

# Burden of illness in Sanfilippo disease (MPS III) – results from an international caregiver survey

Jacqueline Adam<sup>1</sup>, Michaela Weigl<sup>2</sup>, Anna Prähofer<sup>2</sup>, Carmen Kunkel<sup>3</sup>, Tabea Friedel<sup>3</sup>, Dragana Miletic Lajko<sup>4</sup>, Jordi Cruz<sup>5</sup>, Fredi Wiesbauer<sup>6</sup>, Takeyuki Akiyama<sup>7</sup>, Nalan Yilmaz<sup>8</sup>, Vanessa Ede-Scott<sup>9</sup>, Kim Angel<sup>10</sup>, Prof Christian J Hendriksz<sup>11</sup>, Dr James Davison<sup>12</sup>, Alexandra Morrison<sup>1</sup>

<sup>1</sup>MPS Commercial\*, MPS House, White Lion Road, Amersham, UK; <sup>2</sup>Gesellschaft für MukoPolySaccharidosen und ähnliche Erkrankungen, Finkham 90, A - 4612 Scharn, Austria; <sup>3</sup>Gesellschaft für Mukopolysaccharidosen e.V, Herstattstraße 35, 63739 Aschaffenburg, Germany; <sup>4</sup>MPS Serbia, Vukasoviceva Str. 50, Belgrade, 1100, Serbia; <sup>5</sup>Asociación MPS España, Anselm Clavé, 1, 08787 La Pobla de Claramunt, Barcelona, Spain; <sup>6</sup>MPS Schweiz, Knonaerstr. 29, CH 6330 Cham, Switzerland; <sup>7</sup>The Japanese Society of the Patients and the Families with Mucopolysaccharidoses, 4998-1 Kusiro Soja, Okayama 710-1201, Japan; <sup>8</sup>MPS L H Derneği, Hakimiyeti Milliye cad., No 58 Vedat Kadri Kancal iş merkezi 46/A, Üsküdar/İstanbul, Turkey; <sup>9</sup>MPS & Related Diseases Society National Office, Upwey, Victoria 3158, Australia; <sup>10</sup>The Canadian Society for Mucopolysaccharide & Related Diseases Inc., 218 – 2055 Commercial Drive, Vancouver, B.C., V5N 0C7, Canada; <sup>11</sup>FYMCA Medical Ltd., 10 Bankside Place, Radcliffe, Manchester, M26 1RW, UK; <sup>12</sup>Great Ormond Street Hospital NHS Foundation Trust, London WC1N 3JH, UK

## Background

- Patients with MPS III (mucopolysaccharidosis III) usually appear normal at birth, with developmental delay becoming evident by the age of 2–5 years (1)
- Mental and motor development reach a peak by age 3–6 years, followed by progressive behavioural disturbances and intellectual decline (1)
- During the final stage, nursing care needs take precedence as children lose mobility, the ability to swallow, and seizures and incontinence have to be managed (2)
- There is great variability in the rate of progression, but on average, death occurs at around 15–20 years of age (1)
- Current treatment is limited to symptomatic and supportive care (1)

## Study aim

- To understand further the burden of illness posed by MPS III on the patients, healthcare resources, families and education

## Methods

- We developed a questionnaire to determine diagnostic pathway and burden of disease
- Only disease burden results are presented here (see poster LB-01 for diagnostic pathway)
- Patient organisations distributed the questionnaire and conducted parent/caregiver interviews in their own countries
- Parents or caregivers of individuals with MPS III were eligible to take part and provided informed consent
- Questionnaires were completed via face to face or telephone interview, by post or online

## Results

- A total of 174 responses were received of which 149 were suitable for analysis
- Of the 40 countries approached, responses were received from 13: Australia, Austria, Bosnia, Canada, Germany, India, Indonesia, Japan, Serbia, Spain, Switzerland, Turkey and UK
- Individuals were aged between 1 and 48 years (mean 18 years); male (47.7%), female (47.0%)

## Burden of disease

- Aspects of disease burden are shown in Figures 1–4

Further disease burdens included:

