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

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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%)

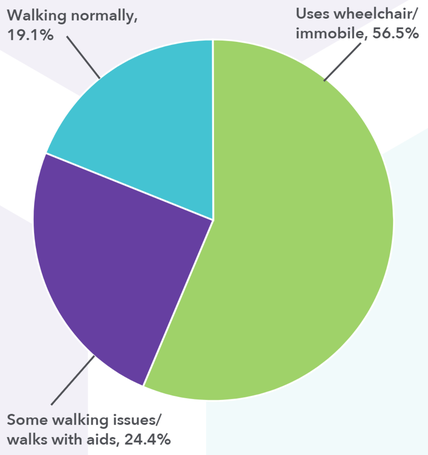
Burden of disease

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

Further disease burdens included:

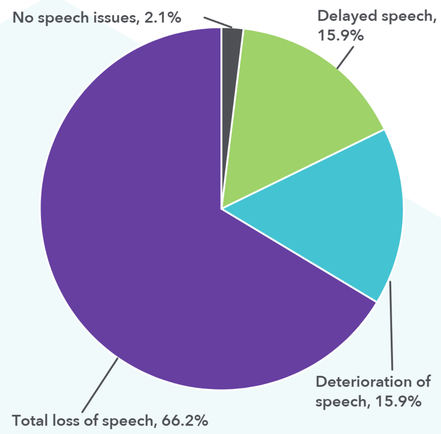
- 80.1% joint/muscle problems :
  - Stiff/painful joints (36.8%)
- 54.2% urinary and faecal incontinence
- 53.5% seizures
- 51.4% frequent upper respiratory infections
- 43.8% night-time waking
- 43.1% hearing loss
- 39.4% impaired eyesight
- 34.8% frequent constipation
- 33.6% difficulty falling asleep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

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



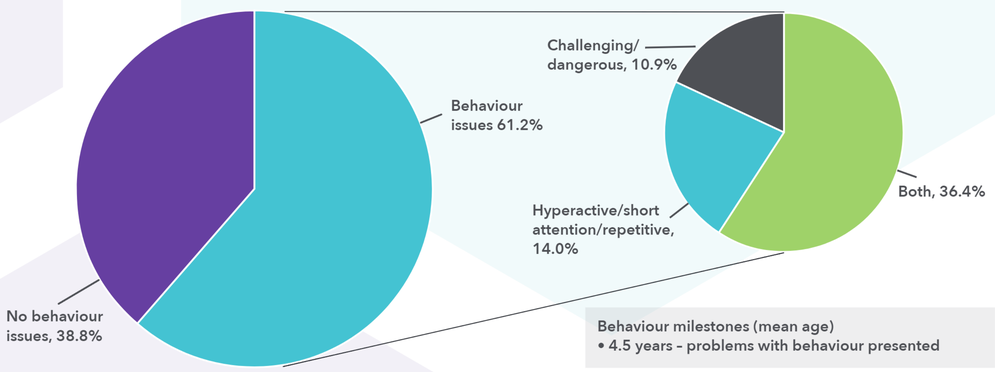
Walking milestones (mean age)
- 12.0 years - started to lose ability to walk
- 12.7 years - started to use wheelchair
- 17.5 years - became immobile

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



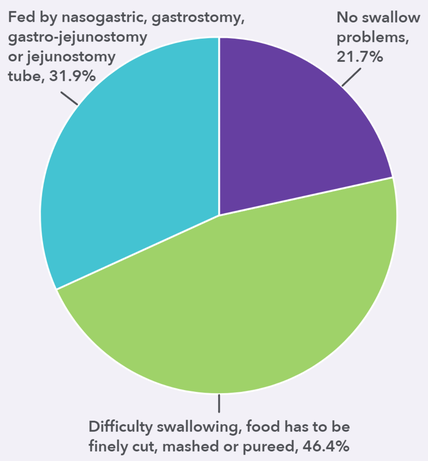
Speech milestones (mean age)
- 8.0 years - started to lose speech
- 10.8 years - complete loss of speech

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



Behaviour milestones (mean age)
- 4.5 years - problems with behaviour presented

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



Swallow milestones (mean age)
- 13.4 years - difficulty swallowing
- 16.5 years - gastrostomy/ gastro-jejunostomy/ jujenostomy tube insertion

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%)
  - T tubes/grommets insertion (50.3%)

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%)

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

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