

Incontinentia Pigmenti: A Case Report

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Introduction

Incontinentia pigmenti, also known as Bloch-Sulzberger disease, is a rare X-linked dominant condition characterized by abnormalities in skin, eyes, teeth, bones and the central nervous system. It appears most commonly in girls. There are four stages of the disease: vesicular; verrucous; hyperpigmented and hypopigmented. Here we present a case of Incontinentia Pigmenti in which all the stages of the disease were present, as, it is not a common presentation in our OPD.

The Case

A seven year old female child from Jumla District (a part of Karnali Zone, situated in the mid-western part of Nepal), studying in grade one presented with 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.

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 built weighing 25 kgs. Her systemic examination was normal. On cutaneous examination, there were multiple vesicles, bullae, erosions on different parts of the body as well as verrucous hyperkeratotic papules and plaques, hyper- and hypopigmented streaks distributed in bizarre pattern following the lines of Blaschko. The lesions were more on the trunk and extremities. Head and neck, palms and soles were spared [Figure 1 and 2]. However there were no extracutaneous manifestations like the teeth, CNS, bone and eye changes.

The haematological parameters were within normal limits. Histopathological examination showed keratinized stratified squamous epithelium showing verrucous acanthosis, compact hyperkeratosis, spongiosis within the spinous layer and foci of intraepidermal keratinization. Reduced melanin along with vacuolations was noted in the basal cells in some foci. Eosinophils were found in the upper dermis and few within the epidermis. The dermis was infiltrated by mixed inflammatory cells. Few melanin pigments were also noted within the dermis.

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



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 CNS¹. This syndrome mainly affects females^{2,3} and is lethal in males⁴.

The genetic defect lies on the X chromosome, localized to Xq28. It is related to the NF kappa B essential modulator (NEMO) gene^{5,6}. Rabia HS et.al has reported 25 adults with IP and nuclear factor- κ B essential modulator gene rearrangement or mutations⁷.

Four distinct clinical stages are recognized⁸:

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 life⁹.

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

Nail changes may also be present in cases with incontinentia pigmenti¹⁰. Aggarwal K, Jain VK, Dayal S has reported a case of incontinentia pigmenti, with nail dystrophy and onycholysis¹¹.

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^{12,13}.

2. In ophthalmologic findings, there may be blindness, strabismus, cataracts, retinal detachment, optic atrophy and microphthalmia^{14, 15, 16}. Minić S et.al has done meta-analysis of 1931 incontinentia pigmenti patients with ocular abnormalities¹⁷.
3. The central nervous system defect includes seizures, mental retardation, spastic paralysis, microencephaly and slow motor development^{18,19,20}.
4. Sketal abnormalities can present with skull deformities, dwarfism, spina bifida, club foot, extra ribs, cleft palate and cleft lips.

Leukocytosis and eosinophilia²¹ 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 achromians²². Treatment is not necessary other than the control of secondary infection as the lesions spontaneously subside in adulthood.

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