Early Onset of Chronic Renal Failure in Infantile Nephropathic Cystinosis

A 1-year-ten-month-old girl, first issue of a non-consanguineous marriage was referred to us for further care. The child was born full term, with birth weight of 3.5 kg. She was noticed to have polyuria, polydipsia, photophobia and failure to thrive from 6 months of age. On evaluation, she was detected to have renal insufficiency at the age of 9 months (blood urea 93.5 mg/dL, serum creatinine 2.1 mg/dL). Renal ultrasound was normal and slit lamp examination of the eyes was suggestive of cystinosis. The patient was started on conservative management of chronic renal failure.

At presentation, the patient weighed 6.2 kg (expected 12 kg). She had fair skin, sparse hypopigmented hair, pallor and clinical evidence of rickets. She was able to sit independently and walked with support. Investigations revealed features suggestive of proximal tubular dysfunction like glucosuria, natriuria, phosphaturia and aminoaciduria; hemoglobin level 7g/dL, peripheral smear showed normocytic hypochromic red cells, blood urea 109 mg/dL and serum creatinine 2 mg/dL. Creatinine clearance by Schwartz formula was 18.42 mL/min/1.73m^2, serum sodium 137 mEq/L, potassium 5.5 mEq/L, calcium 9.6 mg/dL, phosphate 4.4 mg/Dl and alkaline phosphatase 407 U/L. Arterial blood gas showed uncompensated normal anion gap metabolic acidosis. Liver function tests and thyroid function tests were normal. Auditory evaluation was normal. On ultra sound, right kidney measured 4.8 cm and left kidney 5.1 cm with loss of corticomedullary differentiation. *X*-ray of the wrist showed rachitic changes. Slit lamp examination revealed pigmented mottling in the mid-peripheral fundus with anterior segment showing crystalline deposits allover cornea, more in the periphery, consistent with the diagnosis of cystinosis (*Fig. 1*). The patient received a high salt intake, calcium, vitamin D, iron and bicarbonate supplements (Shohl's solution). Dietary advice for higher caloric and protein intake was given.

Infantile nephropathic cystinosis is an autosomal recessively inherited cystine storage disorder characterized by intra-lysosomal accumulation of cystine(l). It is an important cause of inherited Fanconi syndrome(2) presenting with failure to thrive, poor feeding, rickets, photophobia, polyuria and polydipsia between 6 to 12 months of age. The condition progresses to end stage renal disease between 5-10 years of age(3). The youngest patients previously reported with cystinosis showed ESRD at 30 months(4) and CRF at 18 months(5). Our patient developed chronic renal failure at 9 months of age. The diagnosis was confirmed by slit lamp examination, which showed characteristic cystine crystals. We suggest that the diagnosis of cystinosis should be considered in young children with chronic renal failure, as outcome of treatment with cysteamine is satisfactory if started early(5).

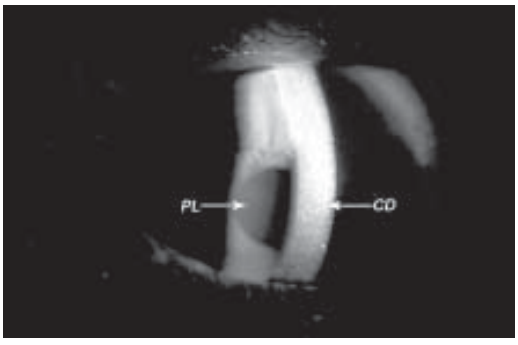

Fig. 1. Slit Lamp Examination of the eye, showing corneal deposits (CD) of cystine. PL: pupil.

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Perforative Duodenal Tuberculosis

A six-year-old female presented with severe abdominal pain and bilious vomiting since 6 hours. For the last 6 months she was being treated with antihelminthics, antispasmodics and antacids for intermittent/ colicky abdominal pain.

Examination showed a cachexic, pale child with tachycardia and tachypnea. Abdomen was mildly distended with no visible loops. Palpation revealed tenderness and guarding in upper abdomen. Liver and spleen were not palpable. Per rectal examination showed bogginess.

Investigations showed hemoglobin of 7.8 g/dL Erythrocyte sedimentation rate was 31 mm/hr. All other serum investigations were within normal limits. *X*-ray abdomen showed massive gas under diaphragm. *X*-ray chest was normal. Ultrasonography showed free fluid in Morrisons and Douglas pouch.

Exploration revealed a stricture in the terminal part of duodenum, tubercles and a (*Fig. 1*) perforated ulcer proximal to the stricture. Mesenteric lymph nodes were enlarged and appeared caseating. Rest of the viscera were normal. The perforation was closed in two layers and omentopexy was done. Side to side duodeno-jejunostomy followed, for non-passable stricture. Mesenteric lymph nodes were biopsied. Histopathology confirmed tuberculosis. Antitubercular treatment was initiated. Postoperative course was uneventful.

Tuberculosis of stomach and duodenum is rare(1) even in patients with pulmonary tuberculosis. Duodenal perforation proximal to tubercular stricture is exceptional. Extreme variety of gastric and duodenal tuberculosis is attributed to factors like sparsity of lymphoid