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Quiz in Hematology

A 14-year-old female patient was admitted to our hospital because of paleness and icterus. She had been investigated at different centers for chronic anemia, icterus, and splenomegaly since age 6. She had no history of blood transfusion, drug usage, or fever. There was no consanguinity between parents, and our patient's sister had similar clinical findings. On physical examination she was pale and icteric and she had markedly enlarged liver and spleen, 3 cm and 8 cm below the costal margins, respectively. Respiratory, cardiovascular, musculoskeletal, and other examinations were normal. Informed consent was obtained.

The hemogram revealed Hb: 8.0 g/dL, RBC: $2.59x10^{12}$/L, Hct: 24.5%, MCV: 94.6 fL, RDW: 17.1, WBC: $7.65x10^{9}$/L, and platelets: $383x10^{9}$/L. Reticulocyte count was 3.4% and peripheral blood smear revealed anisocytosis and poikilocytosis. The liver and kidney function test results were within normal limits. Serum total and indirect bilirubin and LDH levels were 5.22 mg/dL, 0.46 mg/dL, and 545 IU/L respectively. Direct Coombs test was negative. Hemoglobin electrophoresis, pyruvate kinase and G6PD, vitamin B12, and folic acid levels were normal. She was immune to HBV infection and her serological tests were negative for HCV and HIV. Bone marrow examination under light microscopy showed erythroid hyperplasia, with binucleated or multinucleated and megaloblastic normoblasts (Figures 1A and 1B):

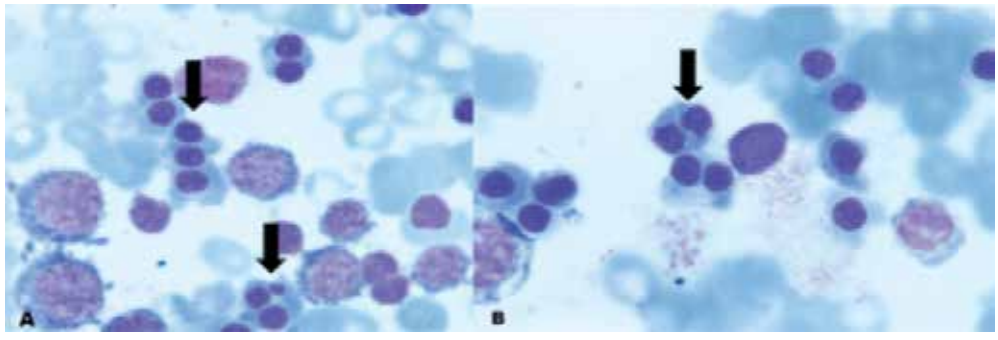

Figure 1. A and B: Marked dyserythropoiesis, nuclear bridging, binucleation, and multinucleation of the erythroid precursors.

Diagnosis: Congenital Dyserythropoietic Anemia Type 2 Due to Compound Heterozygote Mutation in *SEC23B* Gene

Bileşik Heterozigot *SEC23B* Gen Mutasyonu Olan Konjenital Diseritropetik Anemi Tip 2 Olgusu

A clinical diagnosis of congenital dyserythropoietic anemia (CDA) type 2 was established after exclusion of other causes associated with dyserythropoiesis and ineffective erythropoiesis, including thalassemia syndromes, vitamin B12 and folate deficiencies, myelodysplastic syndrome, and sideroblastic anemia. Molecular analyses for CDA type 2 revealed compound heterozygote mutations in the *SEC23B* gene (c.325G >A, p.Glu10^{9}Lys in exon 4 and c.938 G >A, p.Arg313His in exon 8).

CDA is a heterogeneous group of rare hereditary disorders of ineffective erythropoiesis and dyserythropoiesis characterized by morphologically abnormal erythroid precursors in the bone marrow. Non-erythroid hematopoietic cell morphology is normal [1]. Type 2 (CDA 2), the most common type, is autosomal recessive and typically shows marked dyserythropoiesis characterized by binucleated and multinucleated normoblasts with erythroid hyperplasia [2,3,4]. Most patients with CDA 2 show variable degrees of anemia, usually have jaundice, and may have hepatosplenomegaly. Our patient was diagnosed as having CDA type 2 by the presence of anemia, icterus, hepatosplenomegaly, binucleated and trinucleated erythroid precursors in the bone marrow, and positive *SEC23B* mutation.

Conflict of Interest Statement

The authors of this paper have no conflicts of interest, including specific financial interests, relationships, and/or affiliations relevant to the subject matter or materials included.

Key Words: Anemia, Congenital dyserythropoietic anemia type 2, *SEC23B* gene

Anahtar Sözcükler: Anemi, Konjenital diseritropetik anemi tip 2, *SEC23B* geni

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