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CASE REPORT

Chediak–Higashi syndrome presenting in accelerated phase: A case report and literature review

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Abstract
Chediak–Higashi syndrome (CHS) is a rare autosomal recessive lysosomal disorder characterized by frequent infections, oculocutaneous albinism, bleeding diathesis, and progressive neurologic deterioration. In 85% of cases, CHS patients develop the accelerated phase characterized by pancytopenia, high fever, and lymphohistiocytic infiltration of liver, spleen, and lymph nodes. Treatment of accelerated-phase CHS is difficult and the prognosis is poor. Here, we report a case of CHS in a 2-year-old boy who presented in the accelerated phase of the disease. CHS diagnosis was made on the basis of clinical characteristics, hair analysis, and identification of pathognomonic giant azurophilic granules in peripheral blood and bone marrow.

Chediak Higashi syndrome (CHS) is a rare autosomal recessive disorder with fewer than 500 cases published worldwide over the last 20 years [1]. The largest CHS study to date included 15 patients [2]. In Tunisia, Bouatay et al. (2014) [3] reported a single case of CHS. The clinical features of this syndrome include partial albinism, photosensitivity, severe recurrent bacterial infections, bleeding diasthesis, and late onset neurological manifestations (central and peripheral neuropathies, sensory loss, muscle weakness, parkinsonism, cerebellar ataxia, and cognitive impairment) [4,5]. Approximately 85% of cases develop a fatal accelerated phase characterized by pancytopenia,

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hemophagocytosis, and marked infiltration of organs by lymphocytes, leading to multi-organ dysfunction [6].

Owing to the rarity of the condition and the characteristic clinical and hematological findings, we report a case of Chediak–Higashi syndrome, which presented as accelerated phase.

Case report

The patient is a 2-year-old male that presented with a month-long history of fever, abdominal distension, and cough. There was a prior history of repeated attacks of low respiratory-tract infections and he developed osteomyelitis at 1 year of age. He is the third child from a consanguineous marriage, displays normal psychomotor development, and there is no family history of the disease. On examination, the patient was febrile, of average build, weighed 14 kg, and had blond hair and hypopigmentation of the skin (Figure 1). The child was anemic, with cervical and axillary lymphadenopathy. Respiratory system examination revealed moderate respiratory distress with bilateral coarse crepitations. The patient had a protuberant abdomen with massive hepatosplenomegaly. The cardiovascular and nervous systems were normal.

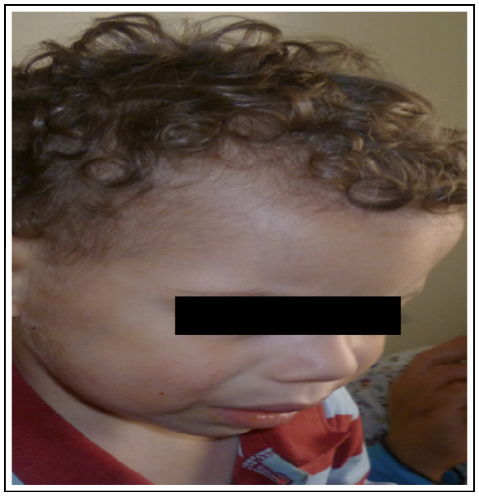

Figure 1 Face of the patient. Note skin hypopigmentation and blond hair.

Laboratory investigations showed elevated C-reactive protein (132 mg/L), hyponatremia (124 mmol/L), high ferritin levels (2685 ng/mL), low fibrinogen levels (<1.5 g/L), and hypertriglyceridemia (3.57 mmol/L). The relevant hematological findings were hemoglobin 5.2 g/dL, leucopenia at 3.56×10^9/L, and thrombocytopenia (Platelet count 26×10^9/L). Peripheral blood smear showed several abnormal giant granules in most leukocytes. Bone marrow aspirate revealed prominent granules within the lymphocytes and myeloid cells (Figure 2). Phagocytosis of red blood cells and red-blood-cell precursors hemophagocytosis was also observed (Figure 3).

The patient fulfilled the diagnostic criteria for hemophagocytic lymphohistiocytosis (HLH), e.g., prolonged fever, splenomegaly pancytopenia, high ferritin levels, low fibrinogen levels, hypertriglyceridemia, and hemophagocytosis. Thus, the diagnosis of accelerated phase of CHS was

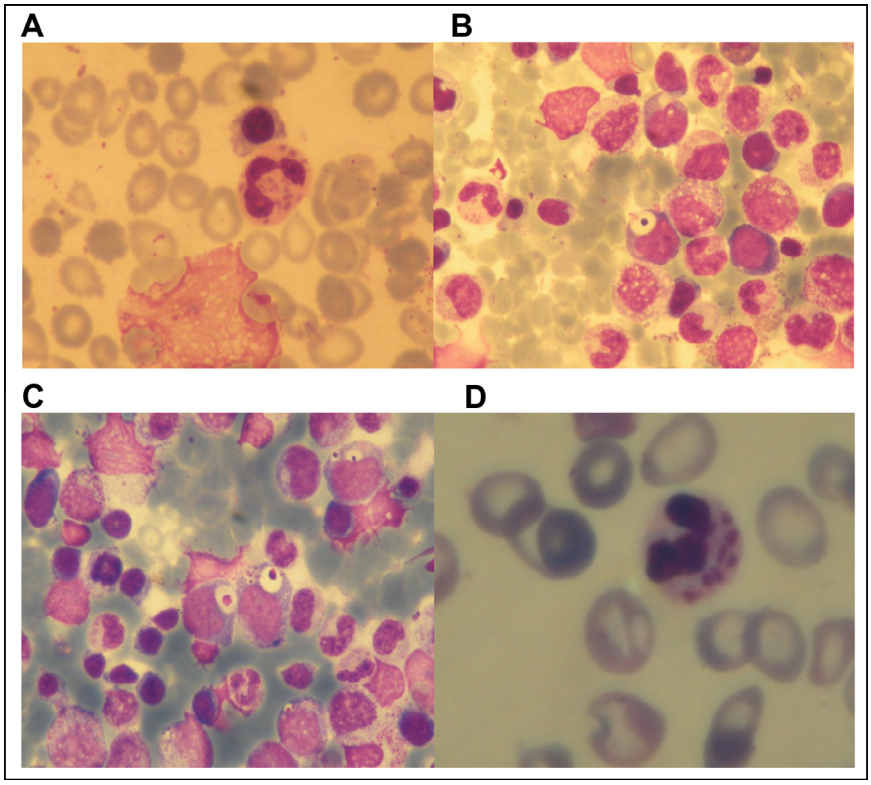

Figure 2 Bone marrow aspirate (A–C) and peripheral blood (D) smear showing myeloid precursor and lymphocytes with abnormal granules.