Plain Language Summary

One-year results of a clinical trial of olipudase alfa enzyme replacement therapy in pediatric patients with acid sphingomyelinase deficiency

Diaz, G.A., Jones, S.A., Scarpa, M. *et al.* One-year results of a clinical trial of olipudase alfa enzyme replacement therapy in pediatric patients with acid sphingomyelinase deficiency. *Genet Med* **23**, 1543–1550 (2021). https://doi.org/10.1038/s41436-021-01156-3

https://www.nature.com/articles/s41436-021-01156-3

Plain Language Summary: Results of an open-label clinical trial of olipudase alfa enzyme replacement therapy in pediatric patients with ASMD after one year of treatment

What was this study about? This study, called ASCEND-Peds, evaluated olipudase alfa in children and adolescents with Acid Sphingomyelinase Deficiency (ASMD). ASMD is a genetic disease-causing fatty material buildup in organs.

Who participated in the study? 20 pediatric patients (from 1 to 17 years old at the start of the study) with ASMD Type A/B or B participated.

What was tested? Olipudase alfa, a replacement for the missing enzyme in ASMD, was tested. The aim was to assess the safety of the treatment in children and to determine if the drug had any impact of the symptoms of ASMD.

How was the study done? This was an open-label study. All patients received olipudase alfa through an IV infusion every two weeks for 52 weeks. Doctors and patients knew the treatment being given.

What were the main findings? After 52 weeks of treatment:

- **Organ Size:** Spleen and liver volumes significantly decreased.
- **Lung Function:** Lung function (DLCO) improved.
- **Growth:** Younger patients showed improvements in height and weight.
- **Blood Counts:** Platelet counts increased.

Safety Information: Olipudase alfa was generally well-tolerated. Most common side effects were mild to moderate infusion-related reactions (e.g., fever, rash, headache). No new safety concerns emerged.

What do these findings mean? Olipudase alfa effectively reduced organ size and improved lung function, growth, and platelet counts in pediatric ASMD patients. This supports its use as a treatment for ASMD in children and adolescents.