Article published online: 2021-06-23

Letter to Editor

Chediak–Higashi Syndrome Presented With Hemophagocytic Lymphohistiocytosis

Sir,

Herein, we report a case of Chediak–Higashi syndrome (CHS) presented with hemophagocytic lymphohistiocytosis (HLH). A 3-year-old girl presented with recurrent infections, fever, and icterus. Physical examination revealed hepatosplenomegaly, bilateral cervical and inguinal lymphadenopathy, silvery gray hair, strabismus, and bilateral nystagmus. Giant granules in lymphocytes and monocytes were seen on blood smear. Bone marrow aspirate exhibited erythrophagocytosis and numerous giant granules of predominantly myeloid lineage [Figure 1a]. Examination of the hair showed an irregular distribution of large and small pigment clumps [Figure 1b]. Magnetic resonance imaging of the brain showed diffuse cerebral atrophic [Figure 1c]. The girl's parents were consanguineous. On laboratory examination, urinary analysis was unremarkable. Hemoglobin, white blood cell count, and thrombocyte count were 7, 6 g/dL, 2300/mm^3, and 76.000/mm^3, respectively. Sedimentation rate was 2 mm/h (N: 0–20 mm/h), and serum triglyceride 390 mg/dL (N: 32–99 mg/dL), fibrinogen 67 ng/dL (N: 200–400 ng/dL), and ferritin was 129.000 ng/mL (N: 7–140 ng/mL) [Table 1]. Prothrombin time was 21 s (N: 11–15 s), and activated partial thromboplastin time was 62 s (N: 25–35 s).

CHS is a rare, autosomal recessive inherited disorder characterized by variable degrees of oculocutaneous albinism, recurrent respiratory system and pyogenic infections and intracytoplasmic giant granules in leukocytes, monocytes, platelets, melanocytes, and erythroid precursors.[1,2] Abnormal granules in the granulocyte cells are pathognomonic for the diagnosis of CHS, and these can be easily seen in blood smear and bone marrow aspiration of all patients with CHS. There are two phases of the disease. In the stable period, clinical situation is milder. In accelerated phase, lymphohistiocytic infiltration, fever, peripheral neuropathy, hepatitis, hepatosplenomegaly, lymphadenopathy, pancytopenia, coagulopathy, and hemorrhage are seen. On gingivitis and buccal mucosa, pseudomembranous may occur. Accelerated phase occurs in 85% of cases.[3,4] HLH, also termed the "accelerated phase," is a hyperinflammatory syndrome with a high mortality rate. In accelerated phase, hepatosplenomegaly, thrombocytopenia, hypertriglyceridemia, and hypofibrinogenemia are detected.[5] In the previous report from our region, Akbayram *et al*. reported a case of 14-month-old girl patient with CHS presented with HLH.[1] The particular aspect of our study is that the patient has a very high ferritin level which presented in HLH. In our case, fever, anemia, neutropenia, thrombocytopenia, low fibrinogen level, high transaminase level, hypertriglyceridemia, hyperbilirubinemia, histiocytes demonstrating erythrophagocytosis in bone marrow aspiration were detected.

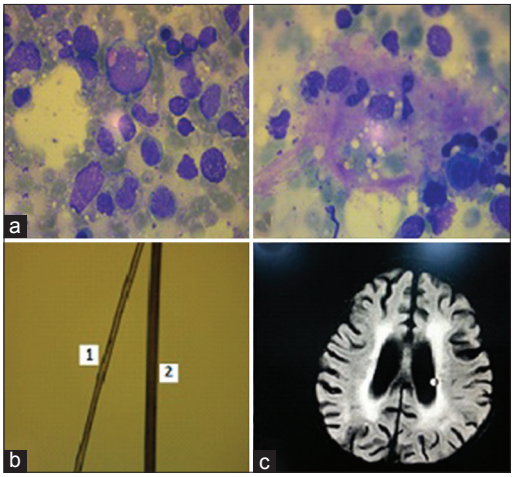

Figure 1: (a) Bone marrow aspirate smears showing giant cytoplasmic granules with in myeloid precursors and hemophagocytosis. (b) 1 patient's hair, 2 Normal hair. (c) Magnetic resonance imaging images show diffüz cerebral atrophy

Table 1: Laboratory findings of the patient

	Result
Hemoglobin (g/dL)	7.6
WBC (/mm^3)	2300
PLT (/mm^3)	76.000
Sedimentation (mm/h)	2
Triglyceride (mg/dL)	390
Fibrinogen (ng/dL)	67
Serum sodium (mEq/dL)	122
Ferritin (ng/mL)	129.000

WBC – White blood cell; PLT – Platelet

Financial support and sponsorship

Nil.

Conflicts of interest

There are no conflicts of interest.

Kamuran Karaman, Sinan Akbayram, Hadi Geylan, Mecnun Çetin[1]

Departments of Pediatric Hematology and [1]Pediatrics, Yuzuncu Yil University, Van, Turkey

Address for correspondence:
Dr. Kamuran Karaman,
Department of Pediatric Haematology, Yuzuncu Yıl University, 65100, Van, Turkey.
E-mail: kamuran_karaman@hotmail.com

References

1. Akbayram S, Akgun C, Basaranoglu M, Kaya A, Balta G, Ustyol LN, *et al*. A case of Chediak Higashi syndrome presented with hemophagocytic lymphohistiocytosis. Int J Hematol Oncol 2011;21:196-9.
2. Pujani M, Agarwal K, Bansal S, Ahmad I, Puri V, Verma D, *et al.* Chediak-higashi syndrome – A report of two cases with unusual hyperpigmentation of the face. Turk Patoloji Derg 2011;27:246-8.
3. Page AR, Berendes H, Warner J, Good RA. The Chediak-Higashi syndrome. Blood 1962;20:330-43.
4. Rubin CM, Burke BA, McKenna RW, McClain KL, White JG, Nesbit ME Jr., *et al.* The accelerated phase of Chediak-Higashi syndrome. An expression of the virus-associated hemophagocytic syndrome? Cancer 1985;56:524-30.
5. Barak Y, Nir E. Chediak-higashi syndrome. Am J Pediatr Hematol Oncol 1987;9:42-55.

Access this article online

Quick Response Code:

Website: www.ijmpo.org

DOI: 10.4103/ijmpo.ijmpo_54_16

How to cite this article: Karaman K, Akbayram S, Geylan H, Çetin M. Chediak–Higashi syndrome presented with hemophagocytic lymphohistiocytosis. Indian J Med Paediatr Oncol 2018;39:122-3.