CASE REPORT

Schimmelpenning syndrome with didymosisaplasticosebacea in an infant

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Introduction: Schimmelpenning syndrome may encompass abnormalities of the cardiovascular, skeletal, ophthalmologic and urogenital systems. Nevus sebaceous is a hallmark finding and ophthalmologic findings are seen in 59% of the cases which include colobomas and choristomas.

Case Summary: A 1-month-old female presented with a verrucous plaque over the scalp and right zygomatic area upon birth. Physical examination reveals a linear yellowish alopecic verrucous plaque over the right frontal region, yellowish alopecic verrucous plaque topped with a skin colored papule over the right zygomatic region, conjunctival mass over the right eye and an atrophic patch with areas of circular erosion over the right occipital region. Ballard score and reflexes were appropriate for gestational age. Newborn screening was normal and otoacoustic-emission-test revealed no hearing loss. She was referred to an ophthalmologist and was assessed to have a lipodermoid, right upper eyelid and optic nerve coloboma. Cranial CT scan is unremarkable. Histopathology showed an increase in number of sebaceous glands with malformed hair units. She was managed holistically and does not have seizures and no secondary development of tumors in the nevus sebaceous.

Conclusion: Schimmelpenning syndrome is usually associated with the clinical triad of nevus sebaceous, mental retardation and seizures. In this case, seizures were absent, however, there is an associated lipodermoid, right upper eyelid and right optic nerve coloboma. In addition, she also presented with aplasia cutis congenita. Hence, it is important to look for other manifestations when patients present with nevus sebaceous because management requires collaboration with different specialties.

Keywords: Schimmelpenning syndrome, nevus sebaceous syndrome, didymosisaplasticosebacea, linear nevus sebaceous syndrome, nevus sebaceous

INTRODUCTION

Schimmelpenning syndrome encompasses a broad spectrum of abnormalities that include the central nervous system, cardiovascular, skeletal, ophthalmologic and urogenital systems.[1] Nevus sebaceous is a hallmark finding [2] and ophthalmologic findings are seen in 59% of the cases which include colobomas and choristomas. [3]

We present a unique case of Schimmelpenning syndrome that does not have seizures, however, is associated with lipodermoid, right upper eyelid coloboma, right optic nerve coloboma and aplasia cutis congenita.

The distinct findings of this case increase the dermatologist's awareness of this illness and will help physicians make an early diagnosis and initiate appropriate management.

CASE REPORT

A 1-month old female presented with a yellowish alopecic verrucous plaque over the scalp and soft mass over the right zygomatic area which presented upon birth. Maternal and neonatal histories are unremarkable. Ballard score is appropriate for gestational age. Physical examination reveals a linear yellowish alopecic verrucous plaque over the right frontal region,atrophic patch with areas of circular erosion over the right occipital region, yellowish alopecic verrucous plaque topped with a skin colored papule over the right zygomatic region and conjunctival mass over the right eye. (Figure 1 A-D). The atrophic patch was assessed to be aplasia cutis congenita, hence biopsy was not done over the said lesion. She was

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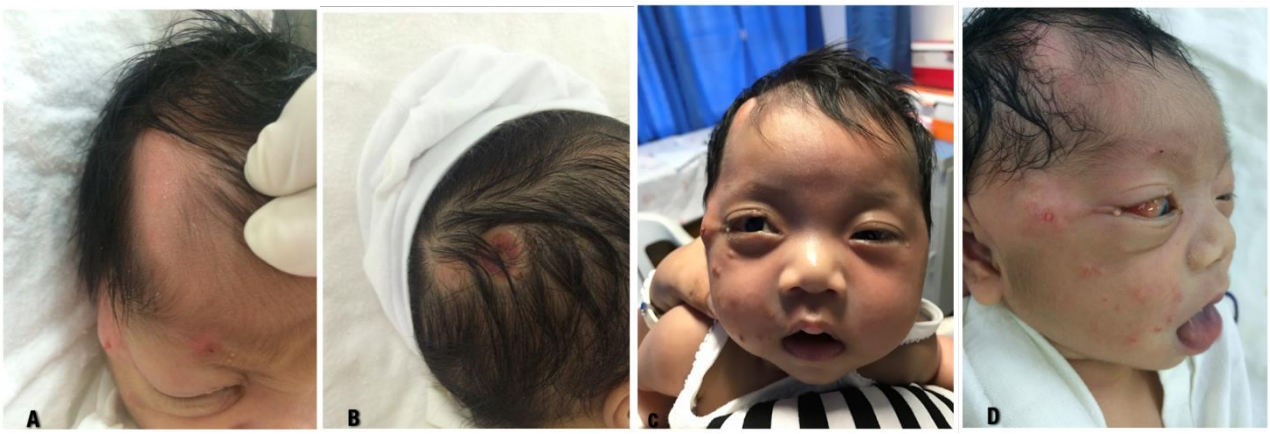


Figure 1 Linear yellowish alopecic verrucous plaque over the right frontal region (A), an atrophic patch with areas of circular erosion over the right occipital region (B), yellowish alopecic verrucous plaque topped with skin colored papule over the right zygomatic region and conjunctival mass over the right eye (C&D).

then referred to an ophthalmologist and was assessed to have lipodermoid, right upper eyelid coloboma and right optic nerve coloboma.

