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Brief Communication

Shabbir Syndrome: Case Report of a Rare Disease

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Abstract

Purpose: To report a case of Laryngeal Onycho Cutaneous Syndrome in a 10 year old child presenting with an ocular surface mass. Methods: A 10 year old boy presented in the out-patient department with a painless mass in left eye. There was history of airway obstruction requiring tracheostomy in early childhood and recurrent skin ulcerations. Aslit lamp examination revealed fleshy mass arising from the supero-nasal conjunctiva of left eye. Systemic examination revealed ulcerative lesions on cheeks and ears with dystrophic nailsin hands and feet.Results: The ocular surface lesion was treated successfully with topical steroid drops. Conclusion: Steroids may be useful in treating early lesion of ocular surface in LOCS. The importance of regular follow must be emphasised for timely management of frequent granulation tissue outgrowth in bid to avoid development of severe symblepharon and blindness.

Introduction

LOCS (Laryngeal Onycho Cutaneous Syndrome) also known as LOGIC (Laryngeal and Ocular Granulation in children from the Indian subcontinent) syndrome was first described by and named after the renowned dermatologist professor Syed Ghulam Shabbir. It is a rare autosomal recessive condition with only a handful of cases reported in literature. The syndrome is mainly confined to Punjabi Muslim population is characterised by epithelial erosion, nail dystrophies and abundant vascular granulation tissue formation on ocular surface and in larynx. It can not only be lethal form airway obstruction in early childhood but may also cause severe morbidity from skin and ocular affection in cases that survive.

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Case

A 10 year old muslim boy was referred to our outpatient department with the complaint of growth in left eye since 2 weeks. The child was a first born childout of a consanguineous marriage. The parents gave history of multiple recurrent ulcerations on face hands and feet of the child with hoarseness of voice noted in infancy. An episode of stridorin 2nd year of life was treated with urgent tracheostomy.

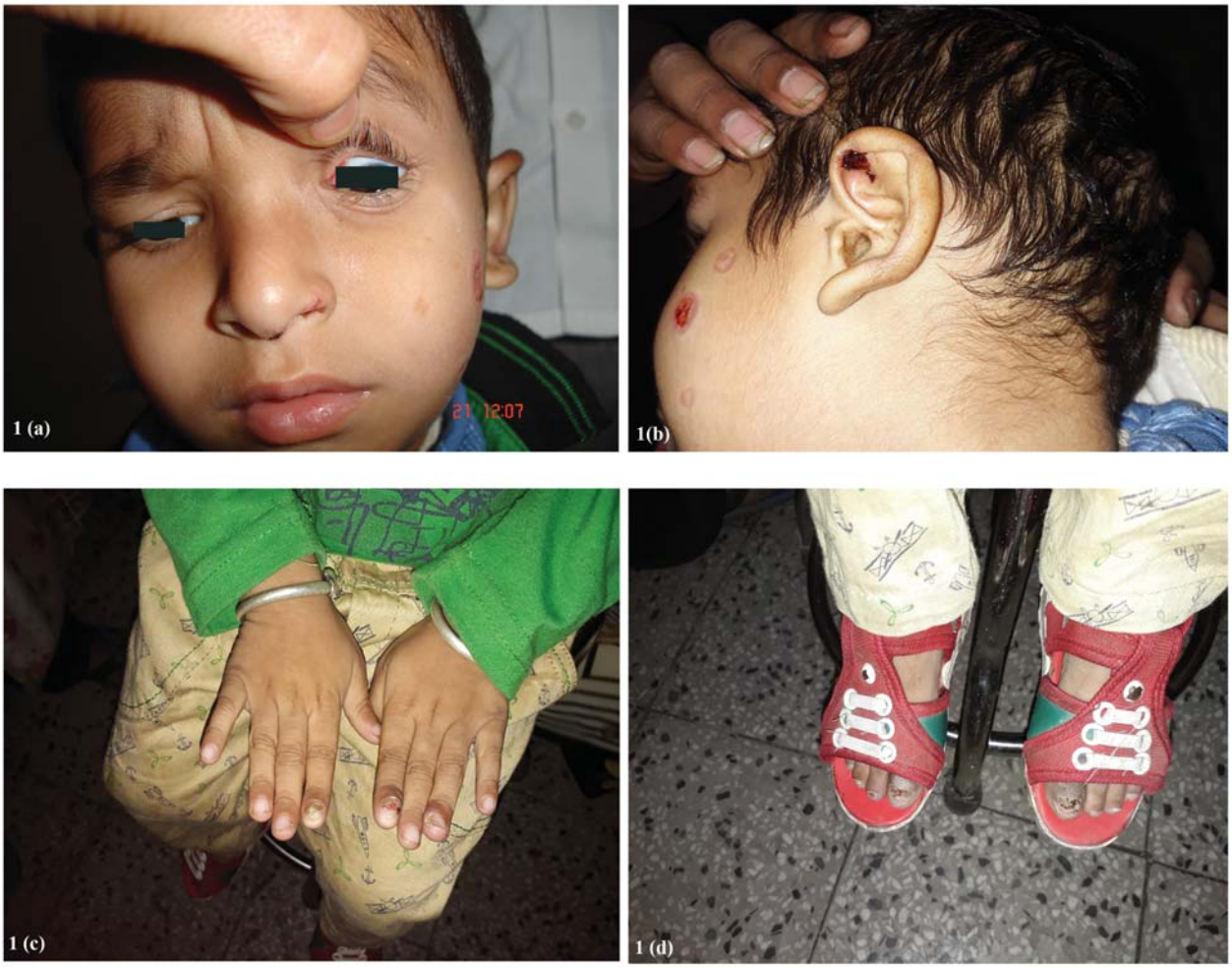
Onocular examination, visual acuity was 6/6 in both eyes. Slit lamp examination revealed apink, fleshy mass arising from the supero-nasal conjunctiva of left ocular surface, sparing the limbus and measuring 2x2 mm.. Anterior segment examination and posterior segment

