Case Report

An Unusual Presentation of Desquamation at Birth

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ABSTRACT

Background : Incontinentia Pigmenti is a rare, X-linked dominant genodermatosis with dermal, ocular, neurological and dental anomalies. **Case Characteristics :** A female newborn presented at birth with excessive desquamation turned out to have Incontinentia Pigmenti. **Message :** Desquamation at birth is an unusual presentation of IP.

INTRODUCTION

Incontinentia Pigmenti (IP; OMIM#308300) is a rare X-linked dominant multisystemic genodermatosis that is usually lethal in males before birth. It occurs in approximately 0.0025% cases at birth. [1] It is characterized by cutaneous, neurologic, ophthalmologic, and dental abnormalities.

CASE REPORT

A female child, born through LSCS, presented at the time of birth with excessive desquamation. Considering it as a variant of normal physiological neonatal desquamation, a dermatologist's opinion was taken. The baby presented on 9th day of life at the Paediatric OPD with neonatal physiological jaundice. On examination, the baby was found to have blistering lesions of the skin of trunk, limbs, back and buttocks but the face and upper limbs were spared. These blisters appeared in crops and did not follow any dermatomal distribution. They burst on their own without any discharge and hyperpigmented verrucous skin lesions were left behind. This process continued for over a week. The child was otherwise asymptomatic. Other than having skin lesions, there were no systemic features of infection/inflammation. Sepsis screen was negative. There was no family history of similar complaints, including her elder sister. The ante-partum and Postpartum period of the mother was also uneventful.

Hence the following differentials of Neonatal Blistering Diseases were arrived upon: 1) transient pustular melanosis, 2) cutaneous mastocytosis, 3) neonatal pemphigus and 4) Incontinentia Pigmenti. As amongst these, the prognosis of IP was worst, a CNS and Ophthalmological screening was done. She was found to have bilateral abnormal retinal vascularisation by 1 month age, more in right eye for which intra-vitreal Anti-Vegf was given and Laser treatment had to be performed subsequently. An MRI at 3 months age revealed no abnormalities. At about 9 months of age, she developed febrile Generalised tonic-clonic convulsions followed by multiple episodes of unprovoked GTC convulsions. Hence the suspicion of IP grew stronger. So she was subjected to further investigations.

A skin biopsy, at 1 year age, was suggestive of mild papillomatosis, mild pigment incontinence and occasional dyskeratotic cells. The NEMO gene deletion test for IP turned out to be positive.

Currently her development is normal for age, vision is preserved and skin shows classical hyperpigmented whorls.

DISCUSSION

About 75% cases of IP are sporadic occurring by a denovo genetic mutation, the deletion produces loss of IKBKG exon, causing an alteration in nuclear factor kappa light (NF-κβ) essential modulator gene located at Xq28.[2] The clinical evolution of the skin lesions has 4 stages: 1) vesiculobullous lesions in a linear pattern on eyethematous area distributed in Blaschko's lines occurring in neonatal period; 2) verrucous hyperkeratotic lesions extending for about 4 weeks; 3) hyperpigmented skin lesions; and 4) hypopigmention and skin atrophy.[3] Diagnosis is achieved by one major and two minor criteria. The major criteria are: Neonatal vesicular rash feature, typical lesions of hyperpigmentation with distribution along the lines of Blaschko and hypochromic

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and atrophic linear lesions in extremities. [4] The minor criteria include neurological (seizures, motor impairments), ophthalmic (optic atrophy, blindness) and dental abnormalities (hypodontia, partial anodontia, delayed eruption, etc). Skeletal and nail involvement have been noted in few patients.

There is no treatment for this disorder and management of the patient involves multidisciplinary medical team to improve the quality of life of the patient and Genetic counselling of the Parents.

In a case like this, we may miss the condition on initial presentation. But a regular and religious follow up of the patient, along with keeping the major disabling outcomes in mind and screening for the same, led to an early intervention for visual presentation in this child and an anticipated development of seizures led to early neurological treatment initiation for the child.

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