- 80.1% joint/muscle problems :
  - Stiff/painful joints (36.8%)
- 54.2% urinary and faecal incontinence
- 53.5% seizures
- 51.4% frequent upper respiratory infections
- 43.8% night-time waking
- 43.1% hearing loss
- 39.4% impaired eyesight
- 34.8% frequent constipation
- 33.6% difficulty falling asleep
- 27.4% frequent diarrhoea
- 23.5% required suctioning
- 22.0% sleep apnoea

Figure 1. Burden of disease: walking (N=131)

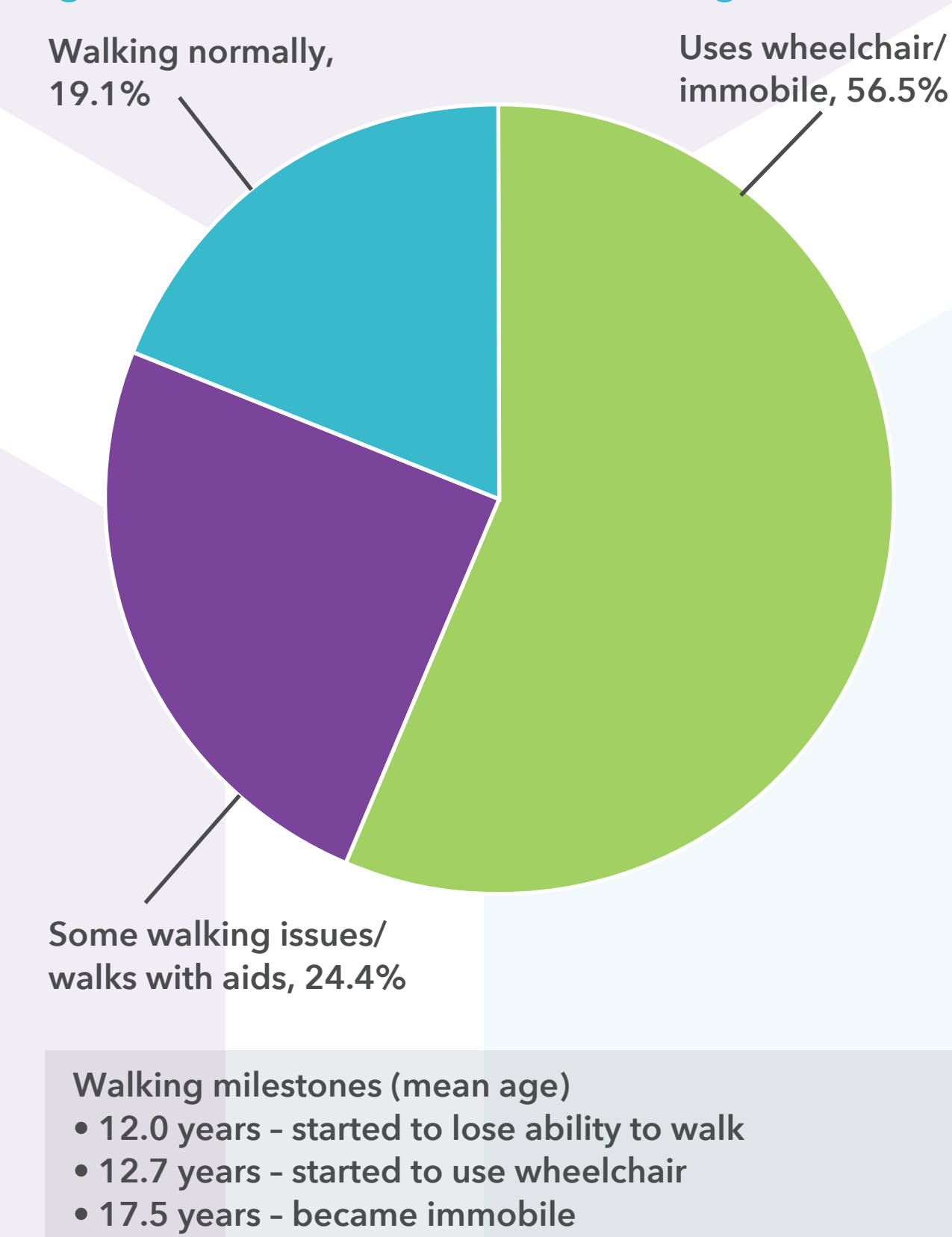


Figure 2. Burden of disease: speech (N=145)

