International Journal of Science and Research (IJSR)
ISSN: 2319-7064
Impact Factor 2025: 7.089

# Schimmelpenning Syndrome - A Rare Multisystem Mosaic

**Dr . Shridevi C Vijayakumar[1], Dr. Vinayak Katkar[2]**

[1]Junior Resident , Department of Dermatology , D Y Patil Medical College , Kolhapur , Maharashtra, India
Corresponding Author Email: *shridevivijaykumar19[at]gmail.com*

[2]Senior Resident, Department of Dermatology, D Y Patil Medical College, Kolhapur, Maharashtra, India
Email: *katkarvinayak2012gmail.com*

**Abstract:** ***Schimmelpenning syndrome is a rare neurocutaneous disorder characterized by the presence of nevus sebaceous along with multisystem involvement. We report a case of a pediatric patient presenting with linear sebaceous nevi distributed along Blaschko's lines, associated with neurological and ocular abnormalities. Early recognition is essential due to potential complications, including seizures and developmental delay. This case highlights the importance of a multidisciplinary approach for diagnosis, management, and long-term follow-up.***

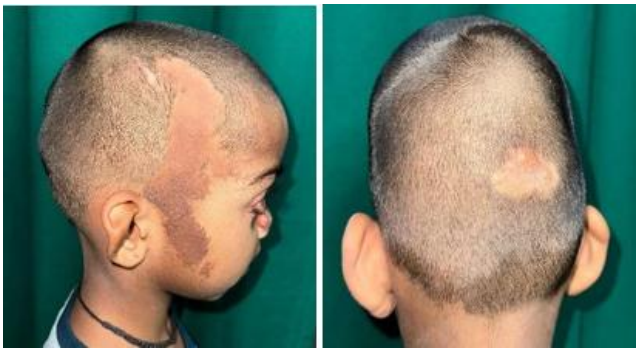
**Keywords:** nevus sebaceous, multisystem, neurological, ocular

## 1. Introduction

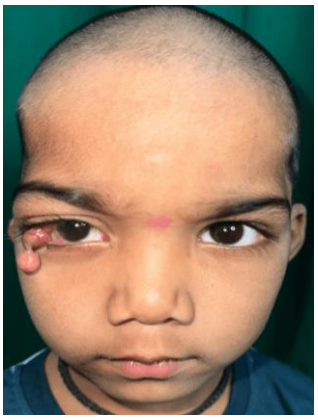
Schimmelpenning syndrome is a rare sporadic neurocutaneous disorder within the spectrum of epidermal nevus syndromes, characterized by nevus sebaceous associated with extracutaneous abnormalities involving the central nervous system, eyes, skeleton, and other organs[1]. Cutaneous lesions are usually present at birth or early childhood as linear or verrucous plaques over the scalp, face, or neck. Neurological features such as seizures and developmental delay, along with ocular and skeletal anomalies, may also occur[2]. Due to its rarity and diverse presentation, early diagnosis and multidisciplinary evaluation are important for timely management and long-term follow-up.

## 2. Case Report

A 6 year old boy with asymptomatic yellow-brown, hairless plaques over the scalp and periocular mass since birth presented to the Dermatology OPD . Examination showed irregular, hyperpigmented tan-brown, slightly verrucous hairless plaque with sharply defined margins over the right side of scalp extending from the fronto-temporal scalp down across the temple, preauricular region and cheek (3x8cm) and a smooth yellow patch over the vertex (3x3cm). A pedunculated pink fleshy nodule near the right periocular area present. The lesions are present since birth slightly increasing in size since then. Based on the history and examination a diagnosis of Schimmelpenning Syndrome was kept. The dermoscopic examination of the plaque was done and biopsy from the periocular mass was sent along with CT paranasal sinuses and MRI Brain with the final diagnosis fitting into Schimmelpenning Syndrome.



Tan-brown, Verrucous hairless plaques present over scalp

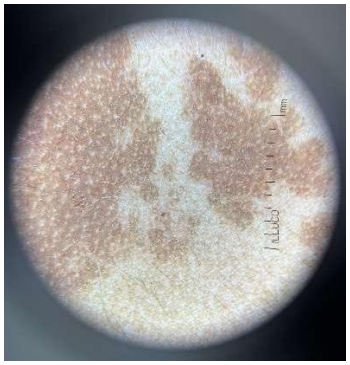


Pedunculated fleshy nodule near right periocular area

## 3. Dermoscopic Findings

Polarized dermoscopy revealed yellowish-white structureless background with irregular light-to-dark brown globular/clod-like areas forming a patchy cerebriform pattern consistent with sebaceous hyperplasia. Pigment network is absent. Focal whitish scales are present.

