P-54 A case of hereditary spherocytosis with no family history

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Introduction

Hereditary spherocytosis (HS) is the most common congenital hemolytic anemia; autosomal dominant inheritance occur in approximately 75 % of patients, with recessive inheritance in most of the remaining. HS contains the word “hereditary”, but most of HS patients have no family history.

Case description

An 17-years-old half American and half Japanese man was admitted to our hospital with fever for four days. He had noticed jaundice when he was sick such as fever or insufficient sleep since a child. A blood test of the medical examination was normal at about 10 years old, and had not been run since then. There were no familiy history of blood disorders and cholelithiasis, and his parents were normal in the medical examination. His father was Irish American descended from Italian and French. On examination, the temperature was 37.6℃, the blood pressure was 114/65 mmHg, the pulse was 112 beats per minute. He had jaundice and splenomegaly. Abdominal ultrasound showed biliary sludge. The Blood test revealed spherocytosis with hemolytic anemia; ID-BiL 6.2 mg/dl, LDH 285 IU/l, RBC count of 208/μl, Hb 6.6 g/dl, MCV 86.5 fl, MCHC 36.7 g/dl, Haptoglobin 58 mg/dl. Coombs test was negative, and red blood cell osmotic fragility test was positive. He was diagnosed with HS. Fever and hemolytic anemia resolved in one week follow-up.

Discussion

He had no family history of HS. A significant number of patients with hematologically normal parents prove to harbor de novo mutations that will exhibit dominant inheritance in subsequent generations. Also, the inheritance pattern of sever HS is almost always recessive, and the parents of an affected patient are usually asymptomatic. The case emphasizes the importance of suspecting HS regardless of no family history.