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CASE REPORT

Linear nevus sebaceous syndrome with bilateral ocular involvement in a Bangladeshi child: A case report with literature review

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The linear sebaceous nevus is one of a variety of epidermal nevi, which is a congenital ectodermal hamartoma that occurs due to post-zygotic mosaic mutation. Such sebaceous nevus is one of the commonest nevi, and up to 30% are associated with syndromic associations, which are rare. After cutaneous manifestation, central nervous system and ocular anomalies are common and found in almost every case. Multiorgan involvement can lead to various minor or major morbidities. Adequate knowledge and a high degree of suspicion can identify this rare disease and can minimize unwanted morbidity.

Keywords: linear nevus, epidermal nevus, dermolipoma, CNS involvement, hamartoma

Introduction

The linear sebaceous nevus (LSN) is one of a variety of epidermal nevi. When linear nevus is associated with multiple organ anomalies, especially the eye, central nervous system (CNS), and bone, it is called linear nevus syndrome (LNSS). This syndrome has a variety of synonyms, like nevus sebaceous syndrome, nevus sebaceous of Jadassohn, organoid nevus syndrome, Schimmelpenning syndrome, Solomon syndrome, and so on (1). In 1895, the linear nevus was first described as a separate entity by Jadassohn and he named this pigmented nevus organoid nevus (2). Schimmelpenning in 1957 first reported the LNSS. This syndrome was described as a triad of midline facial linear nevus sebaceous, convulsion, and delay in development by Feuerstein and Mims in 1962. After review of 60 cases, Solomon and Esterly expanded the spectrum of LNSS in 1975. They found involvement of multiple systems like ocular, central nervous, skeletal, cardiovascular, and urologic systems (3). Literature search showed that along with LN, ophthalmic presentation is found in about 10–30% cases (4). The ophthalmic presentations are squint, colobomas, choriostoma, retinal degeneration, cataract, vascularized cornea, scleral ossification, optic nerve hypoplasia, and so on (3, 4). CNS anomalies are most common after cutaneous involvement, which may be functional or structural or both. Functional anomalies are intellectual deficit (about 80%) and convulsion (60%), which may be generalized, tonic-clonic, focal motor, or infantile spasm. The structural CNS anomalies are hemiatrophy, hemimegacephaly, vascular, posterior fossa, or gyrate anomaly. Interestingly, 75% of patients with CNS findings have no detectable anomaly in imaging (1). Due to common involvement of skin, ophthalmic, and neurological symptoms in almost every case, Lambert and colleagues suggested the syndrome as oculo-neuro-cutaneous syndrome in 1987 (5).

In this article, we analyzed the phenotypic features of a case of LNSS in a Bangladeshi child who presented with bilateral ocular involvement and her treatment outcome.

Case report

A 10-year-old female girl from a non-consanguineous family presented with decreased visual acuity and a lid anomaly. Her family history and birth history were normal. Her intellectuality was normal, but she had a history of convulsion 2 to 3 times without fever during her first 3–4 years of life. Her ocular examination revealed vision of counting finger (CF) 1 meter in the right eye, which did not improve with refraction, and 6/18 in the left eye, which improved to 6/9 with +0.25 cylinder in 90 degrees. She had ptosis, inferiorly placed globe, upper lid coloboma with small nodular lesion, correctopia (upward placed pupil), and up gaze restriction in the right eye (**Figure 1B, C**). Dermolipoma was present in the super lateral quadrant of the right eye, and limbal dermoid-like lesions were present in both eyes (**Figure 1A**). The posterior segment showed coloboma of the optic disk with choroidal osteoma-like lesion in the superior nasal quadrant of the right eye (**Figure 1D**). The left eye disk was normal, but choroidal osteoma-like lesion was also present in the same quadrant. Her intraocular pressure was 16 and 14 mm of Hg. She had a papillomatous pigmented lesion over the entire nose in an elongated shape and another bigger lesion on the right-sided cheek, extending from the angle of the mouth to the base of the external ear (**Figures 2-A1, A2**). On gentle squeezing of the skin lesion, there was a sebaceous expression. Her right-sided forehead was steep and bulged more forward than her left side with some flat nevus (**Figure 2B**). She had a small alopecia (hair less patch) like area in scalp (**Figure 2C**). No gait abnormality and no skeletal abnormality or history of fractures was found. Her height was 49 inches. She had no other cardiac, urogenital, or dental abnormalities. Her computed tomography (CT) scan showed bilateral scleral calcification, which suggests scleral ossification or choroidal osteoma, cerebral atrophy in the temporal lobe, and an enlarged lateral ventricle on the right side (**Figure 3**). Excavation of the right optic nerve was also found in B-scan ultrasonography, and the left eye was normal. Her serum calcium, phosphate, and SGPT were normal.

