

A qualitative study to understand caregivers' burden of acid sphingomyelinase deficiency

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Introduction

- Acid sphingomyelinase deficiency (ASMD), historically known as Niemann–Pick disease, is a rare, progressive, and potentially fatal lysosomal storage disease, which consists of three distinct subtypes:^{1,2}
 - Type A is characterized by severe progressive neurodegeneration during childhood and early mortality
 - Type B is a milder, more variable, form of disease that occurs later in adulthood; common manifestations include hepatosplenomegaly and dyslipidemia
 - Type A/B is an intermediate form with a slower, chronic neurodegenerative disease course.
- Despite the varied symptoms experienced by patients, limited research exists focusing on how ASMD symptoms impact caregiver quality of life.³

Objective

- The objective of this study was to gain a better understanding of the humanistic and socio-economic burden on caregivers of patients living with chronic forms (type B or A/B) of ASMD.

Methods

- This was an observational, qualitative study using one-on-one interviews with caregivers (e.g., parents or other family members) of adult and pediatric patients diagnosed with ASMD.
 - Building on a previous study that focused on identifying core concepts integral to the patient and caregiver experience,³ this study focused on assessing caregiver burden.
- Caregivers were recruited and interviewed between August 2019 and December 2019.
 - Recruitment was led by patient advocacy groups complying with good clinical practices in the US and UK
 - Interviews were conducted using a semi-structured interview guide (approved by an institutional review board) focused on assessing caregiver burden.
- To be eligible for inclusion, caregivers had to be ≥18 years old, and were required to have lived with a person diagnosed with a chronic form of ASMD (type B or A/B), and be the primary guardian, for ≥6 months prior to study.
- The interview was based on two major themes:
 - Disease understanding, symptoms, level-of-severity, and caregiver training, to better understand the background of the patient living with ASMD and that of the caregiver
 - Open-ended questions about areas of impact (where 0 = least bothersome; 10 = most bothersome) to better understand the caregiver perspective when caring for an individual living with ASMD.
- Sociodemographic data was obtained via a self-administered questionnaire, completed at the end of the interview to reduce potential stereotype activation/bias.

Data analysis

- Audio recordings collected during the interviews were transcribed, de-identified, and systematically analyzed using a qualitative analysis software program, ATLAS.ti (Version 8.2).
- Descriptive statistics (mean; standard deviation [SD], frequency) were used to characterize the participants in terms of sociodemographic and clinical characteristics.

Results

Caregiver population

- Seven caregivers (USA, n=4 and UK, n=3) participated in the study, demographics are presented in **Table 1**.
 - Mean (SD) age of caregivers was 49.1 (10.68) years, and 71% of caregivers reported spending >21 hours weekly caring for someone living with ASMD.
- Six of seven caregivers (86%) were asked if they received any formal training to fulfil their caregiver role, with all six reporting that they had not completed any formal training.
- The most reported caretaking activity was medication management (n=4/7, 57%).
 - This included staying on top of medication routines (n=1/7; 14%), giving medication reminders (n=1/7; 14%), purchasing (n=1/7; 14%), preparing (n=1/7; 14%), and administering medications (n=1/7; 14%)
 - US caregiver of pediatric patient: *'Well, I take him to his medical appointments, of course, and I usually get his medications ready for—and that includes purchasing, ordering...'*

- Other caregiving activities included taking the patient with ASMD to all their doctor appointments (n=3/7, 43%), providing transportation (n=3/7, 43%) preparing meals (n=3/7, 43%), and providing regular healthy dietary reminders (n=2/7, 29%).
- Many caregivers (n=5/7, 71%) were the sole caretaker of the patients with ASMD.
- Caregivers mentioned getting support from family members (n=4/5; 80%), support groups (n=4/5; 80%), significant others (n=2/5; 40%), parents (n=2/5; 40%), and friends (n=2/5; 40%).

Table 1 Demographics of caregivers

Characteristic	USA (N=4)	UK (N=3)
Age, years		
Mean (SD)	52.5 (12.48)	44.7 (7.51)
Median [range]	53 [39–65]	45 [37–52]
Sex		
Male	1	0
Female	3	3
Ethnicity (US only)		
Not Hispanic or Latino	4	0
Missing	0	3
Racial background		
White	3	2
Asian or Asian British	0	1
Other*	1	0
Marital status		
Single, never married	0	1
Married	3	0
Divorced	1	1
Missing	0	1
Employment status		
Employed, full-time	2	0
Homemaker	1	2
Unemployed and looking for work	0	1
Retired	1	0
Highest level of education		
Some college – US	1	0
Four-year college – US	2	0
Graduate school – US	1	0
University degree (BA, BSc) – UK	0	2
Post-graduate degree (MA, PhD, PGCE) – UK	0	1
Employment affected because of individual living with ASMD		
No	3	0
Yes	1	3
Time spent per week caring for patient with ASMD, hours		
0	0	0
1–5	1	0
6–10	1	0
11–15	0	0
16–20	0	0
>21	2	3

Number of caregivers are reported for each characteristic, unless otherwise specified. *1= Half White & Half Asian (Korean). ASMD, acid sphingomyelinase deficiency.

