CASE REPORT

Chediak–Higashi Syndrome – A Rare Case Report

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ABSTRACT

A 4-year-old female child born out of a consanguineous marriage, full-term normal vaginal delivery, presented with multiple hypopigmented macules giving mottled pigmentation over the face and neck. There was a history of swelling and pain in the right ear and recurrent skin infections. On examination, she had a silvery gray hair, mottled pigmentation over the face and neck, perichondritis of right ear, impetiginous lesions over the lower limbs, hepatomegaly. Ophthalmological examination revealed partial oculocutaneous albinism and nystagmus. Routine blood investigations were within normal limits. Peripheral smear examination revealed prominent granules within the leukocytes. Hair shaft examination revealed regularly arranged small clumps of melanin. It was diagnosed as a case of Chediak–Higashi syndrome. This case is presented for its rarity.

KEY WORDS: Chediak–Higashi, silvery hair, syndrome.

Introduction

Chediak–Higashi syndrome (CHS) is a rare lysosomal disorder characterized by defect is in the gene lysosome trafficking regulator, resulting in defective vesicular transport to and from the lysosome. Chediak–Higashi gene product has been identified and mapped on chromosome 1 q 43.[1] It is characterized by oculocutaneous hypopigmentation, photophobia, nystagmus, neutropenia, and an abnormal susceptibility to cutaneous and respiratory infections.[2,3] Hepatosplenomegaly, lymphadenopathy, pancytopenia, jaundice, and gingivitis with bleeding tendency are other common features.[4] The diagnosis can be confirmed by recognition of the characteristic large cytoplasmic inclusions in leukocytes by peripheral smear examination.[1] Morbidity results from patients to frequent bacterial infections or to an accelerated phase, consisting of a lymphoproliferative syndrome with hemophagocytosis and infiltration of most tissues.[3]

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Case Report

A 4 year old female child born at full term by normal delivery to consanguineous parents presented with a history of pigmentary disturbances in skin from the age of 2 years. There was a history of recurrent skin infections, swelling, and pain in the right ear for 15 days. No history of respiratory infection, bleeding, and similar complaints in the family. On examination, she had silvery gray hair, multiple ill-defined hypopigmented macules over the face, neck, perichondritis of right ear, multiple erosions covered with crusts over both upper limbs and lower limbs, hepatomegaly (Figures 1 and 2). Blood investigations were within normal limits. Hair shaft examination revealed regularly arranged clumps of melanin (Figures 3 and 4). Peripheral smear showed giant granules in leukocytes (Figure 5). It was diagnosed as a case of CHS. Appropriate antibiotics were used to treat the infection. The patient was referred to a higher center for further management.

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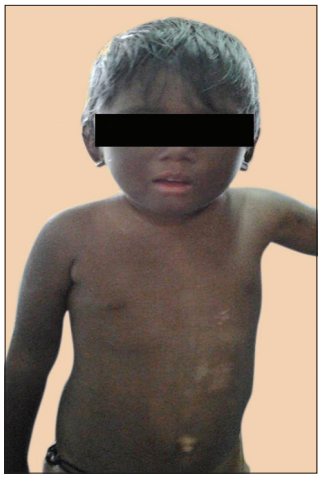

Figure 1: Photograph showing silvery gray hair, mottled pigmentation

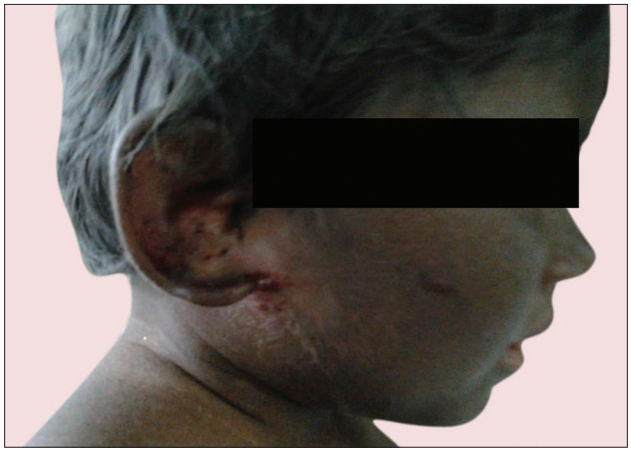

Figure 2: Photograph showing perichondritis of right ear

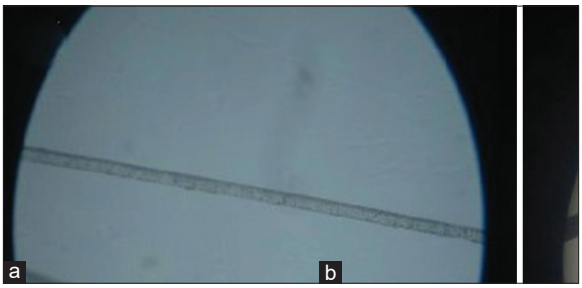

Figure 3: Light microscopic examination of hair shaft ×10 (a) normal hair (b) patient hair

Discussion

Patients with CHS exhibit hypopigmentation of the skin, hair, and eyes due to the presence of giant melanosomes which cause pigment dilution, possibly secondary to impaired melanin trans-port.[5] There are recurrent staphylococcal and streptococcal infections as a result of neutropenia and other

