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Case Study

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HYPOMELANOSIS OF ITO: A CASE PRESENTATION

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ABSTRACT
Hypomelanosis of Ito is a rare condition characterized by distinctive hypopigmented whorls and streaks distributed along the lines of Blaschko and is often associated with neurological and musculoskeletal abnormalities. We hereby report a 2-year-old boy who presented with extensive hypopigmented whorls and patches along the lines of Blaschko present on both sides of the trunk since birth. The child had no other systemic abnormality. The early diagnosis and timely work up to rule out any systemic involvement is important for timely management and better quality of life.

INTRODUCTION
Hypomelanosis of Ito is characterized by hypopigmented whorls and streaks following the lines of Blaschko.[1] Also known as Incontinentia Pigmenti Achromians (IPA), is a rare neurocutaneous syndrome clinically characterized by macular hypopigmented worls, patches and streaks resembling fountain spray splatters.[2] These skin lesions become apparent at birth or during childhood and normally are present on the trunk, occasionally on the extremities and rarely on the face.[3] Most patients have multisystem involvement and may show chromosomal mosaicism, the most common being neurological, muscular, skeletal and ocular.[2]

The early diagnosis and timely work up to rule out any systemic involvement is important for timely management and better quality of life.

CASE REPORT
A 2 year old boy from Kerala presented with asymptomatic whitish patches over the body since birth. On cutaneous examination he had multiple, asymmetric, hypopigmented macules distributed bilaterally along the Lines of Blaschko. over the trunk, upperlimbs and legs. General physical exmination revealed no abnormality seen. The investigations Ultrasound abdomen revealed featur. Systolic murmur was present in the mitral and pulmonary areas. His complete haemogram was normal except for eosinophilia (70%) and nonmocytic hypochromic to microcytic hypochromic anemia with mild degree of anisopoikilocytosis.

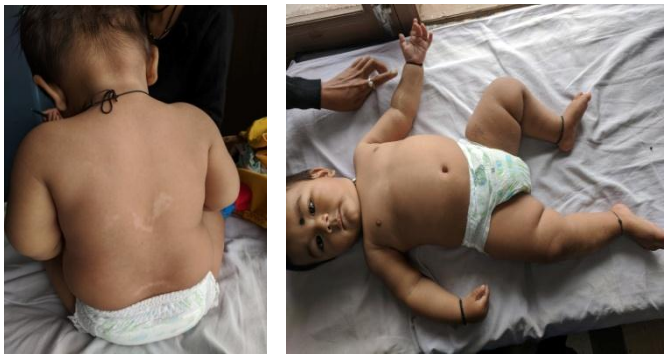

Figure 1&2: Whorls and streaks of hypopigmentation over the trunk and extremities.

DISCUSSION
Hypomelanosis of Ito is a neurocutaneous disorder characterized by a bizarre, bilateral, irregularly shaped leukoderma afecting the trunk and extremities and often associated with neurologic and musculoskeletal abnormalities.[4] Though originally described as a purely cutaneous disease subsequent reports have included 33% to 94% association with multiple extracutaneous

manifestations mostly of the central nervous and musculoskeletal systems leading to frequent characterization as a neurocutaneous disorder.[5] The hypomelanotic macules of Hypomelanosis of Ito are usually present at birth but may appear in early infancy or childhood.[4] Clinically the lesions appear as a negative image of incontinentia pigmenti, distributed with a libateral, asymmetric, whirled or streaked "marble cake" pattern often in parallel arrway and along Blaschko" lines.[4] The most severe complications concerned with the central nervous system are mental retardiation and epilepsy and both of these are present in the more than 50% of cases.

Moreover other complications can be observed in some patients which consists of ocular, musculoskeletal and oral alterations, hypotonia, macrocephalia, microcephalia, congenital cardiac malformations, urological and genital malformations and other rarer disorders.[6] Of 115 reported cases, abnormal chromosomal patterns have been found in 60.[7] The most common alterations include mosaic trisomy 18, diploidy/triploidy, mosacism for sex chromosome aneuploidey and tetrasomy 12 p.8 According to earlier studies on this pathological condition a marked reduction of melanocytes is detected in the hypopigmented areas.[8]

In the present case report, the child was thoroughly investigated with classical pigmentary cutaneous lesions of HI for any systemic association. No systemic abnormality was found.

There is no definite treatment for pigmentary disorders. Meticulous systemic examination should be carried out to detect additional abnormalities.. Even in our case, the child was thoroughly investigated with classical pigmentary cutaneous lesions of HI for any systemic involvement.

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