

X-linked ichthyosis along with epidermolysis bullosa

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Abstract

Ichthyoses are a heterogenous group of hereditary keratinization disorders that share in common the accumulation & shedding of large amounts of hyperkeratotic epidermis. Early reports of ichthyosis in the Indian and Chinese literature date back to several hundred years. X-linked recessive ichthyosis (XLI) is a common disorder of keratinization and affects males who inherit an X-chromosome having a steroid sulphatase genetic mutation. In the present communication we report a case of XLI and dystrophic epidermolysis bullosa in the same patient. To the best of our knowledge it has been reported only once before.

Keywords: Epidermolysis bullosa, Ichthyosis, Keratinization

Introduction

Ichthyoses are a heterogenous group of hereditary keratinization disorders that share in common the accumulation and shedding of large amounts of hyperkeratotic epidermis.^[1,2] The name ichthyosis is derived from the Greek ikthys meaning “fish” and refers to the similarity in appearance of the skin to fish scales. Early reports of ichthyosis in the Indian and Chinese literature date back to several hundred years.^[3] X-linked recessive ichthyosis (XLI) is a common disorder of keratinization and affects males who inherit an X-chromosome having a steroid sulfatase genetic mutation. This mutation causes deficiency of steroid sulfatase.^[4] In majority of the XLI cases, the Steroid sulfatase gene is totally depleted as confirmed by polymerase chain reaction techniques.^[5] It typically presents as dark-brown polygonal scales on different parts of the body surface. The lesions are usually distributed symmetrically and are generally more evident on the extensor aspects of the limbs, particularly on the lower extremities.^[6] Dystrophic epidermolysis bullosa (DEB) is inherited as an autosomal recessive or dominant trait and is due to COL7A 1 gene mutations.^[4] It presents as large blisters, both spontaneously and secondary to mechanical forces, typically on trauma-exposed sites or

over bony prominences. The blisters heal with scarring and sometimes with hypo or hyperpigmentation. In the present communication, we report a case of XLI and DEB in the same patient. To the best of our knowledge, it has been reported only once before.^[3]

Case Report

A 20-year-old student reported to the Department of Oral Medicine and Radiology with history of bleeding and swelling of gums since 3 months [Figure 1]. In addition, he also complained of loss of nails from the hands and feet of long duration. He gave a history of extreme dryness of the skin all over the body. Further questioning revealed that there was history of blisters occurring over the extremities since early infancy. He was diagnosed to have DEB. A skin biopsy performed at 8 months of age showed subepidermal blistering. The patient's parents were non-consanguineous and he was born of a normal vaginal delivery. However, the mother also had loss of nails of both hands and feet since a long time. Definite history of blistering in the mother was also obtained.

Examination of the oral cavity revealed swollen gums with hypoplastic teeth. There was periodontitis, transposition of 13, and Class III malocclusion. There were no blisters in the oral cavity. Panoramic radiological examination revealed nothing significant.

Examination of the skin revealed large dark-brown polygonal scales, most marked on the limbs, especially the lower limbs [Figure 2]. There were hypopigmented atrophic scars on dorsa of hands and elbows [Figures 3 and 4]. Nails from all the digits were absent. A dermatological consultation was sought. A diagnosis of X-linked ichthyosis was made and confirmed on histopathology. In view of the history of blistering since early infancy and absence of nails with scarred areas, diagnosis of DEB was agreed to by the dermatologist. However, in view of lack of investigations, namely, transmission electron microscopy and immunofluorescence microscopic mapping, no further investigations were done for epidermolysis bullosa.

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Figure 1: Extraoral profile of the patient



Figure 2: Hypopigmented scars



Figure 3: Epidermolysis bullosa on hands



Figure 4: Epidermolysis bullosa on feet



Figure 5: Figure showing swollen gums



Figure 6: Intraoral view

Discussion

This is the second report of a patient with XLI and DEB, both of which are genetically unrelated. The oral lesions in DEB range from mild to extremely severe [Figure 5 and 6]. Most

individuals with dominant dystrophic forms of inherited EB develop oral mucosal lesions that are characteristically larger (>1 cm), and more numerous than those observed in EB simplex, often more erosive, and are usually quite painful.^[7] These are more severe in the recessive type of DEB. In

dominant DEB, as in our patient, there is infrequent blistering. However, one has to be careful as blistering can be induced easily, and dental treatments should be approached with extra diligence to reduce soft tissue trauma. Oral ulcerations in DEB can affect all areas of the oral mucosa and heal with scarring except in dominant type.^[8] This can result in marked changes in the oral architecture. Over time, oral blistering may lead to obliteration of the vestibule, ankyloglossia, and microstomia.^[9] Swollen and bleeding gums in our patient were probably not related to the DEB. This could have been due to poor oro-dental hygiene. Despite normal salivary secretion in most patients with DEB, the oral cavity tends to be inoculated with high numbers of bacteria and there tends to be excessive tooth plaque formation, which further promotes dental caries. In patients who are severely affected, there is tremendous difficulty performing normal oral hygiene due to their extreme soft tissue fragility. Moreover, there is an added risk of developing oral squamous cell carcinoma in individuals with recessive DEB.^[10]

Our patient, fortunately, had a recessive form of DEB. However, the scales of XLI were quite typical. Regular use of keratolytics and moisturizers was advised to the patient. However, in view of the coexistence of DEB, the patient was advised to rub the moisturizers gently. We report this case because the association of these two genetic disorders has not been reported in Indian literature before. Management of oral lesions in such patients requires special skills because minor operative procedures can possibly produce ulcerations and blisters.

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