INTRODUCTION

Feuerstein in 1962 defined the Linear Nevus Sebaceous Syndrome (LNSS) as a neurocutaneous syndrome. Classically, it consisted of a triad of midline facial linear nevus sebaceous, central nervous system and oculal abnormalities. To the best of authors’ knowledge ophthalmic features of LNSS have never been reported in Pakistani population. We report two cases of LNSS, associated with multiple cutaneous nevus sebaceous lesions, complex ocular choristomas and rare bilateral presentation in one patient. Ocular choristomas included limbal dermoids, dermolipomas at superior fornices and choroidal choristoma. Ocular surface was successfully reconstructed by excision of limbal dermoids, partial keratectomy and amniotic membrane transplant.

CASE REPORT

Case 1: A 9 years old female child with subnormal intellect was brought to OPD with progressive protrusion of left eye for the last 2 months. Birth and family history was non-contributory. She had history of fits since 1 year of age. Parents were not concerned about abnormal appearance of the eye which the child had since birth. On examination, she had typical sebaceous nevii involving centre of scalp, left side of forehead, face and neck. These lesions were circumscribed slightly raised yellow colored plaques with linear arrangement and overlying areas of alopecia (Figure 1a). Facial and neck lesions were pigmented too. She also had ipsilateral dental anomalies and facial asymmetry. Ocular examination of right eye was unremarkable. Left eye could barely perceive light. There was dermolipoma at superior fornix, medial corneoscleral limbal dermoid, microcornea, iris coloboma and corectopia (Figure 1b). Left fundus view was precluded by corneal mass. B scan ultrasonography disclosed an echogenic plaque in posterior fundus. CT scan of brain showed left sided hemiatrophy of brain, dilated ventricles and no associated focal lesions (Figure 2). Patient was diagnosed as linear nevus sebaceous syndrome on the basis of oculo-neurocutaneous involvement. These cases provide an interesting constellation of classical phenotypic features in patients, sparingly described dental anomalies and choroidal osseous lesion in one patient, and rare bilateral ocular involvement in other patient.

Ophthalmic Manifestations of Linear Nevus Sebaceous/Organoid Nevus Syndrome

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ABSTRACT

Linear Nevus Sebaceous Syndrome (LNSS) is a rare sporadic oculoneurocutaneous disorder, also classified as Organoid Nevus Syndrome. It consists of a triad of midline facial linear nevus sebaceous, central nervous system and ocular abnormalities. To the best of authors’ knowledge ophthalmic features of LNSS have never been reported in Pakistani population. We report two cases of LNSS, associated with multiple cutaneous nevus sebaceous lesions, complex ocular choristomas and rare bilateral presentation in one patient. Ocular choristomas included limbal dermoids, dermolipomas at superior fornices and choroidal choristoma. Ocular surface was successfully reconstructed by excision of limbal dermoids, partial keratectomy and amniotic membrane transplant.


INTRODUCTION

Feuerstein in 1962 defined the Linear Nevus Sebaceous Syndrome (LNSS) as a neurocutaneous syndrome. Classically, it consisted of a triad of midline facial linear nevus sebaceous, seizures, and mental retardation.1 In 1987 Lambert and colleagues suggested that the triad for this disorder should be changed to include the midline facial linear nevus sebaceous, neurologic abnormalities, and ophthalmologic abnormalities thus redefining the syndrome as an oculo-neuro-cutaneous syndrome. The association is also known as organoid nevus syndrome.

Ocular complications can include tumors/choristomas (of eyelid, conjunctiva, cornea and sclera), coloboma (of lids retina or uveal tissue), squint, nystagmus, vitreous opacities, cornal opacities scleral ossification, retinal anomalies, cataracts, optic nerve hypoplasia and microphthalmia etc.2,3 In addition to ophthalmic features the broad spectrum of anomalies in LNSS may affect multiple organ systems, including central nervous system (brain tumors, hemimegalencephaly and lateral ventricle enlargement). Neurological manifestations are seizures (75%) and intellectual deficit (60%).

