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Case Report

Cutaneous presentation in chediak – Higashi syndrome – A rare case report

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ABSTRACT

A One and half year-old female child born out of 2^{o} consanguineous marriage came with multiple hypopigmented patches over the face, trunk and lower limbs. There was history of recurrent upper respiratory tract infections. On examination she had silvery grey hair, hypopigmented patches and mild hepatomegaly. Ophthalmological examination revealed oculocutaneous albinism. Skin biopsy showed coarse clumps of melanin pigment in the epidermis. Hair mount examination revealed melanin granules in cortex and medulla. Routine blood investigations were within normal limits. Peripheral smear examination showed giant granules in neutrophils and lymphocytes.

1. Introduction

Chediak – Higashi syndrome (CHS), first described by Beguez – Cesar in 1943[1] is also called "Beguez Cesar syndrome". Further reports by Chediak in 1952 and Higashi in 1954 which emphasized the hematological features.[2] It is a rare autosomal recessive disorder characterized by hypopigmentation of skin, eyes and hair. It is mainly characterized by mutation in CHS1/LYST gene located at 1q42, resulting in abnormal protein trafficking and aberrant fusion of vesicles.[3,4] It constitutes a part of "silvery hair syndrome" along with other similar condition like Griscelli syndrome (GS).[5] Generally these patients present with organomegaly along with recurrent infections due to immune deficiency. Skin features as a presenting complain is rarely encountered. Herein we report one case of CHS in one and half year-old girl who came with cutaneous presentation. This case is being reported due to its extreme rarity.

2. Case Report

One and half year-old girl born out of to 2° consanguineous marriage, full term by normal vaginal delivery was referred to our hospital with multiple hypopigmented patches over face, trunk and lower limb (Figure 1a) since past 6 months along with silvery grey hair (Figure 1b). There was a history of recurrent respiratory tract infections for 6 months and intermittent fever in the last 2 months. Meanwhile broad-spectrum antibiotics were started and she became afebrile within 2 days. There was history of prior admissions with similar complains. There was no similar complaints in the family.

Physical examination showed developmental delay. On palpation there was mild hepatosplenomegaly, vital parameters were within normal limits. Further examination revealed patient was having difficulty in opening eyes in bright light and ophthalmological examination revealed oculocutaneous albinism. Skin biopsy showed coarse clumps of melanin pigment in the epidermis and dermal lympho-histiocytic aggregates (Figure 2a,b). Hair mount examination revealed melanin granules in cortex and medulla. This finding has an overlapping feature with other

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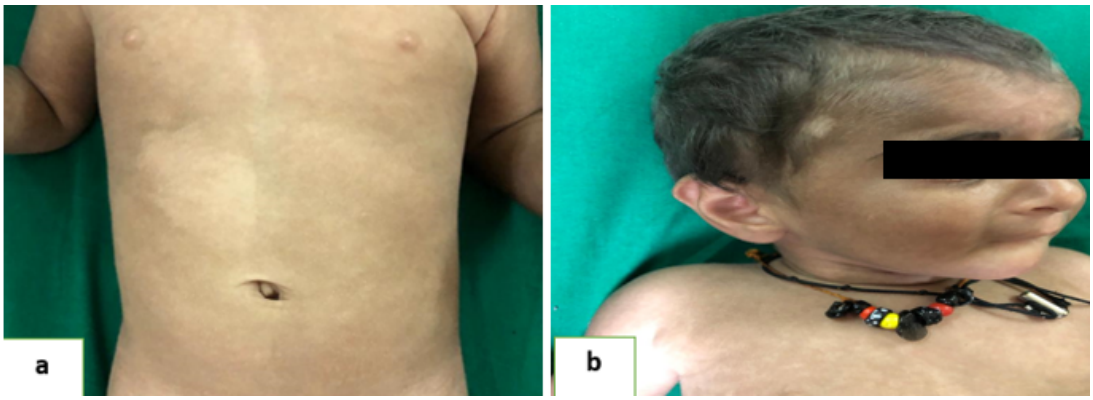

Fig. 1: a: Child of CHS with hypopigmented skin patch; **b:** Child of CHS with silvery grey hair.

