CASE REPORT

Hypomelanosis of Ito with gynaecomastia and dental anomaly

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SUMMARY

Hypomelanosis of Ito is a rare neurocutaneous syndrome. Cutaneous involvement is characterised by streaks and swirls of hypopigmentation arranged in a Blaschkoid pattern. Neural involvement along with other systemic features are seen. We report a case of a 13-year-old boy who presented with the characteristic skin involvement of hypomelanosis of Ito, mental retardation, teeth abnormalities and gynaecomastia along with psoriasis.

BACKGROUND

Hypomelanosis of Ito (HI) was first described as incontinentia pigmenti achromians in 1952.¹ Its prevalence ranges between 1 in 3000 and 1 in 10 000 and is twice more common in girls than boys.¹ It is associated with multiple extracutaneous features in 33%–94% cases, mostly of the central nervous and musculoskeletal systems.² Though there may be multisystemic involvement in HI, associated gynaecomastia has been rarely reported.³

CASE PRESENTATION

A 13-year-old Indian boy presented with red raised mildly itchy scaly lesions over the body for last 1 month. There was presence of unusual asymptomatic hypopigmented streaks on the trunk and upper limbs since birth. The child was born to non-consanguineous parents by normal vaginal delivery with no history of birth asphyxia with a birth weight of 3 kg. On examination, the child was obese with a body mass index of 30.2, weight 60 kg (87

percentile), height of 165 cm (85.4 percentile). On neuropsychiatric evaluation, the child had delayed global milestones, deficit in intellectual and adaptive functioning. There were bizarre, well-defined, bilaterally symmetrical streaks and whorls of hypopigmentation all over the trunk extending over to upper limbs (figures 1 and 2). The patient also had associated bilateral gynaecomastia (figure 2) with no history of any previous drug intake. Testicular size was <2.5 cm, and there was no development of terminal pubic hair, that is, his pubertal development corresponded to Tanner stage G2 and P1. There were associated dental anomalies in the form of extra teeth (figure 3). On the trunk and upper limbs, discrete scaly plaques suggestive of psoriasis were present (figure 1). Hair, nail, palms, soles and mucosa were not involved. There were no musculoskeletal, ocular, cardiac or other neurological involvement. No other pigmentary disorder or cutaneous lesion anywhere on the body was seen. There was no significant history of similar lesion in the family.

On the basis of the characteristic skin findings and associated systemic features, a diagnosis of HI with mental retardation, gynaecomastia and psoriasis vulgaris was kept. The patient was given topical steroids plus salicylic acid preparations to be applied on the psoriatic plaque lesions and was counselled about the course and prognosis of the disease.

INVESTIGATIONS

Routine haematological and biochemical tests were within normal limits. Thyroid function test and fasting blood sugar were normal. For management of gynaecomastia, the child was advised hormonal assay. Follicular stimulating hormone was 11.30 mIU/mL (normal range 0.1–2.3), luteinising hormone 2.1 mIU/mL (normal 0–1.4), serum testosterone 1.45 ng/mL (normal range 2.67–10.12), oestradiol 24 pg/mL (normal range of 5.0–16.0 pg/mL) and serum prolactin 9.20 (normal 1.80–15.90). These features suggested hypogonadism. To rule out central nervous system anomaly, CT head was advised but his parents refused. Facilities for karyotyping are not available at our institute.

DIFFERENTIAL DIAGNOSIS

Cutaneous lesions of HI mimic incontinentia pigmenti (fourth stage) and linear and whorled nevoid hypermelanosis. Incontinentia pigmenti (fourth stage) is preceded by vesiculobullous,

Figure 1 Well-defined, bilateral streaks of hypomelanosis along the Blaschko's lines on the trunk with psoriatic plaques.

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Figure 2 Well-defined, bilateral streaks of hypomelanosis along the Blaschko's lines on the front of the trunk extending to the arms with gynaecomastia.

verrucous and hyperpigmented swirls and is lethal in males. HI though more common in females is seen in males also and change in morphology of lesion is not seen. Linear and whorled nevoid hypermelanosis manifests as hyper pigmented macules in streaky configuration along the Blaschko's lines and may be associated with congenital anomalies but not associated with systemic abnormalities and is gradually progressive. HI is usually associated with systemic abnormality and is non-progressive.

