

## SPOT DIAGNOSIS (IMAGE GALLERY)



### NEUROCUTANEOUS SYNDROME

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A two year old boy born of a third degree consanguineous marriage, came with complaints of developmental delay and abnormal skin lesions, noticed since birth. There was no similar family history. Antenatal history was uneventful. On examination, he had hypertelorism, high arched palate, delayed dentition, pectus carinatum, genu valgum with

knock knees, hypotonia and peculiar skin lesions all over the body, characterized by multiple asymmetrical hypopigmented whorls, streaks and reticular patches on trunk, upper and lower limbs.

### What is the diagnosis?

Hypomelanosis of Ito. It is a rare neurocutaneous syndrome with hypopigmented whorls of skin along the Blaschko lines (1), first described by Ito in 1951 (2) with a female: male ratio of 2:1 and no racial predilection. It was earlier also known as incontinent pigmenti achromians, because it appears as the negative image of incontinent pigmenti. The varied phenotype is probably due to genetic mosaicism. (3) Sporadic mutations also contribute to it. It is present at birth, or soon after and is classically described as hypochromic lesions in distinctive patterns (e.g. whorls, patches, streaks). The lesions may be unilateral (46 percent) or bilateral and usually show a midline cutoff mostly on the ventral surface of the trunk and flexor surface of the limbs and are easily demonstrable by wood lamp examination. The skin lesions may later become pigmented and blend with the rest of the skin. It is associated with various other congenital abnormalities with mental retardation and seizures being the most commonly reported in literature. (4) Other abnormalities include hypertelorism, visual disturbances, cleft palate and lip, anodontia, renal anomalies, various musculoskeletal malformations like scoliosis, limb hypertrophy and syndactyly. Neuro imaging should be done to rule out associated structural abnormalities. There is no specific treatment and comprises of management of co-morbidities by a multidisciplinary approach. Traditional depigmenting agents (e.g. hydroquinone, corticosteroids, kojic acid) may be used. (5)

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