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Case Report

Ocular Manifestations of Schimmelpenning-Feuerstein- Mims syndrome - A Rare Phakomatosis

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Abstract

Schimmelpenning-Feuerstein-Mims (SFM) syndrome is a rare phacomatosis characterized by sebaceous nevus of Jadasson associated with extracutaneous abnormalities affecting the brain, eyes and bones. Though Nevus sebaceous is the most common type of organoid epidermal nevus, but its association with Schimmelpenning-Feuerstein-Mims syndrome is rarely established. The purpose is to report the ocular manifestations of Schimmelpenning-Feuerstein-Mims syndrome of an 11-year-old boy. The patient had the classic triad of epibulbar choristomas, lid and retinal coloboma, and nevus sebaceus of Jadassohn. His vision in right eye was 1.00 and left eye was 0.30 in LogMAR chart. He had lid coloboma, multiple limbal and scleral choristoma, cloudy cornea, retinal and optic disc coloboma in right eye. Left eye had limbal dermoid but cornea was relatively clear with a normal fundus. A brownish black verrucous plaque on the right facial area was discovered spreading ipsilaterally to the cervical region, combined with cicatricial alopecia, microtia, periocular nodule and pigmentation. The retinal and optic disc coloboma was confirmedwith B scan ultrasonography of right eye.

Keywords: Nevus sebaceous of Jadassohn; Phacomatosis; Limbal dermoid; Coloboma; Epibulbar choristomas

Abbreviation

SFM: Schimmelpenning-Feuerstein-Mims

Introduction

Linear nevus sebaceus of Jadassohn, also known as organoid nevus syndrome or Schimmelpenning - Feuerstein-Mims syndrome (SFM) is a rare congenital phacomatosis initially defined by the dermatologist Josef Jadassohn in 1895 [1]. It is a congenital hamartomatous lesion characterized by cutaneous sebaceous nevus, seizures, and ocular abnormalities [2]. Numerous cutaneous, skeletal, neurologic, cardiovascular and ocular neoplasm and malformations have been termed as a part of SFM syndrome [3] Epibulbar choristomas is the most common ocular association [4-6]. The ocular, cutaneous and CNS manifestations are listed in table 1-3 respectively. The different collaboration of the nevus sebaceous of Jadassohn with these findings is quite comparable to the oculo-neuro- cutaneous syndromes, or phakomatosis and described as organoid nevus syndrome [5,7]. The sebaceous nevi strictly follow the 'lines of Blaschko' (Figure 3) which is a prototype of dermatological 'nevus lines' ofhead and neck named after Blaschko [3]. The ocular manifestations of Schimmelpenning-Feuerstein-Mims syndrome were rarely reported by the ophthalmologist. We report an 11 year old boy with organoid nevus syndrome giving emphasis on the ocular findings.

Ocular structure	Features
Globe	Microphthalmos
Orbit	Proptosis
Lid	Naevus, Coloboma, Hemangioma
Conjunctiva/episclera	Choristoma (most common), Dermoid Epidermoid, Complex choristoma, Vascular hemangioma
Sclera	Epi/intra-scleral cartilage, Epi/ intra-scleral bone, Posterior scleral choristoma
Cornea	Choristoma, Cloudy cornea
Anterior chamber	Axenfeld anomaly, Posterior embryotoxon
Lens	Cataract
Retina	Coloboma, Coats disease
Choroid	Coloboma, Osteoma
Optic nerve	Coloboma, Pit, Hypoplasia, Glioma

Table 1: Ocular manifestations of Schimmelpenning -Feuerstein-Mims syndrome.

Linear nevus usually respecting the midline, most commonly found on the scalp, retro auricular area andneck [5]
Alopecia
Sebaceous lobules
Malignant degeneration in 10% - 20% of cases [2]
Basal Cell Carcinoma [9]
Verrucous plaque

Table 2: Cutaneous manifestations of Schimmelpenning -Feuerstein-Mims syndrome.

Seizures
Mental retardation
Structural brain defects
Various deficits
Strabismus
Nerve palsies
Pupillary abnormalities

Table 3: CNS manifestations of Schimmelpenning -Feuerstein-Mims syndrome.

