The long-term family impact of MPS III: A qualitative study

Authors: B Pentecost[1], A Wilson[1], S Thomas[1]

Affiliations: [1] MPS Society, Amersham, UK

Overview of the condition

Sanfilippo syndrome (MPS III) is a rare, progressive neurodegenerative disorder causing severe cognitive, behavioural, and physical decline.

Children typically progress through:

Early stage: developmental delay, hyperactivity, safety risks
Mid stage: cognitive regression, loss of communication, extreme behaviours
Late stage: total physical dependency, seizures, feeding impairment, premature death

Care needs evolve from constant supervision → 24-hour medical care over time.

Data collected

Daily care and supervision
Employment & finances
Emotional wellbeing
Access to services and education
Sibling impact
Experiences with healthcare systems

Aims & objectives

Aim:
To explore the socioeconomic burden of MPS III across disease stages.

Objectives:

- Identify caregiving demands
- Assess impact on employment and income
- Evaluate access to support and services
- Capture emotional burden
- Compare parent and clinician perspectives
- Identify gaps in care and policy

Method

Qualitative study: using semi-structured interviews

Participants: parents/caregivers + specialist clinicians

Interviews: conducted via video and transcribed

Thematic analysis across key burden domains

Recruitment

Recruited via MPS Society UK

3 parents (representing disease stages):
Early (age 4)
Mid (age 10)
Late (teenager)

Multidisciplinary clinical team also interviewed

Overview of need across disease stages

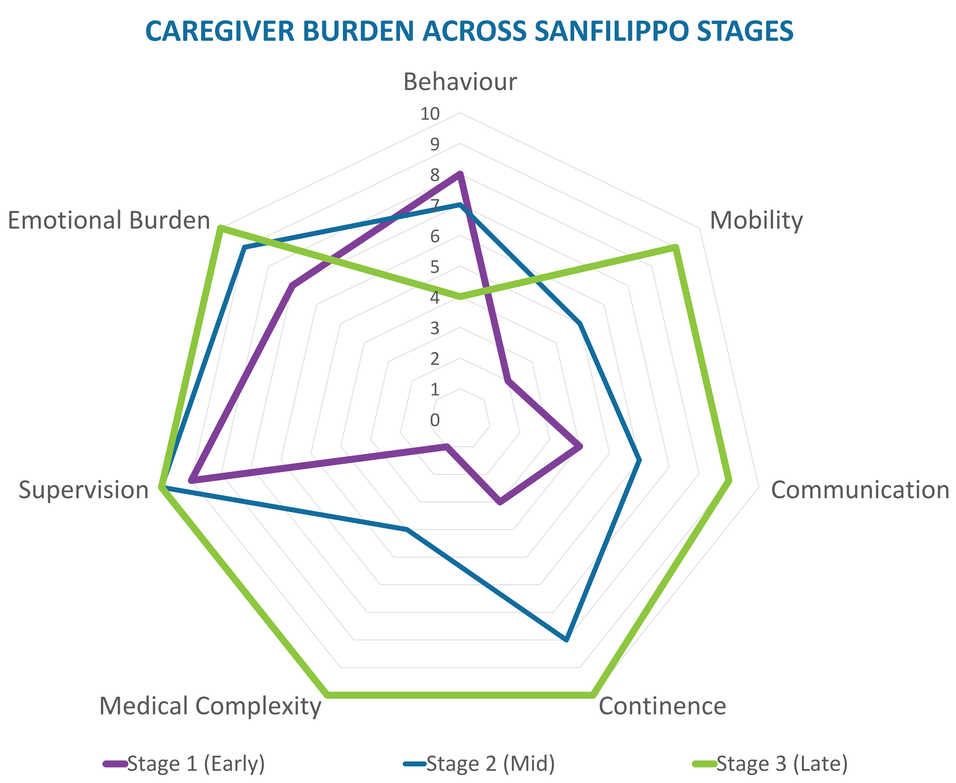
Domain	Early	Mid	Late
Supervision	Constant (behaviour)	Constant (safety)	24/7 medical
Mobility	Fully mobile	Unsafe mobility	Wheelchair/hoist
Communication	Delayed	Minimal	Non-verbal
Continence	Emerging issues	Full incontinence	Full + complex care
Care hours	Several/day	15–18 hrs	18–24 hrs

