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# INCONTINENTIA PIGMENTI – A CASE REPORT

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**ABSTRACT**
Incontinentia pigmenti, an uncommon genodermatosis, primarily affects female infants. Classically, it manifests with linear vesicular lesions evolving into verrucous lesions within a few weeks and followed by characteristic swirl like pigmentation that persists for many years. Here we report a case of a 4-year-old girl, brought by her parents with complaints of linear lesions in the left lower limb.

## INTRODUCTION

Incontinentia pigmenti (IP) was first described by Garrold et al in 1906. It is a dominantly inherited, X-linked syndrome characterized by cutaneous lesions (vesicular, verrucous and pigmented), associated with developmental defects of the eye, central nervous system and skeletal system. It more commonly affects females and found to be embryonic lethal in males. Incontinentia pigmenti is caused by mutations in the NEMO/ IKK-γ gene, which is located on chromosome Xq28.325. Typical skin lesions along the Blaschko's lines follow four cutaneous stages (vesicular, verrucous, hyper pigmented and hypo pigmented).

## CASE REPORT

A 4-year-old was brought by her parents with complaints of altered skin color in a linear pattern over the left lower limb. The child was asymptomatic at birth. On day 4, her parents noticed multiple vesiculo-bullous eruptions in linear groups over both lower limbs and appearance of crops of lesions continued for about a week. Few scattered lesions were also noted on the trunk. The lesions healed in a week, leaving behind warty lesions, which flattened at most of the places in 6-8 weeks and left slaty brown pigmentation in characteristic linear, bizarre and spiral fashion, which became static. Systemic examinations were normal. Routine blood investigations showed eosinophilia of 10% and absolute eosinophil count was 780/cubic mm. The VDRL test of the patient and her parents was non-reactive. History of consanguinity was not present. Parents were normal. Obstetric history of mother was normal, her blood group was `O' positive, fasting blood sugar was 83 mg% and serum creatinine was 1.3 mg%. Since parents were not willing for biopsy of skin, histopathological examination was not done.

## DISCUSSION

Bloch–Sulzberger syndrome, Bloch–Sulzberger disease, melanoblastosis cutis, nevus pigmentosus systematicus. Bloch–Siemens syndrome. [1-3]. The "vesiculobullous" stage usually begins between 0 and 2 weeks of age and persists for 8 weeks. Vesicles may or may not coalesce to form bullae and may remain scattered. They are usually clear and tense and appear in crops. When arranged linearly over the trunk and extremities. The sites commonly involved are the lateral trunk, extremities and perimammary region. The next stage, "verrucous" stage usually starts between the 2nd and 6th week of life as smooth red nodules or plaques, often having an irregular linear appearance, on the trunk and extremities.

Sometimes they have a bluish-purple hue & ulcerate. Lesions particularly those situated on the dorsal aspect of hands and feet develop a warty or verrucous appearance. The "pigmentary" stage usually starts between the 3 to 7 months of age, progresses steadily up to the second year and ultimately fades by the second and third decade of life. In some cases, it may be the only cutaneous change noticed. The pigmentation, blue-grey or slate-brown, has a characteristic capricious, bizarre, whorled or splashed distribution. The next stage, "Hypo pigmented," atrophic/ sclerotic in which anhidrotic streaks may be detected as sequelae in the later stages. Sometimes, multiple linear and macular telangiectasias may also be found. Hair changes in the form of localized cicatricial alopecia at or near the vertex of the scalp may be present at birth or subsequently develop in infancy in 25% of cases. The nails may become thin, flat or spoon-shaped with longitudinal and transverse ridges.

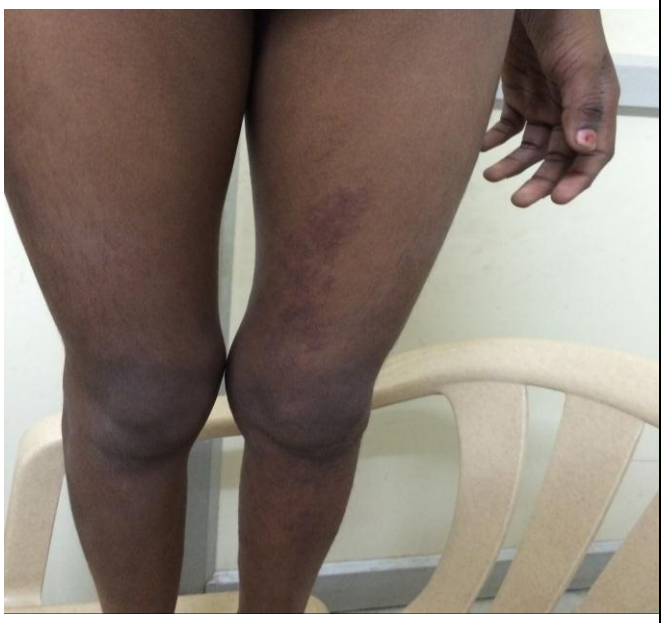
- Dental defects are very frequent and include delayed dentition, cone- or peg-shaped crowns, malformation, and missing teeth, particularly the premolars and upper lateral incisors
- Ocular defects, some causing blindness, are encountered in about one-third of the patients and include strabismus, nystagmus, blue sclerae, cataract, exudative chorioretinitis, optic atrophy, retinal vascular abnormalities, retinal pigmentation, retinal detachment, retrolental fibroplasia- like condition and microphthalmia.
- Central nervous system disorders in the form of mental retardation slow motor development, epilepsy, spastic paralysis and microcephaly may be detected in about one-quarters of cases.
- Skeletal abnormalities like skull deformities, dwarfism, shortened extremities, syndactyly, spina bifida, clubfoot, extra ribs, cleft palate, cleft lip and ear abnormalities may be detected in some cases.

Histopathology: The early inflammatory phase of IP, shows eosinophilic spongiosis and scattered dyskeratotic keratinocytes. In verrucous lesions, the epidermis is acanthotic with hyperkeratosis and focal dyskeratosis. In third stage (pigmentary), there is pigmentary incontinence and the fourth stage (hypo pigmented, sclerotic or atrophic) is characterized by a thinned epidermis and dermis devoid of appendages [5,6].

Diagnosis: The triad of vesiculobullous eruptions, warty lesions and pigmentation in a female infant is clinically suggestive of incontinentia pigmenti.

Treatment: Family counseling may prevent occurrence of new cases. As the skin lesions spontaneously subside in adulthood, no treatment is necessary other than prevention of secondary infection and assurance.

**Figure 1. Clinical picture showing slaty brown pigmentation in characteristic bizarre, linear and spiral fashion in the left lower limb**



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**CONFLICT OF INTEREST:**
The authors declare that they have no conflict of interest.

**STATEMENT OF HUMAN AND ANIMAL RIGHTS**

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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