Generalized Vesicles and Plaques in a 2-day-old Female Infant

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CASE REPORT

A 2-day-old female infant was found to have some rice-grain to pea-sized vesicles scattered on trunk and extremities since birth (Fig. 1, 2). In addition, erythematous patches and plaques over face, neck, trunk and extremities with agenesis of right toes were also noted (Fig. 3). Laboratory data showed leukocytosis with marked eosinophilia. Echocardiogram revealed patent ductus arteriosus and type II ventricular septal defect. Computed tomography of brain showed dysgenesis of corpus callosum. Ophthalmoscopic examination demonstrated retinal hemorrhage. A skin biopsy was taken from a vesicle on the right calf and the histopathology of the tissue specimen was shown as follows (Fig. 4). There was no family history.

Fig. 1&2
Discrete rice-grain to pea-sized vesicles with erythematous patches and plaques over face, neck, trunk and extremities.

Fig. 3
Agenesis of right toes.

Fig. 4
Skin biopsy specimen of the vesicular lesion shows intraepidermal spongiotic vesiculation filled with eosinophils, individual necrotic keratinocytes, and infiltration of eosinophils in the dermis. (H & E, 100X)

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**DIAGNOSIS: Incontinentia Pigmenti**

**DISCUSSION**

Incontinentia pigmenti (IP), also known as Bloch-Sulzberger syndrome, is an uncommon X-linked genodermatosis that affects mostly females and is usually lethal in males in utero. Cutaneous manifestations are classically subdivided into 4 stages: vesicular, verrucous, hyperpigmented, and atrophic.

Approximately 80% patients with IP are associated with several systemic abnormalities such as CNS, CVS, eyes, hair, nails and teeth. Vertex alopecia is the most common hair manifestation. Subungal keratotic tumor and nail dystrophy may appear at a later stage of IP. The most common abnormality of teeth is partial anodontia or absence of teeth. Ocular findings include strabismus, optic nerve atrophy, foveal hypoplasia, avascular retina, retina hemorrhage and vitreous hemorrhage. Clinical manifestations of CNS include infantile spasms, seizure, spastic paralysis and motor retardation. Cardiac abnormalities, such as VSD, PDA and tricuspid insufficiency, have been described. However, these associated developmental defects may not regress and may result in a less favorable outcome. Our case had PDA, VSD, dysgenesis of corpus callosum and agenesis of toes.

Histopathologic findings of the IP, stage 1 exhibit spongiotic dermatitis with massive intraepidermal and dermal eosinophilia, as well as eosinophil-filled intraepidermal vesicles. Multiple dyskeratotic cells can be appreciated in the epidermis. The verrucous stage is characterized by papillomas formed by epidermal hyperplasia with a growing number of dyskeratotic cells and hyperkeratosis. Melanin deposition in the melanophages of a thickened papillary dermis, leading to pigment incontinence is the most common finding during stage 3. The histopathologic findings of stage 4 presents as atrophic epidermis with a loss of rete ridges and dermal sweat coils.

More recently, the gene for incontinentia pigmenti has been mapped to Xq28 and mutations in the *NEMO/IKKγ* gene located at Xq28 have been found to cause expression of the disease. Eosinophil recruitment through eotaxin release by activated keratinocytes and endothelial cells of dermal vessels was described.

The first stage IP may mimic other blistering diseases of the newborn. Infectious diseases, such as varicella, herpes simplex and bullous impetigo can be differentiated from IP by means of bacterial, viral cultures and Tzanck smear. Bullous drug eruption may be associated with a period of drug history. Immune-mediated bullous dermatoses, such as bullous pemphigoid, neonatal pemphigus vulgaris, linear IgA dermatosis, dermatitis herpetiformis, and epidermolysis bullosa acquisita have characteristic pathological and immunofluorescence findings. In addition, bullous mastocytosis and Langerhans cell histiocytosis should be ruled out by skin biopsy.

The skin lesions of IP can have spontaneous improvement and resolution. Further management include preventing bacterial superinfection, follow-up ophthalmologic examinations, complete neurologic examination, radiologic studies, referral for radiologic evaluation and dental intervention by the age of 2 years.

**REFERENCES**