The Journal of the Association of Physicians of Tamil Nadu,
Vol. 3, Issue 2, English Quarterly, April – June 2024

A Case of Schimmelpenning Syndrome

Dr. Gokul Babu[1], Dr. Nikita Narayanan[2], Dr. Soundarya S[3], Dr. Jayakar Thomas[4]

[1,2]Resident, [3]Associate Professor, [4]Emeritus Professor

Department of Dermatology Venereology and Leprosy, Chettinad Hospital and Research Institute

Abstract

Schimmelpenning syndrome is a neurocutaneous condition characterized by one or more sebaceous nevi, usually appearing on the face or scalp, associated with CNS anomalies, the ocular system, skeletal system, cardiovascular system and genitourinary system. Here we present to you the case of 12 yr old female presented to the outpatient department with schimmelpenning syndrome

Keywords*: nevus sebaceous, Schimmelpenning syndrome, acanthosis, mosaicism*

1. Introduction

Schimmelpenning syndrome is a congenital neurocutaneous disorder encompassing cutaneous nevi and extracutaneous manifestation (CNS anomalies, ocular system, skeletal system, cardiovascular system and genitourinary system). The classical triad of schimmelpenning syndrome involves sebaceous nevi, seizures and mental retardation. It is actually thought to result from genetic mosaicism, possibly an auto-dominant mutation arising after conception. It actually includes sebaceous nevus which is associated with extracutaneous abnormalities mainly affecting the brain, eyes, and bones [2].

2. Case Report

A 12-year-old female presented to the outpatient department with hairless yellowish verrucous plaque on the occipital region of the scalp and over the left temporal area extending towards the lower occipital region of the scalp associated with seizures. The patient gives a history of medication for the seizures for the past 2 years. No history of involvement of eyes and bones. No significant family history.

On Examination

Hairless yellowish verrucous plaque on the occipital region of the scalp pigmented verrucous plaque seen over the left temporal area extending to the lower occipital region of the scalp. Histologically the specimen shows hyperkeratosis, acanthosis and papilomatosis. Based on the clinical examination and histopathology the patient has been advised to remove the nevus sebaceous as a prophylactic measure to prevent the risk of developing malignant neoplasia. The patient was advised to take a neurology opinion for the seizures, since no other extracutaneous manifestation involving eyes and bones was not reported further interventions were not needed [3,4].

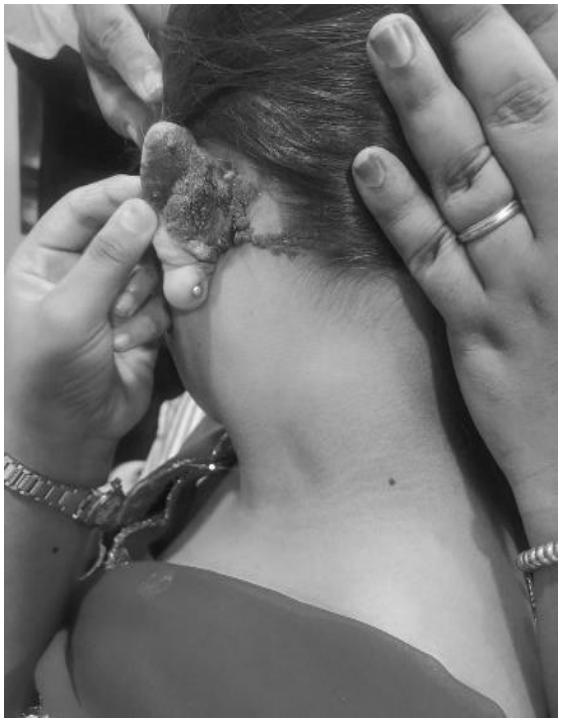

Figure 1: *Pigmented verrucous plaque is seen over the temporal area extending to the lower occipital region of the scalp.*

