The impact of olipudase alfa on QoL in pediatric ASMD patients with neurologic disease

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BACKGROUND:

Acid Sphingomyelinase Deficiency (ASMD) is an ultra-rare autosomal recessive lysosomal storage disorder characterized by intracellular lipid accumulation in multiple organ systems resulting in significant impacts on morbidity and mortality. Olipudase alfa, an enzyme replacement therapy, was recently approved by several agencies for the treatment of the non-neurologic manifestations of ASMD. Studies demonstrate improvement in organomegaly, pulmonary function and lipid profiles with olipudase alfa.

ASMD is a spectrum disorder with the variability in neurologic symptoms accounting for that spectrum. It is unknown what effect olipudase alfa has on ASMD pediatric patients and families with self-reported neurologic manifestations.

OBJECTIVE:

The objective is to better understand the real-life impact of ASMD on pediatric patients and their caregivers with neurologic manifestations and assess how olipudase alfa impacts quality of life (QoL) for that population.

> *"I feel very hopeful. This treatment has given us a lot of hope in regard to our kids' health. We know we still need to find treatment for the neurological aspect of things, but it's given us a lot of hope. We just feel like it's taking care of their bodies and keeping them as healthy as possible."*

METHODS:

Pediatric patients (≤18 years of age) with a confirmed diagnosis of ASMD and receiving olipudase alfa for at least 12 months were recruited in early 2022 through national patient organizations to participate in a global online questionnaire followed by semi-structured interviews. The parents of ten patients with ASMD who had utilized olipudase alfa as an experimental therapy for pediatric patients participated in the study. Quantitative analysis of the results was undertaken. Interview transcripts were analyzed using an inductive thematic approach.

RESULTS:

All 10 patients in this study perceived significant benefit in the non-neurologic symptoms of the disease including improvements in organomegaly and abdominal symptoms (i.e., early satiety, abdominal discomfort). Interestingly, 40% of the study participants who perceived this benefit also reported neurologic manifestations associated with ASMD (figure 1) including ataxia, hypotonia, seizures and neuropathy.

> *"I don't know what the neurological portion of things will look like for him, but I know that he has a chance now and we can see what it is because of this drug."*

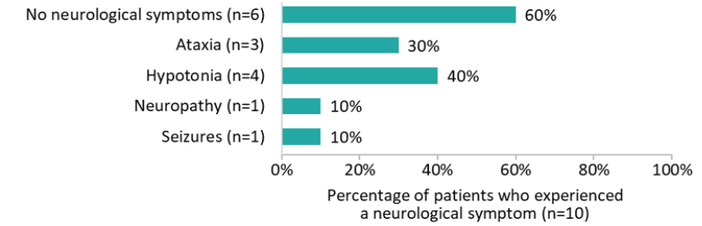

Figure 1. Percentage of patients (n=10) who reported a neurological symptom. Values add up to more than 100% as patients could report multiple symptoms.

Caregivers reported their own lives improved because of the positive impacts of olipudase alfa on their children with 80% of participants noting an improvement in their own mental health because of olipudase (figure 2). While on olipudase, eight participants felt that their disease manifestations improved, one participant noted a slowing of disease progression, and one patient had disease stabilization (figure 3).

CONCLUSION:

Olipudase alfa improved QoL for patients and caregivers, including those with neurologic disease. These results suggest olipudase alfa is associated with meaningful improvements in QoL for pediatric ASMD patients and families, including those with neurologic manifestations.

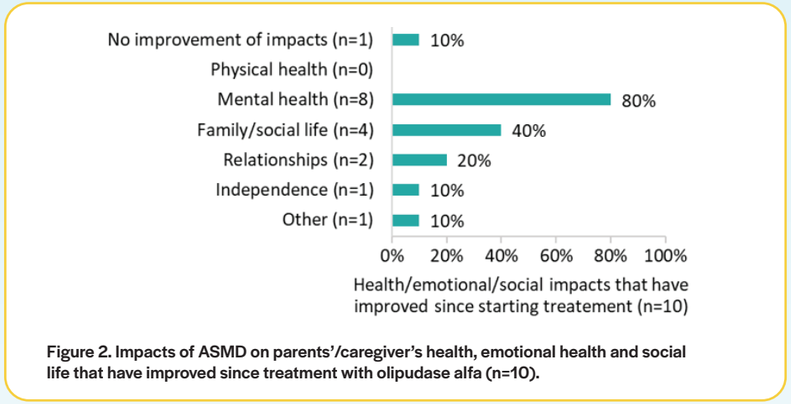

Figure 2. Impacts of ASMD on parents'/caregiver's health, emotional health and social life that have improved since treatment with olipudase alfa (n=10).

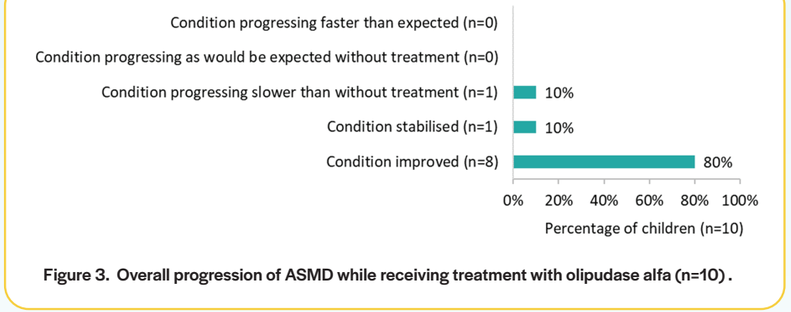

Figure 3. Overall progression of ASMD while receiving treatment with olipudase alfa (n=10) .

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