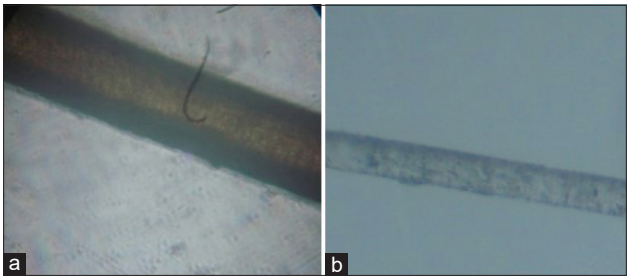

Figure 4: Light microscopic examination of hair shaft ×40 (a) normal hair (b) patient hair

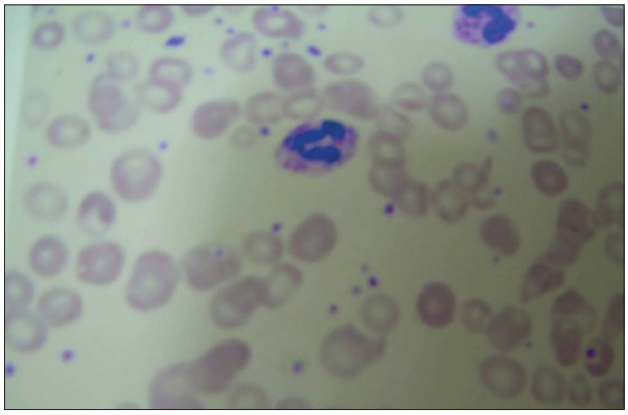

Figure 5: Peripheral smear showing giant granules in leukocytes

complications such as anemia, thrombocytopenia, and lymphoma can also occur in these patients.[3,4] In our case, the child had silvery gray hair, recurrent skin infections, hepatomegaly, perichondritis of the right ear, and typical peripheral smear examination showing giant granules within the leukocytes and hair shaft light microscopy with findings consistent with a diagnosis of CHS. The condition has to be differentiated from other conditions such as Griscelli syndrome and Elejalde disease. Certain laboratory investigations help to differentiate between the three entities [Table 1].

The prognosis of CHS is generally not good.[2-4] Clinicians should be familiar with the severity of the disease because death often occurs in the first decade of life as a result of overwhelming infections, hemorrhage, or development of the accelerated lymphoma-like phase.[2-4] Treatment is limited to allogeneic bone marrow transplantation (BMT), although the accelerated phase may respond to etoposide plus systemic steroids and intrathecal methotrexate, the disease relapses invariably.[3]

Table 1: Investigations to differentiate between Griscelli syndrome, CSH, and Elejalde syndrome.[6]

Investigations	Griscelli syndrome	CHS	Elejalde syndrome
Peripheral blood smear		Prominent granules in leukocytes and giant granules	
Light microscopy of hair	Small and large clumps of melanin in irregular pattern	Small clumps of melanin in regular pattern	Small and large clumps of melanin in irregular pattern.
Histopathology of skin	Excess pigmentation of skin in basal layer and scanty pigmentation in surrounding area	Entirely normal but may show melanin macroglobules and perhaps sparse dermal melanin	Irregular sized melanin granules dispersed in basal layer.
Electron microscopy of skin	Mature melanosomes in melanocytes and to some extent in keratinocytes	Large melanosomes in both melanocytes and keratinocytes	Melanosomes at different stage of formation in the melanocytes.

CHS: Chediak–Higashi syndrome

Allogeneic BMT from a human leukocyte antigen-matched sibling is the therapy of choice and should perform early. If no matched family donor is available, an unrelated donor or a placental blood graft is a good alternative. Without BMT, children with Chediak–Higashi syndrome usually die before age 10 years.[7]

References

1. Atherton DJ. The neonate. In: Champion RH, Burton JL, Burns DA, Breathnach SM, editors. Rook's Textbook of Dermatology. 6th ed. London: Blackwell Science; 1998. p. 449-518.
2. Paller AS. Genetic immunodeficiency diseases. In: Freedberg IM, Eisen AZ, Wolff K, Austen KF, Goldsmith LA, Katz Sl, *et al*, editors. Fitzpatrick's Dermatology in General Medicine. 6th ed. Toronto: McGraw-Hill; 2003. p. 1119-29.
3. James WD, Berger TG, Elston DM. Disturbances of pigmentation. In: Andrew's Diseases of the Skin. 10th ed. Philadelphia, PA: WB Saunders; 2006. p. 853-68.
4. Fitzpatrick TB, Ortonne JP. Normal skin color and general considerations of pigmentary disorders. In: Freed-berg IM, Eisen AZ, Wolff K, Austen KF, editors. Fit-zpatrick's Dermatology in General Medicine. 6th ed. Toronto: McGraw-Hill; 2003. p. 819-35.
5. du Vivier A. Disorders of the hair and scalp. In: Atlas of Clinical Dermatology. 3rd ed. Philadelphia, PA: Churchill Livingstone; 2002. p. 628.
6. Cahali JB, Fernandez SA, Oliveira ZN, Machada MC, Valente NS, Scotto MN. Elejalde syndrome: Report of a case and review of literature. Pediatr Hematol 2004;21:479-82.
7. Liang JS, Lu MY, Tsai MJ, Lin DT, Lin KH. Bone marrow transplantation from an HLA-matched unrelated donor for treatment of Chediak-Higashi syndrome. *J Formos Med Assoc* 2000;99:499-502.

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