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HYPOMELANOSIS OF ITO: SPECTRUM OF THE NEUROCUTANEOUS SYNDROME

Dermatology

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ABSTRACT

Hypomelanosis of Ito, also known as Incontinentia Pigmenti Achromians (IPA), is a rare neurocutaneous syndrome clinically characterized by hypopigmented linear,whorls, patches and streaks following Blaschkoid lines. It was originally described as a purely cutaneous disease, but later studies reported its association with multiple extracutaneous features, mostly of the central nervous and musculoskeletal systems. This leads to spectrum of Hypomelanosis of Ito as a neurocutaneous disorder. We report a nine-months-old female baby who presented with constellation of cutaneous and neurological disorders.

KEYWORDS

Hypomelanosis of Ito, Blaschkoid lines, Neurocutaneous syndrome

INTRODUCTION:

The clinical picture of Hypomelanosis of Ito was first described in 1952 by a Japanese doctor named Ito. He described it as 'incontinentia pigmenti achromians' and in 1973 Jelinek was able to differentiate this from that of Block-Sulzberger syndrome .[1] Hypomelanosis of Ito is a rare genetic disorder, considered to be a neurocutaneous syndrome and characterized by streaky, patchy, whorl-like, or linear hypopigmented macules occurring on any part of the body.[2] Lesions first appear as small 0.5-1 cm hypopigmented or depigmented macules that merge to form larger patches. Macules cover more than two dermatomes and are often on both sides of the body. Patches are not symmetrical. Recent studies showed associated abnormalities occur in 30-50% of patients with cutaneous lesions.[3] Thorough history-taking and physical examination with attention to neurological and ophthalmological findings are necessary to detect associated abnormalities.

CASE REPORT:

A nine-months-old female baby born of non-consanguineous marriage presented with light-coloured lesions over trunk since birth. There was no family history or similar lesions in the other siblings. The lesions were increasing in size with the progression of age.There was history of multiple episodes of generalised seizures at the age of six months. General physical examination of the patient revealed no other abnormality. On cutaneous examination, multiple bizarre, asymmetric hypopigmented macules are present in whorls & patches over trunk bilaterally along Blaschkoid lines.[Figure 1,2,3] Palms and soles are not affected. Examination of hair and teeth revealed no abnormalities. On auscultation there was systolic murmur in the mitral and pulmonary area but had no cardiac anomalies on cardiological examination. On thorough examination for any associated abnormalities, we found no neurological, musculoskeletal, ophthalmological, and any other rare associations.. The child underwent magnetic resonance imaging of brain, which did not reveal any abnormality Other routine investigations were within normal limits.. A 3mm skin biopsy was taken from the hypopigmented macule and histopathological examination showed decreased content of pigment granules in the basal cell layers of the epidermis. No other abnormality was seen histologically and dopa reaction was not done.

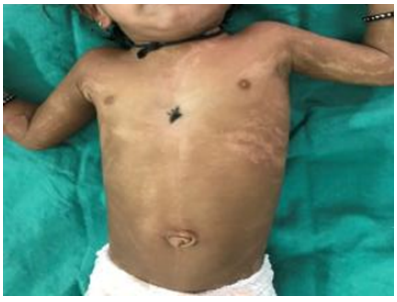

Figure 1: Hypopigmented macules arranged in blaschkoid pattern

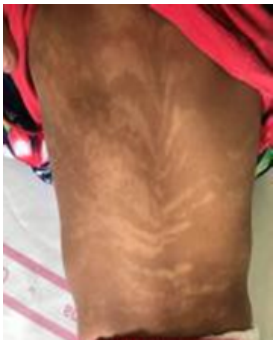

Figure 2: Hypopigmented macules over back

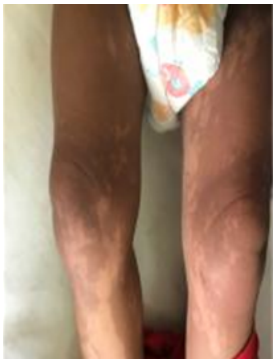

Figure 3: Hypopigmented macules over legs

DISCUSSION:

Hypomelanosis of Ito(HI) is rare neurocutaneous syndrome characterized by macular hypopigmented whorls, patches, and streaks either U/L OR B/L following Blaschkoid lines.[4] Pattern of hypopigmentation in HI is the result of the migration of two clones of primordial melanocytes, each with a different pigment potential.[5] Most patients have multisystem involvement and may show chromosomal mosaicism, the most common being neurological, muscular, skeletal, and ocular.[6] There is a higher frequency (94-100%) of central nervous system abnormalities reported in association with hypomelanosis of Ito. The commonest of these were developmental delay, mental retardation of various degrees ranging between mild to severe, poor school performance and autistic- like behavior especially in those who suffered from either infantile spasm or drug resistant seizures. Developmental delay is reported in 75% of affected patients and moderate to severe cognitive deficits (IQ <70) in 57%; with only 20% of patients having an IQ above 85% [7] The association between seizures and mental retardation is frequent and reported in 60-70% of cases. In general, seizures occurred in 50%, commonly appearing early

in the first year of life with the most frequent seizures type including generalized tonic or tonic-clonic, complex partial, myoclonic seizures and infantile spasm .While some patients suffer from generalized seizures that are well controlled with anticonvulsant therapy, many have severe, pharmaco-resistant focal seizures, and of those some patients may benefit from surgery [8,9,10] our patient has similar history of multiple episodes of generalised toni-clonic type of seizures at the age of six months but without any neurological abnormality in MRI of brain. Since our patient was still nine months old there may be chance of Developmental delay,mental retardation and autistic behaviour in future age for which treating physician should be vigilant enough to detect as early as possible to avoid these complications.

CONCLUSION:

Any condition with skin manifestations along Blaschko's lines may be confused with hypomelanosis of Ito. Incontinentia pigmenti is marked by preceding vesicular or verrucous phases. Goltz focal dermal hypoplasia along Blaschko's lines may cause confusion, but focal absence of dermis, multiple papillomas of mucous membranes, and linear hypo- and hyperpigmentation suggest the existence of pigmentary mosaicism No definite treatment for hypomelanosis of Ito present. Meticulous proper systemic examination should be carried out to detect additional abnormalities. Seizures should be managed with proper anti-epileptics. Orthopedic and physical therapy should be instituted as and when required. Even in our case, the child was thoroughly investigated with classical pigmentary cutaneous lesions of HI for any systemic association. The prognosis was explained to the parents as there were no other abnormalities and was advised for regular follow-up every six months.

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