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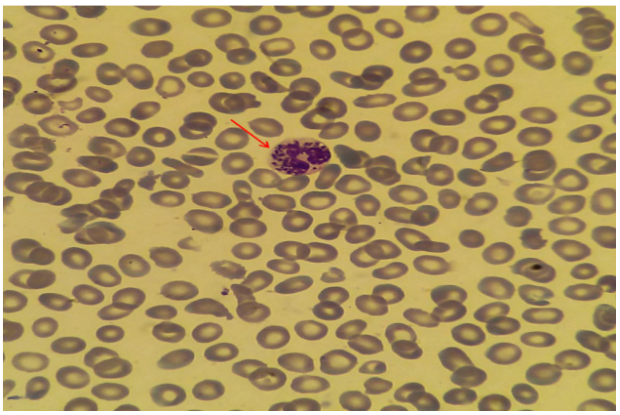

Figure 1. Peripheral Blood Smear of the Patient (Wright staining × 100).

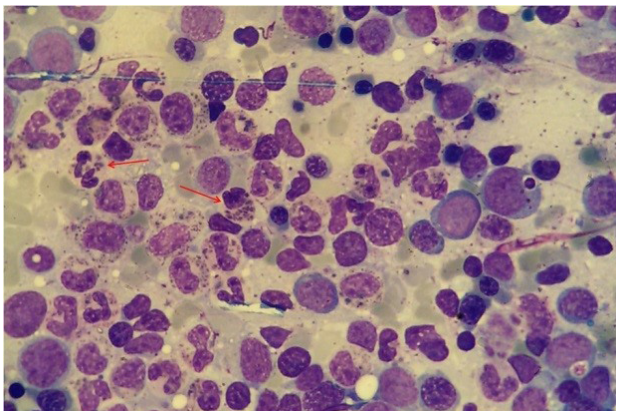

Figure 2. Bone Marrow Aspiration of the Patient (Wright staining × 100).

A 27-years-old man was referred to the emergency department of the Tohid hospital of Sanandaj with fever, tachypnea, tachycardia and purulent sputum-producing cough.

He mentioned a history of a hereditary disorder from infancy with an initial presentation of recurrent respiratory infection, hepatosplenomegaly, speckled hypopigmentation of face and extremities and strabismus. His two brothers died in infancy due to a similar disorder. On physical examination, hepatosplenomegaly without lymphadenopathy and coarse, bibasilar end-inspiratory crackles were noted. White cell count was 2500 per mm^2, hemoglobin was 8.5 mg/dL and platelet count was 40 000 per mm^2. Abdominal ultrasound sonography showed huge splenomegaly (249 mm) and hepatomegaly (172 mm). Based on history, physical examination and pancytopenia, a peripheral blood smear was prepared which showed giant granules in the cytoplasm of neutrophils (Figure 1). Bone marrow aspiration and biopsy were performed that revealed approximately 70 percent cellularity with vacuoles containing large azurophilic granules in myeloid precursor cells without lymphohistocytic infiltration (Figure 2).

What is your diagnosis?
See the next page for your diagnosis.

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Photoclinic Diagnosis

Chediak-Higashi Syndrome

Chediak-Higashi Syndrome (CHS) is a rare autosomal recessive disorder, which has been reported in less than 500 cases worldwide.[1] The patients are typically identified in infancy or early childhood, characterized by diverse clinical manifestations such as partial oculocutaneous albinism, sensory and motor neurologic defects, hematologic manifestation (anemia, thrombocytopenia, leukopenia and abnormal bleeding time) which lead to immunodeficiency, bleeding tendency and frequent bacterial infections.[2] It is also associated with 'accelerated phase' which is a lymphoproliferative disorder and is characterized by lymphocytic infiltration of the major organs of the body.[3]

The diagnosis of CHS is based on examining peripheral blood smear and bone marrow for the characteristic cytoplasmic giant granules in neutrophils, eosinophils, and other granulocytes. The diagnosis can be confirmed by genetic testing for mutations in the CHS1/LYST gene.[4]

CHS is treated with prophylactic antibiotics for acute bacterial infections and opportunistic pathogens. This can control recurrent infections but cannot prevent the other complications of CHS. Granulocyte colony stimulating factor (G-CSF) is used to correct neutropenia and decrease infection. High-dose glucocorticoids and splenectomy can be used to induce transient remission in the accelerated phase. Hematopoietic cell transplantation (HCT) is the choice treatment for correction of the immunologic and hematologic manifestations of CHS and is the only curative treatment.[5]

Most patients with CHS die from pyogenic infection before seven years of age if not transplanted,[6] but our case was 27-years-old despite lack of HCT.

Authors' Contribution

All authors contributed equally to this study.

Conflict of Interest Disclosures

None.

Ethical Statement

Informed written consent was obtained from the patient to participate in the study.

Acknowledgements

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