Clinical Case Reports International

Case Report
Published: 22 Dec, 2020

Chédiak-Higashi Syndrome: Case Report and Review of the Literature

Safa Suleman Elfaituri[1] and Idris Matoug[2]*

[1]*Department of Dermatology, Benghazi University, Benghazi, Libya*

[2]*Department of Pediatric, Benghazi University, Benghazi, Libya*

Abstract

A case of Chediak-Higashi syndrome is reported in a two-year-old girl who presented with fever, hepatosplenomegaly, and pancytopenia without any history of previous infections. Parental consanguinity was present. Presence of giant granules in leucocytes and melanocytes of the hair confirmed the diagnosis. Though most patients undergo a variable period of recurrent infections before going into the accelerated phase, this case was unusual in that her primary presentation was in the accelerated phase.

Introduction

Chédiak-Higashi Syndrome (CHS) is a rare autosomal recessive multisystem disease, characterized by partial oculocutaneous albinism, blond hair, severe immunodeficiency and presence of abnormal large cytoplasmic granules in leukocytes and other granule containing cells. The accelerated phase of CHS consisting of a lymphoproliferative syndrome manifested by hepatosplenomegaly, lymphadenopathy and pancytopenia may occur shortly after birth or several years later [1]. Morbidity results from frequent cutaneous and respiratory pyogenic infections or from an accelerated phase. Most patients undergo a variable period of recurrent infections before going into the accelerated phase. Therefore, primary presentation in the accelerated phase is unusual [2].

Case Presentation

Our patient was born at full term to consanguineous parents after normal pregnancy and labor. She was normal at birth and her symptoms began at the age of two years when she started having progressive abdominal distension, associated with fever and loss of appetite for about 1 month before seeking medical advice. Parents and siblings; 2 sisters and 4 brothers are healthy; they have neither fair skin nor blond hair. One sibling with fair skin and silvery hair was died due to similar disease. The onset of his illness was started at 3 months of age with recurrent chest infections, developed accelerated phase and died at the age of 1.5 year.

On examination she had fair skin and blond hair with photophobia and nystagmus. Her weight and height were at 3rd percentile. Chest was clear. Abdominal examination revealed hepatosplenomegaly. Ophthalmic examination showed pale retina and papilledema bilaterally. Lab investigations results were anemia (HB=6), platelet =27, WBC=6 with neutropenia, ESR=40,

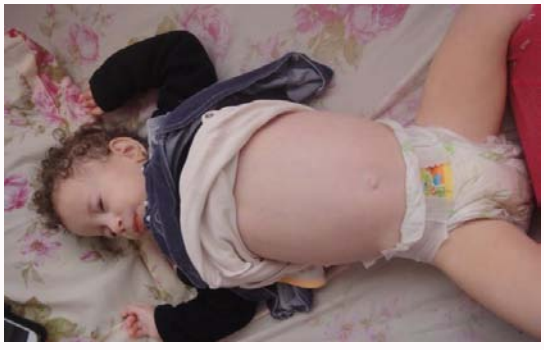

Figure 1: Fair skin & hepatosplenomegaly.

OPEN ACCESS

Correspondence:
Safa Suleman Elfaituri, Department of Pediatric, Benghazi University, Benghazi, Libya,
E-mail: selfaitoury@yahoo.co.uk

Received Date: *18 Nov 2020*
Accepted Date: *17 Dec 2020*
Published Date: *22 Dec 2020*

Citation:
Elfaituri SS, Matoug I. Chédiak-Higashi Syndrome: Case Report and Review of the Literature. Clin Case Rep Int. 2020; 4: 1204.

Copyright *© 2020 Safa Suleman Elfaituri. This is an open access article distributed under the Creative Commons Attribution License, which permits unrestricted use, distribution, and reproduction in any medium, provided the original work is properly cited.*

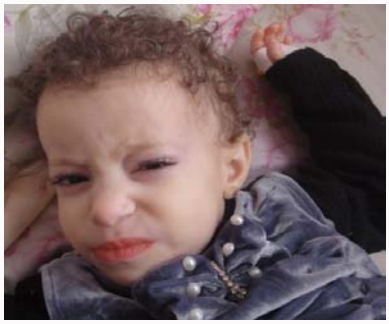

Figure 2: Fair hair & photophobia

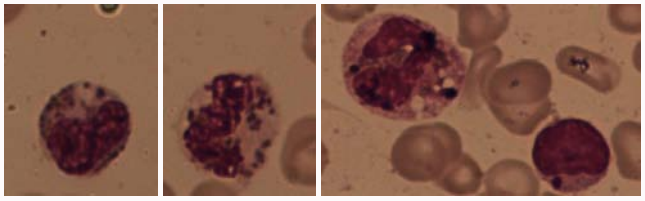

Figure 3: Bone marrow with granular cells. Peripheral blood with giant granules WBC.