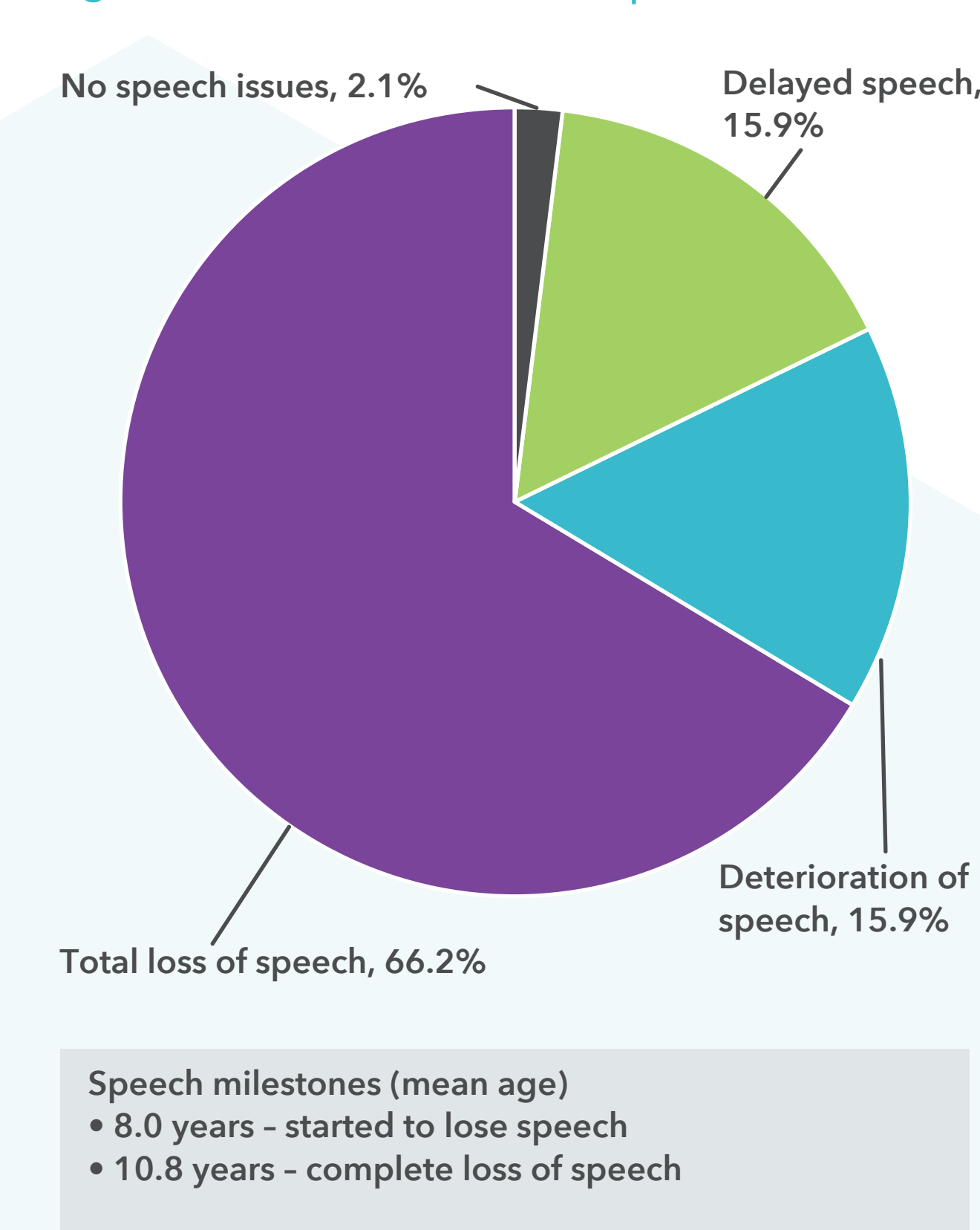


Figure 3. Burden of disease: behaviour (N=129)

