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## Late Diagnosed Fanconi's Anemia Presenting As Acute Renal Failure

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Figure 1. Absence of radius in right forearm.

A 30-year-old woman was admitted to the internal medicine clinic with acute, chronic renal failure. Her past history revealed high BUN levels during the last 3 months. She developed weakness, vomiting, tachypnea, convulsions and unconsciousness 3 days before admission. Physical examination revealed dehydration with Kussmaul type respiration, fever and uremic encephalopathy. She had no thumbs bilaterally. There was no consanguinity between the presents. There was no previous history of blood transfusion or any other medication related to anemia.

Her blood pressure was 60/30 mmHg, her skin dry and hyperpigmented. Microphthalmia was present. Laboratory findings revealed hemoglobin to be 5.3 g/dl, WBC 57 700/mm<sup>3</sup> and platelets 509 000/mm<sup>3</sup>. Peripheral blood smear showed 90% neutrophils, 5% lymphocytes, and 5% monocytes. MCV was 81µ<sup>3</sup>, MCH 30,3, and the erythrocyte sedimentation rate 80mm/hr.

Serum BUN was 200mg/dl, creatinine 6 mg/dl, glucose 201 mg/dl, Na 120mEq/L, K 5.3 mEq/L, Calcium 8.6 mg/dl, Phosphate 6 mg/dl, Uric Acid 8.8 mg/dl, total protein 5.3 g/dl, albumin 2.3 g/dl, serum iron 18 (N:60-150), total iron binding capacity 359 (N: 250-400), folic acid 7.8 ng (N: 3.1-12.4), and B<sub>12</sub> 2000 pg/ml (N: 223-1132). Bone marrow aspiration was normocellular with normal megakaryocytes. Blood pH was 7.16, HCO<sub>3</sub><sup>-</sup> 3.2 mmol/L, and pCO<sub>2</sub> 112 mmHg. Urinalysis revealed abundant erythrocytes and pyuria. Urinary sodium was 176 mEq/L, C<sub>cr</sub> 15 ml/min/1.73 m<sup>2</sup>. Urine and blood cultures were negative. Extremity radiology showed no thumbs bilaterally and no radius in the right forearm (Fig. 1). Abdominal ultrasonography showed left agenetic kidney, and the right kidney 120mm in size with pelvicalycial dilatation. 99Tc DMSA revealed the absence of the left kidney. There was reduced uptake at the lower pole of the right kidney. 99mTc DTPA showed left renal agenesis with delayed excretion and reduced GFR in the right kidney. Cystoscopy revealed multiple diverticules in the bladder. Peritoneal dialysis was performed. Administration of dopamine (2 µg/kg), NaCO<sub>3</sub>, 3rd generation cephalosporine and diazepam I.V. was begun. The patient recovered from the sepsis. During follow-up she developed pancytopenia with Hb 7.3g/dl, WBC 800 mm<sup>3</sup>, and platelets 29 000mm<sup>3</sup>. The second bone marrow was markedly hypocellular. Blood and urine cultures revealed a significant candida growth. C<sub>cr</sub> was at 7 ml/min/1.73m<sup>2</sup>. Fluconazol 400 ng bd and digoxine were given. Cardiopulmonary arrest developed and she died 12 hours later.

### Discussion

Fanconi's Anemia (FA) is a rare clinical syndrome

which typically presents in childhood with a variety of physical abnormalities and progressive bone marrow failure and is associated with a markedly increased risk of malignancy (1, 2). It was first described in 1967 in three brothers with hypoplastic anemia associated with a spectrum of physical abnormalities involving the skin, central nervous system and gonads (7). Since then a large spectrum of congenital malformations, including defects of the thumb, radius and radial artery, have been found in association with FA<sup>2</sup>. Most patients are diagnosed during childhood, at a median age of 7 years, and in 94% of cases the initial hematological abnormality consists of cytopenia affecting at least one lineage (1, 3). Actuarial risk of myelodysplastic syndrome (MDS) or acute myelogenous leukemia (AML) has been reported to be 52% (37% to 67%) by 40 years of age (2, 4), but renal failure has not been reported previously in the literature as a presentation of FA.

Increased chromosomal breakage in lymphocytes following treatment with a DNA cross-linking agent such as Diepoxybutane (DEB) is now absolutely essential in order for an FA diagnosis to be confirmed (5, 6, 7). We were unable to perform a DEB test on the patient, but a

possible diagnosis of FA was made based on her short stature, original face shape, microphthalmia, absence of thumbs and radius and renal agenesis and the hematological findings which later developed.

The three-month history of high BUN levels in our patient have led us to consider the possibility of chronic renal failure. It is also possible that it was an acute exacerbation of chronic renal failure secondary to sepsis originating from the bladder diverticula and infection. We conclude that the diagnosis of FA should be considered in patients with morphological abnormalities presenting with acute renal failure even in the absence of typical hematological abnormalities.

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