CASE REPORT

Family of Three Generations with Incontinentia Pigmenti

Johny Vakayil Francis[a], Naveen Thomas[a], Aruna Chandra Babu[a]

a. Department of Pediatrics and Neonatology, Medical Trust Hospital, Kochi, Kerala, India.

ARTICLE INFO

KEYWORDS

Incontinentia Pigmenti, Bullous Skin Lesion
Linear and Whorled Pigmentation
Genetic Disorders of Pigmentation
Blaschko's Lines

CORRESPONDING AUTHOR

Dr. Johny Vakayil Francis, MBBS, DCH, MD
Consultant Pediatrician and Neonatologist and Head
Department of Pediatrics, Medical Trust Hospital
Kochi, Kerala, India. 682016
Phon e: +919656332219 Email: drjohnyvf@yahoo.com

PUBLISHED ON

9th May 2021

DOI

https://doi.org/10.52314/fnb.2021.v1i1.8

CITE THIS ARTICLE AS

Francis JV, Thomas N, Babu AC. Family of Three Generations with Incontinentia Pigmenti. FNB. 2021 May 7;1(1):25–7.

AUTHOR INFORMATION

1. Dr. Johny Vakayil Francis, MBBS, DCH, MD, Consultant Pediatrician and Neonatologist and Head Department of Pediatrics, Medical Trust Hospital Kochi, Kerala, India.
2. Dr. Naveen Thomas, MBBS, MD, DNB Consultant Dermatologist, Department of Dermatology Medical Trust Hospital, Kochi, Kerala, India
3. Dr. Aruna Chandra Babu, MBBS, DNB Trainee in Pediatrics, Department of Pediatrics, Medical Trust Hospital Kochi, Kerala, India

CONFLICT OF INTEREST

None

FINANCIAL SUPPORT

Nil

Access this Article Online

Scan this QR Code

ABSTRACT

Incontinentia pigmenti, (IP) also known as Bloch-Sulzberger syndrome, is an uncommon X-linked dominantly inherited syndrome, characterized by various cutaneous manifestations which include vesicular, verrucous and pigmented lesion, associated with developmental defects of the central nervous system, skeletal system and the eye.[1] We are presenting a family of three generations with Incontinentia Pigmenti and a brief review of clinical updates.

A term female infant born by cesarian section to a fourth gravida mother with history of previous 3 abortions, presented with multiple linear vesiculobullous lesions over the upper and lower limbs. Mother previously had an ectopic pregnancy, followed by spontaneous abortion and a third twin pregnancy with cystic hygroma on anomaly scan which was medically terminated.Infant's mother, grandmother and maternal aunt also had similar lesions over the limbs and abdomen with healed rash presently. Maternal aunt also had a female infant born with similar skin lesion **(Figure 1 – Pedigree Chart).** There are no surviving male children in the family.

Child had vesicular / pustular lesions resembling bullous impetigo predominantly on the upper limb and lower limb **(Figure 2a & 2b).** There were similar lesions on the trunk as well. Lesions on the upper limb and lower limb were arranged in a linear pattern. Gram stain from the lesion did not reveal any organisms. Tzanck smear done revealed absence of any multi-nucleated giant cells. In view of the lesions following Blaschko's line and positive family history, a diagnosis

