Hypomelanosis of Ito with an unusual pulmonary abnormality in an infant

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ABSTRACT

Hypomelanosis of Ito (HI) is a neurocutaneous syndrome characterized by hypopigmented cutaneous lesions and extracutaneous manifestations frequently affecting the nervous system and the musculoskeletal system. Dysmorphic features, dental, ophthalmic, gastrointestinal, cardiac, and renal abnormalities are described in a minority of patients. The authors describe a 4-month-old infant having HI with unusual pulmonary hypoplasia that has not been reported so far.

Key words: Computed tomography thorax, hypomelanosis of Ito, infant, pulmonary hypoplasia

Access this article online

Website: www.idoj.in

DOI: 10.4103/2229-5178.131109

Quick Response Code:

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INTRODUCTION

Hypomelanosis of Ito (HI) is a rare neurocutaneous syndrome characterized by hypopigmented skin lesions with a peculiar pattern of streaks, whorls, swirls, and patches.[1,2] The associated systemic abnormalities predominantly affect the nervous and musculoskeletal system and less commonly gastrointestinal, renal, and cardiac systems.[1-5] So far, as per our knowledge, no major pulmonary abnormality has been reported in association with this syndrome. We report a case of HI in an infant with segmental pulmonary hypoplasia for the first time.

CASE REPORT

A 4-month-old female infant was referred to the Department of Pediatrics with a history of persistent chest indrawing since birth. She was born at full term by cesarean section, cried immediately after birth, and had a birth weight of 2700 g. She was found to have respiratory distress with chest indrawing on the first day of life and required admission in the neonatal intensive care unit for 2 weeks and needed supplemental oxygen. She was reported to have persistent chest indrawing since then. She had two episodes of respiratory infection subsequently with worsening chest indrawing, which were treated with oral medications. There was no history of seizure in the past or family history of skin disorders. The child was referred to us at 4 months of age for the evaluation of unresolved chest indrawing. On examination, she weighed 4320 g with length of 54 cm and head circumference of 36 cm. Respiratory rate was 70/minute. Other vital signs were normal. Diffuse hypopigmentation, mainly streaks and few whorls on the skin were found on the left side of the chest, abdomen, and upper and lower extremities [Figure 1a and b]. Consultation with dermatologist confirmed these skin lesions as HI. Respiratory system examination revealed bilateral subcostal retractions with no added sounds on auscultation. Other systemic examination was normal. Chest radiograph was normal [Figure 2a]. Echocardiography revealed patent foramen ovale, mild pulmonary artery hypertension, and normal biventricular systolic functions. Gastroesophageal nuclear scan was negative for gastroesophageal reflux. Neurosonogram was normal. As no etiology could be identified for persistent chest indrawing, computed tomography (CT) scan thorax was ordered. CT thorax revealed hypoplasia of

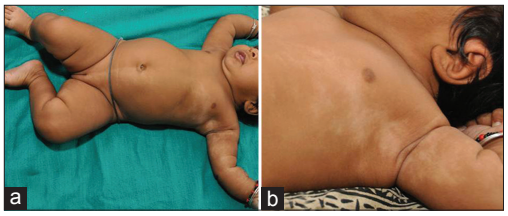

Figure 1: (a) Hypopigmented lesions involving left side of the chest, abdomen, and upper and lower extremities; (b) irregular margins of lesions on close-up view

the inferior lingular and posterior segment of the left upper lobe [Figure 2b and C]. The child was treated symptomatically for viral superinfection and discharged with an advice to have close follow-up. The cytogenetic studies could not be carried out due to financial constraints.

