Hypomelanosis of Ito

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Introduction

Hypomelanosis of Ito (HOI) is a neurocutaneous disorder characterized by bizarre, bilateral and irregularly shaped leukoderma affecting the trunk and extremities, often associated with neurological and musculoskeletal abnormalities [1]. Ito first described this condition in 1951 [2]. In 1967, Hamada et al confirmed the association between skin lesions and systemic abnormalities, including mental retardation. Ruiz-Maldonado et al proposed diagnostic criteria in 1992 [3]. We report two patients with this disorder.

Case Reports

Case 1

A four-and-a-half month old male infant presented with gradually increasing size of the head and depigmented areas over the trunk and limbs since birth. Born at 30 weeks gestation there was no contributory antenatal or neonatal history. There was no history of intraventricular haemorrhage or meningitis in the neonatal period. An elder sibling aged 5 years was normal.

Dermatological examination revealed hypopigmented macules in a linear, whorled pattern distributed bilaterally over trunk and extremities arranged along the lines of Blaschko (Fig. 1). A solitary neurofibroma over the right elbow was noted. The hair, nails, palms, soles and mucosae were normal.

Fig. 1 : Hypopigmented macules in linear whorled pattern

Musculoskeletal examination revealed macrocephaly with occipito-frontal circumference of 45 cm at 4½ months of age. The infant had ocular hypertelorism and hypotonia of limbs with a small umbilical hernia. No deformity of spine was evident. Slit lamp and fundus examination were normal. Neurological examination revealed developmental delay with head lag and persistent grasp and Moro's reflexes.

Ultrasonography of the abdomen and 2D-echocardiography were normal. Radiological examination of the spine and neurosonogram were normal. MRI brain revealed delayed myelination with abnormal operculum on the right side and increase in brain size. The child is under monthly follow up.

Case 2

A 7-year-old boy presented with complaints of hypopigmented lesions over the left upper limb and left side of the trunk since first year of life.

Dermatological examination revealed hypopigmented macules in a linear, whorled pattern distributed unilaterally over the left upper arm and left side of trunk arranged along the lines of Blaschko. There was no associated neurological or musculoskeletal abnormality. Examination of the eye by slit lamp and fundoscopy revealed no abnormality. Radiological examination of the spine and echocardiography were normal.

Discussion

HOI is the third most common neurocutaneous disorder, after neurofibromatosis and tuberous sclerosis [4,5]. It is diagnosed in 1 per 8000-10000 unselected patients in general paediatric outpatient clinic and 1 out of every 790 in a paediatric dermatology clinic [6,7].

Some believe HOI is related to autosomal dominant inheritance [8] and others attribute it to chromosome instability and mosaicism [3]. Chromosomal abnormalities, particularly translocation and mosaicism have been reported in approximately 50% of cases [5]. It is suggested that HOI is not a single condition but a

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phenotype of chromosomal mosaicism [4].

Clinically, lesions appear at birth or infancy as asymmetric, whorled or streaked lesions in a “marble cake pattern” along the lines of Blaschko (lines of migration of embryonic skin cells, invisible under normal conditions). Multiple extracutaneous abnormalities involving the central nervous system, eyes and musculoskeletal structures occur in 76-94% of patients [3,4,9]. Convulsions and mental deficiency are common. Other associated disorders include macrocephaly, microcephaly, hemi-hypertrophy, kyphoscoliosis, coarse facial features, genital anomalies, inguinal hernia, congenital heart disease, hypertelorism and abnormalities of the teeth and eyes [10]. The severity of manifestations appears similar in both sexes. 1st patient had skin and central nervous system involvement, while the 2nd had isolated skin involvement.

Differential diagnosis includes pigmentary anomalies along the lines of Blaschko such as linear and whorled nevoid hypermelanosis, naevus depigmentosus and incontinentia pigmenti. Linear and whorled nevoid hypermelanosis manifests as hyperpigmented macules in streaky configuration along Blaschko’s lines gradually after birth and may be associated with congenital anomalies. Naevus depigmentosus is characterized by circumscribed achromic macules at birth, with no changes thereafter. Systemic abnormalities are rare in nevus depigmentosus. Incontinentia pigmenti is an X-linked dominant disorder, lethal in males, characterized by three stages – vesiculobullous, verrucous and hyperpigmented. In a minority (13%) of cases there is a fourth stage with swirls of hypopigmentation which may be confused with HOI.

Skin lesions with altered pigmentation are an important marker of underlying neuropediatric disease.

The aim of reporting these two cases is to highlight the systemic defects associated with HOI which affect three-fourths of patients. An active search for neurological defects is therefore mandatory in all cases of HOI.

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