First Case of Infantile Cystinosis in Indonesia Presenting with Fanconi Syndrome: Management Challenge in Developing Countries

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Introduction

Cystinosis is a rare autosomal recessive metabolic disorder due to impaired carrier-mediated transport of cystine out of cellular lysosomes. The cystinosis gene, CTNS, resides on chromosome 17p13, encodes an integral lysosomal membrane of the protein called cystinosin, which functions as a cystine carrier.¹

Case

The Journey to Diagnosis

• A 16-month-old-boy came to Emergency Room with fever, cough, and profuse vomiting.
• Physical examination showed sunken eyes and dry mouth, but massive polyuria was noticed
• Laboratory exams: metabolic acidosis, hyponatremia, severe hypokalaemia (1.3 mmol/L), hypophosphatemia (0.9 mg/dL), hypouricemia. Urinalysis showed albuminuria and glucosuria
• History of failure to thrive since age 9 months (Figure 1)
• His elder sister experienced failure to thrive, metabolic acidosis, and died due to dehydration at age 2 years, without knowing underlying diagnosis

Fanconi syndrome

The most common cause in infancy: infantile cystinosis

Genetic analysis confirmed cystinosis: compound heterozygous for CTNS gene, one pathogenic variant, c.681+1G>A in intron 9 & one likely pathogenic variant, c.757_758del in exon 10

¹Elmonem AM, et al. Orphanet J Rare Dis. 2016;11:47
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Treatment

Cysteamine was started at age 2 years, through special access scheme. At that time, glomerular filtration rate (GFR) was 59 mL/min/1.73 m2, stage 3 chronic kidney disease, and renal ultrasound already revealed nephrolithiasis. Nutrient requirement was ensured by tube feeding. Supplementation of phosphate, sodium, potassium, vitamin D, and bicarbonate are continuously given with regular laboratory monitoring.

Discussion

As a consequence of late treatment, patient had arrested linear growth (Fig. 2 and Fig. 3). Leukocyte cystine assay to monitor treatment response was not available in our country thus we adjust cysteamine dosage based on body surface area and rigorous monitoring of electrolyte, acid base, and GFR. Awareness to cystinosis must be developed when dealing with Fanconi syndrome.

He developed corneal cystine deposit that requires cysteamine eye drop which is unaffordable. National health policy should underscore the need for management for orphan disease.