A Case of Incontinentia Pigmenti

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Incontinentia pigmenti is an uncommon neurocutaneous genodermatosis characterized by three stages; vesicular lesions, verrucous lesions and hyperpigmentation. A two-week-old female infant showed grouped erythema-based vesiculopustules on the whole body since birth. One month later, vesiculopustular lesions began to disappear gradually. At this time, she developed linear verrucous plaques on the dorsum of the feet and hands and reticulated hyperpigmented patches on the lower extremities. At 2 months of age, vesicular lesions completely disappeared and the pigmented patches spread to the abdomen which became darker with time. The verrucous lesions diminished at 3 months of age. There were no neurological or ocular defects. We describe a case of incontinentia pigmenti with typical clinical and pathological features at each stage. (Ann Dermatol 9:(1)46~50, 1997).

Key Words : Incontinentia pigmenti

Incontinentia pigmenti(IP) is a rare genodermatosis typically involving three stages; vesicular lesions, verrucous lesions and hyperpigmentation. IP is also known as Bloch-Sulzberger syndrome and was first described by Bloch in 1926 and later by Sulzberger in 1928[1]. The disorder is inherited as an X-linked dominant trait and is probably fatal in male fetuses, which accounts for the female : male ratio of 37:1[2].

CASE REPORT

A two-week-old female had recurrent erythematous vesiculopustules on the trunk and extremities since birth. The vesiculopustules(Fig. 1A) progressed to linear verrucous plaques(Fig. 2A) on the dorsum of feet and hands during the first 6 weeks. At two month of age, reticulated hyperpigmented patches(Fig. 3A) appeared on the lower extremities and gradually spread to the abdomen, which became darker with time. After 3 months of age, the verrucous lesions lasted for about 3 months and then disappeared. At 5 months of age, we found alopecia on the vertex of the scalp. At 6 months of age, pigmentation of the extremities began to decrease. A complete blood cell count demonstrated leukocytosis (29440/μl) and the eosinophilia (7770/μl) at the age of 2 weeks and eosinophil count decreased with time. The child was born at term by spontaneous vaginal delivery. Her mother had had one spontaneous abortion of a fetus whose sex was unknown. We could not find any cutaneous lesions suggesting incontinentia pigmenti from the patient's mother.

At the vesicular lesion site, a skin biopsy was taken which revealed spongiosis and intraepidermal blisters. There were numerous eosinophils within the blisters and in the upper dermis(Fig. 1B). Histopathological study from the verrucous lesion revealed acanthosis, irregular papillomatosis, hyperkeratosis, basal cell vacuolization and mild, chronic, inflammatory infiltrates in the dermis(Fig. 2B). The hyperpigmented third stage of IP showed melanophages in the dermis and corre-

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Table. Characteristics of 26 cases of incontinentia pigmenti reported in Korean literature

Case	Sex	Onset	Distribution	Stage
1. Lee et al[13]	F	Birth	Trunk & extremity	3rd
2. Lee HW[14]	F	Birth	Trunk & extremity	2nd
3. Cho et al[15]	F	3weeks	Trunk & extremity	3rd
4. Cho et al[15]	F	Uncertain	Trunk & extremity	not done
5. Cho et al[15]	F	Uncertain	Trunk & extremity	not done
6. Cho et al[15]	F	Uncertain	Trunk & extremity	not done
7. Kim et al[16]	F	3 days	Trunk & extremity	1st
8. Kim et al[15]	F	3 days	Trunk & extremity	2nd
9. Kim et al[16]	F	Birth	Trunk & extremity	2nd
10. Kim et al[16]	F	4 days	Tronk & extremity	1st
11. Kim et al[16]	F	3 yrs	Trunk & extremity	3rd
12. Suh et al[17]	F	7 days	Trunk & extremity	3rd
13. Suh et al[18]	F	Birth	Trunk & extremity	2nd
14. Kim et al[19]	F	Birth	Trunk & extremity	1st
15. Kim et al[19]	F	Birth	Trunk & extremity	3rd
16. Park et al[20]	M	Birth	Trunk & extremity	2nd
17. Park et al[21]	F	3days	Trunk & extremity	1st
18. Chun et al[22]	F	Birth	Extremity	2nd
19. Chun et al[22]	F	2days	Extremity	3rd
20. Chun et al[22]	F	Birth	Trunk	3rd
21. Chun et al[22]	F	3days	Trunk & extremity	2nd
22. Chun et al[22]	F	4months	Extremity	2nd
23. Chun et al[22]	F	2days	Extremity	1st
24. Chun et al[22]	F	Birth	Trunk & extremity	1st & 2nd
25. Chun et al[22]	F	2days	Ttunk & extremity	1st
26. Chun et al[22]	F	Birth	Trunk & extremity	1st