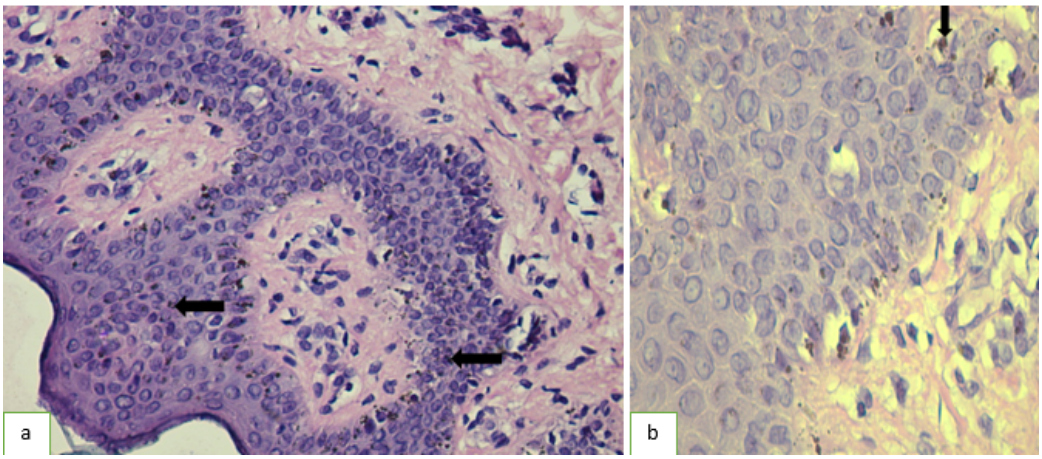

Fig. 2: a: Low power (10x) micro photograph of H and E section of skin biopsy showing coarse clumps of melanin pigment in epidermis; **b:** High power (40x) micro photograph of H and E section of skin biopsy showing coarse clumps of melanin pigment in epidermis.

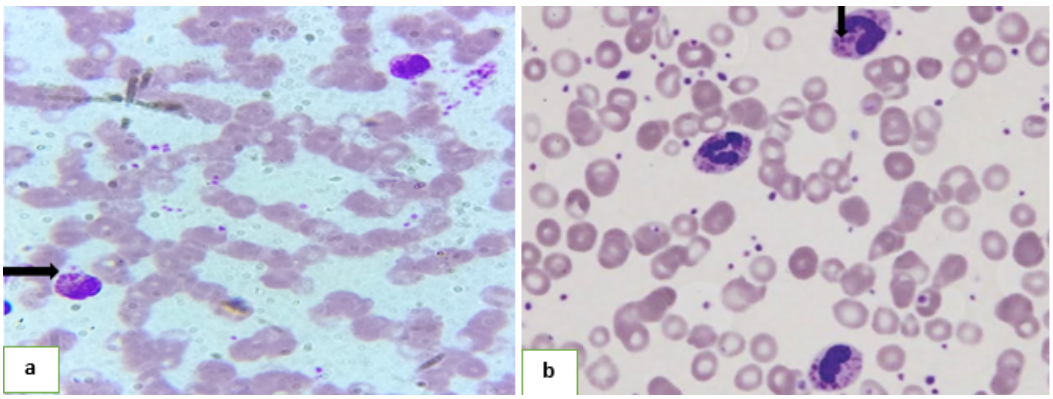

Fig. 3: a: 100x Leishman stained peripheral smear showing giant cytoplasmic granules in lymphocytes; **b:** 100x Leishman stained peripheral smear showing giant cytoplasmic granules in neutrophils**.**

silvery hair syndromes. But peripheral smear findings of giant granules in leucocytes helped us to achieve final diagnosis (Figure 3a,b). Routine blood investigations were within normal limits. Other investigations including genetic testing could not be done because patient was not affordable. Based on clinical presentation, skin manifestation and hematological findings a diagnosis of CHS was made.

3. Discussion

CHS and GS represent disorders of syndromic albinism with silvery grey hair in which the primary defect is in the transfer of melanosomes.[5] Clinically classical case of CHS present as recurrent pyogenic infections, mild bleeding diathesis, partial oculocutaneous albinism, hepatosplenomegaly and peripheral neuropathy.[4] GS also have same clinical manifestation. In both of these conditions increased susceptibility to infection especially skin and respiratory tract is due to the defective function of neutrophils i.e., poor mobilization from bone marrow, decreased deformability resulting in defective chemotaxis and delayed phagolysosome fusion resulting in impaired bactericidal activity.[6] Histopathological features are characterized by large, irregularly shaped melanin granules scattered in the upper dermis within melanophages. Hair shafts also demonstrate abnormal aggregates of melanin.[7] However CHS and GS cannot be differentiated solely on histopathology. To make correct diagnosis and to differentiate between CHS and GS, it requires peripheral blood smear evaluation. On peripheral blood smear giant cytoplasmic granules in leucocytes is a hallmark of CHS. This giant cytoplasmic granules in leucocytes are never seen in GS.[1]

Present case indicated provisional clinical diagnosis of both CHS and GS. Histopathology also shows considerable degree of overlapping. But giant coarse granules in leucocytes pointed us towards diagnosis of CHS.

It is recommended that parents and all the siblings of the patients of CHS should be screened for the presence of giant granules in the leucocytes for early diagnosis. Prenatal diagnosis can be done by examination of hair from the foetal scalp biopsy specimens and leucocyte from the foetal blood samples.[8]

4. Conclusion

Clinical and histopathological features of CHS and GS are overlapping but further peripheral smear findings helped us to cling to our diagnosis of CHS.

5. Source of Funding

None.

6. Conflicts of Interest

There is no conflict of interest.

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