examination of both eyes was within normal limits.There were multiple ulcerative lesions on cheeks and ears with dystrophic nails in hands and feet. He was diagnosed as a case of Laryngo-oculo cutaneous syndrome (LOCS) and already receiving treatment in department of dermatology and otorhinolaryngology (ENT).

For the ocular lesion topical steroid drops (Flouromethanol) were prescribed three times a day. The ocular surface lesion completely resolved at a follow up of 4 weeks. The child was advisedregular followup with ophthalmology, dermatology and ENT. The parents were also counselled about the genetic nature of the disease the risk of occurrence in further children.

Discussion

LOCS was first described in 1986, by Shabbir and his colleagues. LOCS syndrome is related to a genetic defect at locus 18q11.2. It is now classified as a subtype of junctional epidermolysis bullosae (JEB) resulting from a characteristic mutation inLM332; laminin which plays a crucial role in regulation of wound healing and formation of granulation tissue (Shaheen,2010). It is an epithelial disorder affecting mainly the conjunctiva, skin and ocular surface. It ischaracterised by recurrent skin ulceration, conjunctival erosions, exuberant granulation tissue formation and scarring, and loss of toe and finger nails beginning in early months of life. The voice is effected secondary to vocal cord thickening. The life-



threatening complication of Shabbir syndrome is airway obstruction resulting from a similar process of recurrent erosions and formation of laryngealgranulation tissue which if untreated may lead to premature death (Shabbir,1986). The ocular surface in these patients undergoes recurrent minimal blistering and granulation tissue formation, that may present as ocular surface mass, as in the current case.The conjunctival lesions are reported to start in the lateral portion of the eye and may cause symblepharon formation. The conjunctival granulation tissue may be so extensive that it may result in total palpebral symblepharon, corneal scarring and hence blindness.To date there is no effective pharmaco-therapy available for LOCS (Strauss, 2006). The disease is mostly managed with surgical interventions like excision of granulation tissue, ocular surface reconstruction with AMG for eye and granulation tissue excision and tracheostomy, for airway disease (Goyal,2006). Thalidomide and amniotic membrane transplantation have been found to reduce corneal scarring. The disease is fatal in a majority of patients in early childhood. In patients who survive through first decade there may be disease remission in the second decade of life.

In current case, the boy initially manifestedulcerative lesions on ear, cheeks and limbs and airway obstruction for which tracheostomy had been performed. The ocular affection was milder and later in onset with isolated ocular surface granulation tissue at one location in left eye which resolved completely with topical steroids. There was no need for any surgery for the ocular lesion.

To conclude the ocular lesions of LOCS may vary in severity. In milder and early ocular surface disease, topical steroids may be useful, thus avoiding the need for surgery and consequent scarring and symblepharon formation.The parents should be made aware of the need of regular followup in ophthalmology department for timely management of recurrent granulation tissue.

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