Her mother provided history of debulking of dermolipoma and upper lid reconstruction in her right eye at the age of 4 years. She had an irregular eye checkup. Currently, she is treated with spectacles. Her parents were counseled about the disease and referred to a dermatologist for treatment of her skin lesions.

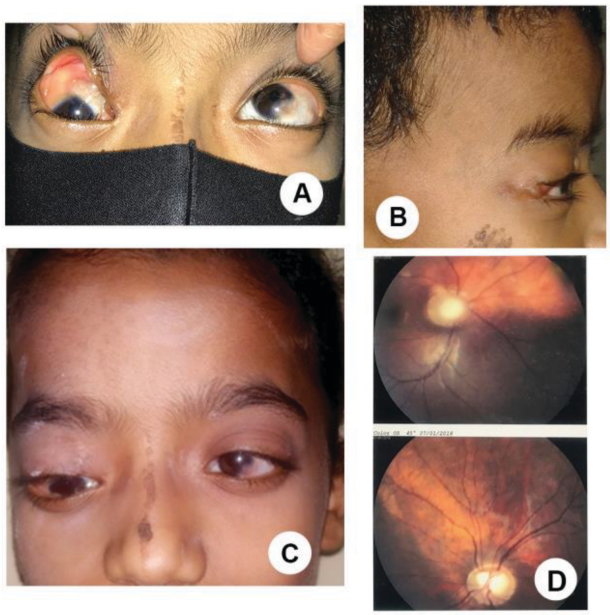

FIGURE 1 | Ocular findings. **(A)** Bilateral limbal dermoid-like lesion with dermolipoma in right eye and restricted upgaze. **(B)** Right upper lid coloboma with nodular lid lesion. **(C)** Right eye ptosis with hypotropia with linear nevus at nose. **(D)** Right optic disk coloboma with bilateral orange-colored choroidal lesion at supero nasal retina.

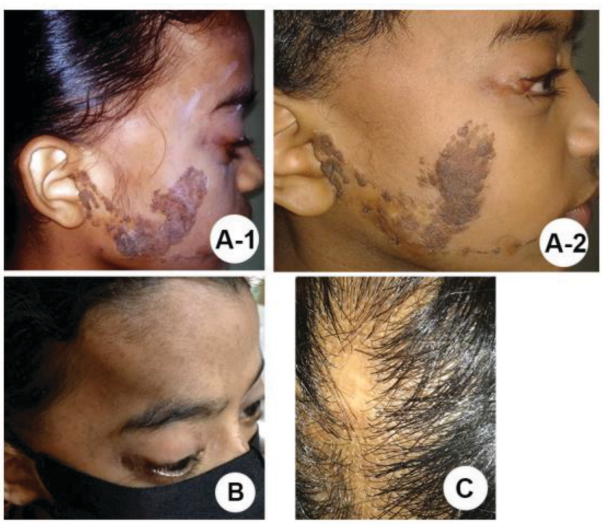

FIGURE 2 | Cutaneous features. **(A1)** Papillomatous pigmented lesion in right-sided cheek, extending from angle of the mouth to root of external ear – at presentation. **(A2)** Same lesion at the age of 4 years. **(B)** Steeped forehead on right side with some flat nevus. **(C)** Alopecia-like lesion on scalp.

Discussion

In LNSS, there is syndromic association of epidermal nevus (EN), which is congenital hamartomas of ectodermal origin, and the sebaceous part is predominant. Though the epidermal nevus prevalence is 1–3 in 1000 people, there is no known prevalence of LNSS (6).