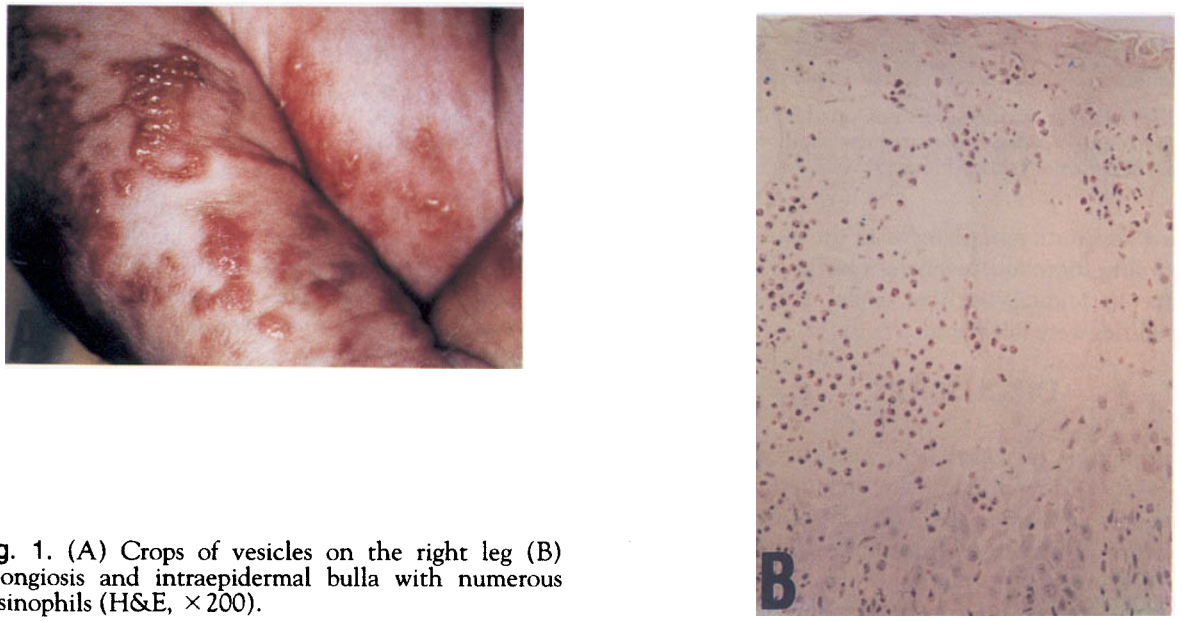

Fig. 1. (A) Crops of vesicles on the right leg (B) Spongiosis and intraepidermal bulla with numerous eosinophils (H&E, ×200).