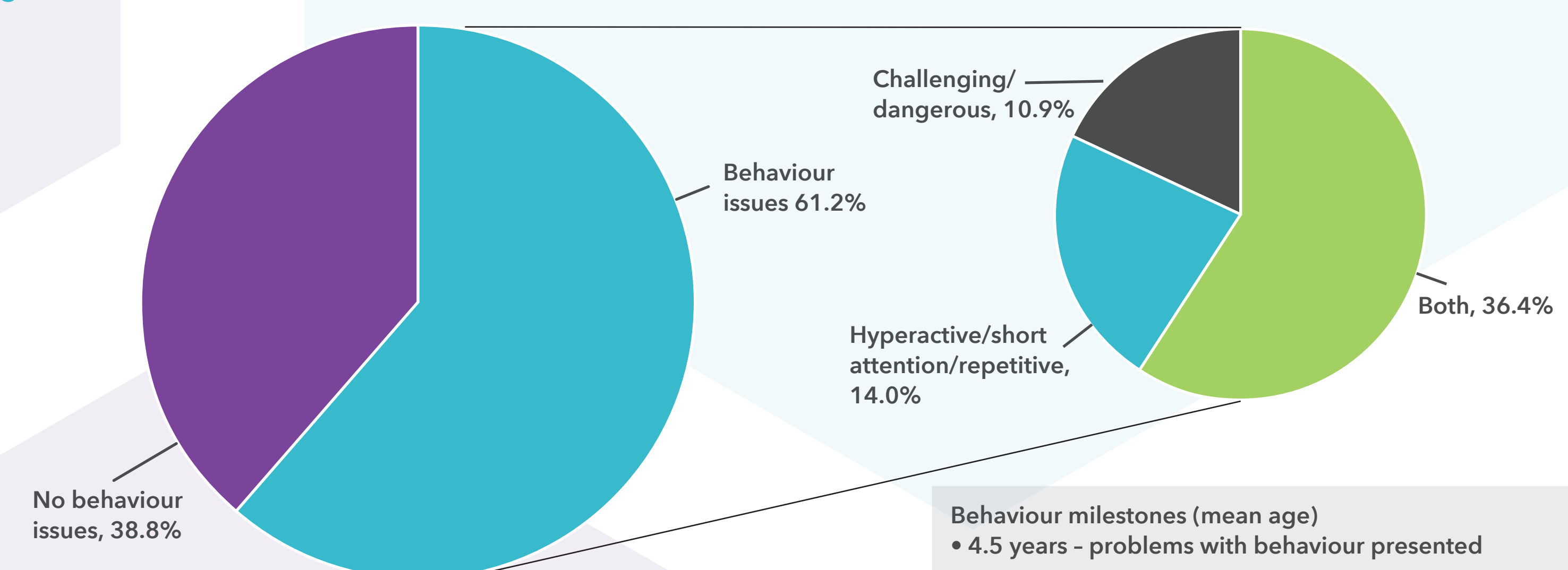
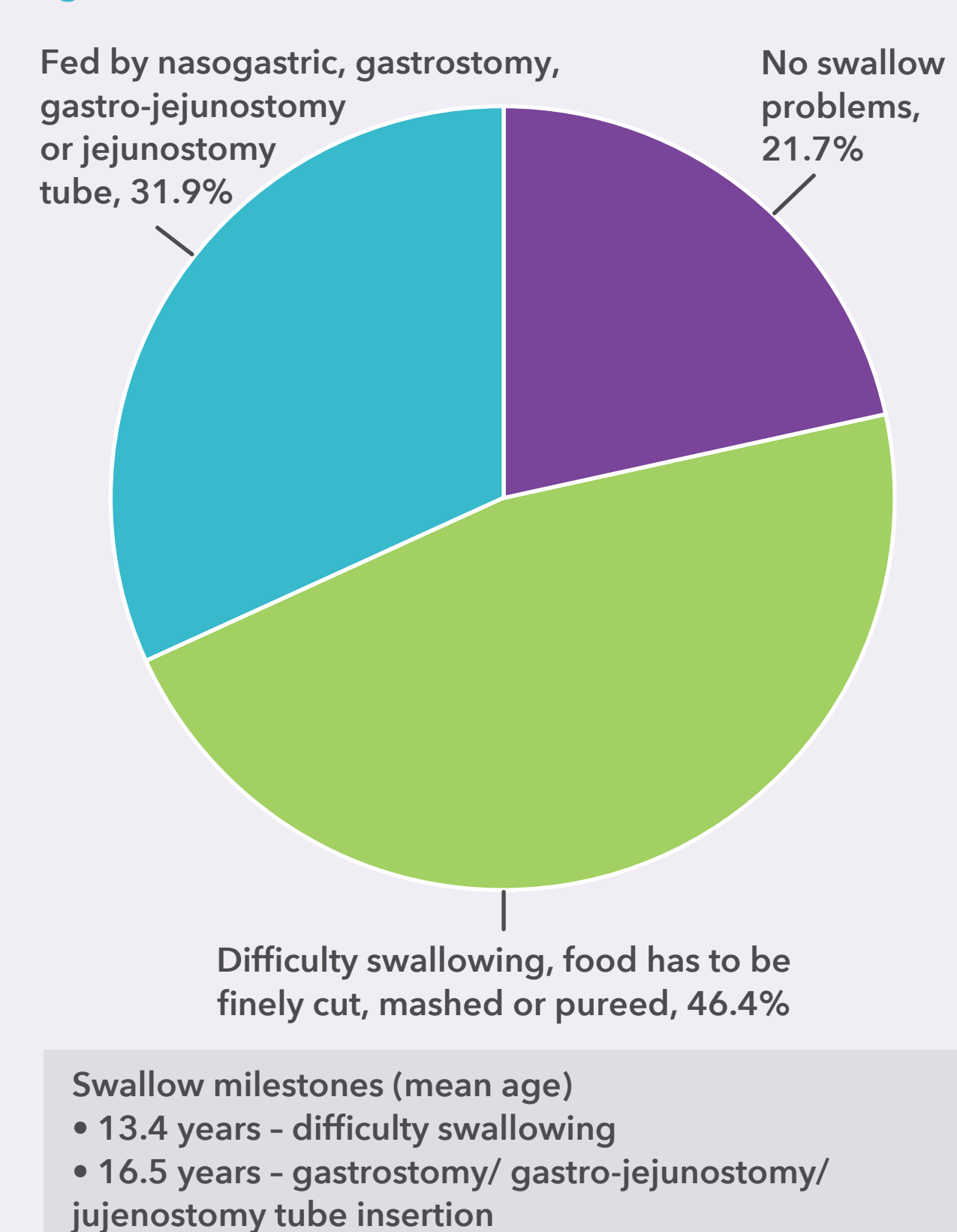