TREATMENT

There is no satisfactory treatment for HI at present. The patient needs to be counselled about the prognosis. Sometimes, patients are not bothered about the pigmentation but a thorough examination to rule out other associated conditions should always be performed in a patient with HI. The patient was referred to endocrinologist for hypogonadism but the parents were only bothered about psoriatic lesions for which he was prescribed topical steroids with salicylic acid.

Figure 3 Extra teeth with spacing abnormality.

OUTCOME AND FOLLOW-UP

The patient did not turn up for follow-up.

DISCUSSION

In 1952, Minor Ito described HI in a Japanese girl as a pure cutaneous entity. The systemic associations of HMI were described later.¹ It is known by various terms like incontinentia pigmenti achromians, pigmentary dysplasia and pigmentary mosaicism. It is usually sporadic but occasional reports of an autosomal dominant and recessive inheritance have been seen. Our patient also had no family history of similar disease. HI is characterised by the presence of widespread symmetrical depigmented or hypopigmented whorls or streaks arranged in a Blaschkoid pattern. Lesions may be present since birth or appear within the first year of life. Other patterns like checkerboard, dermatomal, phylloid, spiral and plaque-like patterns have also been reported.⁴ The extent of the disease is variable with unilateral or bilateral symmetrical patterns of involvement following Blaschko's lines which may be segmental or involve the whole body. Other cutaneous lesions associated with HI include café-au-lait spots, nevus marmorata, angiomatous nevi, nevus of Ota and Mongolian spot. Various systemic manifestations include neurological, musculoskeletal, ocular, dental, cardiac, renal, genitourinary, breast and so on.^{5 6}

CNS involvement is the most common and can manifest as mental retardation, microcephaly, macrocephaly, seizures, autism, psychomotor impairment, learning difficulties, dyskinesias or Sturge-Weber syndrome like leptomeningeal angiomatosis.^{2 5} Our patient had delayed milestones and difficulty in learning suggestive of mental retardation.

Various anomalies of teeth have been found in association with HI such as hypodontia, enamel defects and caries.^{6 7} There was irregular arrangement of teeth and doubling of mandibular row of incisor, spacing defects and altered dental cusp and canines. Extra teeth and spacing abnormality were seen in our patient.

Asymmetric development of breast and gynaecomastia have been rarely reported. Most of the cases in literature have been described in early childhood which could be the reason why this association has been rarely mentioned.

The association of various systemic manifestations in addition to cutaneous features could be due to multiple genetic defects. It is thought that HI has phenotypic variability and genetic difference.⁸ Various chromosomal alterations like translocation aneuploidy, mosaicism, trisomy and deletion may be seen. Though karyotyping could not be done in our patient, the association of obesity, increased height, gynaecomastia and features of hypogonadism suggest a chromosomal abnormality. Ruiz-Maldonado *et al* proposed diagnostic criteria of HI in 1992.⁹ Hypopigmented streaks are the essential criteria. Our patient had the essential criteria along with one major and two minor criteria so was labelled as a case of definite HI.

Histopathologically in HI the number of melanocytes may be normal or reduced or there may be a reduction in the intracellular content of melanin, and there are no melanophages. In incontinentia pigmenti, there is pigment incontinence and melanophages are seen in the dermis.

Association of psoriatic plaques to the lesions of HI was described by Pavithran which was attributed to the Koebner phenomenon of psoriasis.¹⁰ Though in our case the distribution of the lesions was discrete, not particularly related to the lesions of HI and few in number, it could have been a chance occurrence rather than related to the primary disorder of HI.

Learning points

- ▶ Hypomelanosis of Ito (HI) is a neurocutaneous syndrome.
- ▶ Other pigmentary disorders along the Blaschkoid pattern should be ruled out.
- ▶ HI has a multisystem involvement but gynaecomastia is a rare association.
- ▶ HI is a phenotypic variable disease with genetic differences.

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