CASE REPORTS
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LOC SYNDROME - A CASE TO UN"LOC" OUR MINDS

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Abstract

Laryngo-onycho-cutaneous syndrome (LOCS) or Shabbir's syndrome is an inherited autosomal recessive disorder affecting consanguineous Muslim families of Punjabi origin. In this condition excessive dermal and submucosal granulation tissue formation leads to hoarse/weak cry, respiratory obstruction, pterygium and symblepheron in the eye. It represents a distinctive form of junctional epidermolysis bullosa (JEB) affecting laminin alpha-3 (LAMA3) gene. All the patients reported so far are from Muslim community and of consanguineous parentage. But our patient is born of non-consanguineous parentage, is a non- Muslim (Hindu) and hails from Chattisgarh province in India with a long survival age.

Key words: Junctional Epidermolysis Bullosa (JEB), Shabbir's syndrome, Laryngo- onycho- cutaneous syndrome (LOC)

Introduction

Laryngo- onycho- cutaneous syndrome (LOC) or Shabbir's syndrome, is an inherited autosomal recessive disorder that affects mainly the offspring of consanguineous Muslim families originating in the Punjabi region of Indian subcontinent.[1] The disease presents with hoarseness of voice, blisters, erosions, ulcerations, dystrophic nail changes, eye changes and deformed teeth. In this condition excessive dermal and submucosal granulation tissue formation leads to hoarse/weak cry, pterygium and symblepheron in the eye and respiratory obstruction which may lead to premature death.

Case Report

A female aged 36yrs from Chhattisgarh, born of non-consanguineous marriage, developed multiple, painful fluid filled lesions over elbows, knees, trunk, back, scalp and extremities since age of 2 months. They used to occur on and off up to the age of 12yrs and used to heal in 2-4 weeks forming scars. Nail changes and dental abnormalities were seen since the age of 15yrs. (Figure 1-7)

She has history of feeble cry and hoarse voice since childhood and recurrent episodes of difficulty in breathing and consulted an ENT surgeon. She was found to have vocal cord thickening and nodules. As it became increasingly difficult for her even to breathe, emergency tracheostomy was done 2 years back at the age of 34 yrs. Since 2 years she started developing redness of eyes with watering, swelling and obstruction of vision.

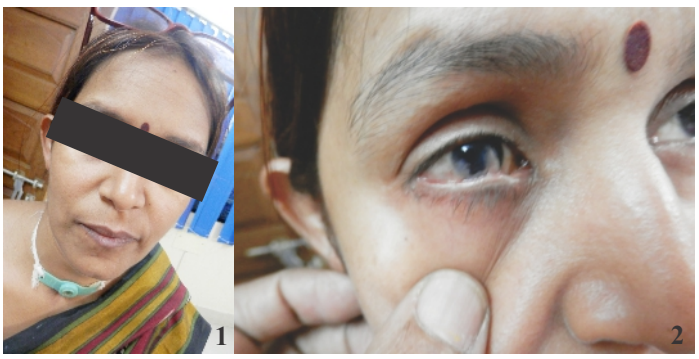

Figure 1 & 2: 1: A permanent tracheostomy. **2:** Pterygium encroaching on to the cornea (rt) and granulation

Routine investigations were all normal. Biopsy showed unremarkable epidermis with patchy dermal lymphocytic infiltration. Immunohistochemistry for G71 and GB3 was requested but it was not done as they were not available.

On the basis of these distinctive clinical features final diagnosis of laryngo-onycho-cutaneous syndrome was made.

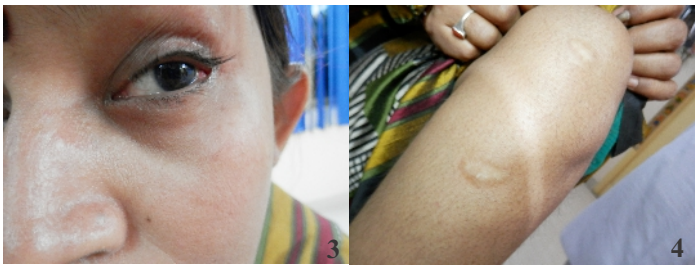

Figure 3 & 4: 3: Symblepharon (adhesion of the palpebral conjunctiva of the eyelid to the bulbar conjunctiva). **4:** Old healed and atrophic scars over lower legs

Discussion

LOC Syndrome (laryngo-onycho cutaneous syndrome) or LOGIC syndrome[2] was first reported by Shabbir[1] in 1986, in Muslim families of Punjab origin; subsequently there were reports of similar cases from UK, Australia and all of these families originally belonged to Punjab Province of Pakistan or India[2,3,4]. The mystery of this syndrome was unravelled in 2003

when McLean et al[5], observed mutations in a candidate gene, laminin alpha-3 (LAMA3) located on chromosome 18q11.2, in which loss of expression mutation also cause Junctional Epidermolysis Bullosa (JEB). In LOC syndrome the causative mutation was frameshift mutation (N-terminal deletion) of laminin 3a isoform. Based on this, it has now been finally established as a subtype of JEB[6,7].

