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Case report

Incontinentia Pigmenti (IP) in Late Preterm Infant

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Abstract
Incontinentia pigmenti (IP) is a rare X-linked dominant neurocutaneous disorder that predominantly affects females and is often lethal in males. It is caused by mutations in the IKBKG (NEMO) gene, leading to dysregulation of NF-κB signalling and multisystem involvement. We report the case of a female infant born preterm at 34+6 weeks to a mother with type 2 diabetes and hypothyroidism. The infant presented at birth with vesiculobullous skin lesions that evolved into pustular and crusted eruptions along Blaschko lines, consistent with IP. A family history revealed two older sisters with similar cutaneous findings in later stages, strengthening diagnostic suspicion. The neonatal course was complicated by hypoglycaemia requiring intensive glucose therapy and subsequent hypernatremic dehydration managed with careful fluid balance. Empirical antibiotics were initiated for presumed sepsis but discontinued after negative cultures. Dermatology review, genetic counselling, and multidisciplinary follow-up were arranged. This case highlights the diagnostic challenges of early IP presentations, which may mimic neonatal infection, and underscores the importance of recognising Blaschko line distribution and eliciting family history. Vigilant monitoring for metabolic disturbances and systemic complications, alongside coordinated multidisciplinary care, is essential to optimise outcomes in affected neonates.
Keywords. *Incontinentia Pigmenti, Preterm Infant, Case Report.*

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Introduction

Incontinentia Pigmenti is a rare X-linked dominant neurocutaneous disorder that primarily affects females and is lethal in most males, with an estimated prevalence at birth of 0.7/100,000 [1]. It is a skin condition that is caused by genetic abnormalities in the epidermis and follows a distribution pattern representing the migratory pathway of epidermal cells during embryonic development. This distribution pattern is known as Blaschko lines, which are characteristic of a group of conditions that follow patterns of cutaneous mosaicism [2]. Incontinentia Pigmenti is characterized by four cutaneous stages: vesicular, verrucous, hyperpigmented, and atrophic, although not all stages are present in every patient [3]. The condition is caused by mutations in the *IKBKG* (NEMO) gene, which regulates the NF-κB signalling pathway involved in inflammation, cell survival, and immune responses (4). Dysregulation of NF-κB signalling has been implicated in a variety of human diseases. It is a multisystem disorder that can be associated with defects related to the heart, teeth, skeletal system, eyes, and central nervous system. Therefore, early recognition, particularly in neonates, along with multidisciplinary follow-up, is essential [5]. It is also recommended to monitor for potential neurologic and ophthalmologic complications of Incontinentia pigmenti [3].

Case presentation

A female infant was born at 34 weeks and 6 days of gestation with a birth weight of 3.69 kg. The mother had insulin-treated type 2 diabetes and hypothyroidism. She received two doses of antenatal corticosteroids, and no antenatal risk factors for infection were identified. The baby was initially stable but developed grunting and increased work of breathing (suggestive of respiratory distress) at two hours of life, necessitating admission to the neonatal unit for respiratory support. Empirical antibiotics were commenced for presumed sepsis. Respiratory distress resolved within 24 hours, and antibiotics were discontinued after negative cultures and improwas ving inflammatory markers. Significant hypoglycaemia was noted, requiring escalating intravenous glucose concentrations (up to 15%) and glucagon infusion (10mcg/kg/hour). Normoglycaemia was eventually achieved, and the infant was transitioned to enteral formula feeds.
A widespread blistering rash noted at birth (Figure 1) progressed over the first few days of life into pustular and crusted lesions (Figure 2), prompting a dermatology review.

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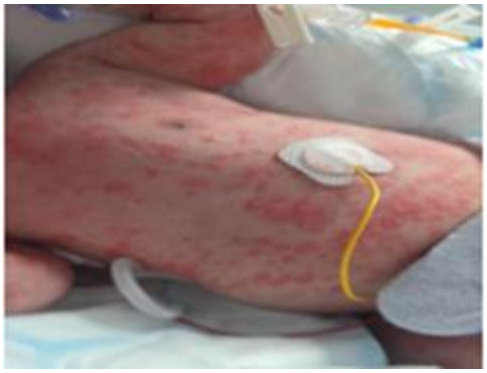

