The Impact of Mucopolysaccharidosis Type III (MPS III) on Family and Finances: A Caregiver Perspective

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Daily Life is Full of Highs and Lows

- Daily life with MPS III (Sanfilippo) is unpredictable and can feel like a rollercoaster
- Children can be vivacious and joyful, but these times are punctuated by periods of distress
- Outbursts of anger and frustration
- Prolonged crying and screaming fits without a known cause can last days to weeks, making it difficult to leave the home
- Child can't communicate what's wrong
- The goal is often to avoid distress and keep the child content and comfortable
- It's like constantly having a newborn
- Constant supervision and support for basic activities of daily living is required

Mental and Emotional Burdens

- Days are packed with speech, physical, behavioral, and language therapy and meetings with teachers and doctors
- Mothers, especially, managed complicated schedules and a large team of healthcare providers and other caregivers
- Lonely, exhausted, overwhelmed, distracted, guilty, and overstimulated
- Mental health and self-care on put on the back burner
- Maintaining other relationships is difficult, and marriages can break down
- Parents worry that other children in the family aren't getting enough attention or that they might be missing out
- Other children in the family worry about how their sibling would be treated and feel isolated
- Worried that their child is suffering
- Feel like a burden to others

Everything is More Expensive

- Earning potential limited by the need for flexible jobs or leaving the work force
- Healthcare costs are high, and wait lists are long
- Insurance coverage is a complex combination of government-funded programs and private insurance
- Insurance providers need to be educated about MPS III
- Insurance and other funding sources are difficult to navigate and not available to all
- Regional differences in insurance coverage can dictate where families live
- Caregiver stipends, when available, are small
- Vacations and travel are harder given the need for specialized equipment and accommodations, so families often take separate trips

Unmet Need

At diagnosis, parents are often told, "Go home and love your child. There's nothing we can do."
But parents need much more than that.

AT DIAGNOSIS:

- Healthcare workers that explain how to best support the child despite a terminal diagnosis
- Urgent need for reliable, simple, multilingual information on what to expect, available resources, and rights
- Connections to support groups
- Child-friendly information

THROUGHOUT JOURNEY:

- Livable wages for caregivers
- In-home support is vital
- A sense of urgency to help their children
- Access to treatment across the child's lifespan
- Education for pediatricians
- Mental health providers who are trained on rare diseases
- Support with IEPs and training school staff

Community Support

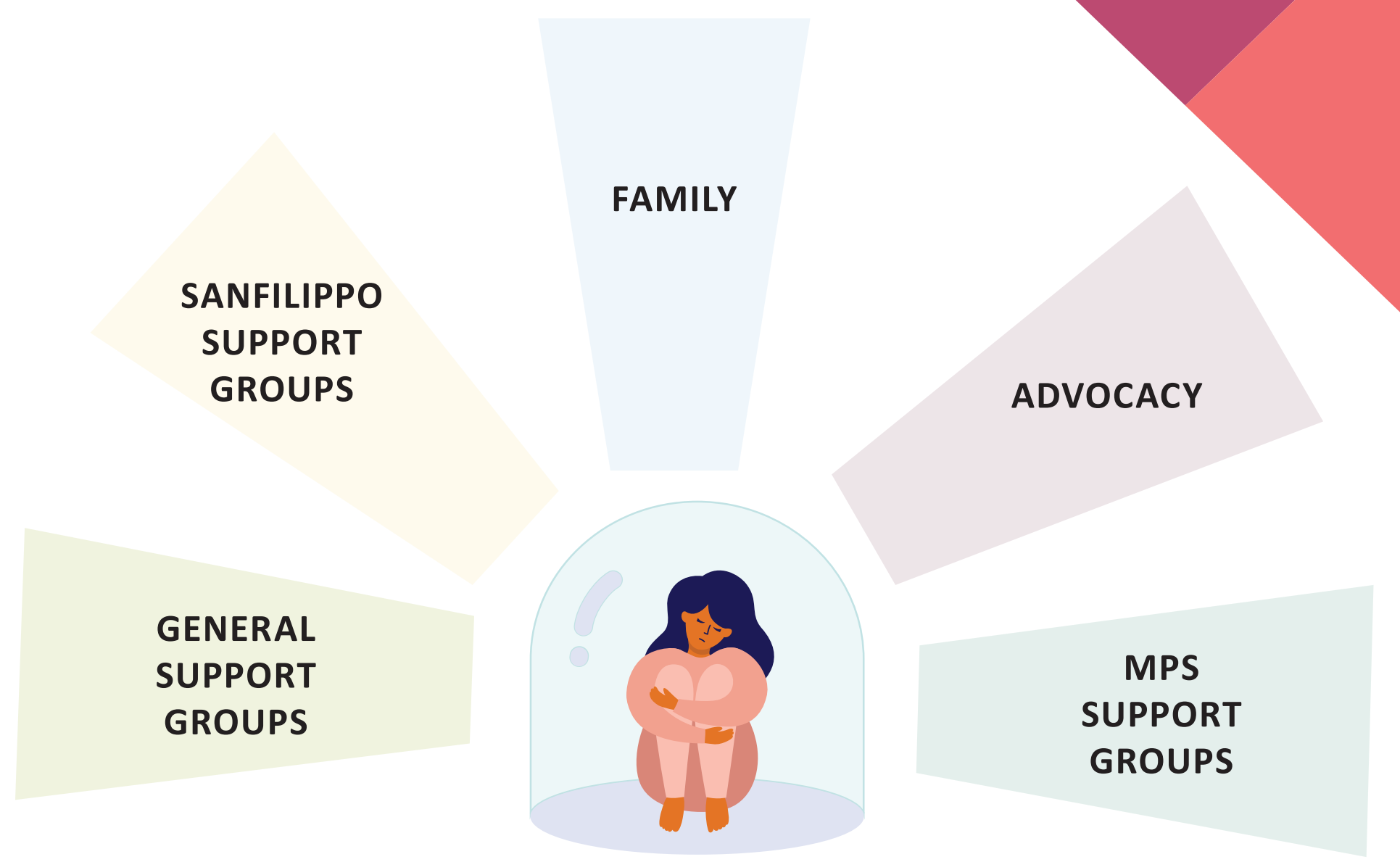
- Families, especially grandparents, were key supports
- But grandparents age and families move away over time
- Specific community support groups were rare
- Feel like the odd one out
- Lack of time and energy limited involvement
- Common sense of isolation
- In response, several caregivers started their own patient advocacy groups

