CASE REPORT

Incontinence pigmenti: A rare genetic disease that affects the skin and more

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ABSTRACT

Incontinentia Pigmenti (IP) is a rare X-linked genetic disorder primarily affecting girls, while it is typically lethal in males. The disease is characterized by variable clinical manifestations affecting the skin, hair, nails, teeth, and central nervous system. It is associated with mutations in the NEMO gene, which impact the function of the NF-kappa-B transcription factor. There is no specific cure, and treatment focuses on symptom and complication management. Early diagnosis is crucial for proper management.

INTRODUCTION

Incontinentia pigmenti, also known as Bloch-Sulzberger syndrome, was first described by Garrod in 1906 and later defined by Bloch (1926) and Sulzberger (1928)[1] as a rare genetic disorder that primarily affects females.[2,3] It is characterized by changes in skin pigmentation, dental issues, nail and hair development abnormalities, as well as ocular and neurological involvement. Although this is an uncommon disease, understanding its symptoms and characteristics is crucial for early diagnosis, which begins at birth and progresses through four stages: vesiculopustular, verrucous, hyperpigmentation, and hypopigmentation phases.[4] These phases can overlap or go unnoticed.

The disease is caused by mutations in the IKBKG gene,[4] also known as the NEMO gene, located on the X chromosome. Because it is an X-linked condition, it typically affects females, while males are rarely affected and may present with milder symptoms.[5] The genetic mutation impacts the production and function of a protein called tumor necrosis factor (TNF), which plays a crucial role in the development and maintenance of skin and other tissues.[2,4]

Diagnosis of incontinentia pigmenti is primarily based on clinical and histopathological evaluation, with genetic testing reserved for uncertain cases.[5]

CASE REPORT

A 2-year-old female presented for a routine check-up, with a personal history of neonatal toxic erythema and seizures at birth. She was born at term via cesarean section, with appropriate weight for gestational age. The mother, a healthy 35-year-old, reported a well-monitored pregnancy.

The patient first presented at 2 months old, with the parents reporting that since birth, she had skin lesions

characterized by papules that later evolved into vesicles following a linear pattern on her upper and lower limbs. She had no fever.

Physical examination revealed localized dermatosis on the inner side of the left arm, thighs, legs, and the sole of the foot, characterized by erythematous vesicular lesions containing serous fluid, some of which were denuded, along with a few keratotic papules arranged in a linear pattern (Blaschko's lines) (Figure 1a, 1b, 1c).

Neurological examination was normal, and the rest of the physical examination showed no abnormalities. A skin lesion biopsy was performed on the right leg, with histopathological results revealing: acanthotic epidermis with hyperkeratosis and numerous dyskeratotic cells in the upper third of the epithelium with adjacent horn swirl formation (Figure 2).

PROGRESSION

At the age of 2 (current age), the patient returned for a follow-up visit accompanied by her parents, showing only a single verrucous papule approximately 2mm in diameter on the left anterior thorax (Figure 3).

DISCUSSION

From a dermatological perspective, the characteristic skin lesions of incontinentia pigmenti typically manifest in early childhood and may progress over time, necessitating continuous monitoring and an individualized therapeutic approach. There is currently no cure for incontinentia pigmenti, so treatment focuses on symptom management and supportive care.[5,7] Treatment options vary depending on the specific symptoms each person experiences. A multidisciplinary

