

### Case 21

A new born Thai girl from Bangkok

#### Chief complaint:

Multiple erythematous papules in linear pattern at left leg

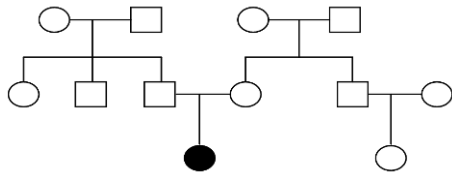
#### Present illness:

The nearterm (GA 36 wks) neonate patient with history of meconium stained amniotic fluid had developed group of vesicles on erythematous base at left leg. At birth, she presented APGAR score 10 and BW 2,580 grams.

2 days later, the skin lesion changed to multiple discrete skin-color hyperkeratotic papules on erythematous base in linear pattern along left leg to left foot

**Maternal history** : ANC at GA 29 wks x 2 times  
: Blood group A/Rh+  
: Anti-HIV, VDRL, HBsAg :- negative

**Family history:** No history of similar skin lesions in her family, Illustrated in the genetic inheritance pedigree chart below:



#### Physical examination

A Thai infant, alert, not pale, no jaundice. HC 33 cm, AF 2x2 cm, PF 0.5x0.5 cm., no cleft lip and cleft palate.

HEENT, Chest-H/L and Abdomen : With in normal limit.

Genitalia : normal female type; Anus/Rectum: Patent anus

Extremities : No deformities; LN : No superficial lymphadenopathy

NS : Grossly intact ; Rooting/ Suckling reflex : Positive

#### Dermatological examination

Multiple discrete skin-color hyperkeratotic papules on erythematous base in linear pattern along left leg to left foot

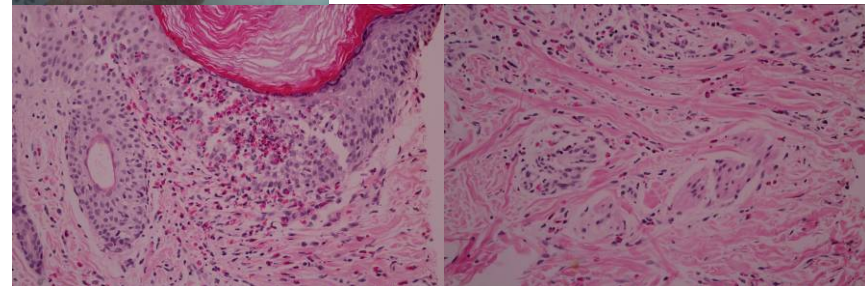
**Ophthalmic examination** : Normal

**Laboratory** : TSH 3.4981 uIU/ml (0.35-4.94)

**Histopathology** (S10-010675A)

Focal spongiosis with numerous eosinophils and occasional dyskeratic cells

Superficial perivascular inflammatory-cell infiltrate of lymphocytes and numerous eosinophils in the upper dermis



**Diagnosis:** Incontinentia pigmenti

**Treatment:** Observation and measure skin lesion with symptomatic treatment

**Presenter:** Natthanej Luplertlop

**Consultant:** Amornsri Chunharas

**Discussion:**

Incontinentia pigmenti (IP) or Bloch-Sulzberger syndrome is a rare, dominant, X-linked genodermatosis caused by a mutation in the NF-kappa B essential modulator gene, NEMO/IKK $\gamma$ , has been mapped to Xq28.<sup>1</sup> The range of diseases caused not only NEMO mutations, the newest research findings on eosinophil recruitment through eotaxin release by activated keratinocytes.<sup>(1-3)</sup> The term incontinentia pigmenti originates from the microscopic appearance of the lesions in the third phase of the disease, which is characterized by the presence of loose pigment in the basal layer of the epidermis, as if the melanocytes showed melanin-incontinence.<sup>(1-2)</sup> IP syndrome was credit to Bloch in 1926 and Sulzberger in 1928. Some of the older names used in the past for this condition include *Asboe-Hansen disease*, *Bloch-Siemens syndrome*, *Siemens-Bloch pigmented dermatosis*, *melanoblastosis cutis linearis*, and *nevus pigmentosus systematicus*.<sup>(2-3)</sup> The condition affects predominantly female newborns. When males are affected, it is generally lethal, resulting in spontaneous abortion in the majority of cases.<sup>(3-4)</sup> The syndrome involves organs and tissues whose embryonic origin is ectodermal and mesodermal. Manifestations may be cutaneous or extracutaneous, the former being more common. In the majority of cases, these abnormalities present in the first few weeks of life and their progression is divided into four phases. Progression of the cutaneous symptoms occurs in four classic phases that may

