

# Hypomelanosis of Ito: Report of two cases

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## ABSTRACT

Hypomelanosis of Ito is a neurocutaneous disorder characterized by hypopigmented whorls, streaks and patches distributed along the lines of Blaschko, often associated with neurological and musculoskeletal abnormalities. We herein report two patients belonging to ethnic Kashmiri origin with this disorder.

**Key words:** Hypomelanosis of Ito; Neurocutaneous; Lines of blaschko

## INTRODUCTION

Ito first described this condition in 1952. Hypomelanosis of Ito is the third most neurocutaneous disorder, after neurofibromatosis and tuberous sclerosis [1]. The characteristic features are the presence of hypopigmented skin lesions arranged in whorls and streaks following the lines of Blaschko. Some believe that Hypomelanosis of Ito is related to autosomal dominant inheritance, others attribute it to chromosomal instability and mosaicism [2]. Chromosomal abnormalities particularly translocation and mosaicism have been reported in approximately 50% cases [3].

## CASE REPORTS

### Case 1

A 7-month old infant was presented by his parents with hypopigmented lesions over his right upper limb which had been there since birth. These lesions became prominent over time and were not preceded by blistering or any verrucous hyperpigmentation. There was no history of seizure disorder or weakness of limbs. The child was a product of non-consanguineous marriage, born as a full term with normal delivery. There was no history of mental retardation or significant learning disability in any other family member.

General physical examination including height, weight and heads circumference were normal. Systemic examination was also non-contributory. Cutaneous examination revealed bizarre hypopigmented linear macules over right arm following the lines of Blaschko (Figure 1). Hair, nails, palms & soles and mucosae were normal. Neurological examination was also normal. There was no musculoskeletal or dental abnormality. Radiological examination of chest, long bones, skull and spine were normal. Ultrasonography of abdomen, 2 D- echocardiography as well as MRI brain revealed no abnormality. IQ testing was not possible because of his age. The child is presently under monthly follow-up.

### Case 2

An 8-year old boy presented with hypopigmented lesions over left side of his trunk from second year of his life. These lesions were progressively increasing in size and degree of hypopigmentation. These lesions were asymptomatic and were not preceded by any vesiculobullous, hyperkeratotic or hyperpigmented lesions. There was no history of seizure disorder, neurological deficit or any behavioral disturbance in the patient. The child was a product of non-consanguineous marriage, born at term, second in birth order. There was no history of mental retardation or any significant learning disability in any of the family members. All the developmental milestones

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were achieved normally with no developmental delay. General physical examination revealed normal height, weight and head circumference as per his age. Systemic examination was also normal. Dermatological examination revealed whorled hypopigmented macular areas over left side of trunk along the lines of Blaschko (Figure 2). Examination of hair, nails, palms & soles and mucosae was normal. Neurological examination was normal. He had acquired a satisfactory comprehensive and expressive language, with no problems in articulation. The boy evidenced a good level of general awareness, with good vocabulary, calculations and reasoning as per his age. Ophthalmological examination including slit lamp examination and fundoscopy was normal. Musculoskeletal and dental examinations were also normal. Radiological examination of spine, ultrasonography of abdomen and echocardiography were all normal. MRI brain as well as ECG revealed



**Figure 1:** Bizarre hypopigmented linear macules over right arm following the lines of Blaschko.



**Figure 2:** Whorled hypopigmented macular areas over left side of trunk along the lines of Blaschko.

no abnormality. IQ testing carried out was also normal. Skin biopsy was not performed as the parents were unwilling for the same. Chromosomal study was not possible in our institute. The child was advised a monthly follow-up.

## DISCUSSION

In 1952, Minor Ito described a Japanese girl with skin of the upper part of the body looking as “if the normal pigment was brushed off”. The depigmented skin lesions were widespread and symmetric, arranged in irregular shapes with “zig zag borders and splash-like spots” on the trunk and in a “linear pattern” down her arms. He defined these lesions as “nevus depigmentosus systematicus bilateralis” [4]. At that time, Ito coined the term “Incontinentia pigmenti acromians” because the pattern of color loss was similar to that of the hyperpigmented changes seen in incontinentia pigmenti of the Bloch-Sulzberger type. Proposed changes in terminology included the terms pigmentary dysplasia, pigmentary mosaicism, pigmentary mosaicism of the Ito type, or hypopigmentation along the lines of Blaschko to reflect the disease pathogenesis or recall the cutaneous patterns [5-7]. Despite these criticisms the term ‘hypomelanosis of Ito’ is still used.

Hypomelanosis of Ito is a multisystem disorder in which most organs of the body show anomalies in addition to the skin. The main features that define hypomelanosis of ito are the cutaneous anomalies. In many instances, patients may present with skin hypopigmentation following the lines of Blaschko, without any other associated anomaly. The pigmentary lesions are either recognizable at birth or become visible during early childhood. Though most of the cases have been reported from Japan, hypomelanosis of ito has been reported from several other countries [8]. Most reported patients are less than 10 years old and sex ratio favored females by 2.5:1. The characteristic clinical features include unilateral or bilateral areas of hypopigmentation with irregular borders arranged in whorls, or linear patterns along the lines of Blaschko. The hypopigmented zones in hypomelanosis of ito can be seen in any part of the body: head, neck, face, trunk or extremities [9,10]. Other cutaneous lesions associated with hypomelanosis of Ito include café-au-lait spots, nevus marmorata, angiomas, nevi, nevus of Ota, and Mongolian blue spot.

Nervous system alterations are the most frequently associated extracutaneous anomalies. Anomalies of CNS

may include microcephaly or macrocephaly, cognitive and motor retardation, seizures, ataxia, hyperkinesias and hypotonia [11]. Musculoskeletal disturbances are usually observed in more severe phenotypes. Skeletal defects include short stature, asymmetry with hemihypertrophy or deformities (pectus carinatum or excavatum) and toe anomalies (clinodactyly, polydactyly, syndactyly, brachydactyly) [12]. Ocular alterations are rarely reported and include strabismus, nystagmus, hypertelorism, ptosis, myopia, amblyopic cataracts, corneal opacity, micro-ophthalmia, macro-ophthalmia, optic nerve hypoplasia and retinal degeneration [13]. Oral manifestation consist of defective dental implantation, partial anodontia, dental hypoplasia or dysplasia, conical teeth and defective enamel. Other systemic anomalies which may be associated include renal disease such as single kidney or ureteral duplication and genitourinary anomalies including cryptorchidism and micropenis [14]. There are a limited number of cases associated with tumors, including cystic teratoma, choroid plexus papilloma, complex mature sacrococcygeal dysembryoma, and dental hamartomas. Rarely, malignancies such as ALL, medulloblastoma and neuroblastoma have been reported.

### Consent

The examination of the patient was conducted according to the Declaration of Helsinki principles.

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