grouping by respiratory support needs

- No ventilation (n=519, 63.4%)
- Non-invasive ventilation (NIV) <16 hours/day (n=207, 25.3%)
- NIV ≥16 hours/day (n=15, 1.8%)
- Tracheostomy <16 hours ventilation/day (n=11, 1.3%)
- Tracheostomy ≥16 hours ventilation/day (n=31, 3.8%).

Disease severity

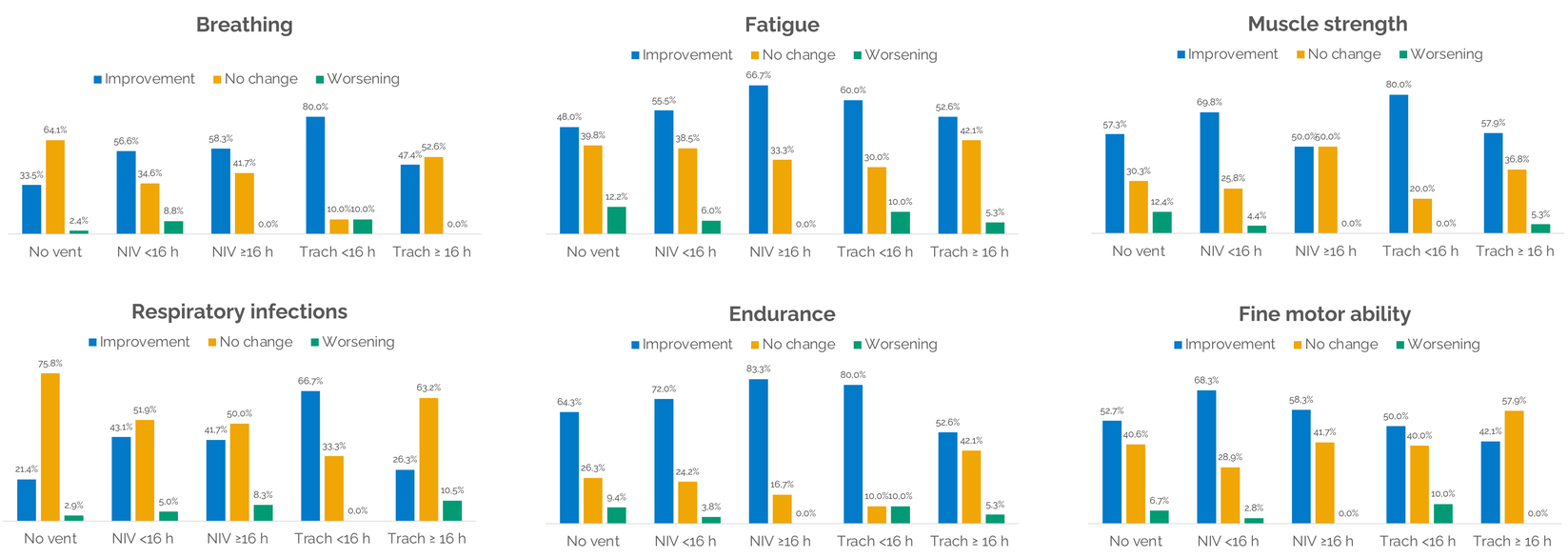


Benefit of treatment through DMTs

Changes in symptoms since starting DMTs

- In **all** ventilation groups, most respondents reported **improvement or stabilisation across domains** when receiving a DMT.
- No meaningful differences between groups were observed.

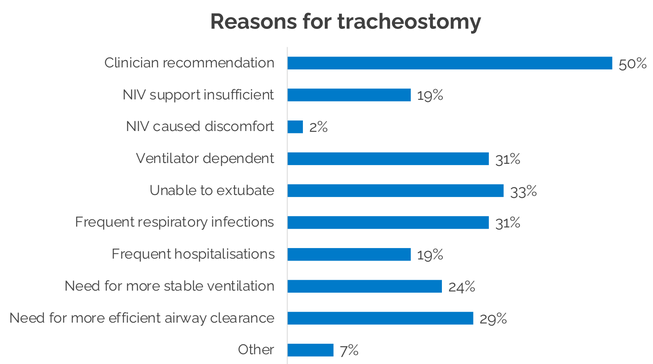
Changes in symptoms since starting a DMT across respiratory need groups

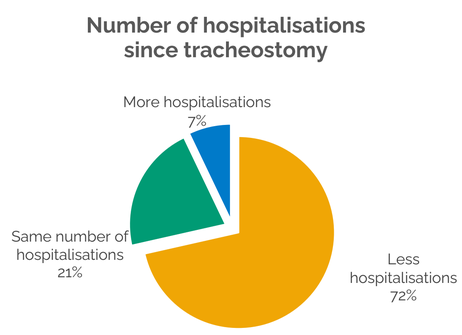


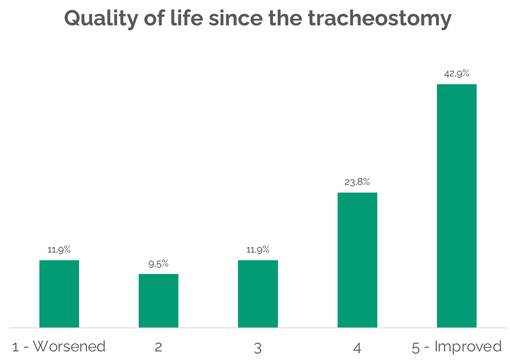
Tracheostomy group

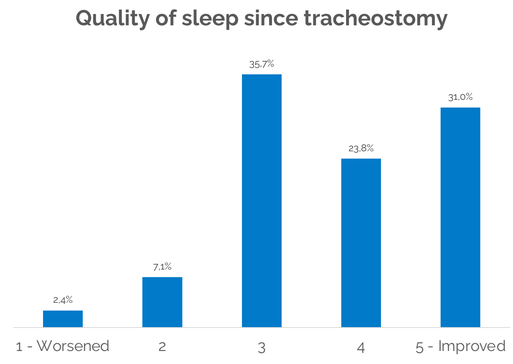
42 individuals

Respondents: people living with SMA (51.2%), legal guardians (48.8%).









Conclusions

- Increased respiratory needs, including tracheostomy, are more common in severe motor phenotypes. However, there is **heterogeneity** in motor function across the respiratory continuum.
- Individuals living with more severe respiratory needs **can benefit from tracheostomy and DMTs**. People living with tracheostomies benefit from treatment similarly to groups not using ventilation or using non-invasive ventilation.
- A **more holistic and multi-domain approach** is needed in guiding SMA treatment and care decisions, so that people with more intensive respiratory needs are **not excluded** from treatment through DMTs.
- Rethinking the use of respiratory status as an exclusion criteria from DMTs will ensure that **people across the motor continuum**, including those with higher motor function, do not fear seeking the respiratory support that they can benefit from.

Acknowledgements

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