be concomitant or sequential: 1) vesicles or linear inflammatory blistering that appear at birth or during the first two months of life and may last weeks or months; 2) hyperkeratotic, verrucous, linear plaques that may last several months; 3) brown or greyish-blue hyperpigmentation, distributed in Blaschko's lines or in swirling patterns that appear in infancy and fade slowly until they disappear sometime in adulthood; 4) hypopigmented linear macules with no skin appendages on the trunk and limbs, appearing in adulthood.<sup>(5-6)</sup> Some investigators describe only three phases, since the fourth phase may occur later and remain underdiagnosed.<sup>(5-7)</sup> Extracutaneous manifestations occur in around 70-80% of cases and involve: the central nervous system (seizures, mental retardation, ischemic cerebrovascular accidents, hydrocephaly and anatomical abnormalities); eyes (strabismus, cataracts, anophthalmia, microphthalmia, optic atrophy and others); teeth (hypodontia and partial anodontia, among others); bone and musculoskeletal structure (syndactylia, skull deformities, nanism, supernumerary ribs, hemiatrophy and shortening of the legs and arms).<sup>(7-8)</sup> There have also been reports of immunological abnormalities such as leukocytosis and eosinophilia in the first phase, as well as poorly functioning lymphocytes and defective neutrophil chemotaxis.<sup>(5,7)</sup>

**Pathological diagnosis:** Diagnosis is based on clinical status and histological findings, which differ in each phase of progression of the disease.<sup>(1-2)</sup> In the vesiculobullous phase, blisters full of eosinophiles are present; in the verrucous phase, hyperkeratosis, dyskeratosis, acanthosis and papillomatosis are present; in the hyperpigmentation phase, there is evidence of pigment leakage and numerous melanophages are present in the dermis; and in the hypopigmentation phase, there is a thinning of the epidermis and an absence of skin appendages.<sup>(3, 6-8)</sup>

**Treatment :** The management of the skin and systemic abnormalities is based on symptomatology alone. <sup>(1-2, 5-8)</sup>

**Follow-up** : The skin lesions generally regress spontaneously. However, there have been reports of the appearance of ophthalmological, dental and neurological abnormalities after childhood, which needs the continued follow-up of these patients.<sup>(5-8)</sup>

## References

1. Lee Y, Kim S, Kim K, Chang M. Incontinentia pigmenti in a newborn with NEMO mutation. *J Korean Med Sci* 2011;26(2):308-11.
2. Jabbari A, Ralston J, Schaffer JV. Incontinentia pigmenti. *Dermatol Online J* 2010;16(11):9.
3. Ormond DT, Gamonal SB, Gamonal A, Carvalho MT. Condiloma de Buschke Lowenstein em paciente com incontinência pigmentar. *An bras Dermatol* 1998;73:29-32.
4. Tristão OC, Baraky AKC, Carvalho MTF, Pereira CAC. Incontinentia pigmenti - síndrome de Bloch-Sulzberger: relato de caso. *An Bras Dermatol* 1995;70:231-3.
5. Pereira MA, Mesquita LA, Budel AR, Cabral CS, Feltrim Ade S. X-linked incontinentia pigmenti or Bloch-Sulzberger syndrome: a case report. *An Bras Dermatol* 2010;85(3):372-5.
6. Motamedi MH, Lotfi A, Azizi T, Moshref M, Farhadi S. Incontinentia pigmenti. *Indian J Pathol Microbiol* 2010;53(2):302-4.
7. Emre S, Firat Y, Güngör S, Firat AK, Karıncaoğlu Y. Incontinentia pigmenti: a case report and literature review. *Turk J Pediatr* 2009;51(2):190-4.
8. Goldberg MF. The skin is not the predominant problem in incontinentia pigmenti. *Arch Dermatol* 2004;140(6):748-50.