

Incontinentia Pigmenti in the First Year of Life

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The authors report the case of a 1-week-old full-term girl presenting a non-symmetrical blaschkoid vesiculobullous dermatosis located on the head, trunk as well as upper and lower limbs (Fig. 1). Her mother recalled a personal history of scarce vesicles in the first months of life, and, on examination, she exhibited linear hypopigmented macules on her lower limbs. A cutaneous biopsy showed superficial dermis with perivascular and mononuclear infiltrates, numerous isolated apoptosis of epidermal cell and eosinophilic exocytosis, confirming the clinical diagnosis of *incontinentia pigmenti*. Two months later, some of the lesions evolved into hyperkeratotic verrucous papules and later to hyperpigmentation along Blaschko lines (Fig. 2). The patient, currently 28 months old, has right eye esotropia with no other neurologic symptoms to date.

Incontinentia pigmenti is a rare genodermatosis due to a mutation of the X-linked *IKBKG* gene that is usually lethal in males, thus affecting mostly girls. The clinical characteristics include the distribution of skin abnormalities along Blaschko lines and four different sequential stages. These are the sole major diagnostic criteria, which are found in most patients. Stage I (Fig. 1), a vesiculobullous eruption, usually appears in the neonatal period, evolving to the verrucous stage (stage II, Fig. 2) within months to years, followed by the hyperpigmented stage (stage III, Fig. 2), and finally the hypopigmented stage (stage IV), which persists throughout life.

Incontinentia pigmenti can affect not only the skin but also other ectodermal derivatives (hair, fingernails, and teeth). Skin alterations are invaluable to the diagnosis of this multisystemic disease. Stages I and II appear in 99% and 96.6% of cases, respectively, before the first year of life, thus enabling early recognition. Central nervous system and ocular alterations determine the prognosis of the disease and should promptly be sought by a multidisciplinary approach including dermatologists, pediatricians, neurologists, ophthalmologists, and dentists.



Figure 1. Vesicles and bullae linearly distributed on both lower limbs.



Figure 2. Verrucous papules over the dorsal aspect of the right hand and hyperpigmentation following the lines of Blaschko along the torso.

Keywords: Infant, Newborn, Incontinentia Pigmenti

WHAT THIS REPORT ADDS

- *Incontinentia pigmenti* has a typical presentation following four different stages, with stage I appearing before the first year of life in 99% of all cases.
- The early recognition of this disease is very important because *incontinentia pigmenti* can affect the central nervous system in 30% of children, most commonly with seizures and a development delay.
- Patients with *incontinentia pigmenti* should be evaluated by a multidisciplinary team and undergo genetic counseling.

Conflicts of Interest

The authors declare that there were no conflicts of interest in conducting this work.

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Confidentiality of data

The authors declare that they have followed the protocols of their work centre on the publication of patient data.

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