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# A case of cystic fibrosis presenting with severe anemia in infancy

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## Introduction

Cystic fibrosis occurs with disruption in chlorine transport as a result of mutation in the KFTR gene located on the long arm of chromosome 7, which encodes the cystic fibrosis transmembrane regulator gene protein found in the epithelial cells of the airways, biliary system, intestines, vas deferens, sweat glands and pancreatic ducts. Ultimately, secretions with low water and electrolyte content and increased viscosity lead to obstruction in many organs and cause the known clinical picture of the disease [1-6]. The incidence of the disease is one in 2000-3500 live births [2]. Different mutations in the cystic fibrosis transmembrane regulator gene may lead to various clinical pictures from mild to severe [1-6]. Although hypoalbuminemia and anemia mostly occur secondary to malabsorption in the advanced stages of the disease, they are two rare findings at initial diagnosis [7,8]. Our case was presented to stress that cystic fibrosis and hemolytic anemia secondary to vitamin E deficiency caused by cystic fibrosis should be kept in mind in the differential diagnosis in the presence of hemolytic anemia in infancy.

## Case presentation

A 50-day-old male patient was admitted with failure to gain weight. He was born at term weighing 3000 g without any consanguinity between his parents. Physical examination showed body weight and height <3 percentile and head circumference 10-25 percentile. The patient appeared pale and had extremely white skin and blond hair (incompatible with the parents). The liver was palpable 1 cm under the costa, but there was no evidence of insufficiency. Organomegaly and lymphadenopathy were absent. Other system examinations were normal. Laboratory tests were Hg: 5.4 g/dL, BM: 11000/mm$^3$, MCV: 86.7fL, MCH: 28 pg, MCHC: 32.3 g/dL, RDW: 16%, Plt: 656 000/mm$^3$. Peripheral smear showed marked hypochromia, fragmented erythrocytes and schistocytes. Reticulocyte count was 17%. Direct Coombs: negative, B12: 1512 pg/mL, Ferritin: 350 ng/mL, TS: 75%, Folate: 12.7 ng/mL. In biochemistry, liver and kidney function tests were normal, only albumin was 2.3 g/dL. Prothrombin Time: 20 sec, INR: 2.3; Activated Partial Thromboplastin Time: 30 sec. Other tests were within normal limits. Pyruvate kinase and glucose 6 phosphate dehydrogenase enzyme levels were within normal limits in tests for hemolytic anemias. Hemoglobin electrophoresis from parents, siblings and the patient were within normal limits. Osmotic fragility test was within normal limits. EBV, CMV, Parvo virus serology sent for viral infections was negative. Mycoplasma serology was negative. After the tests, the patient received irradiated and filtered ERT at 5+15 cc/kg. Head radiography was performed and osteopetrosis was excluded. Abdominal USG was within normal limits. Ophthalmologic and cardiologic evaluation was normal. Bone

marrow aspiration was performed after the tests for the etiology of anemia were clear. This was considered as bone marrow with increased heterogeneous cellularity. The patient who also presented with green-colored and frequent stools during follow-up was referred to EÜTF. The sweat testing performed 2 times showed Na: 112 mmol/L and 140 mmol/L and the stool sent for cystic fibrosis was fat +. Genetic examination showed ΔF508 and ΔFI507 heterozygous mutations. Supportive treatment was started for Cystic Fibrosis. Hemolytic anemia was not observed again during follow-up with vitamin E supplementation.

## Discussion/conclusion

Different mutations in the cystic fibrosis transmembrane regulator gene may lead to various clinical pictures ranging from mild to severe [1-6]. Cystic fibrosis causes a wide range of clinical signs, but different clinical findings may also be observed by age groups. While meconium ileus, neonatal cholestasis, growth retardation, bronchiolitis, pneumonia, rectal prolapse and steatorrhea are common clinical signs in infant and infancy, malabsorption, recurrent pneumonia, bronchiolitis, nasal polyps and intussusception are more common in childhood [2-4]. In our case, malabsorption developed in the early period due to the association of two severe heterozygous mutations. Pancreatic insufficiency is observed in 85-90% of cases with CF [7,8] and may present after birth or signs may appear in later years. In the presence of pancreatic insufficiency, complaints such as fatty, foul-smelling and large amounts of stool occur. As a consequence, inability to gain weight and malabsorption findings develop. In patients diagnosed with cystic fibrosis by sweat test, the prevalence of hypoalbuminemia and low serum alpha tocopherol levels has been reported as 36% and 38%, respectively, in the first 3 months of life [9,10]. Hemolytic anemia and edema which are the initial signs of this disease have been reported more frequently in infants with low serum vitamin E levels [11]. In a prospective study in these infants, hemolytic anemia developing due to vitamin E deficiency was reported at 4% [12]. Since hemolytic anemia secondary to vitamin E deficiency was observed in our patient, this case was presented to suggest that cystic fibrosis should also be considered when investigating the etiology of hemolytic anemias observed in infants. In addition, potential oxidant metabolites (iron supplementation, gastroenteritis, hereditary or acquired factors, etc.) may trigger and increase hemolysis in vitamin E deficiency in infants with cystic fibrosis.

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