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Case Report

Incontinentia Pigmenti: An Iranian Case Report

Shirin Mohamadi[1*], Fahimeh Abdollahimajd[2], Soheila Sotoudeh[3]

1.Division of Neonatology, Department of Pediatrics, Shohadaye Tajrish Hospital, Shahid Beheshti University of Medical Sciences, Tehran, Iran

2. Department of Dermatology, Shohada-e Tajrish Hospital, Shahid Beheshti University of Medical Sciences, Tehran, Iran

3. Department of Dermatology, Center of Excellence, Children's Medical Center Hospital, Tehran University of Medical Sciences, Tehran, Iran

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Abstract

Incontinentia Pigmenti (IP) is a rare X-linked dominant genetic disorder primarily affecting the skin, with cutaneous manifestations present in all cases. Other ectodermal tissues—including the central nervous system, eyes, hair, nails, and teeth—may also be involved to varying degrees. In this report, we present the case of a newborn female who exhibited widespread vesiculopustular skin lesions at birth, predominantly affecting the upper and lower extremities. At 9 hours old, the newborn with skin lesions suspicious for generalized impetigo, transferred to the NICU at Children's Medical Center in Tehran, Iran, for further evaluation and management. No abnormalities were observed in the hair, nails, oral mucosa, eyes, or central nervous system during the initial assessment and the final diagnosis was IP.

Keywords: Incontinentia Pigmenti; Neonatal Vesiculopustular Lesions; Neonatal Impetigo; Skin Diseases

***Corresponding Author:** Shirin Mohamadi, MD
Division of Neonatology, Department of Pediatrics, Shohadaye Tajrish Hospital, Shahid Beheshti University of Medical Sciences, Tehran, Iran

E-mail: dr.sh.mohamadi.ped@gmail.com

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Introduction

Incontinentia pigmenti (IP) is a rare X-linked dominant genetic disorder (1). It is a multisystem disease characterized by abnormalities in tissues and organs derived embryologically from the ectoderm and neuroectoderm (2). In healthy individuals, melanin pigment is confined to melanocytes in the basal layer of the epidermis; however, in IP, melanin is aberrantly distributed in the superficial layers of the skin (3).

In addition to the skin, other structures originating from the neuroectoderm may also be affected, including the eyes, central nervous system, teeth, hair, and nails (4). Clinical manifestations encompass a broad range of cutaneous, neurological, ocular, and dental abnormalities (5). The condition predominantly affects female neonates, as it is typically lethal in males due to its X-linked dominant inheritance pattern (3, 6, 7). Classically, the evolution of skin lesions in incontinentia pigmenti (IP) progresses through four distinct stages: vesicular rash, verrucous lesions, hyperpigmented macules, and finally, hypopigmented macules (3, 6).

The first stage, known as the vesiculobullous stage, typically manifests within the first two weeks of life. It is characterized by erythematous streaks, plaques, pustules, or vesicles. Peripheral eosinophilia is observed in approximately 65% of cases during this stage (8).

The second stage involves the development of hyperkeratotic, warty lesions on an erythematous base, most commonly affecting the extremities (9). The third stage, hyperpigmentation, is considered the hallmark of IP. It usually appears between 3 and 6 months of age and presents as asymmetrically distributed hyperpigmented macules following Blaschko's lines (10).

The final stage, hypopigmentation, is typically permanent and manifests as hairless, anhidrotic patches or streaks—often with or without atrophy—commonly seen on the flexor surfaces of the lower legs (5).

We report a case of IP in a female neonate with no positive family history, underscoring the importance of considering IP in the differential diagnosis of neonatal vesiculobullous or neonatal impetigo-like skin lesions, given the rarity and multisystem involvement of this condition.

Case Presentation

The case is a female neonate, the second child of non-consanguineous parents, with no significant family history of genetic or dermatologic disorders. She was born at term (39 weeks of gestation) via normal vaginal delivery (NVD), with a birth weight of 3,140 grams, a head circumference of 36 cm, and an Apgar score of 9. At birth, the infant presented with widespread vesiculopustular skin lesions, predominantly affecting the upper and lower extremities. On clinical examination performed 9 hours after birth, signs of superinfection of the lesions were noted. As a result, the neonate was transferred to the Neonatal Intensive Care Unit (NICU) at Children's Medical Center, Tehran, Iran, for further evaluation and management. On physical examination, Vital signs were stable: heart rate 140/min, respiratory rate 50/min, temperature 37.1°C, and oxygen saturation 97%. In skin examination, the neonate exhibited linear vesicular and pustular plaques on both upper and lower extremities, along with widespread vesicles on the trunk, some of which showed early hyperpigmentation **(Figure 1)**. Several lesions

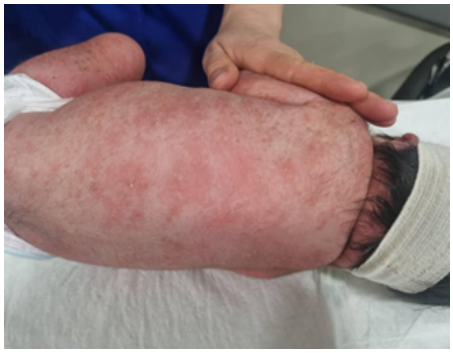

Figure 1. Linear vesicular and pustular plaques on both upper and lower extremities

appeared superinfected, with features resembling impetigo. Additionally, salmon patches were noted on the upper eyelids and occiput.

Examination of the head revealed normal cranial sutures and fontanelles. Cardiopulmonary auscultation was unremarkable, and both the chest and abdomen appeared normal. The infant had normal female external genitalia. Neurological assessment, including sensorimotor function and extremity reflexes, was within normal limits. A summary of the initial laboratory findings is provided in Table 1, which revealed marked eosinophilia (905 cells/μL). A chest X-ray was unremarkable **(Figure 2)**. The results of smear and culture of skin lesions were negative. Blood culture was also negative.

