Incontinentia Pigmenti: A Case Report

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Abstract

Incontinentia pigmenti is a rare X-linked dominant condition characterized by abnormalities in skin, eyes, teeth, bones and the central nervous system. A seven year old female child presented to the Dermatology Out Patient Department (OPD) of Bir Hospital with vesicles, bullae, verrucous papules and plaques and hypopigmented streaks on the trunk and extremities of six years duration. Based on the clinical findings she was diagnosed as a case of Incontinentia pigmenti.

Cause of death was not known. Our patient is the third from his second wife. All the other siblings are healthy. There is no parental consanguinity. There is no history of similar skin lesions in her family or first degree relatives.

On physical examination, the patient is of average build weighing 25 kgs. Her systemic examination was normal. On cutaneous examination, there were multiple vesicles, bullae, hyperkeratotic papules and hypopigmented patches all over the body of five years duration. According to the patient’s father, the child was a full term normal home delivery. At the age of two year they noticed a warty plaque at the medial aspect of left ankle joint which was preceded by vesicles. Gradually she started developing similar lesions on different parts of the body which would start as vesicles and bullae, then become hyperkeratotic and some lesions became hyperpigmented or hypopigmented. The lesions are still increasing in size.

The father of the patient was married twice. He has four children from the first wife; one son and three daughters. All are healthy. From the second wife, he had five children: two sons and three daughters but the 1st child, which was a male, died 2 to 4 months after birth.

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![Image 1 showing linear verrucous lesions with vesicles, bullae and erosions]

**Fig 1 and 2:** Showing linear verrucous lesions with vesicles, bullae and erosions

![Image 2 showing linear hyper-and hypopigmented streaks]

**Fig 3, 4 and 5:** Linear hyper-and hypopigmented streaks

On the basis of clinical and histopathological findings the patient was diagnosed as incontinentia pigmenti.

The secondary infections were treated with oral cefadroxil 250 mg x bid x 7 days and topical fusidic acid.

**Discussion**

Incontinentia pigmenti also known as Bloch-Sulzberger syndrome or Bloch-Siemens syndrome is a rare and complex hereditary syndrome in which vesicular, verrucous and pigmented cutaneous lesions are associated with developmental defects of the teeth, eye, bone and CNS. This syndrome mainly affects females and is lethal in males.

The genetic defect lies on the X chromosome, localized to Xq28. It is related to the NF kappa B essential modulator (NEMO) gene. Rabia HS et al has reported 25 adults with IP and nuclear factor-κB essential modulator gene rearrangement or mutations.

Four distinct clinical stages are recognized:

**Stage 1:** inflammatory macules, papules, vesicles and pustules

**Stage 2:** hyperkeratotic and verrucous lesions

**Stage 3:** grey-brown pigmentation

**Stage 4:** atrophic, hypopigmented and depigmented bands or streaks that are hairless and anhidrotic and fail to tan on sun exposure.

Stage 1 usually begins between 0-2 weeks of age and persists for 2 months. Stage 2 usually starts between the second and sixth week of life. The pigmentary stages usually starts between the 12th and 26th week of life, which progresses upto the 2nd year and fades by the 2nd and 3rd decade of life.

Hair changes may be seen at the vertex in the form of cicatricial alopecia.

Nail changes may also be present in cases with incontinentia pigmenti. Aggarwal K, Jain VK, Dayal S has reported a case of incontinentia pigmenti, with nail dystrophy and onycholysis.

The extracutaneous manifestations are seen in more than half of the patients with incontinentia pigmenti:

1. Dental defects can be seen in the form of partial anodontia, pegged teeth and missing teeth, particularly the upper lateral incisors and the premolars.
2. In ophthalmologic findings, there may be blindness, strabismus, cataracts, retinal detachment, optic atrophy and microphthalmia\textsuperscript{14, 15, 16}. Minić S et al. has done meta-analysis of 1931 incontinentia pigmenti patients with ocular abnormalities\textsuperscript{17}.

3. The central nervous system defect includes seizures, mental retardation, spastic paralysis, microencephaly and slow motor development\textsuperscript{18, 19, 20}.

4. Skeletal abnormalities can present with skull deformities, dwarfism, spina bifida, club foot, extra ribs, cleft palate and cleft lips.

   Leukocytosis and eosinophilia\textsuperscript{21} are common findings. Histopathological features differs with the clinical stage of the disease. In the first stage, there are intraepidermal vesicles with eosinophilic spongiosis. In the second stage, there is acanthosis, hyperkeratosis and papillomatosis with persistent presence of necrotic keratinocytes and the final stages of pigmentation is due to melanin in melanophages in the upper dermis.

**Conclusion**

Incontinentia pigmenti should be differentiated from linear and whorled nevoid hypermelanosis, epidermolysis bullosa and childhood bullous pemphigoid, Naegeli-Franceschetti-Jadassohn syndrome and incontinentia pigmenti achromians\textsuperscript{22}. Treatment is not necessary other than the control of secondary infection as the lesions spontaneously subside in adulthood.

**References**


