Case Series of Maroteaux-Lamy Syndrome in Indonesia:
Dealing with Diagnostic and Treatment Issues

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BACKGROUND

Maroteaux-Lamy syndrome (MPS type VI) is an **autosomal recessive** lysosomal storage disease with progressive multisystem involvement associated with **deficiency of arylsulfatase B**. Symptoms, onset and rate of progressions vary greatly from one to another. We report 2 cases of MPS VI from Indonesia which diagnosed in their pre-adolescence age.

CASE 1

- A 9-year-old-boy who came with seizure
- The patient has communicating hydrocephalus, corneal clouding, glaucoma, short stature, umbilical hernia and hepatosplenomegaly (liver 3 cm, spleen schuffner II).
- **Blood spots MS Assay showed low arylsulfatase B (ARSB) activity (0,03 uM/hr).**
- The patient died due to pneumonia.
The Patient is a 13-year-old girl
- Presented with glaucoma, corneal clouding, short stature, dysostosis multiplex and hepatosplenomegaly (liver 2 cm, spleen schuffner I).
- Blood spots MS Assay showed low arylsulfatase B (ARSB) activity (0.03 uM/hr).
- This patient is now in wheel-chair bound.

We try the possibilities of ERT for her, by contact the galsulphase manufacturer, but until now there was no charity funding for MPS VI in Indonesia.

CONCLUSION
- The symptoms and severity of MPS VI can differ from one to another.
- MPS VI doesn’t affect intelligence, but when symptoms apparent may be observed by an alert physician and lead to accurate diagnosis for optimizing patient management.
- It is necessary to initiate enzyme replacement therapy with galsulphase for preventing the rate of progression but we still have issue for performing ERT for MPS VI.