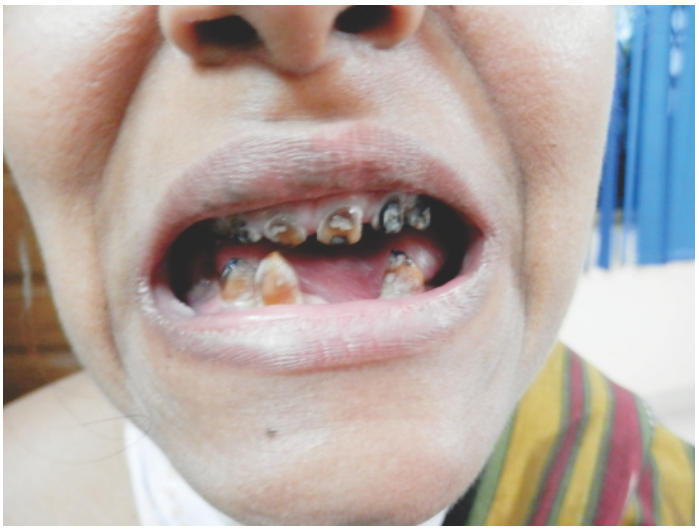

Figure 5: Loss of teeth, discolored and distorted teeth with caries

In contrast to the other JEB subtypes, patients with LOC syndrome have minimal blistering and extensive granulation tissue formation[7,8] which leads to chronic non healing ulcers, dystrophic nail changes, vocal cord thickening and thickening of conjuctival tissue, clinically manifesting as hoarse voice or weak cry at infancy, respiratory obstruction, failure of tooth enamel formation and marked dental malformations.

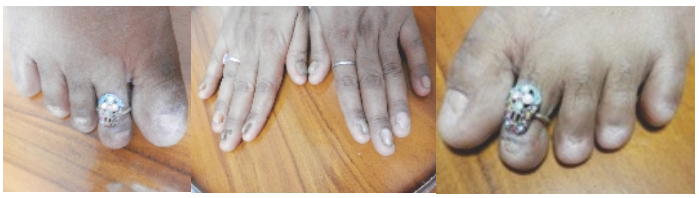

Figure 6: (a, b & c) - Twenty nail dystrophy

Although eye involvement in LOC syndrome was not mentioned in the original description, ocular granulation tissue resembling pterygium was reported in all subsequent patients[2,3,4,8] and is a prominent feature in our patient also. Our patient also had symblepheron, granulation tissue at the lateral border of left eye and her visual acuity was diminished. Nail dystrophies have been reported in all patients but twenty nail dystrophy as seen in our patient is unusual.

All the patients reported so far are from muslim community and of consanguineous parentage. But our patient is born of non-consanguineous parentage, is a non- Muslim (Hindu) and hails from Chattisgarh province in India.

Most of the patients die due to respiratory obstruction and infections and do not survive beyond second decade. Our patient is now 36 years old and she is the longest surviving individual affected with this syndrome so far.

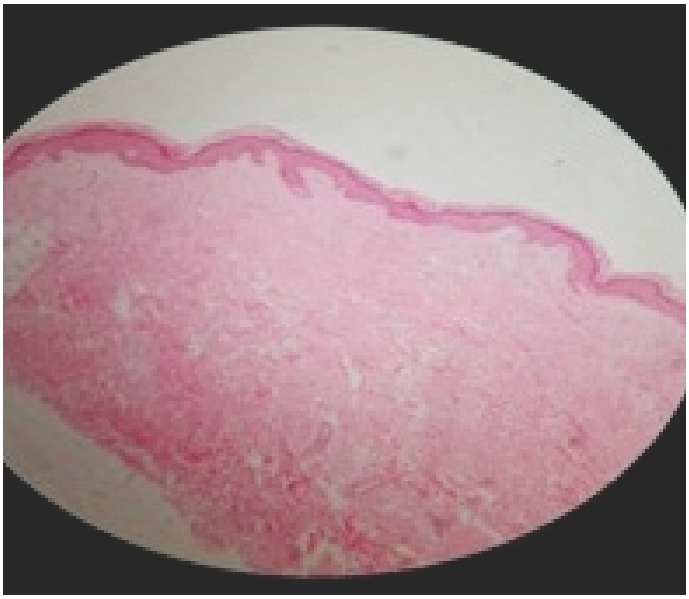

Figure 7: Histopathology of skin (haematoxylin and eosin, 10X)

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