This report documents two cases of LNSS in Pakistani population which were presented to the pediatric ophthalmology clinic of a tertiary care eye hospital.
Case 2: An eight-months infant was brought with complaints of abnormal growth in both eyes since birth. He also had classic multiple nevus sebaceous lesions having smooth / verrucous appearance with areas of alopecia on occipital region and left side of head. Ocular and family history was not significant. He had bilateral limbal dermoids and fleshy papillomatous lesion in the right superior fornix (Figure 3). Visual acuity was 6/15 with Cardiff cards although the child showed preference for the right eye. Fundus examination showed non-specific generalized chorioretinal changes. CT scan brain was normal. On the basis of clinical picture, a provisional diagnosis of linear nevus sebaceous syndrome was made.

Excision biopsy and histopathology of the conjunctival lesion showed complex choristoma of conjunctiva lined by stratified squamous layer, subepithelial dense fibrosis, hair follicles, sebaceous glands and mature cartilage. Bilateral limbal dermoids were removed with partial keratectomy and amniotic membrane transplant was done. The child was followed up to 6 years of age. He had regular follow-ups to dermatologist. Binocularly Snellen’s visual acuity on final visit was 6/12. The limbal mass did not recur. He did not develop seizures but had subnormal intellect.

DISCUSSION

Linear nevus sebaceous (nevus sebaceous of Jadassohn) is a congenital hamartoma of skin and its adnexa, pertaining to group of organoid nevus, first identified by Jadassohn in 1895.4 It develops mainly on head and neck region. These cases had typical sites of skin lesions as reported in literature. The most common site is scalp (66.8%), followed by face (26.7%) and neck (5.5%).5 The female child had pigmented lesions, however, pigmentation is not commonly reported in nevus sebaceous lesions.

Both of the cases documented here had sporadic presentation of the syndrome. No significant family history of any congenital disease (especially ocular, neurological or dermatological) was present. Almost all reported cases of LNSS present sporadically except for a single case reported by Bianchine in 1970 that had positive family history of neurological illness.

Ocular features are present in about 10 - 30% of patients with the syndrome.6 So far only one case of sebaceous nevi has been reported in Pakistani population7 that did not have any reported ocular features. Extensive local literature search shows that ophthalmic features have never been reported in association with LNSS in local population before. Choroidal osseous lesions have also been rarely reported in association with LNSS. The first case here, had posterior osseous ocular choristoma as suggested by CT scan, however, a histological diagnosis could not be obtained as the parents refused enucleation. The ocular osseous lesion, anterior epibulbar choristomas, and nevus sebaceous of Jadassohn in this case exactly match with the cases reported by Traboulsi et al.3

Ocular involvement in LNSS is unilateral, most cases being reported in left eye. One of the patients had bilateral ocular lesions. To the best of authors' knowledge, only 6 cases up till now have been reported with bilateral features.8 So this case would be the seventh reported case of nevus sebaceous syndrome with bilateral ocular involvement.

Nevus sebaceous lesions evolve through three dermatologic stages. The first stage is from birth to puberty, involves underdevelopment of pilosebaceous units, lesions are small and hairless, and may regress in size. Stage-2 starts at puberty, and is characterized by massive gland enlargement and the epidermis becoming verrucous. During stage-3, there is 10 - 20% risk that nevus may develop benign or malignant tumors, with the most common pathology being basal cell epithelioma.9 Tumors usually develop in adulthood or old age.10 Prophylactic excision before puberty is recommended. The cause of proptosis in young girl (Case 1) could be due to glandular hypertrophy associated with puberty or it might have an underlying neoplastic process. Enucleation was planned but the
parents refused surgery and lost follow-up. This case also highlights the dilemma that health of female child is not a priority for patients of poor socio-economic status.

Both cases had conjunctival choristomas and corneal opacities due to limbal dermoids. In first case the left eye had fundus lesion and no useful vision. The vision, however, remained good in second case with no posterior segment involvement. Partial keratectomy and amniotic membrane transplant is good option for ocular surface reconstruction in these cases.

Both cases had neurological involvement i.e. first case had seizures and the other child suffered intellectual deficit. The female patient also had skeletal involvement and fibrous dysplasia of mandible ipsilateral to site of lesion.

Although LNSS affect different organ systems, many clinicians are unaware of it, resulting a delay in diagnosis and treatment. It is suggested that all patients of LNSS presenting to ophthalmology clinics must be diagnosed and managed in collaboration with medical internist and dermatologists / plastic surgeons as there is a significant risk of seizures and malignant transformation along the course of disease.

REFERENCES