ASMD patient population

- Demographics of the patient population are reported in **Table 2**.
 - Most patients had ASMD type B (86%)
 - Four of seven patients (57%) were aged 0 to 10 years old when initially diagnosed
 - Most patients with ASMD had a mild or moderate level of disease (71%).
- Caregivers were asked to comment on the symptoms that the person with ASMD they care for experience.
 - The majority of the symptoms reported centered around respiratory issues (n=6/7, 86%), pain (n=5/7; 71%), and swelling (n=5/7; 71%).

Impacts reported by caregivers

- The most frequently reported impacts by caregivers experienced were emotional (100%), financial (100%) and physical well-being (86%) (**Figure 1**).
- All seven caregivers reported that their caretaking duties affected them in an emotional manner.
 - The most frequently reported emotional impacts included stress (86%), frustrations/bad mood (71%), and anxiety of the condition worsening (57%) (**Table 3**)
 - Caregivers of pediatric patients reported high frequency of stress (57%) and anxiety of the condition worsening (57%), whereas caregivers of adults commonly reported frustrations/bad mood (43%), anger/resentment (29%), and stress (29%).

Table 2 Characteristics of patients living with ASMD

Characteristic	USA (N=4)	UK (N=3)
Current age of patient living with ASMD		
0–10	0	1
11–20	2	1
21–30	0	1
>51*	2	0
Age of patient living with ASMD when first diagnosed with ASMD		
0–10	2	2
11–20	1	1
21–30	1	0
Age of patient living with ASMD when first started showing symptoms		
0–10	4	2
11–20	0	1
ASMD type		
Type B	4	2
Type A/B	0	1
Don't remember/don't know	0	0
ASMD severity		
Mild	1	1
Moderate	2	1
Severe	1	0
Very severe	0	1
Medical device use		
Continuous positive airway pressure	0	0
Bilateral positive airway pressure	0	0
Other: supplemental oxygen, spleen guards, wheelchair, etc.	1	1
Surgical procedures		
Surgery to remove the spleen	0	0
Blood transfusion	1	0
Other	2	0

*There were no participants between 31–50 years of age. ASMD, acid sphingomyelinase deficiency

Figure 1 Caregiver-reported impacts of caring for individuals with ASMD (N=7)

