CASE REPORT

Choroid Osteoma in Schimmelpenning-Feuerstein-Mims Syndrome

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Received: May 09, 2017;
Initial review: June 21, 2017;
Accepted: March 10, 2018.

Background: Schimmelpenning syndrome is a multisystem disorder. **Case characteristics:** A term female neonate with sebaceous nevi of the face had choroid osteoma of the right eye. **Observation:** At one month of age, the infant was observed to have choroidal neovascularization that was successfully treated with laser photocoagulation and anti-VEGF. **Message:** Choroid osteoma and neovascularization are rare associations of Schimmelpenning syndrome, and should be screened for and managed early.

Keywords: *Choristoma, Management, Retinal coloboma, Screening.*

Schimmelpenning-Feuerstein-Mims Syndrome is a rare multisystem disorder characterized by sebaceous nevus associated with other abnormalities of the brain, eyes, and bones. Sebaceous nevi clinically presents as hairless hypopigmented to yellowish circumscribed plaques with a tendency to become more verrucous with puberty. These nevi demonstrate hyperplasia of the epidermis, immature hair follicles, and irregularities of morphology and distribution of sebaceous glands [1]. Sebaceous nevus when associated with multiple organ involvement is the organoid nevus syndrome known as Schimmelpenning syndrome [2]. We report the presence and successful management of choroidal osteoma induced neovascularization in a neonate who had sebaceous nevi associated with ophthalmologic and cardiovascular involvement.

Case Report

A term female neonate, born out of third degree consanguineous marriage with an uneventful antenatal period, presented with hypopigmented-to-yellowish plaques pathognomonic of sebaceous nevi over the forehead and chin in a blaschloid distribution associated with non-scarring alopecia of the scalp (***Fig.* 1**). Ophthalmologic examination demonstrated right corneal haziness with vascularization. Slit lamp examination revealed a calcified lesion over the bulbar conjunctiva with the possibility of choristoma of the conjunctiva; there was no cataract. Fundus examination showed right retinal coloboma. Contrast enhanced computed tomography revealed calcified lesions of the right optic canal. Magnetic resonance imaging was suggestive of right retinal coloboma associated with calcified lesion around the optic nerve suggestive of choroid osteoma. The echocardiogram revealed ventricular septal defect. There was no genitourinary abnormality. Skeletal survey, and serum levels of calcium, phosphorous, alkaline phosphatase, parathyroid hormone and 25-hydroxy vitamin D were normal. The neonate was otherwise

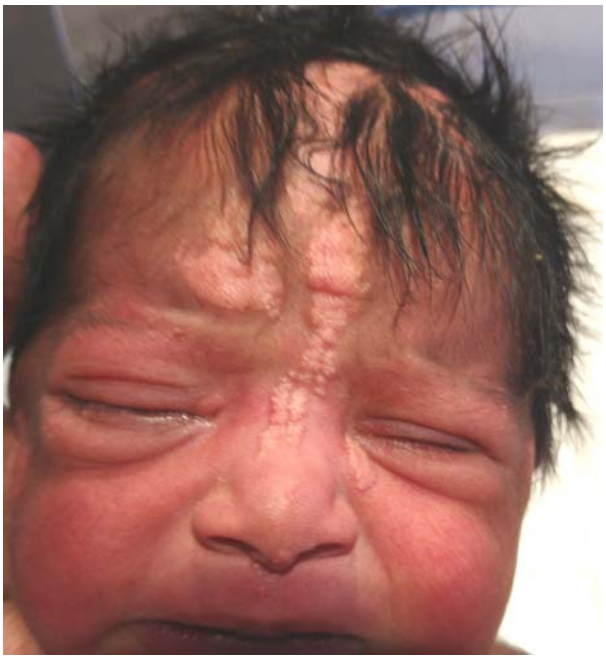

Fig. 1 *Hypopigmented-to-yellowish hairless skin lesion suggestive of sebaceous nevi.*

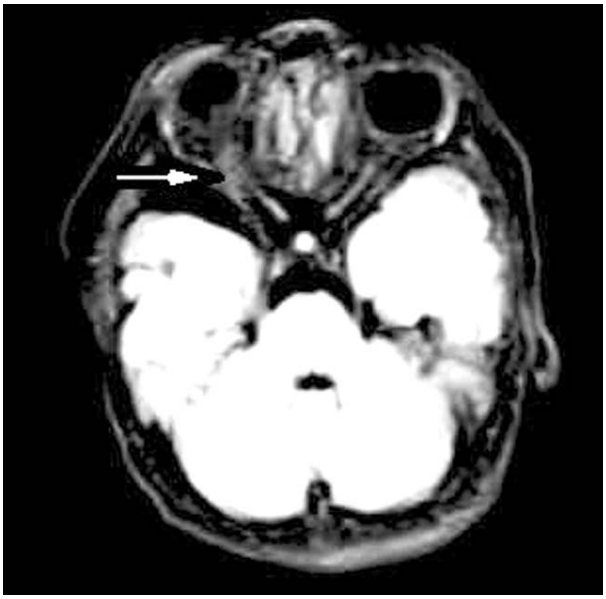

FIG. 2 *MRI showing right retinal coloboma and choristoma (arrow).*

asymptomatic and discharged on day 7 of life. On follow-up at one month of age, the infant was observed to have choroidal neovascularization that was treated with laser photocoagulation and intravitreal anti-VEGF (bevacizumab). At four months of age, there was a gradual regression of choroidal neovascularization, and preserved vision.

DISCUSSION

Schimmelpenning syndrome is characterized by sebaceous nevi of Jadossohn with the eye, brain and skeletal defects. Observed prevalence is 1 to 3 per 1000 live births. The occurrence of the disease is sporadic due to postzygotic mutation. In 60-80% of the cases, the skin lesions are confined to the scalp and face region, and are associated with nonscarring alopecia of scalp [3].

Choristoma of the conjunctiva, coloboma of the eye, epibulbar dermoid and scarring degeneration of the retina are the most common ocular findings in this syndrome [4,5]. In this case, apart from the common ocular findings like choristoma of the bulbar conjunctiva and retinal coloboma, we observed an unusual calcified lesion suggestive of choroid osteoma in the right eye. Choroid osteoma is a benign tumor of mature bone replacing the choroid. Some consider it as choristoma in the region of choroid [6]. Visual loss may result from atrophy of retinal pigment epithelium overlying the osteoma, serous retinal detachment over the osteoma, and choroidal neovascularisation. In the present case, the neonate developed choroidal neovascularization that was successfully treated with laser photocoagulation and intravitreal anti-VEGF (bevacizumab), thus preserving the vision [7].

Neurological abnormalities observed in this syndrome include hemi megalencephaly, corpus callosum agenesis, gyri malformations and Dandy-Walker cyst. Skeletal defects observed are vertebral defects, craniofacial defects, asymmetry of skull bones and bone cysts. Other uncommon association includes coarctation of the aorta, genitourinary abnormalities, and vitamin D resistant rickets. There is a predisposition to develop benign skin tumors in the sebaceous nevi [5]. The overall prognosis depends on the severity of associated anomalies. In the present case, the neonate had ventricular septal defect with no neurological and skeletal defects.

We conclude that choroid osteoma and choroidal neovascularisation are rare ophthalmologic manifestations of Schimmelpenning syndrome which should be screened for so that initiation of early treatment results in better visual outcome.

Contributors: VB, SM, MM and SP: case management and supervision; VT: drafted the manuscript, which was revised and approved by all authors.
Funding: None; *Competing interest*: None stated.

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