PAEDIATRIC DERMATOLOGY

INCONTINENTIA PIGMENTI: A CASE REPORT

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Background: Incontinentia Pigmenti (Bloch-Sulzberger Syndrome) is a multi-system disorder whose name refers to the histopathologic finding of dermal melanophages in the third stage. An incidence of 0.6-0.7/1,000,000 live births has been established with only 42 cumulative cases reported in the Philippine Dermatological Society Health Information System from 2011 to 2018. It is an X-linked dominant disorder caused by a NEMO gene mutation in Xq28. De novo mutations may occur in 65%. This disorder is characterized by four clinical stages namely: Vesicobullous, Hyperkeratotic, Hyperpigmented, and Hypopigmented, however, individual stages may be absent or overlap. Diagnosis is mainly clinical and diagnostic criteria were suggested depending on the presence of a familial history. Management includes topical medications, prevention of secondary complications, and genetic counseling.

Observation: This a case of 1-year old female who presented with multiple hyperpigmented macules and patches some linear, some whorled, following the lines of Blaschko, on the face, trunk and extremities. Her lesions developed at 3 days of age, which were initially inflammatory. She has no family history and other systems were uninvolved except for findings of conical teeth and sparse, dull hair. The clinical presentation fulfills the diagnostic criteria for Incontinentia Pigmenti. Genetic counseling was advised however unavailable locally.

Key message: Incontinentia Pigmenti is a rare genetic condition with various skin, neurologic, ophthalmologic, and dental manifestations. It has four clinical stages, is diagnosed clinically, and may be confirmed by biopsy. Symptomatic treatment is advised for skin lesions and prevention of complications is key. Surveillance is paramount for early detection of other co-morbidities.