

Early severe anemia as the first sign of cystic fibrosis

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Dear Editor,

Sismanlar et al. introduced that early severe anemia may be the first sign of cystic fibrosis (CF). Among their 231 patients with CF, they identified 17 subjects with isolated severe anemia. CF patients with isolated anemia are younger, have a lower level of albumin, and prolonged PT/INR.

Recently, we coped an 82-day Italian male with insufficient growth and pallor. He was recently hospitalized because of bronchiolitis, followed by rotavirus enteritis. Moderate anemia (8.7 g/dL) was interpreted as sideropenic and treated with oral supplementation.

On admission, he was extremely pale, with green, liquid stools. CBC showed RBC $2.12 \times 10^{12}/L$, Hb 6.1 g/dL, MCV 92 fl. No signs of hemolysis or other common causes of anemia were found.

Anemia was treated with erythropoietin, with no success. Transfusion induced transient correction of anemia. A few days later, during an upper respiratory infection with diarrhea, albumin was 18.7 g/L; PT/INR 1.19. Sweat chloride test

(102–106 mmol/L) suggested CF, confirmed by mutation analysis (Phe508del/2183AA > G) despite negative screening for CF at birth.

Replacement therapy with pancreatic enzymes was started: anemia and stools normalized and the child gained weight.

We retrospectively reviewed our files and identified a 3-month old girl with poor growth and severe dermatitis. CBC showed RBC $1.90 \times 10^{12}/L$, Hb 5.5 g/dL, MCV 101 fL. Her brother was heterozygous for CF Phe508del mutation.

A single erythrocyte transfusion induced transient correction of anemia. Concurrent hypoalbuminemia (16.2 g/L) was treated with replacement therapy. Since dermatitis progressed, fecal chymotrypsin level was found 1 U/g. CF was diagnosed based on sweat chloride test (95–94 mmol/L) and evidence of Phe508del. Replacement therapy with pancreatic enzymes improved anemia and dermatitis.

Phe508del was present in 7/17 Sismanlar's patients and in our cases. This may suggest the need for a wider genotype-phenotype evaluation.

D. Degani and C. D'Orazio equally contributed to this paper.

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