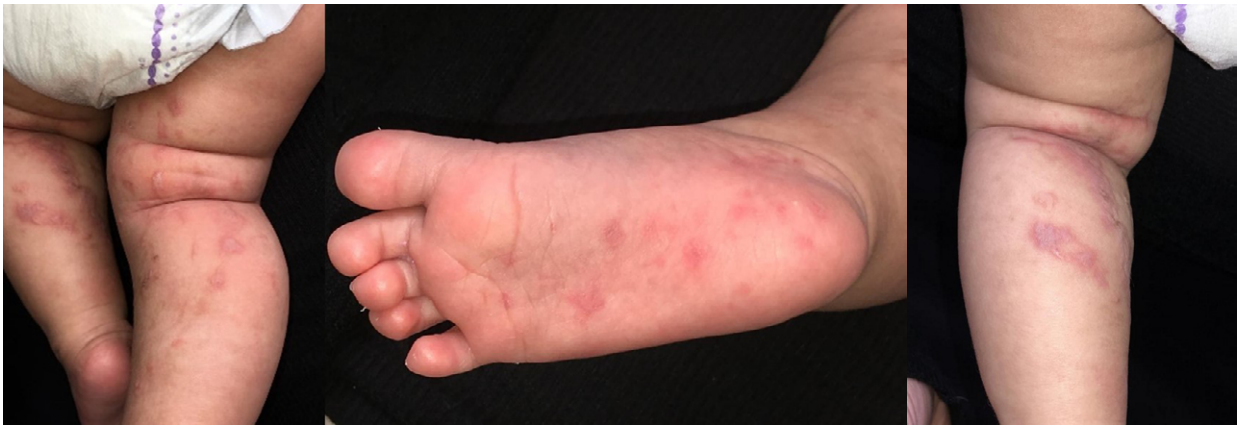

Figure 1 a,b,c: Erythematous vesicular lesions with serous fluid, some denuded, and a few keratotic papules following a linear pattern (Blaschko's lines).

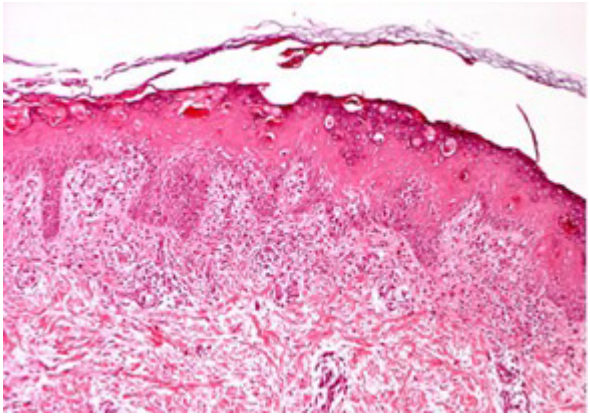

Figure 2. Acanthotic epidermis with hyperkeratosis and numerous dyskeratotic cells in the upper third of the epithelium, with adjacent horn swirl formation.

Figure 3: Verrucous papule approximately 2mm on the left anterior thorax.

healthcare team, including dermatologists, ophthalmologists, dentists, and neurologists, can work together to provide comprehensive.[8] Our patient was managed by a multidisciplinary team due to the occurrence of seizure episodes at birth and presented at two months old to our clinic with skin lesions. These lesions were treated with topical agents, and at the two-year follow-up, her skin condition showed significant improvement.

Treatment for skin issues may include the use of creams or ointments to reduce irritation and keep the skin hydrated.[9] In some cases, surgery may be necessary to address cosmetic effects or correct dental anomalies. Eye problems may require glasses or contact lenses, cataract surgery, or treatment for specific eye-related complications.[10,11] For neurological disorders, treatment may involve physical and occupational therapy, as well as antiepileptic medications to control seizures.[10]

FOLLOW-UP

It is essential for patients with pigmentary incontinence to receive regular medical follow-ups to monitor disease progression and address any potential complications.

CONSENT

Informed consent was obtained from the patient's representative for the publication of data and images.

CONCLUSION

Incontinentia pigmenti is a rare disease that presents challenges for both patients and healthcare professionals. Due to its genetic nature, genetic counseling and psychological support are important aspects of managing this condition. Patients and their families can benefit from education about the disease, access to support groups, and connection with specialized organizations.

While incontinentia pigmenti can significantly impact the quality of life, it is essential to note that symptoms often improve over time. With appropriate support and comprehensive symptom management, many individuals with incontinentia pigmenti can lead fulfilling and satisfying lives.

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