Picture Story

Incontinentia Pigmenti

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(Key words: incontinentia pigmenti)

Linear skin lesions were noticed at birth in a baby girl, first born to a young mother. The lesions consisted of vesicles, papules and verrucous plaques on all 4 limbs and lateral aspects of trunk (Figure 1).

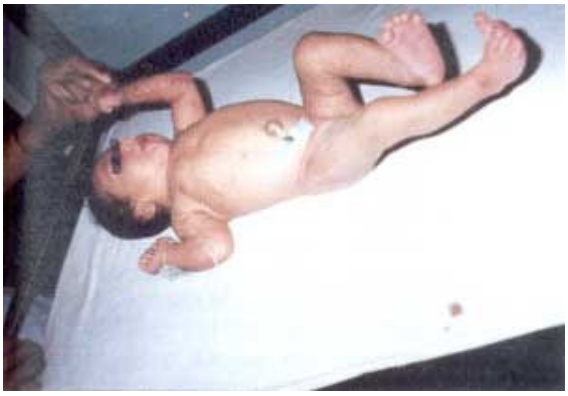

Figure 1. *Vesicular linear lesions at the age 28 hours*

Vesicular lesions increased over the first week of life and gradually disappeared leaving streaks of hyperkeratotic areas. There were no associated ocular or nail abnormalities (Figure 2).

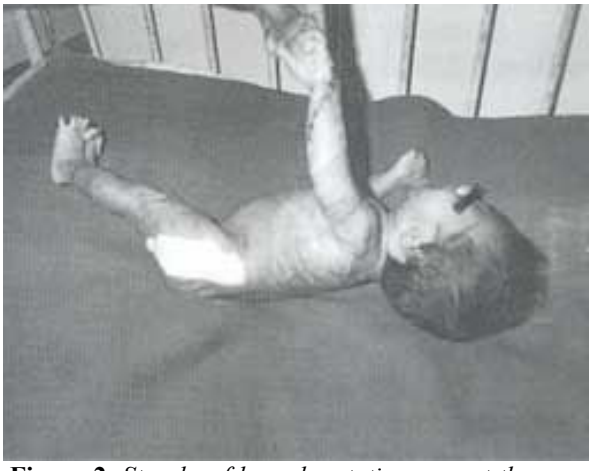

Figure 2. *Streaks of hyperkeratotic areas at tha age 10 days*

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The mother, who was examined at delivery, did not have hyperpigmented linear streaks over the abdomen or thighs. A clinical diagnosis of Incontinentia Pigmenti was made and it was later confirmed by histology.

Incontinentia Pigmenti (IP) or "Bloch Sulzberger Syndrome" is a rare X-linked dominant disorder, usually lethal in males, characterized by abnormalities in the ectodermal tissues including, skin, CNS, teeth, hair and eyes[1] (Table 1).

Table 1
Associated abnormalities of IP[1,2,4,5,6]

Ocular (25-35%)
- Strabismus (commonest)
- Cataract
- Microphthalmia
- Optic atrophy
- Proliferative vitreoretinopathy
- Retinal detachment
- Iris hypoplasia
- Blue Sclera

CNS (33%)
- Cognitive development retardation (12%)
- Seizures (13%)
- Spasticity (11%)

Dental (65%)
- Late dentition
- Conical teeth (30%)
- Hypodontia
- Anodontia (43%)

Skeletal
- Syndactyly
- Microcephaly
- Dwarfism
- Spina bifida
- Club foot
- Hemiatrophy

Others
- Nail dystrophy
- Alopecia (scarring/patchy/diffuse)

IP is caused by mutations in a gene called NF kappa-B essential modulator (NEMO) which encodes a regulatory component of the I-kappa-B-kinase complex required for activating the NF-kappa-B pathway[2,3,4].

There are generally 3 stages of evolution[1,5,6]. Onset of the vesiculobullous stage of the disorder is at birth, or within 2 weeks of birth, in most cases. The verrucous stage may begin from 2nd to 6th week and the pigmentary stage, the hallmark of IP, appears from the 12th-26th weeks of age. Usually all 3 stages occur in an individual but up to 14% have only hyper-pigmentation along the lines of Blaschko. Recently a 4th stage with hypopigmented or atrophic linear lesions has been identified[6].

Lesions of IP at birth may be confused with those of herpes virus infection and bullous impetigo but the linear configuration of the lesions in a female infant is the unique feature[1,5,6].

Although the skin lesions may constitute the only manifestation, approximately 80% of affected children have other defects (Table 1).

Choice of management depends on the occurrence of non-cutaneous abnormalities since the skin lesions are benign. The high incidence of associated major anomalies warrants genetic counselling[1,5,6].

Incontinentia Pigmenti was first reported in Sri Lankan children in 1985[8].

References

1. Champion R H, Burton J L, Burns D A, Breathnach S M.(editors) Textbook of Dermatology, 6th ed. vol.2; London; Blackwell Science, 1998.

2. A recurrent deletion in the ubiquitously expressed NEMO gene. *Human Molecular Genetics* 2001; **10 (19)**: 2171-9

3. A retrospective study of IP seen at the National skin center, Singapore over a 10 year period. *Ann Acad Med Singapore* 2001; **30(4)**: 409-13

4. 55 atypical forms of IP in male individual results from mutations of a cytosine tract in exon 10 of NEMO (Ikk-gamma) *American Journal of Human Genetics* 2001; **68 (3)**: 765-71.

5. Fitzpatric T B, Eisen A Z, Wolff K, Friedberg T M, Austen K F. (editors) Dermatology in general medicine. 3rd ed. vol 1; New York; Mc GrawHill, 1987.

6. Odom R B, James W D, Berger T G. (editors) Diseases of the skin. 9th ed.; Philadelphia; Harcourt Health Sciences, 2000.

7. Laser photocoagulation in preproliferative retinopathy of incontinentia pigmenti. *JAAPOS* 2001; **5 (4)**: 258-9.

8. De Silva D G H, Chularatne W, Perera D J B. Incontinentia pigmenti in Sri Lanka. *CMJ* 1985; **30 (4)**: 179-82.