A 10 Year-old Male Presenting with Generalized Edema

A 10-year-old male was transferred to hospital because of generalized edema and vomiting. He was healthy until 1 month ago when malaise, anorexia and edema developed. There were positive history of short stature and poor weight gain since childhood in this patient. Physical examination revealed a pale, lethargic, edematous and ill child with temperature of 37.5°C, pulse of 92/min, respiratory rate of 20/min and blood pressure of 160/100 mmHg. His body weight was 23 kg. Auscultation of the chest revealed rales in lower parts of lungs, hyperkinetic heart, tachycardia with a III/VI systolic murmur in LSB. The patient had normal abdominal examination. Hypochromic anemia, hyperphosphatemia, increased level of creatinine, potassium and PTH and mild metabolic acidosis were seen in laboratory values. Chest x-ray and abdominal x-ray are shown in figure 1 and 2.

What is your diagnosis?

![Figure 1. Patient’s Chest X Ray](image1)

![Figure 2. Patient’s Abdominal X Ray](image2)
Photo Quiz Answer

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Chest X Ray (CXR) (Fig.1) reveals enlarged cardiac silhouette with acute angle cardiophrenic recesses. Costophrenic recesses are patent. These findings are mostly suggestive of pericardial effusion. There is evidence of right paracardiac infiltration as well. Abdominal x-ray (Fig.2) reveals calcifications in the region of either kidneys in favour of nephrocalcinosis. Abdominal sonography (not shown) reveals multiple calcifications mostly on renal pyramids on either kidney. Combination of edema, hypertension, and growth failure in association with hyperphosphatemia, anemia, hyperparathyroidism and mentioned radiological findings are highly suggestive of chronic renal insufficiency associated with nephrocalcinosis. The association of nephrocalcinosis and renal impairment and positive family history may bring additional information for diagnosis of hyperoxaluria. Fundus examination in this patient showed retinal oxalate deposits.

Primary hyperoxaluria (PH1), in general, has several presentations: infantile form with early nephrocalcinosis and kidney failure [1]; childhood or adolescence form with recurrent urolithiasis and progressive renal failure [2], a late-onset form with occasional stone passage in adulthood [3] and asymptomatic forms [4,5].

References