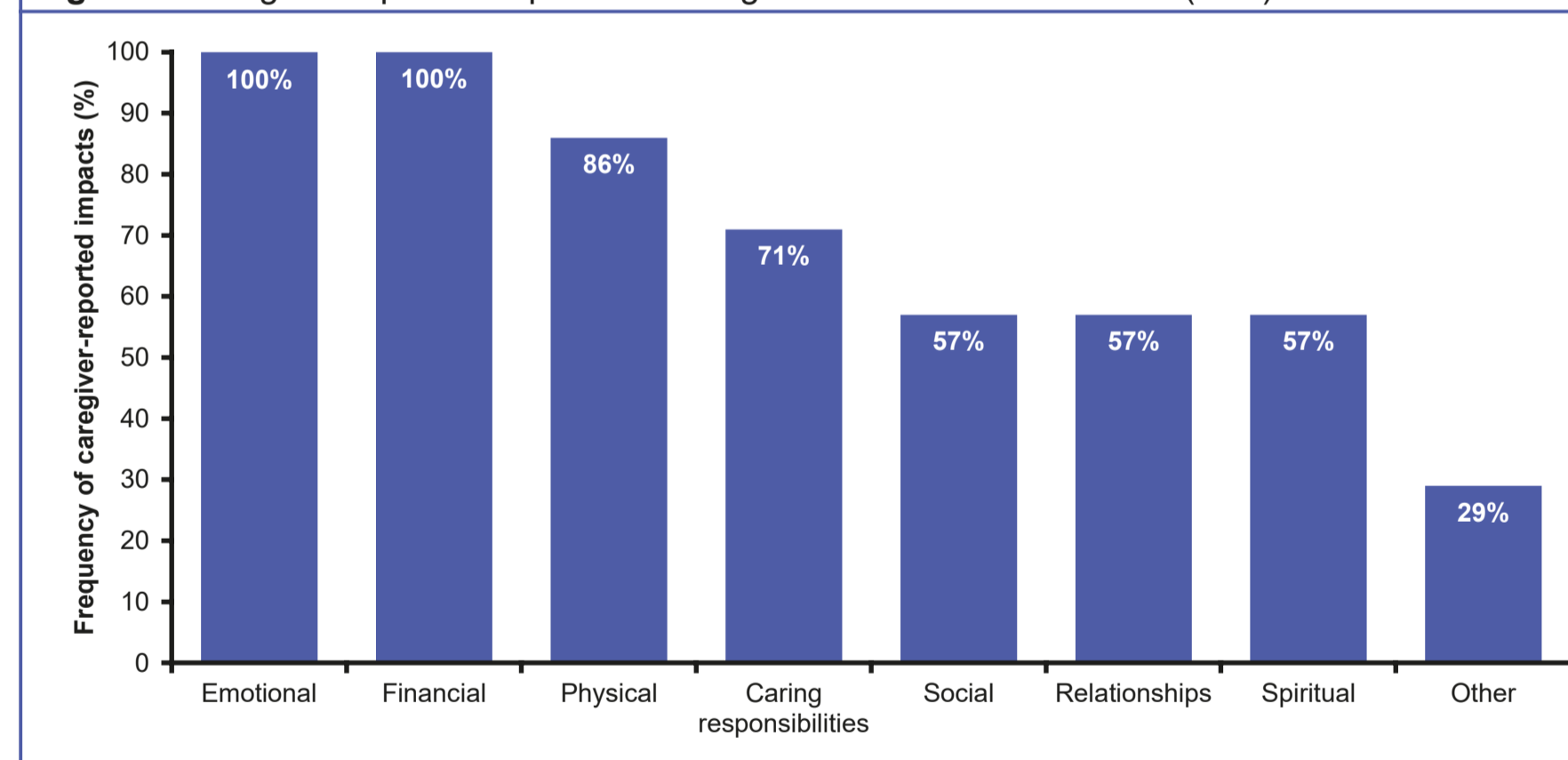


Table 3 Emotional impacts of ASMD reported by caregivers of pediatric/adult patients (N=7)

Impact, n	Caregivers of pediatric patients who endorsed impact	Caregivers of adult patients who endorsed impact
Stress	4	2
Frustrations/bad mood	2	3
Anxiety about the potential of the condition worsening	4	0
Depression	2	1
Anger/resentment	0	2
Loss of confidence/inability to share emotions	1	1
Suicidal thoughts and attempts	0	1
Grief	0	1
Uncertainty when partner is depressed	0	1
Feeling sad for partner	0	1

ASMD, acid sphingomyelinase deficiency

- When asked how they felt about the impact of ASMD, caregivers reported:
 - UK caregiver of pediatric patient: *'...you go through a lot of emotions. You go through hate, blame, anger, emotion.'*
 - US caregiver of pediatric patient: *'It does affect it negatively, because I don't think it's good to be that stressed out all the time or worried.'*
- Caregivers also ranked how bothersome these impacts were, with all seven ranking emotional impacts as 8 or above (**Table 4**).
- All seven caregivers also reported that their finances were impacted as a result of caring for someone with ASMD.
 - The most frequently reported financial impacts included having more expenses (n=3/7; 43%), needing to refinance their mortgage (n=1/7; 14%), and applying for social assistance (n=1/7; 14%)
 - US caregiver of adult patient: *'We certainly have less disposable income than we otherwise would have had.'*

Table 4 Ranking of emotional impacts reported by caregivers (N=7)

Impact, n	Ranking
Stress	8 out of 10, n=2
Anxiety about the potential of the condition worsening	9 out of 10, n=1
Depression	8 out of 10, n=1 9 out of 10, n=1
Loss of confidence/inability to share emotions	9 out of 10, n=1
Grief	8 out of 10, n=1

Conclusions

- Overall, caregivers reported pervasive impacts of ASMD on daily life including their emotional, financial, and physical wellbeing, as well as daily activities.
- Although the sample of caregivers was small, the results displayed the varied areas of caregiving that are impacted because of ASMD, and highlighted that caregivers managed a spectrum of ASMD symptoms and impacts faced by the patient as well as their own burdens because of ASMD.
- There is a need for more training for caregivers of patients with ASMD.

References

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Disclosures

RPJ is a current employee of Sanofi Genzyme and may hold shares and/or stock options in the company. RP is an employee of Evidera which was paid by Sanofi Genzyme for work related to this study. JC declares no competing interests.

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