Figure 1. Vesiculobullous rash lesions at birth

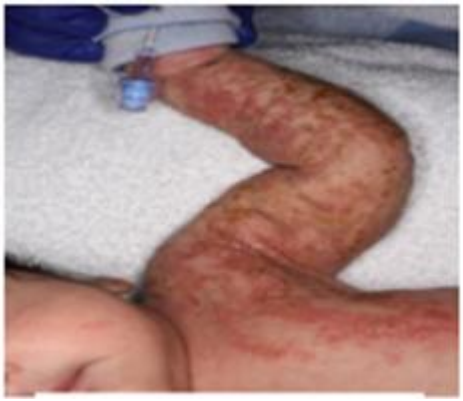

Figure 2. Pustular and crusted lesions by day 3

A diagnosis of Incontinentia pigmenti was made based on the clinical presentation of the rash following Blaschko lines, along the abdominal region(Figure 3) . As the lesions were following narrow bands of Blaschko, this aided in differentiating this condition from other conditions that present with patterns of cutaneous mosaicism (Figure 4).

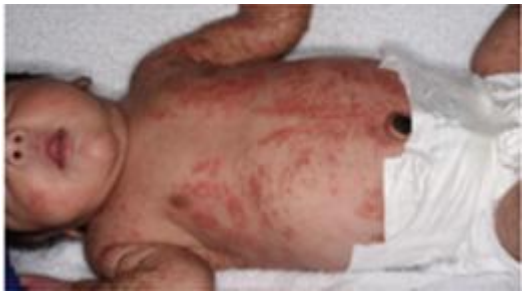

Figure 3. Lesions following Blaschko lines over the abdomen and chest

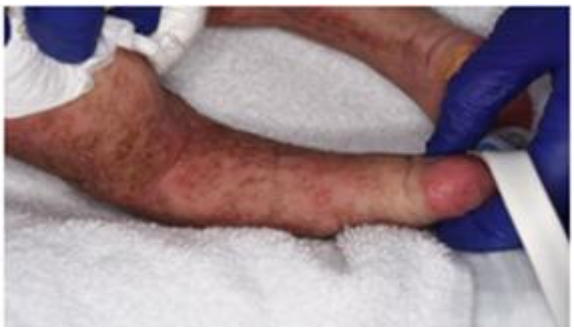

Figure 4. Pattern of cutaneous mosaicism forming narrow linear bands

It was also noted prior to diagnosis that the patient's two older sisters exhibited similar skin findings, except that they currently have atrophic hypopigmented streaks (a later stage of Incontinentia Pigmenti). The patient has a brother who did not inherit the skin condition. This family history, along with the developing pattern of the skin lesions, was crucial in identifying this rare skin condition.

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On day 10 of life, the infant developed severe dehydration and hypernatremia. Intensive fluid therapy was initiated, which included careful fluid balance, daily weights, and serum electrolyte monitoring. The infant responded well to treatment and was discharged on day 15 of life with dermatology follow-up, local neonatology follow-up, and genetics counselling in place. The infant underwent serial blood glucose monitoring, inflammatory marker assessment, including C-reactive protein and blood cultures, and serum electrolyte analysis. Dermatological assessment with clinical photography was performed to document the progression and distribution of skin lesions. Further evaluation included genetic testing and a metabolic screen to exclude underlying metabolic or inherited disorders.
Empirical intravenous antibiotics were commenced initially for presumed neonatal sepsis and discontinued once blood cultures were negative and inflammatory markers improved. . Supportive skin care with regular emollients was instituted, and ongoing dermatology follow-up was arranged. The infant stabilised clinically and was discharged home on day 15 of life with improving skin lesions and no further complications identified at the time of discharge (Figure 5).