Complete blood count and newborn screening were unremarkable. She was referred to ENT for otoacoustic emission test which revealed that patient currently has no hearing loss. Cranial CT scan revealed no abnormal density changes in the brain and brainstem parenchyma.

Skin punch biopsy was done on 2 sites. Site A consist of the linear yellowish verrucous plaque over the right frontal region which showed increase in the number of sebaceous glands with malformed hair units (Figure 2A and B) which is consistent with nevus sebaceous. Site B consist of the yellowish alopecic plaque topped with a skin colored papule over the right zygomatic area which showed a pedunculated fibrous stroma with increase blood vessels and increase in the number of sebaceous glands with malformed hair units (Figure 2 C and D) consistent with angiofibroma and nevus sebaceous.

The patient was discharged and was advised to follow up when seizures arise.

Upon follow up, after 1 year, she was still

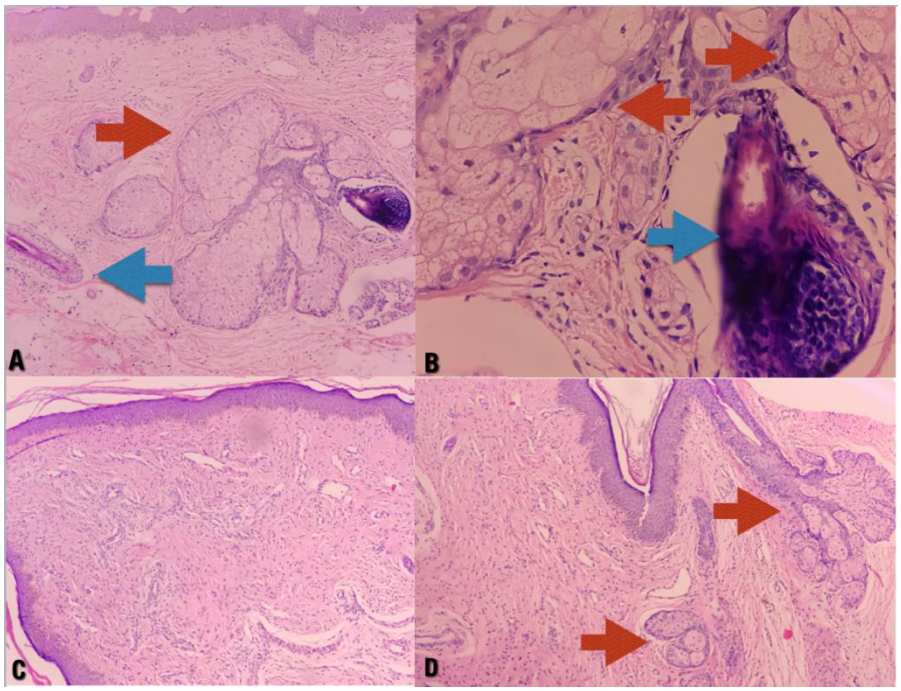
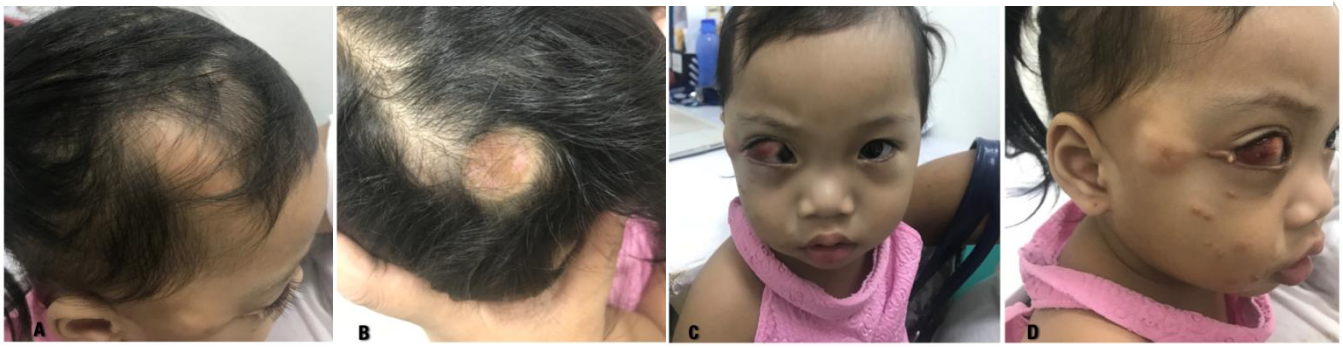


