

A Retrospective Study of Incontinentia Pigmenti Seen at the National Skin Centre, Singapore Over a 10-year period

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Abstract

Introduction: *Incontinentia pigmenti* is a rare X-linked dominant disease which affects the ectodermal tissues, usually lethal in males. **Materials and Methods:** A retrospective analysis of clinical data obtained from the photographic documentation and casenotes of patients diagnosed to have *incontinentia pigmenti* at the National Skin Centre. The study covered the period from January 1990 to December 1999. **Results:** Twenty-six patients were diagnosed to have *incontinentia pigmenti* of the Bloch-Sulzberger type; 23 (88.5%) were females and 3 (11.5%) were males. There were 20 Chinese, 3 Malay and 3 Indian patients. Most patients had cutaneous manifestations at birth or within the first week of life. Cutaneous features included vesicles, papules, verrucous plaques and splash-like hyperpigmentation along the lines of Blaschko. The cutaneous lesions were widespread in 21 (81%) and localised in 5 (19%) patients. In some cases, hypopigmented atrophic streaks (2 patients) or whorled scarring alopecia (4 patients) were seen. Extracutaneous manifestations, seen in 5 (19%) patients, included neurological, dental and ocular defects. One Malay girl had severe neurological involvement associated with ocular abnormalities. A positive family history was present in 6 (23%) patients. The 3 male patients were Chinese without any family history. **Conclusions:** Each stage of the disease comes with its own set of differential diagnosis, including infections e.g. herpes virus infection and other types of genodermatoses e.g. linear and whorled nevoid hypermelanosis. The phenomenon of whorled scarring alopecia, hitherto unreported in the literature, corresponded to the lines of Blaschko. In the 3 Chinese male patients, the disorder probably originated from a new mutation. X chromosome inactivation in females during early embryogenesis results in a mosaic population of cells and this explains the linear and patchy manifestations of *incontinentia pigmenti*.

Ann Acad Med Singapore 2001; 30:409-13

Key words: Functional X-chromosome mosaicism, *Incontinentia pigmenti* in males, Lines of Blaschko, Whorled scarring alopecia, X-linked dominant inheritance

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