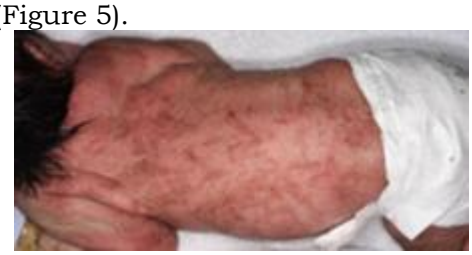

Figure 5. Improving and healing of skin lesions at day 15

Discussion

Incontinentia pigmenti (IP) is a rare X-linked dominant neurocutaneous disorder that predominantly affects females and results from mutations in the *IKBKG* (NEMO) gene, which plays a critical role in the NF-κB signalling pathway involved in cell survival and immune regulation. Incontinentia pigmenti (IP) is an important cause of vesiculobullous and pustular eruptions in the newborn. They emphasise that IP often presents at birth or within the first days of life, commonly during the vesicular/vesiculopustular stage [6], exactly as seen in this patient. The condition primarily affects ectodermal tissues, including the skin, eyes, central nervous system, and dentition. Although classically regarded as a dermatological condition, IP is a multisystem disorder with potentially significant neurological and ophthalmological sequelae, underscoring the importance of early recognition and appropriate follow-up. Early neonatal presentations may be diagnostically challenging, as initial cutaneous manifestations can be subtle or atypical and are frequently mistaken for neonatal infection or inflammatory pustular dermatoses.
This case is particularly instructive because the infant presented during the early vesiculopustular stage of IP, prior to progression to the more recognisable hyperpigmented or atrophic phases. At this stage, the rash may closely resemble neonatal sepsis-related skin changes, herpes simplex infection, bullous impetigo, or transient neonatal pustular conditions, often leading to empiric antibiotic treatment and delayed dermatological diagnosis. Careful clinical observation revealed a linear and whorled distribution of lesions along Blaschko lines, a hallmark of cutaneous mosaicism that reflects the embryonic migration of epidermal cells. Recognition of this characteristic pattern was central to establishing the diagnosis and differentiating IP from other neonatal blistering disorders [7].
A detailed family history further strengthened the diagnostic suspicion, revealing similar cutaneous findings in two older female siblings who had progressed to later-stage hypopigmented and atrophic streaks [8,9]. This inheritance pattern is consistent with the X-linked dominant nature of IP and highlights the importance of eliciting a thorough family history when evaluating neonatal dermatological presentations. Similar neonatal presentations have been described in published case series [3], who reported that many affected patients present in the neonatal period with vesiculobullous eruptions that evolve through the classic stages of IP [10], further emphasised the need for early recognition of IP due to the risk of associated neurological and ophthalmological complications, which may not be apparent at birth but can have long-term consequences.
In addition to the dermatological findings, this case was complicated by significant metabolic disturbances, including hypoglycaemia and hypernatremic dehydration, both of which are highly relevant to neonatal practice. As an infant of a diabetic mother, this patient was at increased risk of hypoglycaemia due to fetal hyperinsulinaemia, necessitating intensive glucose management. The subsequent development of hypernatremic dehydration likely reflected a combination of factors, including feeding difficulties and increased insensible fluid losses related to extensive skin barrier disruption. Neonates with blistering or inflammatory skin conditions are particularly vulnerable to fluid and electrolyte imbalance, and this case reinforces the importance of vigilant fluid monitoring, daily weight assessment, and early recognition of dehydration in such patients.

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This case also underscores the critical role of early genetic counselling in families with known or suspected inherited conditions. Despite clinical features suggestive of IP in two older siblings, the family had not previously received formal genetic evaluation or counselling. Earlier recognition could have enabled anticipatory guidance, informed family planning, and early surveillance for potential systemic complications. Genetic counselling should therefore be offered proactively whenever an inherited disorder is suspected, particularly in families with multiple affected children or characteristic patterns of inheritance.
Early multidisciplinary involvement was pivotal to the successful management of this infant. Collaboration between neonatology, dermatology, and genetics facilitated timely diagnosis, appropriate metabolic management, and coordinated follow-up planning. This case highlights the broader principle that dermatological findings in neonates may provide essential diagnostic clues to rare but clinically significant multisystem conditions. Maintaining a high index of suspicion, particularly when rashes follow patterned distributions or are accompanied by a suggestive family history, can lead to earlier diagnosis, targeted management, and improved outcomes for affected infants and their families.

Conclusion

This case highlights that Incontinentia pigmenti may present subtly at birth and evolve over several days into vesiculopustular and crusted lesions, a pattern that differs from its more recognisable presentation in later childhood and may delay diagnosis. A detailed family history proved to be a powerful diagnostic tool, as enquiring about similar skin findings in siblings or parents can reveal previously unrecognised inherited conditions, particularly in X-linked disorders. The development of hypernatremic dehydration in this infant underscores the vulnerability of hospitalised neonates with skin barrier dysfunction and the importance of meticulous fluid balance monitoring, including daily weight measurements. Accurate recognition of dermatological findings across diverse skin tones is essential to ensure timely diagnosis and equitable care. Early and accurate identification of IP allows for timely genetic counselling, which is critical for family planning, anticipatory guidance, and psychological support; this case also reflects a missed opportunity for earlier counselling following affected siblings. Finally, coordinated multidisciplinary input involving neonatology, dermatology, genetics, and endocrinology is key to optimising outcomes in neonates with complex multisystem conditions.

Conflict of interest. Nil

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