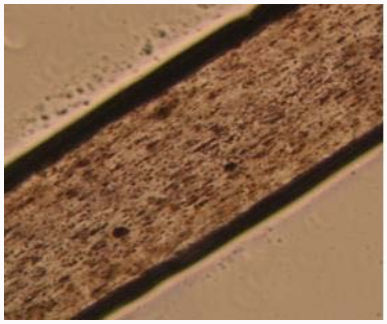

Figure 4: Giant melanosomes in hair under light microscope.

normal RFT, elevated liver enzymes and hypergammaglobulinemia. Microscopic examination of Wright-stained blood and bone marrow films showed giant granules of leukocytes and their precursors respectively whereas large clumped melanosome were seen by hair examination; confirming the diagnosis of CHS. On the basis of the clinical presentation, hematologic, and cytological findings, a diagnosis of accelerated phase of CHS was made. Molecular testing could not be performed due to unavailability. Patient was treated with antibiotics, ascorbic acid and blood transfusion. Currently the patient is under observation with continued symptomatic treatment.

Discussion

CHS is a lysosomal disease in which there is aberrant fusion of primary lysosomes with gross enlargement of lysosomes in all tissues. The mutated gene for CHS, lysosomal trafficking regulator is located at chromosome 1q2-q44 [3]. The exact incidence of CHS is unknown, less than 500 cases have been reported worldwide in the past 20 years [4].

The pathologic hallmark of CHS is the presence of giant lysosomal granules in the myeloblasts and promyelocytes of the bone marrow and in white blood cells of peripheral blood. The inclusions in melanocytes and keratinocytes consist of clumped melanosomes [5]. CHS can be diagnosed prenatally by examining a sample of hair from a fetal scalp biopsy or testing leukocytes from a fetal blood sample [5].

Patients with CHS exhibit hypopigmentation of the skin, hair and eyes, possibly secondary a feedback inhibition of melanin synthesis, which results from accumulation and clumping of melanosomes (Destruction of the giant melanosomes may dilute skin color. However, abnormal packing of normal-sized melanosomes into large lysosome-like structures in the epidermal cells may provide a more likely basis for the hypopigmentation [3]. Other features include photophobia and nystagmus, easy bruisability and peripheral neuropathy [4].

Our case had fair skin and blond hair since birth unlike her relative parents and siblings but similar to her died brother, bone marrow and peripheral blood smears showed giant granules in leukocyte precursor cells and leukocyte respectively. Hair examination under light microscope showed large clumped melanosome. She had no bleeding history but had photophobia, nystagmus, pale retina and papilledema most probably is due to infiltration of the optic nerve by lymphocytic cells.

Cell-mediated defect including deficient natural killer cell activity, humoral immune response abnormalities as well as impaired neutrophil chemotaxis and neutropenia in CHS leads to increased susceptibility to infections. Infections are most commonly due to *Staphylococcus aureus* and B-hemolytic streptococci. Approximately 85% of patients develop an accelerated phase which is usually fatal. The widespread visceral infiltration of lymphohistiocytic cells gives rise to fever, jaundice, hepatosplenomegaly, lymphadenopathy, pancytopenia, a tendency to bleed and neurologic changes. Viral infections, especially with Epstein-Barr virus, could be the cause of this lymphomatous phase [6]. Most patients undergo a variable period of recurrent infections before going into the accelerated phase [2]. Therefore, primary presentation in the accelerated phase in our patient was unusual.

CHS is considered a fatal disease, death often occurs in the first decade of life as a result of overwhelming infections, hemorrhage or development of the accelerated lymphoma-like phase [1]. The treatment of CHS is difficult, and has limited to bone marrow transplantation [7] although the accelerated phase may respond to etoposide plus systemic steroids and intrathecal methotrexate but the disease relapses invariably [1].

Conclusion

We report a rare case of CHS, to the best of our knowledge, she is the first case reported from Libya. It is unusual case in that the first presentation was in the accelerated phase at 2 years of age. As bone marrow transplantation is the only solution to the disease, early diagnosis is required before the accelerated phase has developed. Genetic counseling and family education about the disease and the recurrence risk in the subsequent pregnancy is needed.

References

1. Masood A, Nadeem M, Aman S, Kazmi AH. Chediak-Higashi syndrome – A case report. Ann King Edw Med Univ. 2008;14(3):119-22.
2. Nargund AR, Madhumathi DS, Premalatha CS, Rao CR, Appaji L, Lakshmidevi V. Accelerated phase of chediak higashi syndrome mimicking lymphoma--a case report. J Pediatr Hematol Oncol. 2010;32(6):e223-6.
3. Reddy RR, Babu BM, Venkateshwaramma B, Hymavathi CH. Silvery hair syndrome in two cousins: Chediak-Higashi syndrome vs. griscelli

syndrome, with rare associations. Int J Trichology. 2011;3(2):107-11.

4. Ghaffari J, Rezaee SA, Gharagozlou M. Chédiak–Higashi syndrome. J Pediatr Rev. 2013;1(2):80-87.
5. Stolz W, Graubner U, Gerstmeier J, Burg G, Belohradsky BH. Chediak-Higashi syndrome: Approaches in diagnosis and treatment. Curr Probl Dermatol. 1989;18:93-100.
6. Carnide EM, Jacob CM, Pastorino AC, Bellinati-Pires R, Costa MB, Grumach AS. Chédiak-Higashi syndrome: Presentation of seven cases. Sao Paulo Med J. 1998;116(6):1873-8.
7. Haddad E, Le Deist F, Blanche S, Benkerrou M, Rohrlich P, Vilmer E, et al. Treatment of Chédiak-Higashi Syndrome by allogenic bone marrow transplantation: Report of 10 cases. Blood. 1995;85(11):3328-33.