Case Report

An 11 year old boy came to the cornea clinic of Ispahani Islamia Eye Institute and hospital with the complaints of dimness of vision in both eyes, right more than left since childhood. On examination, we found that his best corrected visual acuity (BCVA) in right eye was 6/60, 20/200, 1.00 and in left eye was 6/12, 20/40, 0.30 in Snellen chart, 20/20 chart and LogMAR chart respectively. In right eye, the clinical features included eyelid coloboma (Figure 1), multiple limbal and scleral dermoid (Figure 2) known as epibulbar choristoma, cloudy cornea, early cataract (Nuclear sclerosis 1) retinal coloboma, and optic disc coloboma (Figure 6 and 7). In left eye, the clinical features were limbal dermoid, partial corneal opacity with a normal fundus. The intraocular pressure was 17 mmHg and 14 mmHg in right and left eye respectively bynon-contact tonometry. The systemic features involved a brownish black verrucous plaque on the right fronto- parietal area spreading ipsilaterally to the cervical region described as Linear Nevus of Jadasson that respects the 'lines of Blaschko' (Figure 4). There was Right fronto-parietal cicatricial alopecia (Figure 4), microtia, combined with periocular nodule and pigmentation (Figure 5). We did the complete neurological examination of the boy. His IQ scoring was average (90 - 109) [8] and gave no history of Seizure. His birth history was insignificant with a normal vaginal delivery at home. None of his family member were affected by a similar disease. The diagnosis was mostly clinical and the optic disc and retinal coloboma was confirmed by B scan ultrasonography of right eye (Figure 6) which showed hyper echogenic posterior pole lesion suggestive of definitive coloboma and orbital shadowing suggestive of posterior scleral choristoma. Color fundus photography of right eye showed optic disc coloboma and retinal coloboma (Figure 7). We prescribed spectacle for the vision improvement and tear substitute as supportive treatment. No definitive treatment was given. We planned to observe the patient regularly for development of further ocular complications like cataract or secondary glaucoma and systemic complications like seizure or any malignant transformations of the skin lesions.

Discussion

The ocular manifestations of the SFM syndrome is one of the fundamental features of the disease [11]. Epibulbar choristoma is the commonest features reported till date of this disease which ranges from a single dermoid to a complex choristoma [12,13]. Clarissa Patias [14] has reported a case of twin sisters who presented with organoid naevus syndrome with epileptic seizure and arachnoid

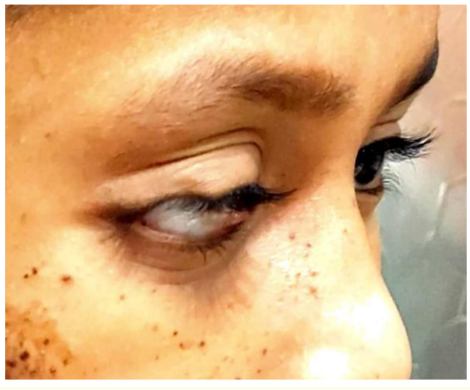

Figure 1: Lid coloboma.

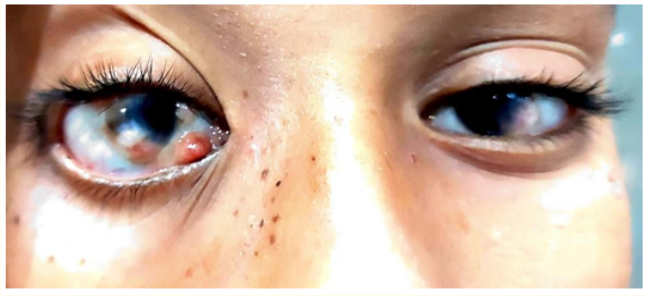

Figure 2: Multiple limbal and scleral dermoid with cloudy cornea.

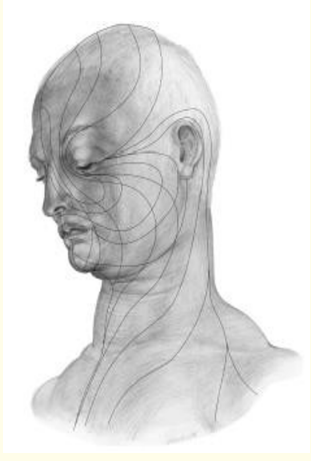

Figure 3: The 'lines of Blaschko' are a prototype of dermatological 'nevus line' of head and neck named after Blaschko respected in many congenital and acquired disorder [10] including SFM syndrome.

