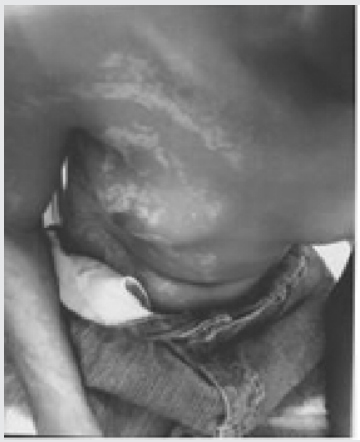
SPOT DIAGNOSIS (IMAGE GALLERY)



>>> Emmanuel Onyekwelu,
Department of Pediatrics, Royal Victoria Teaching Hospital, Banjul The Gambia West Africa. E-Mail:euonyekwelu@hotmail.com.

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 afecting the skin, the central nerv ous system, the bones and the eyes. Most afected children are girls; it is usually associated with in ut ero 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 irst 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 lexural folds in the axilla, the groin and nipples. The fourth stage is the hypopigm entation 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, difuse hypop igmented retinal changes, congenital developmental cataracts or leukocoria, hemihypertrophy, hemivertebrae, scoliosis, spinal dysraphism, syndactyly, ear anomalies, accessory ribs. There is no speciic management require d 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.

E-published: February 2009