School, the Good and Bad

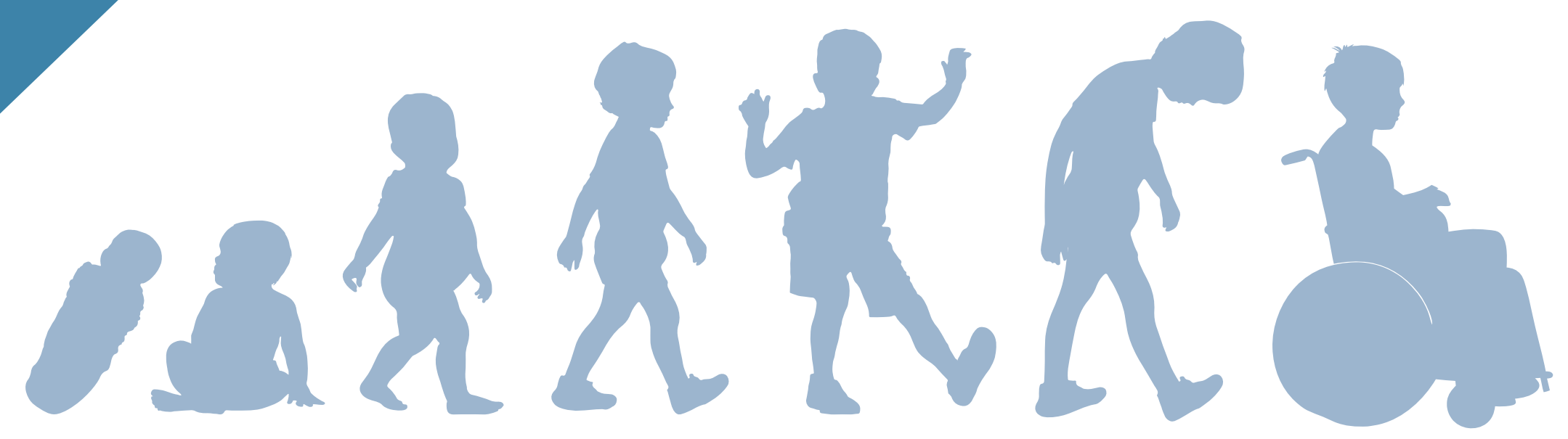
- Most thrived with the social engagement provided by school
- Important support for child and family quality of life
- Comforting to have trusted carers
- Provides routine, familiarity, and new experiences
- Music, social interaction, dance, animals, gardening, playing with peers
- Lack of training or negligence in some schools contributed to regressions

