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 AB 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 socioeconomic burden on caregivers of patients living with chronic forms (type B or AB) of ASMD.

Methods
This was an observational, qualitative study using one-on-one interviews with caregivers. Building on a previous study that focused on identifying core concepts integral to the patient and caregiver experience,4 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 completing 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 AB), and be the primary caregiver, for at least six months prior to study.

Of 12 caregivers interviewed, a subsample of 9 caregivers (USA, n=4 and UK, n=3) participated in the study, demographics are presented in Table 1.

Currently, the majority of patients with ASMD are diagnosed with type B (86%) and the average age of patients living with ASMD when first started showing symptoms is 21–30 years old (57%) (Table 2).

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 being the sole caretaker of the patients with ASMD.

Demographics of the patient population are reported in Table 2.

ASMD severity
Most patients had ASMD type B (86%) and the majority of patients had a mild or moderate level of disease (71%) (Table 3).

ASMD population
Demographics of the patient population are reported in Table 2.

Most patients had ASMD type B (86%).

Five of seven patients (57%) were aged 0 to 10 years old when initially diagnosed.

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ASMD population
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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 caring 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

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

The study is in accordance with the Declaration of Helsinki and with the guidelines of the International Committee of Medical Journal Editors (ICMJE) for conducting research with human subjects. The study was approved by the institutional review board of the sponsor of the study and was conducted in accordance with the ethical principles that have their origin in Declaration of Helsinki. The protocol was registered with the ClinicalTrials.gov database (NCT04341337).

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