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Linear Lesions, Lifelong Implications: A Neonatal Presentation of Schimmelpenning Syndrome

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Abstract

Schimmelpenning syndrome (Linear Nevus Sebaceous Syndrome) is a rare neurocutaneous disorder characterized by nevus sebaceous with variable extracutaneous involvement. We report a term neonate with linear yellowish verrucous plaques over the face, right ear, midline neck, and right lower lip following the Lines of Blaschko. Detailed evaluation, including neuroimaging, ophthalmologic examination, echocardiography, and skeletal survey, was normal. A diagnosis of Schimmelpenning syndrome with isolated cutaneous manifestations was made. The infant was enrolled in multidisciplinary follow-up for surveillance of delayed systemic involvement. This case emphasizes the importance of early recognition and comprehensive evaluation of neonates presenting with characteristic nevus sebaceous lesions.

Keywords: Schimmelpenning Syndrome, Linear Nevus Sebaceous Syndrome, Nevus Sebaceous, Epidermal Nevus Syndrome, Neurocutaneous Disorder, Neonate

Introduction

Schimmelpenning syndrome is a rare congenital neurocutaneous disorder first described by Gustav Schimmelpenning in 1957. It is characterized by the presence of nevus sebaceous in association with developmental abnormalities involving multiple organ systems. The syndrome belongs to the broader group of epidermal nevus syndromes and is caused by postzygotic activating mutations, most commonly involving genes in the RAS/MAPK signaling pathway. [1]

Nevus sebaceous typically presents as a yellow-orange, hairless plaque that follows the Lines of Blaschko, reflecting embryonic mosaicism. Although cutaneous lesions are the hallmark of the syndrome, patients may develop neurological abnormalities such as seizures and developmental delay, ocular defects including coloboma, and skeletal abnormalities. The severity of disease is highly variable, ranging from isolated skin involvement to extensive multisystem disease. [3]

Early diagnosis is crucial to facilitate prompt screening for associated anomalies and establish long-term surveillance strategies. [4]

Case Report

Clinical Presentation

A term neonate was born by normal vaginal delivery to non-consanguineous parents with an uneventful antenatal and perinatal history. Birth weight and anthropometric parameters were appropriate for gestational age.

At birth, multiple congenital cutaneous lesions were noted involving the right side of the face, right ear, midline neck region, and right lower lip. The lesions appeared as well-demarcated, yellowish, verrucous

plaques arranged in a linear pattern corresponding to the Lines of Blaschko.

The infant had no dysmorphic features, seizures, feeding difficulties, or other systemic complaints. General physical examination and neurological assessment were unremarkable.

Diagnostic Evaluation

A multidisciplinary assessment was performed to evaluate for extracutaneous involvement.

1. Cranial ultrasonography and neuroimaging revealed no structural abnormalities.
2. Detailed ophthalmological examination was normal.
3. Echocardiography demonstrated normal cardiac anatomy and function.
4. Skeletal survey showed no evidence of bony abnormalities.
5. Hearing assessment was within normal limits.

Based on the characteristic clinical appearance of the lesions and the absence of alternative diagnoses, a diagnosis of Schimmelpenning syndrome with isolated cutaneous manifestations was made.

Management and Follow-Up

The patient was managed conservatively with regular dermatological care and parental counseling regarding the nature of the condition. A multidisciplinary follow-up plan involving pediatrics, dermatology, neurology, and ophthalmology was established to monitor for the development of delayed extracutaneous manifestations and potential secondary neoplasms arising within nevus sebaceous lesions.

Discussion

Schimmelpenning syndrome is a rare mosaic disorder with an estimated prevalence of less than 1 per 100,000 live births. The hallmark lesion, nevus sebaceous, is usually present at birth and most commonly affects the scalp and face. Extracutaneous manifestations occur in a significant proportion of patients and may involve the central nervous system, eyes, skeleton, cardiovascular system, and genitourinary tract. [3,5]

Neurological abnormalities are among the most frequently reported systemic manifestations and include epilepsy, developmental delay, intellectual disability, and structural brain malformations. Ocular findings may include coloboma, choristoma, and retinal abnormalities. Skeletal defects such as scoliosis, limb asymmetry, and craniofacial anomalies have also been described. [5,6]

Our patient demonstrated characteristic cutaneous findings without evidence of systemic involvement. Such isolated presentations are uncommon but have been reported and likely represent a milder end of the phenotypic spectrum. Nevertheless, extracutaneous manifestations may emerge later in childhood, necessitating long-term surveillance. [6]

This case underscores the importance of recognizing the distinctive cutaneous features of Schimmelpenning syndrome and conducting comprehensive baseline investigations even in asymptomatic neonates.

Conclusion

Schimmelpenning syndrome should be considered in neonates presenting with congenital linear verrucous plaques following the Lines of Blaschko. Early recognition allows timely evaluation for associated systemic abnormalities and facilitates multidisciplinary management. Although our patient exhibited only cutaneous manifestations, long-term follow-up remains essential due to the potential for delayed extracutaneous complications and neoplastic transformation within nevus sebaceous lesions.

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