The literature also showed one third of patients with EN may have involvement of different organ systems, that is, a chance to develop any EN syndrome which may be of different kinds, and six different syndromes with EN have been described up to now (6). These include (1) proteus,

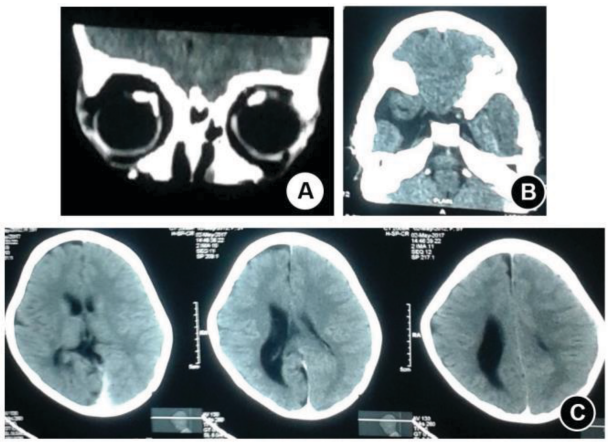

FIGURE 3 | CT scan findings. **(A)** Bilateral ocular calcification at supero-medial part due to either choroidal osteoma or scleral ossification. **(B)** Axial section of CT scan showing hypodense area temporal lobe probably due to brain atrophy. **(C)** Enlargement of right lateral ventricle.

(2) CHILD syndrome (congenital hemidysplasia with ichthyosiform nevus and limb defect), (3) phakomatosis pigmentokeratotica, (4) sebaceous nevus, (5) Becker's nevus, and (6) nevus comedonicus syndromes (7).

In this syndrome, there is a mosaic mutation where a few cells have a mutation and other cells will be mutation-free. It occurs sporadically, and it is thought that the inheritance is not found because germline mutation or post-zygotic whole cell mutation is incompatible for life due to its severity. Mutation is found in HRAS and KRAS genes post-zygotically (8). It is said that this disease is a unique example of developmental mosaic RASpathies of phacomatoses as it involves mainly skin and neurogenic organs (3). This syndrome does not have any sex predilection also.

These nevi are commonly found on the scalp (66.8%), face (26.7), and neck (5.5%) (5). In our case, it was on the face (over the nose and cheek). She also had patchy alopecia in the head. Interestingly, her intelligence was normal in spite of the presence of definite findings in brain imaging, though she had twice a history of convulsion at an early age. She had no bony abnormality; serum calcium and phosphate were also normal, but her height was less than that of the age-matched girl (54.3 inches vs. 49 inches). Our case presented bilaterally, but the right eye was more involved than the left. In her right eye, due to optic disk coloboma, her vision was poor and did not improve with refraction. She had dermolipoma and lid coloboma in her right eye, which was excised and reconstructed at 4 years of age, but she still had the evidence of those lesions during her presentation to us. Both her eyes had limbal dermoid-like flat lesion which was away from the visual axis and not disturbing her vision in the left eye. According to the literature, unilateral ocular presentation is common, and the literature showed left eye predominance. A total of 20 bilateral cases were reported up to 2022 (3, 5, 9), and she is the first Bangladeshi child reported as per our best knowledge.

The nevus usually passes through three stages. The first stage extends from birth to puberty, where lesions are small, hairless, and smaller in size due to underdevelopment of the pilosebaceous unit. At puberty, the second stage is started, where glandular enlargement becomes massive and the epidermis becomes verrucous. The third stage is from adulthood to old age, and there is up to 20% chance of developing a tumor from this nevus either benign or malignant. Basal cell epithelioma is the commonest tumor reported (5). Our patient was in the first stage and will soon enter into the puberty stage. She is referred to a dermatologist for management of nevus as prophylactic excision is recommended prior to puberty.

Neurological involvement is the most common syndromic presentation in LNSS and functional is more prominent than structural abnormality. Sometimes, functional abnormalities like subnormal intelligence and convulsion are found in the absence of a definite structural abnormality. Our patient had right-sided cerebral atrophy in the temporal lobe and an enlarged lateral ventricle with only a two times history of convulsion, but intelligence was good.

Due to multiorgan presentation, sometimes, the syndromic association is diagnosed at delay. She visited to several ophthalmologists from her early childhood and underwent surgical intervention but was diagnosed at near puberty. So, any patient presented with lid coloboma and dermolipoma should go for meticulous search for any skin lesion and CNS abnormality to exclude syndromic association.

Conclusion

LNSS is rare and can present to any physician according to the severity of symptoms. These patients may need combined treatment from an internist, dermatologist, ophthalmologist, and neurologist. Updated knowledge of ophthalmologists about this rare disease can enhance proper referral and timely treatment.

Author contributions

SR: planning and article writing. SB: literature review. FH: literature review and native English correction. MK: editing and photography. All authors contributed to the manuscript and approved the submitted version.

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