Case Report

Hypomelanosis of Ito: Neurocuteneous Syndrome

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Abstract

A 7-year-old girl, 2nd issue of non-consanguineous parents hailing from Rajshahi got attended to our outpatient department of Pediatrics with the complaints of white patches all over the body since early infancy, poor intelligence, and speech problems. There was history of developmental delay but no history of seizures or visual problems or hearing impairment. She also had dental abnormalities. Family history was not significant. For these she treated with several oral and local antibacterial, antifungal and corticostresoid medications, but no improvement. Examination revealed symmetrical hypopigmented patches involving the trunk and upper limbs, and were following Blaschko's lines. Anthropometry and others examination were normal. The hearing assessment was normal. There were no ocular abnormalities but have delayed dentation with mal-alignment of teeth with carries. She have moderate level of intellectual disability (ID). Our diagnosis was hypomelanosis of Ito (HI); a rare type of neurocuteneous syndrome. Routine investigations were normal, CT scan of the brain showed mild cortical atrophy, EEG was normal. The parents were counseled about the recent level of intellectual functioning, asked to allow the child for learning self-help skills, and referred to a child development center, child psychologist, and speech therapist. Our patients were well and no complications were observed till writing this report. We like to share our experience of diagnosing and treating this very rare disease of HI.

Key words: *Hypomelanosis of Ito, Nerurocuteneous Syndrome.*

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Introduction

Hypomelanosis of Ito (HI) is a rare congenital skin disorder affecting children of both sexes that can have associated defects in several organ systems. HI is presented by unilateral or bilateral hypopigmented skin lesions with the multisystem disorder. Skin lesions may be in different textures, such as whorled, linear, or patchy, and are mostly accompanied by abnormalities of the central nervous system, skeletal system, teeth, and eyes. HI is associated with sporadic gene mutations but there is no evidence for genetic transmission, chromosomal mosaicism, and chromosomal translocation have been reported.[1]

Hypomelanosis of Ito (HI) is a rare neuroectodermal disorder mostly associated with intellectual disabilities (ID) and seizures. It is characterized by typical skin lesions. Various chromosomal anomalies have been found in some patients and the current consensus is that the phenotype of hyperpigmentation or hypopigmentation following Blaschko's lines occurs due to cutaneous mosaicism, either for a monogenic or a chromosomal disorder. The nervous system is the most commonly affected in the form of ID (70%), epilepsy (40%), microcephaly (25%), and hypotonia (15%). The musculoskeletal system is the next most frequently involved system, affected by thoracic, limb deformities, and scoliosis. Patients have minor ophthalmologic defects (strabismus, nystagmus) about 25%, and 10% have cardiac defects.[2] Hypomelanosis of Ito was described as a pure cutaneous disease. Extracutaneous manifestations were including skeletal, muscular, ocular, and central nervous system symptoms forming a neurocutaneous syndrome.[3]

Incontinentia pigmentosa achromiance was an old name of HI probably due to the negative image of incontinentia pigmentosa. Hypopigmented skin lesion is associated with developmental and neurological abnormalities. Mental retardation, pyramidal

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tract dysfunction, and seizures. Ophthalmologic disorders are also present.[4]

Case report

A 7-year-old girl, 2nd issue of non-consanguineous parents came to our outpatient department of Pediatrics of Khwaja Yunus Ali Medical College & Hospital, Sirajgonj, Bangladesh with the complaints of generalized hypopigmented patches all over the body noticed since early infancy, poor intelligence, and speech problems with the history of developmental delay. There was no history of seizures or visual problems or hearing impairment but have dental abnormalities. Family history was not significant. Examination revealed symmetrical hypopigmented patches arranged linearly, involving predominantly the trunk and upper limbs, and were following Blaschko's lines (figure 1, 2, 3, 4, 5). Another additional finding was the supernumerary nipple on the right side (figure 2). Anthropometry and others examination were normal. The child was diagnosed with a rare neurocutaneous syndrome named hypomelanosis of Ito. The hearing assessment was normal. There were no ocular abnormalities but have delayed dentation with mal-alignment of teeth with carries. The overall IQ of the patient was found to be 50, indicative of a moderate level of intellectual disability (ID). Routine investigations were normal, CT scan of the brain showed mild cortical atrophy (figure 6), EEG and color Doppler echocardiography was normal. The parents were counseled about the recent level of intellectual functioning, asked to allow the child for learning self-help skills, and referred to a child development center, child psychologist, and speech therapist. Our patients were well and no complications were observed till writing this report.