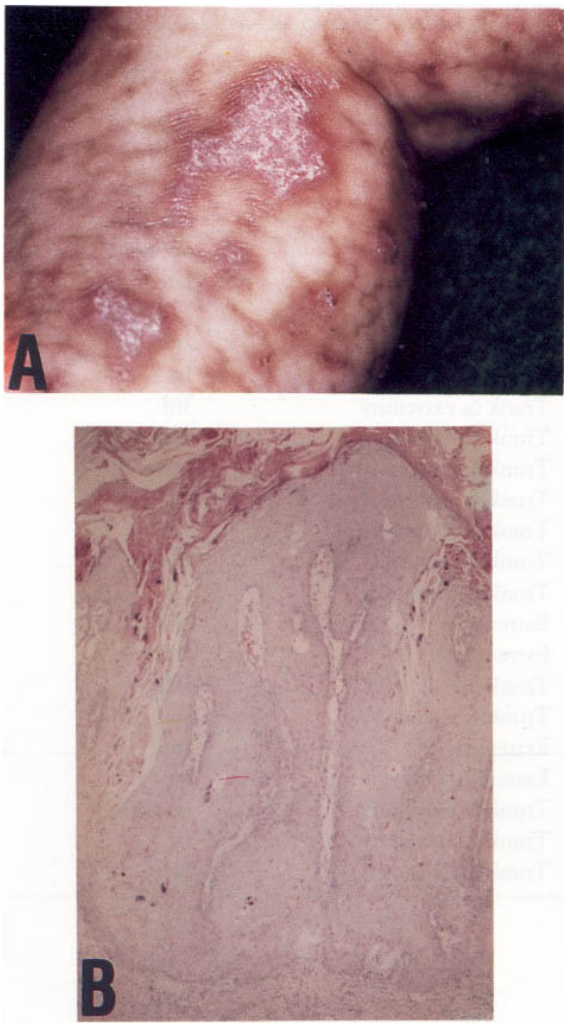

Fig. 2. (A) Linear verrucous lesions on the right leg (B) Hyperkeratosis, acanthosis, irregular papillomatosis and basal cell vacuolization (H&E, ×100).

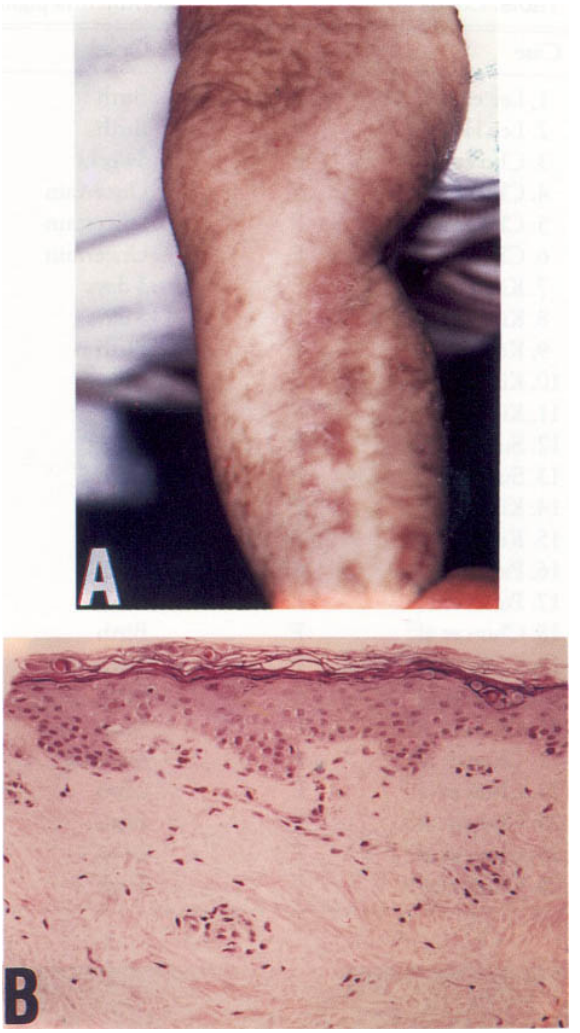

Fig. 3. (A) Reticulated hyperpigmented patches on the right leg (B) Vacuolar alteration of the basal layer and dermal melanophages (H&E, ×40).

sponding diminution of pigment in the basal layer with vacuolization and degeneration(Fig. 3B). Extracutaneous findings such as ocular and dental abnormalities were not found.

