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Syed Ghulam Shabbir (1923-2002) and His Syndrome

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ABSTRACT

Professor Syed Ghulam Shabbir (1923-2002), *is one of the Pakistani well-known dermatologists. In* 1986, *Shabbir and his colleagues described a novel autosomal recessive syndrome*, *which they called laryngoonychocutaneous syndrome. They reported this condition*, *in* 22 *patients in* 12 *families living in Lahore*, *Pakistan. This syndrome is characterized by cutaneous erosions*, *nail dystrophy and exuberant vascular granulation tissue in certain epithelia*, *especially conjunctiva and larynx. This report sheds light on Shabbir and the syndrome that bears his name.*

***Keywords*:** *Laryngo Onycho Cutaneous Syndrome*, *Shabbir Syndrome*, *Genodermatoses*

1. Introduction

Professor Syed Ghulam Shabbir (1923-2002) (**Figure 1**) is one of the Pakistani renowned dermatologists [1,2]. Among his great contributions to dermatology, he is credited for describing, with his colleagues, a syndrome [3], that was later known as Laryngo Onycho Cutaneous Syndrome (LOCS) or Shabbir syndrome [4].

LOCS [5-8] (OMIM 245660) is also known as LOGIC syndrome [5] (Laryngeal and Ocular Granulation in children from the Indian subContinent).

It is a rare autosomal recessive condition with only a few cases reported worldwide [5].

Figure 1. Professor Syed Ghulam Shabbir (1923-2002).

The syndrome is an epithelial disorder, which is confined to the Punjabi Muslim population. It is characterized by cutaneous erosions, nail dystrophy and exuberant vascular granulation tissue in certain epithelia, especially conjunctiva and larynx .The disorder may result in corneal scarring and blindness [4].

The syndrome was mapped [5] to Gene map locus: 18q11.2.

Manifestations appeared during the first months of life and included skin ulceration, recurrent loss of toenails and fingernails, and conjunctival scarring. Other epithelial surfaces were later involved. The voice was sometimes affected because of vocal cord thickening and/or nodules; in some children a weak cry was noted at birth. Some of the affected persons had amelogenesis imperfecta. No impairment of immune function was detected. The main complications, however, occur in the respiratory tract, where a similar process of erosions and subsequent formation of granulation tissue causes airway obstruction which may lead to premature death [8].

Patients with Shabbir's syndrome have minimal blistering and extensive granulation formation .The conjunctival lesions start in the lateral portion of the eye and result in symblepharon. The conjunctival granulation tissue often leads to total palpebral occlusion and blindness. Conjunctival granulation tissue is rare in other variants of junctional epidermolysis bullosa (JEB) [2].

LOCS was reclassified as a subtype of JEB [6] based

on clinical features with a unique mutation affecting the N terminus of the alpha3 chain of LM332. LOCS is now believed to be a nonblistering variant of JEB [8].

Some studies showed that the laminin alpha3a N-terminal domain (LAMA3gene) is a key regulator of the granulation tissue response, with important implications not only in LOC but in a range of other clinical conditions associated with abnormal wound healing [4].

The tissue localization of the laminin α3A corresponds to the clinical manifestations of Shabbir's syndrome *i.e.* with LM332 variant. This applies to the skin, nail, and mucous membrane fragility while with LM311 variant, which is present in the lungs; these patients are also susceptible to pneumonia. Similarly, missense mutations in the tumor suppressor gene encoding p63 protein result in reduced p63 expression in Shabbir's syndrome which might be related to the corneal granulation overgrowth and ocular changes in these patients. Immunofluorescence mapping reveals type IV collagen in the floor and bullous pemphigoid 180 antigen in the roof of blister *i.e.* cleavage occurs in the lamina lucida [2].

To date there are no efficacious treatments available for LOCS [8]. The disease is refractory to pharmacotherapy and often surgical interventions like tracheostomy, suprapubic catheterization are required [2]. Vascular laser therapy showed encouraging results in laryngeal lesions in one case [5]. Thalidomide and amniotic membrane transplantation were successfully used to reduce corneal scarring. The majority of patients succumbs to the disease during childhood; nevertheless, in those who survive the condition remits in the second decade [2].

Some authors [8] report a 16-year-old girl with LOCS who failed to respond to methylprednisolone and cyclophosphamide, but had a partial response to oral thalidomide with marked decrease in granulation tissue and tracheal secretions. Interruption of treatment resulted in prompt resurgence of the granulation tissue which was again controlled by reintroduction of thalidomide. The authors proposed that in the absence of effective therapies for LOCS, a trial of thalidomide in these patients should be considered [8].

Amniotic membrane transplantation with symblepharon lysis have been performed effectively , for ocular surface reconstruction in the management of epidermolysis bullosa and other conditions that cause corneal scarring and symblepharon in children, like LOCS. In one series [7], children with epidermolysis bullosa fared better and the effects of surgery lasted longer compared with patients with other causes of symblepharon and massive pannus [8]. In that report, only the patient with laryngo-onychocutaneous syndrome had recurrence of granuloma , at 9 months after surgery [7].

LOCS is first described by Shabbir and his colleagues, in 1986. Shabbir is an eminent Pakistani dermatologist, while working at Mayo Hospital, Lahore; he observed a new, distinctive, recessively inherited disease which exclusively occurred in Muslim children of Punjab Province of Pakistan. He and his colleagues reported the disorder, in 22 patients in 12 families living in Lahore, Pakistan.

Professor Shabbir was born in Lahore on 20-02-1923. In 1960, He founded dermatology department at, King Edward Medical College, Mayo hospital, Lahore, Pakistan [1]. It is the Largest Skin Unit in Pakistan. He was the head of the department from 1960-1987.

Professor Shabbir is one of the founders of dermatology in Pakistan. He had participated in many assignments related to Pakistan Association of Dermatologists and its journal.

He was, also, involved actively, in teaching dermatology.

He died on 24-08-2002. Currently his son, Professor Atif Hasnain Kazmi is the head of the dermatology department at, King Edward Medical College, Mayo hospital, Lahore and the President of Pakistan Association of Dermatologists (PAD).

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