Figure 4. Burden of disease: swallow (N=138)



## Education

- 82.2% of children attended a specialist school during their education
- Specialist education started at a mean age of 5.9 years
- 70.5% required one to one adult support at school

## Conclusions

This study quantifies many aspects of burden of disease, some of the medical and educational resource impacts and family burden associated with MPS III against which the benefit and cost effectiveness of new treatments may be measured.

## References

1. <https://rarediseases.org/rare-diseases/mucopolysaccharidosis-type-iii/> (accessed on 2 Jan 2018)
2. Cleary MA, Wraith JE. Management of mucopolysaccharidosis Type III. Arch Dis Child 1993;69: 403-6

## Acknowledgments

Louise Cleary and Steven Cotterell, Patient Advocates, MPS Society, for their help with the questionnaire design  
The research was funded by Alexion Pharmaceuticals Inc., BioMarin Europe Ltd., Lysogene, Orchard Therapeutics Ltd., Shire International GMBH and Swedish Orphan Biovitrum AB (Sobi)

Poster available at [mpsccommercial.com/publications](https://mpsccommercial.com/publications)

## Healthcare needs

- Regular medications for seizures (52.8%), sleep (44.4%) and constipation (33.3%)
- Supportive care: physiotherapy (60.4%), speech and language therapy (31.9%) and occupational therapy (16.0%)
- 90% of patients had undergone surgery:
  - Removal of adenoids and/or tonsils (63.4%)

## Impact on families

- One or both parents had to stop working – 34.7%, 3.5%, respectively
- One or both parents had to reduce working hours – 27.8%, 7.6%, respectively
- Adaptations to the family home were often necessary (76.6%)

\*MPS Commercial is a wholly owned, not for profit subsidiary of the Society for Mucopolysaccharide Diseases (MPS Society) whose social objectives are to reinvest any profits for the purposes of education, enhancing needs-led advocacy support, quality of life research and scientific research to the MPS community.