HURLER SYNDROME IN INDONESIA:
DIAGNOSTIC AND THERAPEUTIC ODYSSEY

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INTRODUCTION:

Hurler Syndrome or Mucopolysaccharidosis IH is a rare genetic disorder caused by deficiency of lysosomal enzyme called alpha L-iduronidase. This condition is characterized by multiple organ dysfunction, due to accumulation of glycosaminoglycan degradation products into various organs. However, in Indonesia this condition is difficult to overcome, as the cost to diagnose and treat this disease is very high.

CASE REPORT:

This is a case of six years old boy, presenting with coarse facies, hydrocephalus, corneal clouding, developmental delay, recurrent respiratory tract infection, claw hand, hepatosplenomegaly, umbilical hernia, and short stature. The diagnosis was made after almost three years of consultation with several different physicians, as the parents realized something was wrong with their child. The child was diagnosed with Down Syndrome and cerebral palsy for several years. In seeking a diagnosis, there are several obstacles, like limited diagnostic facilities and not all examination and treatment are covered by national health insurance.
Diagnostic confirmation was made by examining enzyme activity and urine glycosaminoglycans metabolite from overseas laboratory. It takes quite a long time to get donation for enzyme replacement therapy. In Indonesia, there are only a few cases of Hurler syndrome reported. Interestingly, this case will be the first Hurler syndrome in Indonesia to receive enzyme replacement therapy (laronidase).

<table>
<thead>
<tr>
<th>Enzyme and GAG examination</th>
<th>Result</th>
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<tbody>
<tr>
<td>Alpha L Iduronidase</td>
<td>0.03 uM/hr</td>
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<tr>
<td>Heparan sulphate</td>
<td>397 ng.ml</td>
</tr>
<tr>
<td>Dermatan Sulphate</td>
<td>698 ng/ml</td>
</tr>
</tbody>
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Table 1. enzyme and GAG urine

**Therapeutic odyssey resume**
- Laronidase are not widely available
- Very expensive enzyme replacement therapy
- Waiting for enzyme donation
- Drug approval procedure and import procedure
- Shipping

Table 2. Therapeutic odyssey resume

**Odyssey to multiple misdiagnosis in our patient**
- Lack of information and awareness about rare disease
- Limited access to diagnostic tool
- Limited physician who familiar with the disease
- Poor communication between physician
- Limited budget from national health insurance
- Economic factor

Table 3. Diagnostic Odyssey resume

**CONCLUSION**
Diagnosis and management of rare autosomal recessive disorders is still a challenge in Indonesia, thus early recognition and prompt treatment is critical, in order to get a better outcome.