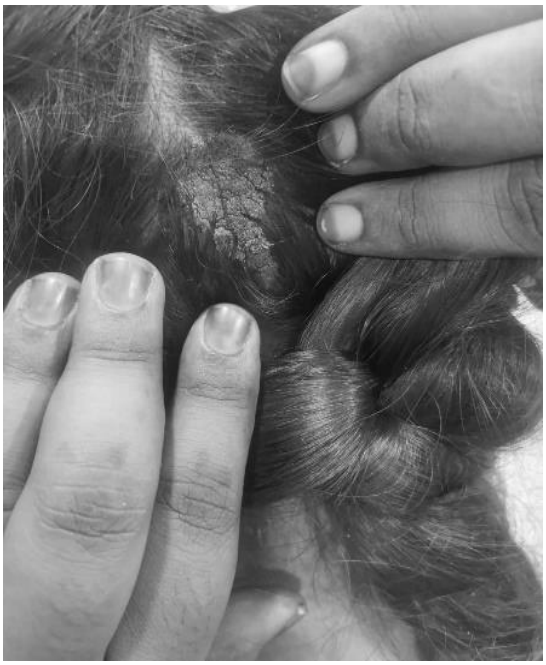

Figure 2: *Hairless yellowish verrucous plaque seen over occipital region of the scalp*

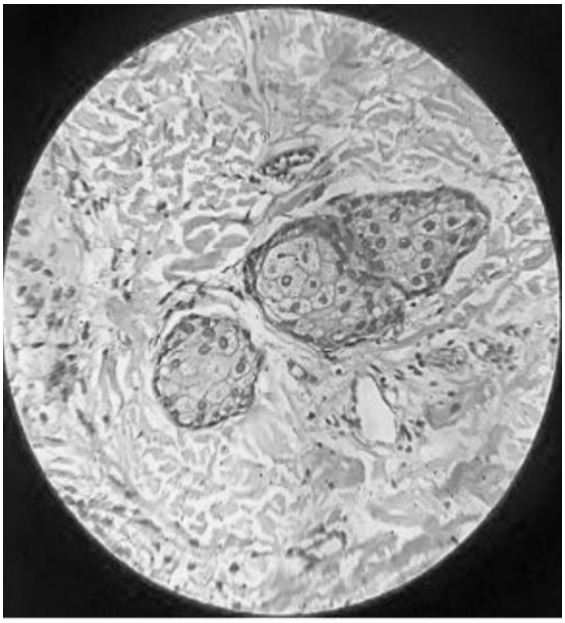

Figure 3: High Power: *Dermis shows sebaceous gland without the presence hair follicle*

3. Discussion:

Nevus sebaceous is the main characteristic and diagnostic criteria for schimmelpenning syndrome. It shows the association of extracutaneous manifestation involving the brain, eyes and bones along with the nevus sebaceous. In brains it could actually result in seizures, but also can have craniofacial defects, and arachnoid cysts, in the eyes it could result in colobamas and other ocular abnormalities and in bones it could result in hypophosphatemic rickets. Remarkably in the epidermal nevus, sebaceous hyperplasia may be minimal or even absent for two reasons: first, the sebaceous glands tend to be underdeveloped before puberty because of the absence of stimulating hormone; second, sebaceous hyperplasia may be minimal or even absent in those parts of a nevus that are localized outside of the head and neck. For differential diagnosis, could include phacomatosis pigmentokeratotic (PPK), nevus comedonicus syndrome and Proteus syndrome. The etiology of schimmelpenning syndrome is still unknown but is actually thought to result from genetic mosaicism. According to this theory, genetic alterations are present in only some of the body cells, most likely to due to the mutation of a gene that occurs after fertilization (post-zygotic mutation). The treatment usually done for the schimmelpenning syndrome is the excision of the nevus sebaceous.

4. Conclusion:

Schimmelpenning syndrome is a congenital neurocutaneous disorder encompassing cutaneous nevi and extracutaneous manifestation (CNS anomalies, ocular system, skeletal system, cardiovascular system and genitourinary system). The treatment usually done for schimmelpenning syndrome is the excision of the nevus sebaceous and treatment according to extracutaneous manifestation involved

References:

1. Shwu-Meei Wanga , Schimmelpenning Syndrome: A Case Report and Literature Review - Taiwan Pediatric Association. Published by Elsevier Taiwan LLC. All rights reserved. http://dx.doi.org/10.1016/j.pedneo.2012.12.017
2. Clarissa Patias Lena , Do you know this syndrome? Schimmelpenning-Feuerstein-Mims syndrome - https://doi.org/10.1590/abd1806-4841.20197661
3. Babar .k. rao (Ed),*Moschella and Hurleys dermatology* (4th edition), Jaypee Brothers Medical Publishers.
4. Jayakar Thomas (Editor), Deepthi Ravi (Co-editor),*Dermpath 101 a guide to residents of dermatology and pathology* , Flexcil.