Case series of Wolman Disease in Indonesia: recognizing diagnostic and treatment challenges

Mars Nashrah Abdullah2, Cut Nurul Hafifah1, Klara Yuliarti1, Damayanti Rusli Sjarif1
1. Division of Pediatric Nutrition and Metabolic Diseases, Department of Pediatrics
Faculty of Medicine Universitas Indonesia / Cipto Mangunkusumo Hospital,
Jakarta, Indonesia
2. Fellowship of Pediatric Nutrition and Metabolic Diseases, Department of Pediatrics
Faculty of Medicine Universitas Indonesia / Cipto Mangunkusumo Hospital

Background

Wolman disease (WD) is a lysosomal storage disease (LSD) which cause deficiency in lysosomal acid lipase, required for degradation of triglyceride and cholesterol esters. Diagnosis was only possible in the last 2 years in Indonesia due to unavailability of enzyme testing. Enzyme replacement therapy (ERT) is still unavailable in Indonesia. We report 2 cases of WD from Indonesia and its challenges in diagnosis and treatment.
1st Case
- A 4-month-old girl
- Abdominal enlargement, mild diarrhea, slow weight gain, and hepatosplenomegaly (liver 5 cm, spleen Schuffner IV).
- Hypertriglyceridemia (3053 mg/dL), hypoglycemia (26 mg/dL), hyperferritinemia (68971 ng/mL).
- Genetic testing showed compound heterozygous pathogenic variant in LIPA gene with low acid lipase (<8 umol/L/h).
- She died within 1 week of hospitalization due to COVID-19 pneumonia.

2nd Case
- A 3-month-old girl.
- Jaundice, relapsing fever, and hepatosplenomegaly (liver 9 cm, spleen Schuffner IV)
- Bilateral adrenal calcification (abdominal X-ray)
- Triglyceride (305 mg/dL) with signs of liver failure.
- Genetic testing showed compound heterozygous pathogenic variant in LIPA gene, with low acid lipase (<26.8 umol/L/h)
- We reached out to the manufacturer of sebelipase alfa to explore the possibilities of ERT, but it was not possible to treat WD in Indonesia
- She died after 1.5 month of diagnosis.
Conclusion

WD is an early onset LSD with devastating outcome without treatment. Patients in middle income countries should also be given equal opportunity to treatment.