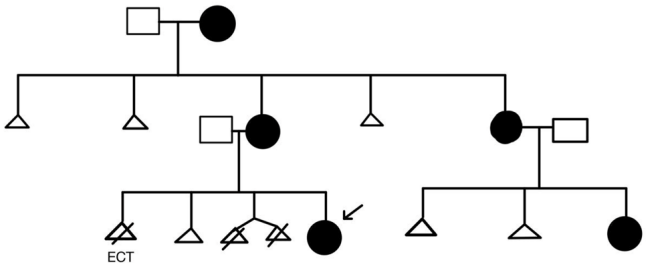

Figure 1. Pedigree Chart

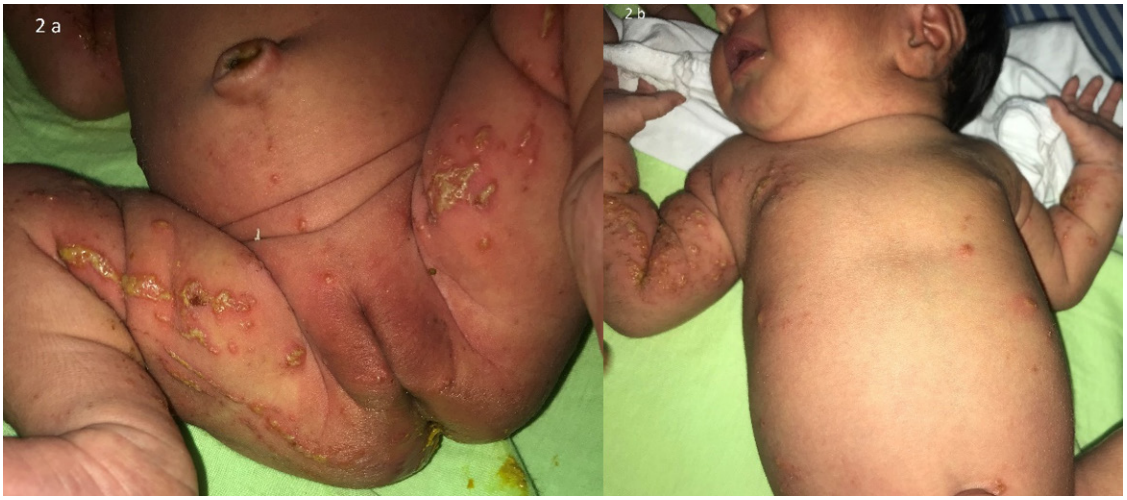

Figure 2. Vesiculo-pustular lesions along the Blaschko's line on the lower limb (2a) and the upper limb. Trunk is relatively spared with few lesions (2b)

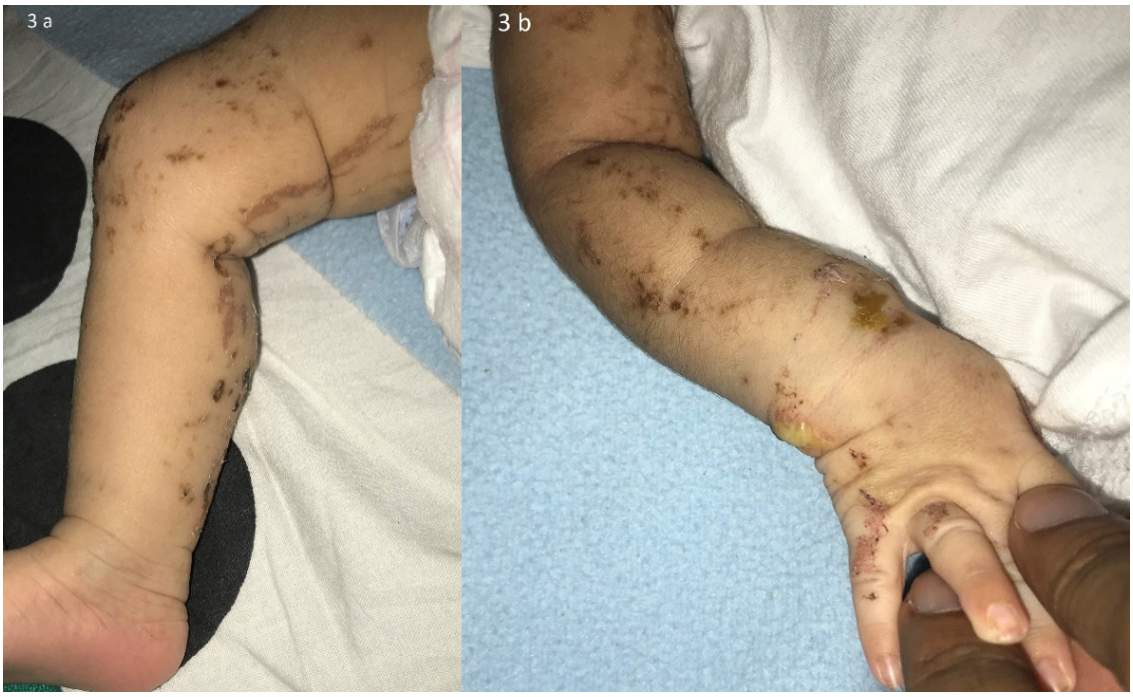

Figure 3a & 3 b. Hyperpigmented hyperkeratotic lesion on the lower limb (3a) and upper limb (3b) at 3 weeks of age corresponding with linear distribution of vesiculo-pustular lesions in the past.

of Incontinentia Pigmenti was made. Patient did not have any ocular or neurological abnormalities. Two weeks later lesions evolved to form hyperpigmented, hyperkeratotic plaques, arranged in a linear pattern following Blaschko's line corresponding to the distribution of the previous rash **(Figure 3a & 3 b).**

DISCUSSION

Incontinentia Pigmenti (IP) is a genodermatosis which presents as four stages of characteristic dermatological polymorphism namely vesiculobullous, verrucous, swirled hyperpigmentation and atretic stage.[2] IP is inherited as an X-linked dominant manner, predominantly affecting females and lethal to males. However there are few case reports of male babies affected with IP.[3] It is a single gene disorder caused by mutations in the NEMO/IKK-γ gene mapped at the distal part of Xq28.[4]

IP typically evolves through four stages.[5,6]

Stage I – **The bullous stage** is characterized by blister- like bullous eruptions that are linear in extremities and / or circumferential on trunk. The eruptions can be erythematous and may appear infectious. Stage I rash can present from birth to 8 weeks and disappears by 18 months.

Stage II – **The Verrucous stage** is characterized by a hypertrophic, wart-like rash that is linear on the extremities and rarely affects trunk or face. This stage is usually seen within the initial few months of life. It arises as stage I begins to resolve, however cases with verrucous stage since birth has also been documented. Stage II typically lasts for a few months, but it can even last for years. This stage also presents with dystrophic nails and abnormalities of tooth eruption.

