Generalized Epidermolysis bullosa with Congenital Synechiae-Associated Malformations and Unusual Ultrastructure: A New Entity

V.P.

Sybert

Children's Hospital and Medical Center, Seattle, Wash., USA

V.P. Sybert, Children's Hospital and Medical Center, 4800 Sand Point Way NE, PO Box C5371, Seattle, WA 98105 (USA)

To the Editor

I read with interest the case report of Taieb et al. (Dermatologica 1988;176:76-82) on an infant born with eyelid synechiae and areas of erosions on the skin and a cleft palate, which only now came to my attention.

I believe this infant fits into the syndrome of AEC (ankyloblepharon filiforme adnatum, ectodermal dysplasia and cleft lip $\pm$ cleft palate). The most characteristic features of this disorder are the adhesions between the eyelids and the clefting defect. In 2 infants with this disorder (also known as Hay-Wells syndrome) we have seen marked erosive changes of the skin which very soon after birth go on to heal. In the second of 2 infants under our care with this condition, a collo-dian membrane was present and peeled at birth leaving areas of erosions. The low-set

and unusual ears were also present in both of our patients. The thickening of the stratum corneum is also typical.

In summary, I believe that this infant has a recognized autosomal dominant disorder and there is a very low recurrence risk. The cleft palate is part of the disorder and not a fortuitous event.

A. Taieb J. Sarlangue

Services de Dermatologie et de Néonatalogie. Hôpital des Enfants, Bordeaux, France

Services de Dermatologie et de Néonatalogie Hôpital Pellegrin-Enfants F-33076 Bordeaux Cédex (France)

The diagnostic proposal of Dr. Sybert for our case published 3 years ago is most probably right. When reviewing photographs published in previous reports [1-3], there are indeed striking fades similarities. At the time of our report, we were mostly concerned by skin fragility and widespread erosions, which had not been noted previously in AEC syndrome. The case was isolated, and the patient died at 3 days of life, increasing diagnostic

Reply

difficulties. Epidermolysis bullosa was the diagnostic proposal, and electron-microscopic studies were consistent with this hypothesis. Clinical slides and histologic sections were circulated, and the case was presented at the World Congress in Berlin in 1987 before a decision of publication was made.

Finally, it should be stressed that AEC syndrome must be added to the list of differential diagnoses in congenital epidermolysis bullosa and that this diagnosis can be made (now quite easily!) on the basis of associated symblepharon, cleft palate and typical facies.

References

Hay RJ, Wells RS: The syndrome of ankyloblepharon, ectodermal defects and cleft lip and palate: An autosomal dominant condition. *Br J Dermatol* 1976;94:277-289. Spiegel J; Colton A: AEC syndrome: Ankyloblepharon, ectodermal defects, and cleft lip and palate. *J Am Acad Dermatol* 1985;12: 810-815.

Happle R: Genetic defects involving the hair; in Orfanos CE, Happle R (eds): *Hair and Hair Diseases*. Berlin, Springer. 1990. pp 330-331.

© 1992 Karger AC. Basel 1018-8665/92/1843-0227

\$ 2.75/0