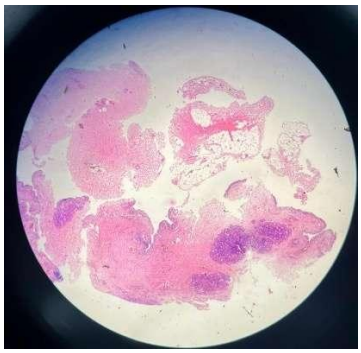
.



Yellow-brown structureless areas in cerebriform pattern

**Histopathological Findings**

The biopsy of periocular mass showed skin covered tissue lined by stratified squamous epithelium. Underlying dermis showed skin appendages with fibrofatty tissue, lacrimal glands and congested blood vessels. Focal areas showed sebaceous glands in high dermis. Section studied from temporal margin showed fibrofatty and fibrocollagenous tissue with chronic inflammatory infiltrates comprising of lymphocytes, plasma cells and histiocytes. Focal area showed conjunctival lining and lacrimal glands. At places congested blood vessels and nerve bundles were noted. Features were of Dermolipoma.



Surface lined by Stratified squamous epithelium overlying fibrocollagenous stroma containing mature adipocytes

**Other Investigations**

CT Paranasal sinuses revealed a Sphenoid Polyp. MRI Brain showed a cystic lesion in the right Merkel's cave consistent with cystic schwannoma.

**Management and follow-up-**

The patient was referred to Ophthalmology for excision of periocular mass. Long-term surveillance was advised for malignant transformation in nevus sebaceous.

## 4. Discussion

Schimmelpenning syndrome is a rare neurocutaneous disorder characterized by nevus sebaceous associated with abnormalities of the ocular, neurologic, and skeletal systems. It is caused by postzygotic mosaic mutations involving the RAS/MAPK pathway, most commonly **HRAS** and **KRAS**, resulting in variable phenotypic expression. [3, 4] Nevus sebaceous typically presents at birth or early childhood as a yellow-orange plaque that may become verrucous with age.[5] Ocular involvement is common and includes epibulbar choristoma, coloboma, strabismus, and dermolipoma. [6] The periocular dermolipoma seen in our patient represents a recognized but uncommon manifestation and highlights the importance of detailed ophthalmologic examination in all suspected cases.

Secondary neoplasms may develop within nevus sebaceous, most of which are benign, while malignant transformation is rare in childhood [5,7] Therefore, management in pediatric patients is usually conservative with regular follow-up, reserving excision for symptomatic, cosmetically significant, or suspicious lesions.

An unusual finding in the present case was a cystic schwannoma. Schwannomas are benign peripheral nerve sheath tumors that are uncommon in children and rarely reported in association with Schimmelpenning syndrome. Its occurrence in our patient may represent either a coincidental lesion or a rare additional manifestation of mosaic dysregulation.[8]

## 5. Conclusion

Schimmelpenning syndrome is a rare multisystem disorder with variable clinical presentation. Our case is notable for the coexistence of nevus sebaceous, periocular dermolipoma, and the uncommon finding of cystic schwannoma in a 6-year-old child. This report expands the recognized phenotypic spectrum of the syndrome and highlights the importance of early diagnosis, thorough systemic evaluation, multidisciplinary management, and long-term follow-up for timely detection of associated anomalies and secondary neoplasms.

## References

[1] Wang SM, Hsieh YJ, Chang KM, Tsai HL, Chen CP. Schimmelpenning syndrome: a case report and literature review. Pediatr Neonatol. 2014;55(6):487-490.
[2] Gupta A, Goel S, Thapar RK. Schimmelpenning syndrome: a neuro-oculo-cutaneous disorder. Indian Dermatol Online J. 2023;14(6):920-921.
[3] Happle R. Schimmelpenning syndrome: an update. *Clin Dysmorphol*. 2008; 17: 219-223.
[4] Groesser L, Herschberger E, Sagrera A, et al. Postzygotic HRAS and KRAS mutations cause nevus sebaceous syndrome. *Nat Genet*. 2012; 44: 783-787.
[5] [5] Cribier B, Scrivener Y, Grosshans E. Tumors arising in nevus sebaceous: a study of 596 cases. *J Am Acad Dermatol*. 2000; 42: 263-268.
[6] Vidaurri-de la Cruz H, Tamayo-Sánchez L, Durán-McKinster C, et al. Epidermal nevus syndromes: clinical findings in 35 patients. *Pediatr Dermatol*. 2004; 21: 432-439.
[7] Moody MN, Landau JM, Goldberg LH. Nevus sebaceous revisited. Pediatr Dermatol. 2012; 29: 15-23.
[8] Rodriguez FJ, Folpe AL, Giannini C, Perry A. Pathology of peripheral nerve sheath tumors. *Acta Neuropathol*. 2012; 123: 295-319.