

SPOT DIAGNOSIS (IMAGE GALLERY)



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A 14 year old negroid girl with non-consanguineously married parents presented with a history of remote symptomatic convulsive seizures, mental subnormality, cognitive impairment since early childhood, and yellowish to brownish irregular skin lesions since infancy. She also has macrocephaly with prominent eyeballs, lat nasal bridge and an epicanthus inversus. Her skin lesions are depicted in the image.

What is the diagnosis?

Incontinentia Pigmenti Type 2 also known as Bloch-Sulzberger syndrome. This is a rare disorder occurring in 1.40,000 births. This child has a whorly, spiral eloquent yellowish, brownish skin lesions involving the face, the trunk, and the limbs along the lines of Blaschkos. Incontinentia pigmenti is an uncommon x-linked dominant congenital multisystemic neurocutaneous syndrome affecting the skin, the central nervous system, the bones and the eyes. Most affected children are girls; it is usually associated with in utero lethality in males. Incontinentia pigmenti refers to the loss of melanin from the basal cells in the epidermis, melanin then collects in the dermis as a free pigment or aggregates of melanophages. Four stages of polymorphic skin lesions with considerable overlap occur in all patients, mostly on the trunk. The first stage is the bullous or vesicular stage, with linear vesicles, pustules, and bullae with erythema along the lines of Blaschko. This stage is present at birth but may recur during childhood with febrile illnesses. The second stage is the verrucous stage, with warty, keratotic papules and plaques. The third stage is the hyperpigmented stage, with macular hyperpigmentation in a swirled, whorly, linear and spiral pattern along the lines of Blaschko. These changes often involving flexural folds in the axilla, the groin and nipples. The fourth stage is the hypopigmentation stage, with hypopigmented streaky and patchy lesional pattern associated with cutaneous atrophy. This stage is consistently present from infancy to adulthood. Other manifestations include onychodystrophy, delayed eruption of the teeth, hypodontia, microdontia, alopecia, mental retardation, seizures, diffuse hypopigmented retinal changes, congenital developmental cataracts or leukocoria, hemihypertrophy, hemivertebrae, scoliosis, spinal dysraphism, syndactyly, ear anomalies, accessory ribs. There is no specific management required for Incontinentia Pigmenti. These skin lesions are better left intact, kept clean or managed symptomatically. Meticulous dental care is very important. Genetic counseling will imply nuclear and extended family involvement for an utmost outcome.

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