DISCUSSION

Incontinentia pigmenti(IP), also known as Bloch-Sulzberger syndrome, is an uncommon neurocutaneous genodermatosis. The cutaneous manifestations of IP consist of three stages. The three stages of IP may be concurrent or some of the stages may occur in utero. The first stage usually begins within 2 weeks of birth but in rare cases may appear after the first year of life. It consists of linear erythema and vesicles that are most often located on the extremities. Crops of vesicles may be recurrent, as in our case, and may persist until 6 years of age.

Stage 2 most often begins between the second and sixth weeks of life and consists of verrucous papules that are often arranged linearly and found predominantly on the extremities. The lesions persist for 6 to 12 months and then resolve spontaneously. However, in a recent report a patient with IP had continuing development of new verru-

cous lesions, one of which resulted in squamous cell carcinoma in situ at the age of 25 years[4].

Stage 3, with age of onset most frequently between 12 and 26 weeks[2], is characterized by whorled, brown to slate-brown hyperpigmented macules and patches on the trunk; the extremities are less often affected. The hyperpigmentation occurs in streaks or whorls which correspond to Blaschko's lines. The distribution of hyperpigmentation is often unrelated to the distribution of the previous vesicular rash. The pigmented lesions remain static for a period of time until they fade during childhood and adolescence. By the age of 16, the majority of these pigmented lesions have disappeared.

The vesicular first stage of our case represented a generalized eruption including the scalp, palms and soles. The second stage, verrucous lesions, continued during the first 3 months. The third stage, hyperpigmentation, had remained on the trunk until the patient was 14 months old. Our case showed classical features of incontinentia pigmenti, where we found the overlapping three stages at one time.

Other cutaneous findings include nail dystrophy, subungal hyperkeratotic tumors, lytic deformities of the phalanges, agenesis of the eyebrows and partial sweat gland aplasia. Hair abnormalities seen with IP include vertex alopecia, hair loss from other body sites, and woolly haired nevus[5]. Major dental abnormalities occur in 65% of patients; the majority of these are identifiable by the age of 2 years[2]. The most common are partial anodontia (43%) and pegged teeth (30%). Eye anomalies have been reported in 25% to 35% of patients with strabismus being the most common (18%)[6]. Retinal detachment and a fibrovascular retrolental membrane are the most frequently reported intraocular abnormalities. The central nervous system is affected in up to 33% of patients[1]. The most commonly reported central nervous system abnormalities are seizures (13%), mental retardation (12%), spastic paralysis (11%), microcephaly and motor retardation. Laboratory findings may show a high peripheral eosinophilia ranging from 5% to 65% and also a high leukocyte count[7].

The patient had suffered from generalized vesiculopustular eruptions on the whole body including the scalp, palms and soles. In infancy, we should differentiate incontinentia pigmenti from other neonatal vesiculopustular eruptions such as erythema toxicum, transient neonatal pustulosis, eosinophilic pustular folliculitis and neonatal herpes simplex infection[8].

Histopathologically, the first stage is characterized by eosinophilic spongiosis, dyskeratotic keratinocytes and a dense dermal infiltrate composed of eosinophils and mononuclear cells[9]. The second stage shows acanthosis, irregular papillomatosis, hyperkeratosis, basal cell vacuolization and a mild chronic inflammatory infiltrate in the dermis. The third stage shows melanophages in the dermis and a corresponding diminution of pigment in the basal layer with vacuolization and degeneration. Ophthalmologic examination should be done within 1 month of birth and should be repeated at 3 month intervals until 1 year of age. All patients with IP should have detailed examinations by the age of 2 years to evaluate partial anodontia and tooth spacing. A detailed family history should be taken. If a woman without history or clinical signs of IP gives birth to a child with IP, the disorder was most likely the result of spontaneous mutation. Many unusual clinical features of have has been reported. In a review of the literature, 26 cases of incontinentia pigmenti have been reported in Korea, all of which showed one or two stages(Table). Our case is characterized by a serial continuous follow-up and clinicopathological matching. From a clinical aspect, a continuous clinical and pathological follow-up is recommended at least for 2 years in the case of IP with initial and subsequent evaluation by specialists (dentists, dermatologists, neurologists and ophthalmologists). In addition, genetic counseling to the parents should be provided[10,11,12].

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