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BLOCH-SULZBERGER SYNDROME: A RARE GENODERMATOSIS - A CASE REPORT

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ABSTRACT Incontinentia pigmenti (IP), also known as Bloch-Sulzberger syndrome, is a rare X-linked dominant genodermatosis affecting primarily females. It presents with characteristic stages of cutaneous manifestations and may involve the eyes, central nervous system, teeth, and hair. This report highlights a neonatal case presenting with vesiculobullous lesions in a blaschkoid distribution, along with maternal cutaneous stigmata and relevant family history. Early recognition and multidisciplinary intervention are essential for improving prognosis and preventing systemic complications.

KEYWORDS : Incontinentia pigmenti, Bloch-Sulzberger syndrome, neonatal dermatoses, genodermatosis, vesiculobullous eruption, X-linked dominant

INTRODUCTION

Incontinentia pigmenti (IP) is a rare X-linked dominant neurocutaneous syndrome predominantly affecting females. The condition is caused by mutations in the **IKBKG gene** (formerly NEMO) located on **Xq28**, responsible for regulating NF-kB pathway activation, critical in inflammation and apoptosis control [1].

IP typically presents in **four cutaneous stages**: vesicular, verrucous, hyperpigmented, and hypopigmented/atrophic. It is associated with **multisystem involvement**, affecting eyes (retinal detachment), CNS (seizures, developmental delay), teeth (hypodontia), and musculoskeletal system (limb anomalies). Male fetuses are often nonviable, leading to spontaneous abortion, making its diagnosis critical for genetic counseling [2].

We present a **rare case of neonatal IP**, with classic vesiculobullous lesions and maternal pigmentary changes, emphasizing the importance of clinical suspicion and genetic insight.

Case Description

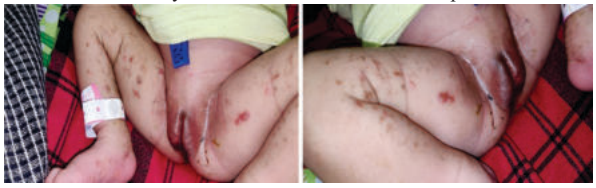
A **5-day-old female neonate**, born at term via normal vaginal delivery to a non-consanguineous couple, presented with **multiple fluid-filled lesions** over the **groin and lower limbs** noted since day two of life.

Maternal History

- Mother had a **history of similar neonatal lesions** and residual pigmentary changes.
- She also reported a **spontaneous abortion** during her first pregnancy.

Cutaneous Examination (Neonate)

- **Multiple vesicles and bullae** on an erythematous base over **bilateral groins and lower limbs**.
- **Blaschkoid arrangement** of lesions with few erythematous plaques.
- No mucosal or systemic involvement observed at presentation.



Cutaneous Examination (Mother)

- **Linear hypopigmented macules** over the **trunk and back**, suggestive of resolved stage 4 of IP.

Investigations And Differential Diagnosis

Differential Diagnoses Considered:

- Incontinentia pigmenti
- Neonatal herpes simplex
- Bullous impetigo
- Epidermolysis bullosa

Tzanck smear and **Gram staining** were negative for acantholytic cells and bacteria, respectively. No family history of immunodeficiency or vesiculobullous disorders.

Given the clinical staging, maternal findings, and family history, a **provisional diagnosis of IP** was made. Genetic testing was advised, and the child was placed under **multidisciplinary evaluation** (dermatology, ophthalmology, neurology, and dentistry).

DISCUSSION

IP is a rare **X-linked dominant disorder** occurring almost exclusively in females. It results from **IKBKG gene mutation**, leading to apoptosis of ectodermal cells in male fetuses (often lethal) and dysregulated NF-kB activation in surviving females [3].

Cutaneous Stages Of IP:

Stage	Clinical Features	Onset
I – Vesicular	Linear vesicles on erythematous base	Birth to 2 weeks
II – Verrucous	Warty papules in linear pattern	2–6 weeks
III – Hyperpigmented	Swirls/whorls of pigmentation	Months 1–6
IV – Hypopigmented	Atrophic or hypopigmented streaks	Late childhood/ adulthood

Cutaneous lesions follow the **lines of Blaschko**, correlating with mosaicism in embryonic development [4].

Systemic Involvement:

- **Ocular:** Retinal vascular anomalies, optic atrophy
- **CNS:** Seizures, spasticity, delayed milestones
- **Dental:** Hypodontia, delayed dentition
- **Hair/Nails:** Alopecia, nail dystrophy

Diagnosis is **clinical**, especially in neonates with maternal stigmata. **Genetic testing** confirms IKBKG mutation but is not mandatory in resource-limited settings. Early **ophthalmologic screening** is essential to prevent irreversible complications.

CONCLUSION

This case highlights the **classic neonatal presentation** of IP with **maternal cutaneous signs** and **positive obstetric history**, which are key to diagnosis. Due to its potential **multisystem complications**, prompt identification and **multidisciplinary care** are crucial. Genetic counseling is recommended to prevent recurrence and facilitate early interventions.

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