DISCUSSION

HI was first described in 1952.[1,2] Because, in 33%-94% of the cases, central nervous system (CNS) and musculoskeletal system were also involved apart from the skin, HI is called as a neurocutaneous syndrome although it was originally described as a purely cutaneous disease.[3] Cutaneous lesions in this rare syndrome include hypopigmented whorls, streaks, and patches along the lines of Blaschko. Ito's hypomelanosis combines structural and functional changes affecting both the melanocytes and keratinocytes in the skin.[1] Extracutaneous abnormalities involve the CNS, musculoskeletal (scoliosis, vertebral anomalies, craniofacial malformations), dental, cardiac, gastrointestinal, or renal system.[2-5] The most commonly associated abnormalities involve the nervous system, including mental retardation (70%), seizures (40%), microcephaly (25%), and muscular hypotonia (15%). The most significant manifestations of the CNS are psychomotor retardation and cognitive deficit. Autism, language disorders, and cerebral malformations have also been explained.

The pigmentary lesions in HI are either recognizable at birth or during early childhood. The lesions arranged in whorls, patches, or linear patterns along the lines of the Blaschko may be unilateral or bilateral in distribution.[2-4] Left side of the body was only affected in the present case. Pigmentary anomalies associated with mosaicism may show several patterns of distribution in the skin: (1) Type 1, typically following the lines of Blaschko. (2) Type 2, checkerboard pattern; (3) Type 3, phylloid pattern; (4) Type 4, patchy pattern without midline separation and (5) Type 5, spiral pattern. Accordingly, other types of cutaneous pigmentary patterns such as the zosteriform or dermatomal or plaque-like arrangement have likewise been observed in HI.[4]

Criteria for the diagnosis of this disease have been proposed by Ruiz-Maldonado *et al*.[4] (1) Congenital or early acquired nonhereditary cutaneous hypopigmentation in linear streaks or patches involving more than two body segments is the sine qua non. (2) One or more nervous system or musculokeletal anomalies form the major criteria. (3) Two or more congenital malformations other than nervous system or musculoskeletal system or chromosomal anomalies constitute the minor criteria. Criterion 1 and in addition one or more major criteria or two or more minor criteria gives a definitive diagnosis of the disease. Criterion 1 alone or in association with one minor criterion is considered a presumptive diagnosis. In the present case, criterion 1 and major pulmonary abnormality and minor cardiac anomalies established a definite diagnosis. The child needs to be monitored for CNS manifestations during the follow-up period.

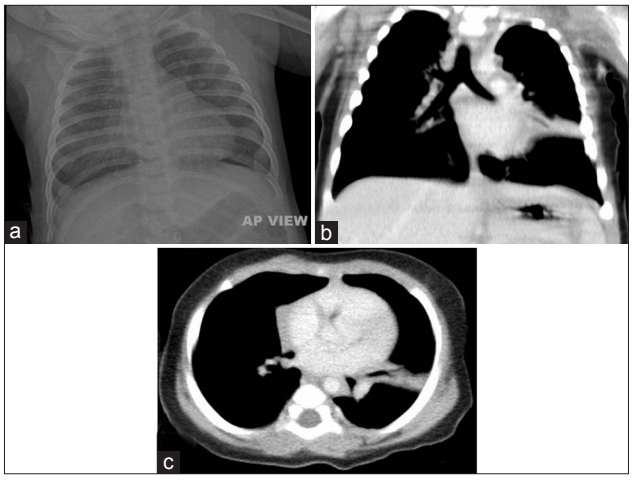

Figure 2: (a) Normal chest radiograph; (b and c) coronal and axial chest computed tomography scan showing hypoplasia of inferior lingular and posterior segment of left upper lobe

Association of congenital cardiac defect with HI is rare and includes atrial septal defect.[5] We described segmental pulmonary hypoplasia with HI that has not been reported in medical literature. Based on our case report, respiratory system involvement could also turn out to be one of the minor criteria for diagnosing HI. We also highlight the importance of thoracic imaging in identifying congenital lung abnormality as the chest radiograph did not reveal any clue in this regard.

In conclusion, in an infant with HI and persistent chest indrawing, especially from birth one needs to consider imaging of thorax to identify underlying congenital abnormality.

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Cite this article as: Bhat RY, Patra S, Varma PC, Prakashini K. Hypomelanosis of Ito with an unusual pulmonary abnormality in an infant. Indian Dermatol Online J 2014;5:196-7.

Source of Support: Nil, **Conflict of Interest:** None declared.