A rare syndrome that can easily be missed: May-Hegglin anomaly

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ABSTRACT

May-Hegglin is a rare disease characterized by macrothrombocytopenia and presence of Döhle-like bodies in white cells. We present a patient treated with acute myeloid leukemia had pale-blue colored inclusion bodies assuming Döhle in his neutrophils.

Key Words: May-Hegglin, Thrombocytopenia, Leukemia.

ÖZET

Tanı zorluğu olabilecek seyrek bir sendrom: May-Hegglin anomalisi

May-Hegglin makrotrombositopeni ve lökositlerde Döhle benzeri cisimciklerle karakterize seyrek görülen bir hastalıktır. Akut miyeloblastik lösemi nedeniyle tedavi ettiğimiz, lökositlerinde soluk-mavi renkli, Döhle cisimcikleri olduğunu düşündüğümüz bir hasta takdim ediyoruz.

Anahtar Kelimeler: May-Hegglin, Trombositopeni, Lösemi.

INTRODUCTION

May-Hegglin anomaly is an autosomal dominant disease characterized by macrothrombocytopenia and presence of Döhle-like bodies in white cells[1-3]. Only about 180 cases have been reported in the literature since its first description by Hegglin in 1945[3]. It is responsible for a mild bleeding tendency in the majority of patients, and is completely asymptomatic in a few. The diagnosis may be missed if a peripheral blood slide is not carefully studied to identify giant platelets and leukocyte inclusions[2]. We present a patient treated with acute myeloid leukemia had pale-blue colored inclusion bodies assuming Döhle in his neutrophils.

A CASE REPORT

A fifty-two years old man who had skin bleeding, fever and dyspnea was admitted to

hospital. He had petechia and echimosis on skin for 3 months. Full blood tests showed a white cell count of 14.5 x 10^9 μL^{-1}, a hemoglobin concetration of 7.2 g/dL^{-1}, a platelet count of 26 x 10^9 μL^{-1}. Peripheral blood smear revealed 48% myeloblast, the presence of pale-blue intracytoplasmic inclusions like Döhle bodies in neutrophils and giant platelets (Figure 1). Inclusion bodies on leukocytes were accepted as Döhle bodies because of fever and infection disease. Diagnosis of acute myeloblastic leukemia (AML) was done by clinical and laboratory means. Daunorubicin and cytosine arabinoside therapy as induction treatment was started. He entered to remission on the 22nd day. Peripheral blood smear revealed inclusion bodies on leukocytes and giant platelets again (Figure 2). His platelet count was 50 x 10^9 μL^{-1}. After consolidation treatments same inclusion bodies, thrombocytopenia and giant platelets persisted although he had remission. Family investigations demonstrated inclusion bodies on leukocytes and macrothrombocytes in his son. In vitro platelet function studies demonstrated normal rates and extent of platelet agregation after collagen, ADP and ristosetin. But he had defective response to epinefrine.

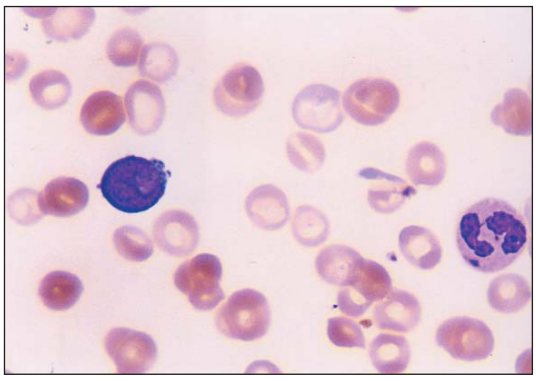

Figure 1. Döhle bodies like inclusion and myeloblast in neutrophils at the diagnosis.

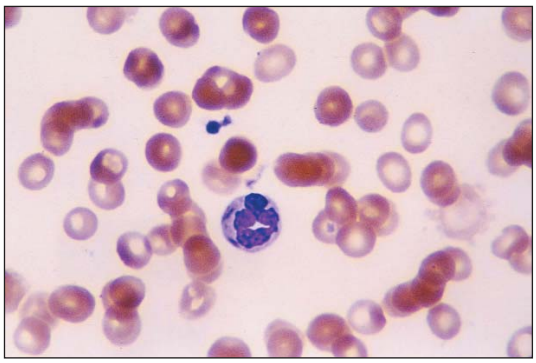

Figure 2. Giant platelets and Döhle bodies like inclusion in neutrophil at the remission.

DISCUSSION

Döhle-bodies are granules seen frequently in neutrophils on the bacterial infectious disease. These are small round or oval pale blue-grey structures, usually found at periphery of the neutrophils. Granules in May-Hegglin anomaly are bigger, and pale-blue coloured. May-Hegglin inclusions occur in all type of leukocytes except lymphocytes. Our patient received chemotherapy with acute myeloid leukemia diagnosis and had febrile neutropenia. We assumed inclusion bodies as Döhle bodies because of the fever at the time of diagnosis and recovery period. Despite the remission period after induction chemotherapy, thrombocytopenia, giant thrombocytes and inclusion bodies in neutrophils persisted. We suspected May-Hegglin anomaly, depending on this data. Further studies confirmed the diagnosis. No bleeding was observed in spite of thrombocytopenia. It has been suggested that the diagnosis of May-Hegglin is easily missed, and its frequency is probably underestimated[3]. Like the presented, establishing inclusion bodies in neutrophils on patients having thrombocytopenia and giant thrombocytes, May-Hegglin anomally should be kept in mind in differential diagnosis.

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