Transitions

- Symptoms can change quickly and unexpectedly
- Less support as children age out of the school system
- Adult day care has long wait lists
- Legal provisions needed to manage money and care decisions for adult child
- End of life planning needed for primary caregiver as well as child
- Some families made end-of-life plans early; others consciously focused on the present
- Having limited time with their child crystallized how precious life was



ISOLATION



INTRODUCTION

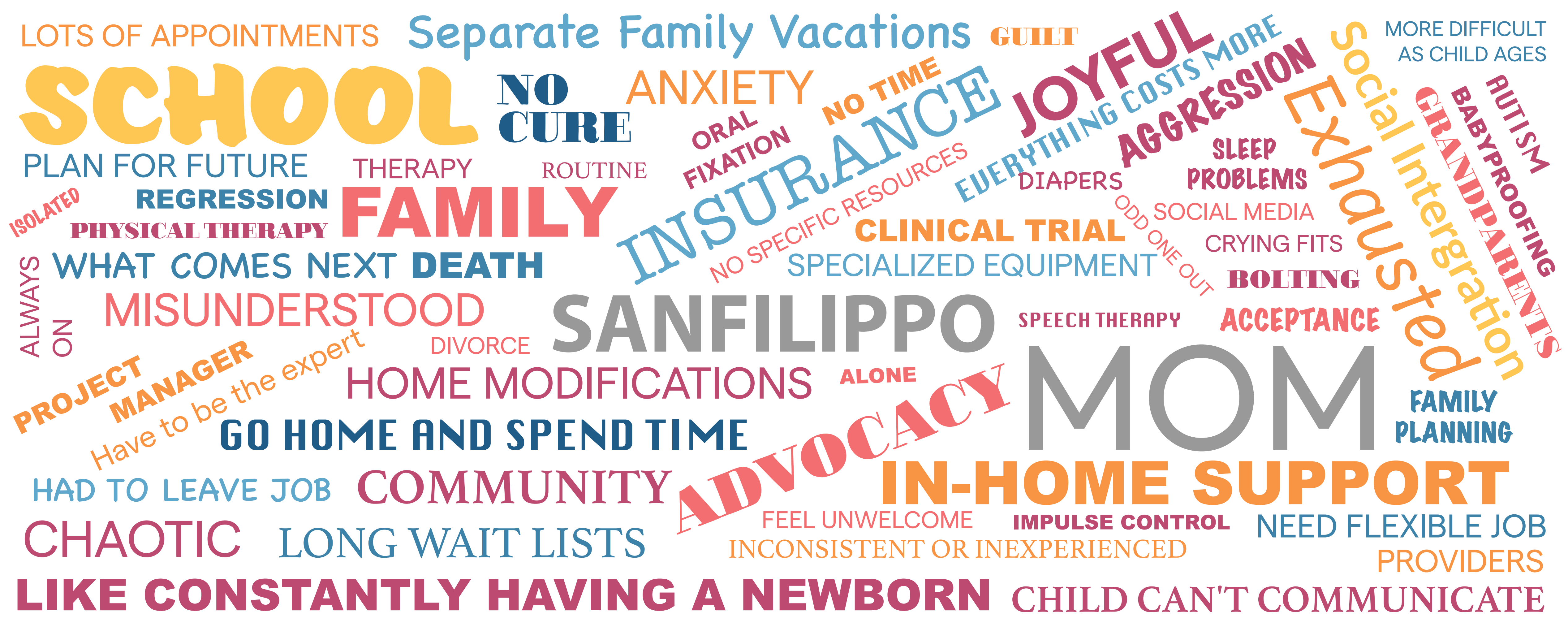
- MPS III (Sanfilippo syndrome) is a group of rare disorders caused by pathogenic variants in genes that encode enzymes needed to break down heparan sulfate
- MPS III is characterized by early neurodevelopmental decline, multi-system disease, and early death
- There is no cure for MPS III and treatment focuses on managing symptoms and quality of life
- Here we describe MPS III from the caregiver perspective