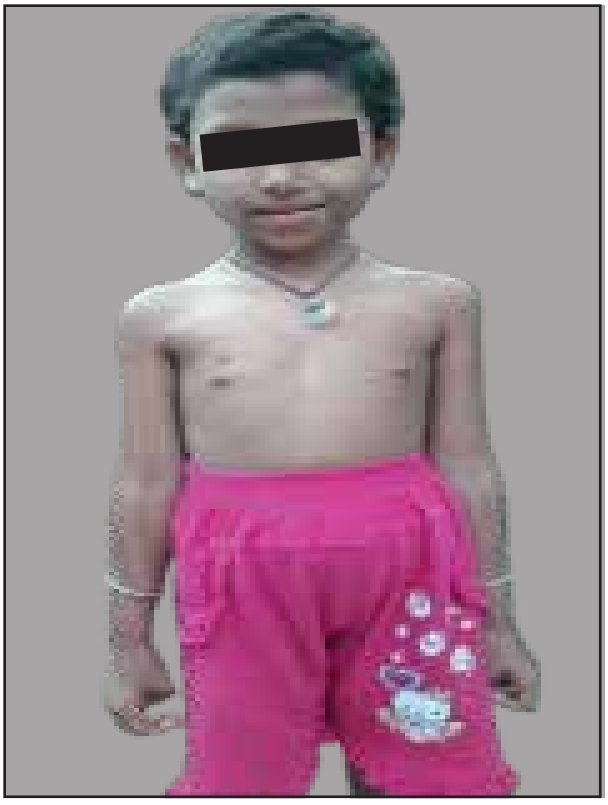


Fig: 1

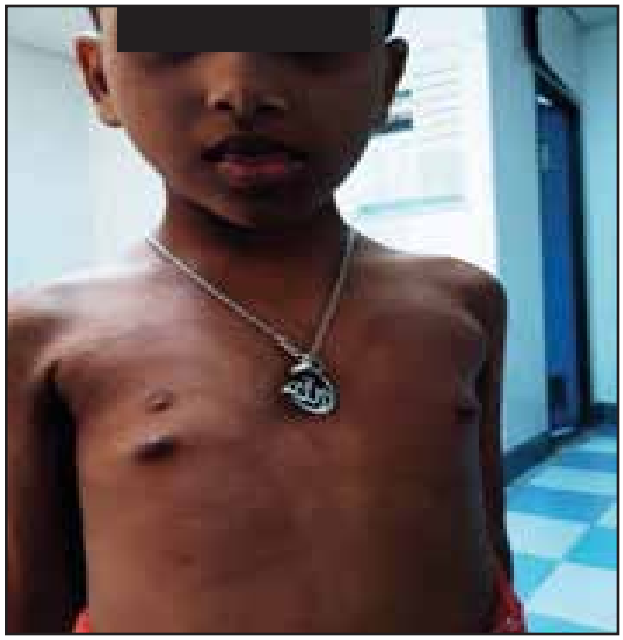
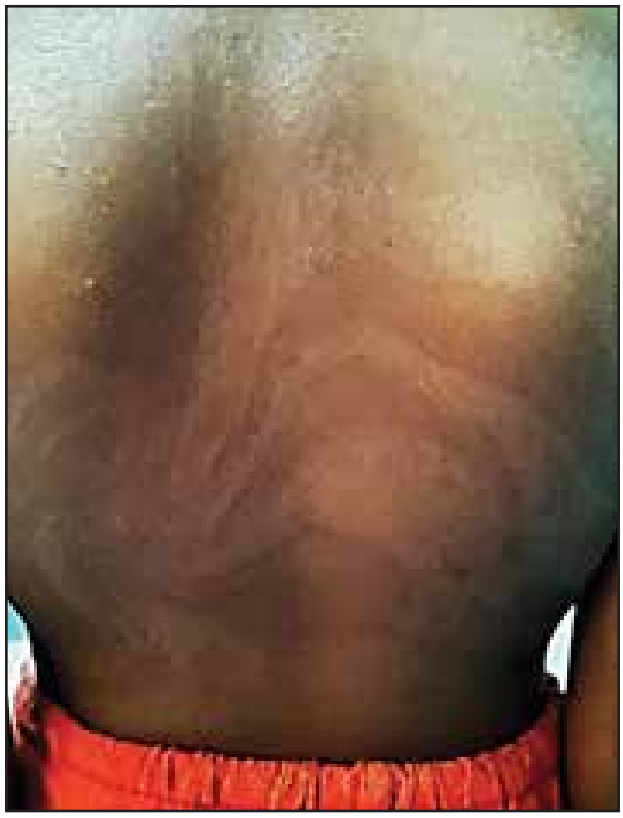


Fig: 2



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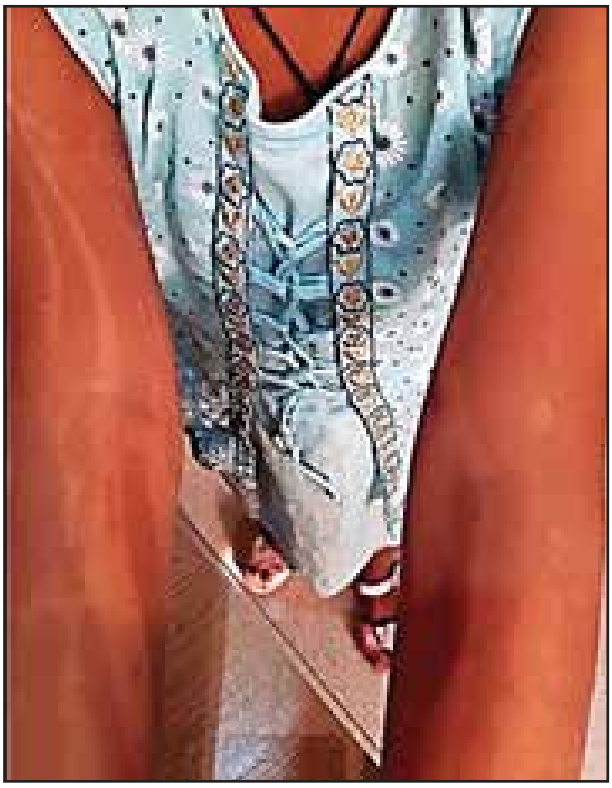