Following dermatology consultation, a clinical diagnosis of incontinentia pigmenti (IP) was suggested.

A skin biopsy was performed to support the clinical diagnosis. Histopathological examination revealed findings consistent with incontinentia pigmenti, including eosinophilic spongiosis, intraepidermal vesicles, and pigment incontinence.

The treatment plan included topical care and supportive measures: application of zinc sulfate 1: 1000 compresses diluted in normal saline on the vesicular skin lesions twice daily for 10 minutes each session, followed by zinc oxide ointment on old and crusted lesions. Regular skin hydration with Eucerin bulk was also initiated. An ophthalmologic evaluation and neurological consultation were also requested to assess for potential extracutaneous involvement, both of which returned unremarkable results. The neonate was discharged on the 7th day of life in stable condition, with improved skin lesions and stable vital signs, continuing the prescribed treatment plan on an outpatient basis.

Follow-up appointments were strongly recommended at the neonatal, dermatology, ophthalmology, and neurology clinics. Additionally, genetic testing was advised to confirm the diagnosis of incontinentia pigmenti. **Figure 3A** and **3B** show the patient's 1-month and 6-month follow-ups.

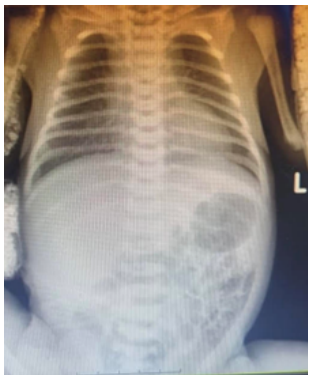

Figure 2. A chest X-ray of patient

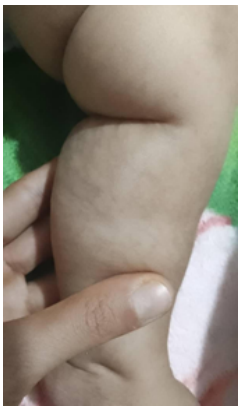

Figure 3A

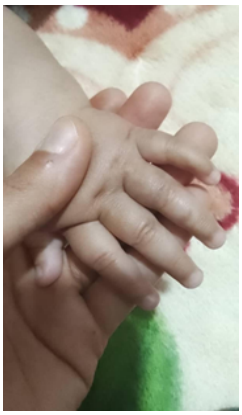

Figure 3B

Figure 3. The patient's 1-month and 6-months follow-up

Discussion

Incontinentia pigmenti (IP) is most commonly inherited in an X-linked dominant manner, which accounts for its strong female predominance—over 95% of affected individuals are female infants (11). This inheritance pattern is typically lethal in males in utero, due to the absence of a second X chromosome to compensate for the mutation.

The cutaneous manifestations are the hallmark of IP and serve as the primary diagnostic criterion. These skin changes typically begin at birth or within the first few weeks of life and may evolve through the characteristic four stages into adulthood (12). In our patient, the presence of widespread vesiculopustular lesions with a linear distribution, particularly on the upper and lower extremities, was the predominant clinical finding, consistent with the first stage of IP.

Similar findings have been reported in the literature. For example, Gray-brown hyperpigmentation was observed on the face, arms, neck, and trunk in the case reported by Rafaatjo et al. (13), illustrating the later hyperpigmented stage of IP. In contrast, Khatami et al. described a familial case of IP in which four family members had a positive history of vesiculopustular skin lesions (2). However, in our patient, there was no reported family history of similar lesions.

Family history can play a crucial role in supporting the diagnosis of IP, although its absence does not exclude the disease, as sporadic cases are also well documented.

Eosinophilia, reported in as many as 65% of IP cases, has been recognized as a major diagnostic criterion; however, it is not pathognomonic and serves primarily as a supportive finding (14). In a study by Hadj-Rabia et al., 23 out of 26 patients exhibited peripheral eosinophil counts ranging from 550 to 15,400 cells/μL (15). Similarly, Poziomczyk et al. reported eosinophilia in 50% of their patients who underwent this test (4). In another case reported by Khatami et al., mild eosinophilia of 18% was noted (2).

Consistent with these findings, laboratory analysis of our newborn, performed within hours after birth, revealed eosinophilia with an absolute count of 905 cells/μL. Neurological manifestations are observed in approximately 13–35% of patients with incontinentia pigmenti (14). Seizures are the most common neurological symptom, occurring in about 40% of affected individuals, followed by motor impairments and intellectual disability (15). However, in the study by Poziomczyk et al., none of the patients exhibited intellectual disability (4). Similarly, in the case reported by Rafatjoo et al., the patient showed no neurological symptoms such as seizures, developmental delay, intellectual disability, ataxia, or motor dysfunction (13). Likewise, our patient had no neurological abnormalities on examination but was referred to a neurologist for further evaluation.

Conclusion

Skin abnormalities are the hallmark and most prominent manifestation of incontinentia pigmenti (IP). Diagnosis is primarily clinical, and while the skin lesions themselves typically do not require specific treatment, the overall prognosis depends largely on the involvement of other organ systems. Given that IP is a multisystem disorder with the potential for serious ocular and neurological complications, a multidisciplinary approach involving dermatologists, neurologists, and ophthalmologists is crucial. Early intervention and lifelong follow-up are essential to monitor and manage potential complications, improve patient outcomes, and enhance understanding of this complex disease.

Conflicts of Interest

The authors declare that there are no conflicts of interest related to this study.

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