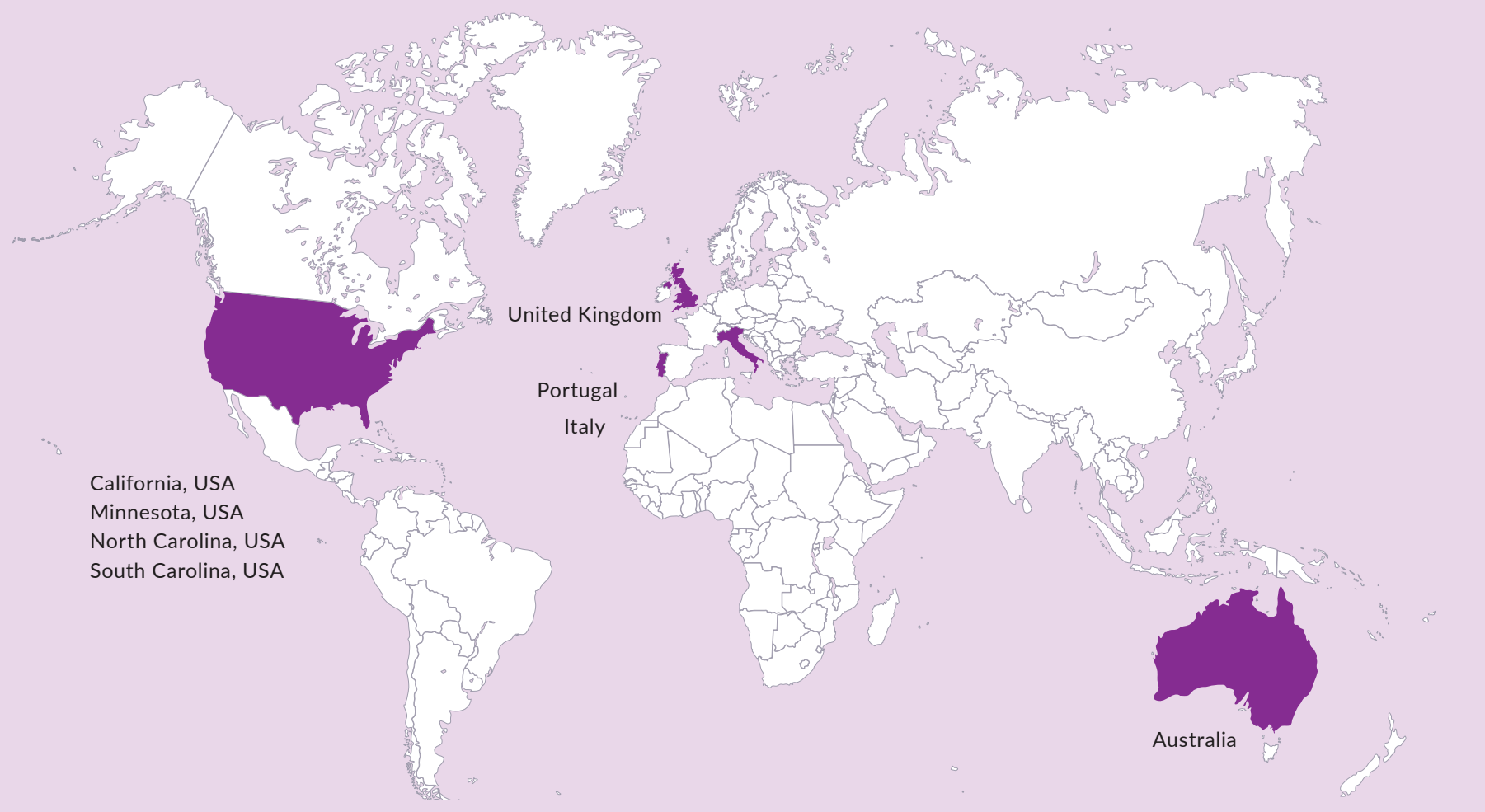
METHODS

- Seven caregivers were recruited world-wide for individual, one-hour interviews in which participants were asked a series of open-ended questions
- Results were summarized manually and authors provided additional insights and discussion
- No compensation was provided
- The text of all the interviews was analyzed to generate the word cloud

DEMOGRAPHICS

- Families were from Australia, Portugal, the UK, Italy, and the US (N Carolina, S Carolina, and Minnesota)
- Participants' children were between 4 and 20 years of age
- Dual-parent families, single-parent families, and blended families were included; some participants had a single child and others had multiple children





CONCLUSIONS

- MPS III (Sanfilippo) impacts all aspects of family life
- Parents are told to take their children home and love them, but far more support is needed from diagnosis and throughout the child's life
- Though caregivers came from different regions with different health care systems, the challenges they face and what they need for support is remarkably similar

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