Fig: 4

Fig: 5

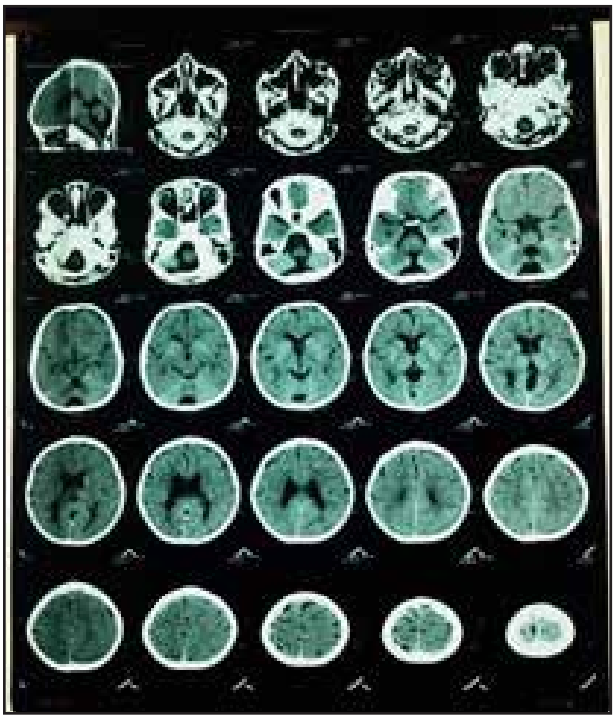

Fig: 6

Discussion

Our case was diagnosed by diagnostic criteria[5]; two major criteria were found: cutaneous hypopigmented linear streaks or patches involving more than two body segments, appearing at early infancy and neurological manifestations (intellectual & learning disabilities and speech problem) without chromosomal and histopathological studies. Similar findings were reported from a various studies done in different geographical locations. A study from India reported that a 12-year-old female child presented with a learning disability. She also had linear and whorled, hypopigmented lesions along the lines of Blaschko distributed over the upper limb, trunk, and face on the left side of the body. She fulfilled the diagnostic criteria for HI with an absence of chromosomal and histopathological studies.[6] Diagnosis of this neurocuteneous syndrome is based on the characteristic hypomelanosis and international criteria of Ito. No pathognomonic laboratory test is available.[4] Another case report from Istambul, turkey mentioned of a 5.5-year-old girl of non-consanguineous parents, with disproportionate overgrowth and lengthening of the right leg with marked hypopigmented lesions over the trunk and legs and delayed speech.[1] Author report a rare case of this disorder in a 9 year old child with complete unilateral skin lesions.[4]

Mild cortical atrophy on a CT scan of the brain was found in this case. Another case report showed diffuse white matter abnormalities and generalized brain atrophy on neuroimaging.[2] A case report of a 10-year-old male child; who presented with multiple congenital anomalies including facial dimorphism, hypopigmentation, musculoskeletal, and nervous system problems. The latter manifested was hypotonia, generalized seizures, and mild mental retardation. Brain MRI revealed normal findings initially, however; follow-up DWI-images were suggestive of an iron accumulation. The facial phenotype

with the bilateral globus pallidi lesions association with HI; possible a novel example of HI.[7]

1 year and 11 months old boy presented with recurrent pneumonia. Skin lesions were asymmetrical, hypochromic, bilateral maculae with irregular borders, distributed in stripes and the form of streaks, whorls, and patches following Blaschko's lines and affecting the trunk and limbs, but not the face, palm, sole, and mucous membranes. These lesions have been present since birth. Also observed that delayed development of teeth. Other siblings and there are no history of similar lesions in other family members. The neurological evaluation indicated a delay in neuropsychomotor development for age according to the 'Denver Developmental Screening Test'. EEG indicated diffuse cortical dysfunction. Brain MRI was compatible with an arachnoid cyst in the left antero-temporal region. An incisional biopsy was done from the lesion. The histologic findings following Fontana-Masson staining and the immunohistochemistry, using avid in biotin method, indicated respectively the absence of melanin and melanocyte in focal areas of epidermis.[8] There is no cure. Therapy is directed towards the associated complications.[4]

Conclusion

Hypo or hyperpigmentation of skin associated with neurological abnormalities could be a neurocutaneous syndrome. Hypomelanosis of Ito is one of the rare type of neurocutaneous syndrome. So, we like to dissemination of our findings of the case study to other physicians.

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