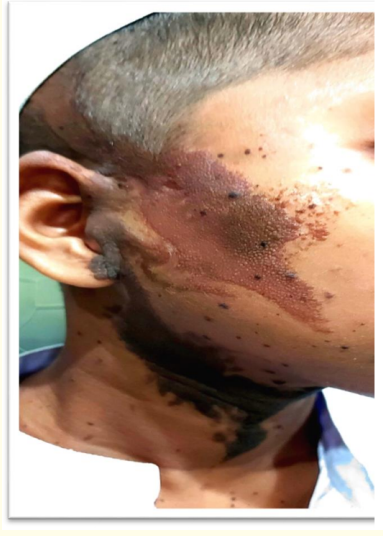

Figure 4: Linear Naevus of Jadasson along with Brownish plaque respecting 'lines of Blaschko', Right fronto-parietal cicatricial alopecia.

cyst. Various type offundus lesions have been observed by different authors that include hypopigmented yellowish-orange wedges of retina, retinal and optic disc coloboma, optic nerve hypoplasia [5,15]. Traboulsi., *et al*. [2] described four cases of nevus sebaceus of Jadassohn which were associated with posterior scleral choristoma and confirmed by histopathological examination. In our case, we report complex choristoma with posterior scleral involvement along with optic disc and retinal coloboma which was confirmed by B scan ultrasonography. We also found unilateral right sided brownish black nevus involving the temporal area spreading to cervical region. Right sided microtia, alopecia and hyperpigmentation were also described in our case. Different case reports from different country has reported similar neurocutaneous features [2,5,16,17]. In our study, the main culprit of vision loss is cloudy cornea along with optic nerve and retinal coloboma rather than epibulbar choristoma which is likewise established by Shields., *et al* [5].

Figure 5: Right microtia, combined with periocular nodule and pigmentation.

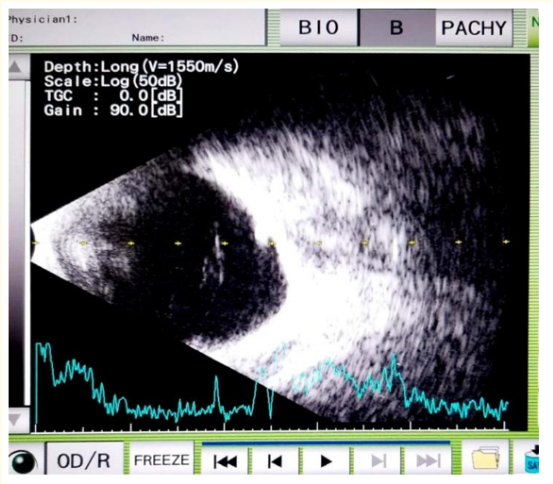

Figure 6: Hyperechogenic posterior pole lesion suggestive of optic disc coloboma with orbital shadowing suggestive of posterior scleral choristoma.

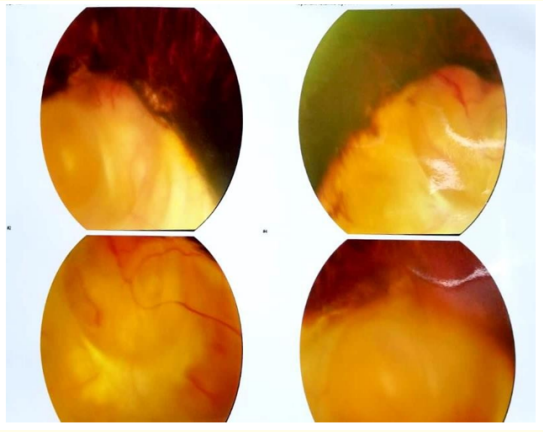

Figure 7: Optic disc coloboma and retinal coloboma.

Conclusion

Patients with Schimmelpenning -Feuerstein-Mims syndrome should be examined for ocular malformations and observe closely for the development of malignant tumors. Though epilepsy is an important feature of the disease, but does not necessarily always present. No treatment is usually necessary but regular follow up is mandatory to manage the further ocular complications like cataract and glaucoma.

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Conflict of Interest

No financial interest or any conflict of interest.

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