Burden across disease stages

Early: high behaviour & supervision burden

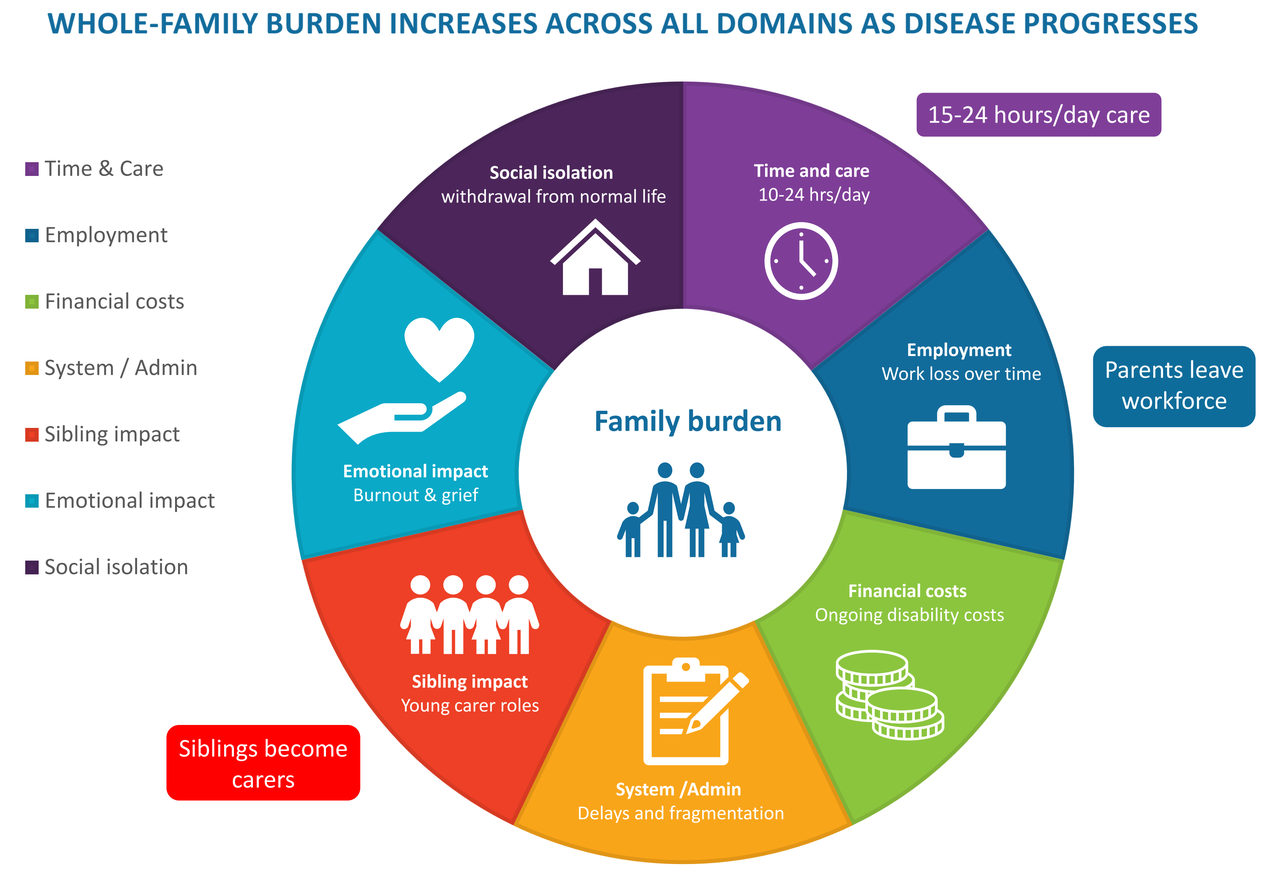
Mid: increased burden across all domains

Late: behaviour decreases but overall burden increases, peak medical, physical, emotional burden



Progressive escalation of caregiver burden across early, mid, and late stages of Sanfilippo syndrome for seven domains

Family burden



Key impacts and burden

Domain	Early	Mid	Late
Time & Care	Parenting with increasing supervision needs	Care intensifies beyond routine parenting	18–24 hrs/day; shifts to clinical-level care
Employment	Reduced or flexible work	Often forced to stop working	Long-term unemployment; loss of income and career identity
Financial	Rising disability-related costs begin	Ongoing costs: continence products, equipment, adaptations	Heating, laundry, travel; costs remain significantly above average
System / Admin	Delays in EHCP and early support access	Benefits and care packages often delayed	System feels slow, fragmented, exhausting
Emotional	Shock, grief, and exhaustion begin	Isolation and burnout intensify	Anticipatory grief alongside persistent distress
Sibling Impact	Understanding and adjustment	Feelings of neglect	Young carer roles; emotional distress, reduced childhood
Social Isolation	Normal life starts to narrow	Families increasingly withdraw	Activities become exhausting or inaccessible

Key take home messages

- MPS III creates extreme, lifelong, and escalating burden on families
- Care needs evolve from behavioural management → 24-hour medical care
- Families act as primary care providers, often at significant personal cost

Current systems are:

- Delayed
- Fragmented
- Reactive (not proactive)

Burden extends beyond the patient → entire family unit

Conclusions

- Sanfilippo syndrome requires recognition as a progressive, high-burden condition needing anticipatory support
- Families provide 15–24 hours of care daily, often sacrificing employment, health, and wellbeing

There is a clear need for:

Earlier intervention → Coordinated services → Reduced bureaucracy → Long term system planning

Without change, families will continue to carry the majority of care and cost burden alone

Acknowledgments: We would like to thank all the families for participating and sharing their insights.