Stage III – **The hyperpigmentation stage** is characterized by macular, slate gray or brown hyperpigmentation that occurs in a "marble cake" or swirled pattern along Blaschko's line. It is the most characteristic stage of IP. It is usually circum-

ferential on the trunk and linear on the extremities. It is more often apparent on the trunks than the limbs. The sites of involvement are not necessarily those of preceding vesicular and warty lesions. Axilla and groin are typically affected. Hyper pigmentation begins between age six months and one year, usually as stage II begins to resolve. It is not present at birth. Stage III can persist into adulthood.

Stage IV – **The Atretic stage** is characterized by linear hypopigmentation of the extremities and when scalp is affected, it results in alopecia. There may not be true hypopigmentation, but rather a loss of hair and epidermal glands. As with the first 3 stages, the pattern follows Blaschko's lines. Stage IV does not occur in all individuals. When present, it arises after the hyperpigmentation fades.

Differential diagnosis for each stage[7,8]

Stage I: Neonatal HSV, herpes zoster,congenital candidiasis, congenital syphilis, bullous impetigo, epidermolysis bullosa simplex, Letterer-Siwe disease, blistering drug eruption, bullous congenital ichthyosiformerythroderma, and neonatal dermatitis herpetiformis.

Stage II: Inflammatory linear verrucous epidermal nevus, lichen striatus, verruca vulgaris, linear psoriasis, linear lichen planus, linea dariers disease, linear porokeratosis

Stage III: Linear and whorled naevoid hypermelanosis, pigmentary mosaicism, Naegeli-Franceschetti Jadassohn syndrome, X linked dominant chondrodysplasia punctata

Stage IV: Hypomelanosis of Ito, other hypopigmentary disorder with localised alopecia, Goltz syndrome.

Diagnostic criteria[9]

Major Criteria: (1) typical neonatal vesicular rash with eosinophilia, (2) typical Blaschkoid hyperpigmentation on the trunk, fading in adolescence; and (3) linear, atrophic hairless lesions.

Minor criteria: (1) dental anomalies, (2) alopecia, (3) wooly hair, and (4) abnormal nails. The presence of any major criterion strongly supports the clinical diagnosis of IP if there is a definitive family history.

Our patient presented with vesiculo-pustular lesions corresponding with stage I of IP since birth, suggesting intrauterine origin. However, transition to stage II evolved in 2-3 weeks.Patient did not have any other neurological or ocular complication at the time of presentation. It is important to distinguish IP from numerous other conditions which can have similar presentation. A positive family history, female gender, loss of male sibling and typical skin lesions in Blaschko's distribution helped us to clinch the diagnosis in this child.

CONCLUSION

IP can have varied clinical presentation and progress. In some babies the disease may originate in intrauterine life and pose a diagnostic dilemma, however detailed history of affected family members and identification of characteristic linear lesions would help to diagnose this condition. It is also important to rule out associated anomalies and provide suitable counselling to the parents.

List of Abbreviations

IP – Incontinentia Pigmenti

REFERENCES

1. Berlin AL, Paller AS, Chan LS. Incontinentia pigmenti: A review and update on the molecular basis of pathophysiology. Journal of the American Academy of Dermatology. 2002 Aug 1;47(2):169–90.
2. Carney RG. Incontinentia pigmenti. A world statistical analysis. Arch Dermatol. 1976 Apr;112(4):535–42.
3. Gupta KD, Padhiar BB, Karia UK, Shah BJ. Case reports of incontinentia pigmenti in males. Indian Journal of Dermatology. 2013 Jul 1;58(4):328.
4. Smahi A, Hyden-Granskog C, Peterlin B, Vabres P, Heuertz S, Fulchignoni-Lataud MC, et al. The gene for the familial form of incontinentia pigmenti (IP2) maps to the distal part of Xq28. Hum Mol Genet. 1994 Feb;3(2):273–8.
5. Scheuerle AE, Ursini MV. Incontinentia Pigmenti. In: Adam MP, Ardinger HH, Pagon RA, Wallace SE, Bean LJ, Mirzaa G, et al., editors. GeneReviews® [Internet]. Seattle (WA): University of Washington, Seattle; 1993 [cited 2021 Apr 29].
6. Kliegman, Robert. Nelson Textbook of Pediatrics. Edition 21. Philadelphia, PA: Elsevier, 2020. Page 12281.
7. Shankar YU, Fatima N, Kumar MA, Prakash KS. Bloch Sulzberger syndrome (Incontinentia pigmenti): A rare case report with dental defects. Journal of Indian Society of Pedodontics and Preventive Dentistry. 2015 Jan 1;33(1):74.
8. Rooks book of Dermatology 9th edition, Chapter 70, page 1885.
9. Greene-Roethke C. Incontinentia pigmenti: a summary review of this rare ectodermal dysplasia with neurologic manifestations, including treatment protocols. Journal of Pediatric Health Care. 2017 Nov 1;31(6):e45-52.