RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA WITH CONGENITAL CATARACT

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case having recessive dystrophic epidermolysis bullosa with severe blistering, scarring, milia, mucosal ulcerations, corneal involvement, teeth anomalies and deformities of hands and feet had the unusual feature of congenital cortical cataract.

Key words : Epidermolysis bullosa recessive dystrophic, Congenital cortical cataract.

Epidermolysis bullosa comprises a group of genetically determined disorders characterised by blistering of the skin and mucosae, the blisters may result from minor mechanical injury or arise spontaneously.[1] Although these disorders have this feature in common and are often described as forms of a single disease, these are genetically distinct diseases mediated by different mechanisms. Recessive dystrophic epidermolysis bullosa (RDEB) is most notable for its severe scarring and deformity and by severe involvement of mucous membranes.[2,3] In this disorder, erosions and blisters are usually manifested at or shortly after birth.[2] Although some blisters may appear to occur spontaneously, most seem to arise at sites of pressure or trauma.[2,3] In RDEB, nails may show extreme involvement with severe dystrophy or complete absence of the nail bed.[4,6] Patients with RDEB may have severe involvement of the oral cavity, oesophagus, eye and destroyed teeth.[5,6] Hair growth may be sparse.[6] The eyes may develop a number of characteristic changes: blepharitis, symblepharon, conjunctivitis or keratitis, or both with associated vesicle formation and corneal opacity.[2,5] Congenital cortical cataract as an associated feature has not been reported earlier in the literature.

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Case Report

An 11-year-old boy was admitted for recurrent blisters which healed with scarring since birth. One elder sibling died due to a similar illness at the age of six months. Another elder sibling was healthy and normal. History of consanguinity was present in his parents. The child was anaemic and short-statured for his age, and there were multiple, large, tense bullae, deep ulcerations with thick crusts and slow healing, mainly seen on the flexures and extremities. The blisters healed with marked scarring and the formation of milia. The hands and feet were deformed and assumed a mitten appearance, with the thumb separate from the four fused fingers. Nails were all lost. There were oral mucosal ulcerations and the molar teeth were deformed and showed severe caries. Patchy cicatricial alopecia was present at the frontal aspect of the scalp. Another interesting aspect noticed was that the child had congenital cortical cataract in the left eye, whereas only light perception was present in the right eye due to keratitis. The general intelligence was normal but the growth of the child was impaired because of deformities.

Comments

A clinical diagnosis of RDEB was made in our patient by the family history of consanguinity, severe blistering and scarring with milia, mucosal ulceration and teeth anomalies and mitten deformities of hands and feet.

Mucosal ulcerations, corneal involvement, teeth anomalies and scarring of conjunctiva have been described[5,6] but not the involvement of lens in the form of cortical cataract.

This child was anaemic and RDEB patients commonly are anaemic and physically small in size.[6] The growth of the child was impaired but the mental development was normal. While growth and development and physical functions may be greatly restricted in patients with RDEB but they can survive to their thirties and forties.[3]

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