**Efficacy and Safety of High-Dose Twice Weekly Sebelipase Alfa in Severe-Onset Wolman Disease: A Case Report**


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**BACKGROUND AND IMPORTANCE**

- **Lysosomal-acid-lipase (LAL) deficiency** is a rare metabolic disease (0.2:10,000) characterized by lysosomal accumulation of cholesterol esters and triglycerides, with a severe and rapidly progressive form, Wolman Disease (WD), usually fatal in the first 6-12 months of life.
- **Sebelipase-alfa (SA)** is a recombinant human LAL administered weekly, initially at 1 mg/kg, with a gradual increase according to response, thus avoiding serious hypersensitivity reactions. Twice weekly dosing with rapid escalation had not been previously described.

**OBJECTIVE**

To describe the efficacy and safety of high dose SA administered twice weekly in severe onset WD.

**METHODS**

- **Diagnosis**: WD with secondary hemophagocytic syndrome.
- **Admission to paediatric critical care unit**.
- **Since admission**: anaemia, thrombocytopenia, hyperferritinemia, altered liver function tests and lipid profile, and massive hepatosplenomegaly.

3-month-old woman

**RESULTS**

- **11 doses of sebelipase alfa over 35 days**
- **No adverse effects were reported**

**CONCLUSION**

Treatment with high-dose twice weekly SA has been an effective and well-tolerated therapy in an aggressive and severe presentation of WD so far, although it is necessary to maintain enzyme replacement for life.