Figure 2 (A to D). Histopathology showing (A) Mild acanthosis with increase in the number of sebaceous glands (Red arrow) with malformed hair units (Blue arrow, H&E, x 100). (B) Increase in the number of sebaceous glands (Red arrow) with malformed hair units (Blue arrow, H&E, x 400). (C) Pedunculated fibrous stroma with increase in blood vessels (H&E, x 100) and (D) Increase in the number of sebaceous glands with malformed hair units (Red arrow, H&E, x 100)

Figure 3. Linear yellowish alopecic verrucous plaque over the right frontal region (A), an atrophic patch with areas of circular erosion over the right occipital region (B), yellowish alopecic verrucous plaque over the right zygomatic region and conjunctival mass over the right eye (C&D).



asymptomatic with no seizures. There was persistence of linear yellowish alopecic verrucous plaque over the right frontal region, an atrophic patch with areas of circular erosion over the right occipital region, yellowish alopecic verrucous plaque over the right zygomatic region and conjunctival mass over the right eye (Figure 3A to D).

DISCUSSION

Schimmelpenning syndrome encompasses a broad spectrum of abnormalities that may affect every organ system, including the central nervous system, cardiovascular, skeletal, ophthalmologic and urogenital system.[4]Nevus sebaceous is a hallmark of Schimmelpenning syndrome, and if physicians are aware of this, an early diagnosis can be made. [1]

Schimmelpenning syndrome may present with epilepsy that occurs in 67% of the cases, with seizures typically beginning during the 1st year of life.[5]It may present with the classic triad of nevus sebaceous, seizures and mental retardation. However, a number of studies revealed that the associated anomalies of the syndrome extend widely beyond the initial triad and not all affected individuals had epilepsy or mental retardation and that additional defects involving ocular, skeletal, cardiovascular and urologic abnormalities may be present; hence the diagnostic criteria of the triad have been abandoned.[6]The most common neurologic abnormalities found on computed tomography are hemimegalencephaly and ipsilateral gyral malformations. [7]

Clinical manifestations such as ophthalmological abnormalities are present in 59% of cases and major ocular abnormalities are colobomas and choristomas.[3] Hypophosphatemic rickets have also been reported to occur.[8] Cardiovascular abnormalities include a patent ductus arteriosus, patent foramen ovale, coarctation of the aorta, hypoplasia of the aorta, ventricular septal defect and atrial fibrillation. [9]

Nevus sebaceous is an uncommon lesions which occurs in 0.3% of neonates.[10]It usually presents as a well demarcated, yellow-orange, alopecic, verrucous plaques and most commonly occurs on the scalp in 59.3%.[3]It has also been reported to be associated with secondary tumors such as syringocystadenomapapilliferum, trichoblastoma and basal cell carcinoma.[11 12 13 14] The risk of malignancy in nevus sebaceous is quite low, with the incidence of basal carcinoma at 1%.[14]

While Schimmelpenning syndrome is one of the well defined epidermal nevus syndrome.[15] Didymosisaplasticosebacea (DAS) is one of the less well defined epidermal nevus syndromes. DAS is a nevus sebaceous that coexist with an aplasia cutis congenita in which the two lesions occur in close proximity to each other. [16] This occurrence may be explained by the non-allelic twin spot phenomenon. [17]

Surgical excision of the nevus sebaceous is the treatment of choice in most cases in which it is a cosmetic problem, however, due to the low risk of malignant degeneration, nevus sebaceous excision is recommended only if suspecting of basal cell carcinoma.[1819] In patients with Schimmelpenning syndrome, a multidisciplinary approach is strongly advised in which the team will be consists of a dermatologist, pediatrician, ophthalmologist, neurologist and orthopedic surgeon.[9 16]

CONCLUSION

Schimmelpenning syndrome is a multisystem dermatological disorder in which a nevus sebaceous may be associated with neurologic, skeletal or ocular abnormalities. Nevus sebaceous is a diagnostic marker of Schimmelpenning syndrome and awareness of this may help physicians make an early diagnosis.

We presented this case because Schimmelpenning syndrome is usually associated with the clinical triad of nevus sebaceous, mental retardation and the presence of seizures. In our case, seizure was not present, however, there is an associatedlipodermoid, right upper eyelid coloboma and right optic nerve coloboma. In addition, the patient also presented with aplasia cutis congenita. Hence, it is important to look for other manifestations when patients present with nevus sebaceous because management requires collaboration with different

specialities which is most essential to prevent . complications.

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