HEREDITARY BENIGN INTRAEPITHELIAL DYSKERATOSIS

K Pavithran

A 21-year-old male had hereditary benign intra-epithelial dyskeratosis. His mother and younger brother also were found to have the same disease. All the three had oral and conjunctival lesions. The histopathological features were characteristic.

Key words : Hereditary benign intra-epithelial dyskeratosis, Dyskeratosis, Leucoplakia.

Hereditary benign intra-epithelial dyskeratosis is a rare congenital syndrome characterized by the development of asymptomatic, soft, whitish patches and plaques on the oral mucosa and bulbar conjunctiva.[1,2] The trait has an autosomal dominant transmission with a high degree of penetrance. We report a case of hereditary benign intra-epithelial dyskeratosis in a young male. His younger brother and mother also were found to have the same disease.

Case Report

A 21-year-old male born to non-consanguinous parents was seen for asymptomatic whitish thickening of the oral mucosa and recurrent attacks of redness of the bulbar conjunctiva since early childhood. It gradually progressed in severity in spite of applying various eye ointments. His younger brother aged 18 and mother aged 45 years also were having the same disease since childhood.

Examination revealed diffuse, soft, white, non-indurated thickening with minute foldings of the buccal and labial mucosae (Figs. 1 and 2). Near the corners of the mouth, buccal mucosa showed soft plaques covered with fine lines that divided the surface into small rectangles. The white macerated superficial cells of the patches could be scraped off easily but the underlying lesion could not be removed in that fashion. When the affected mucosa was stretched, minute, pin-point, opaque elevations on the surface became apparent. The tongue, pharynx, palate, gingiva and fauces remained unaffected. The teeth were normal. Anal, nasal and penile mucosae also were normal.

Fig. 1. Diffuse white thickening of the cheek mucosa.

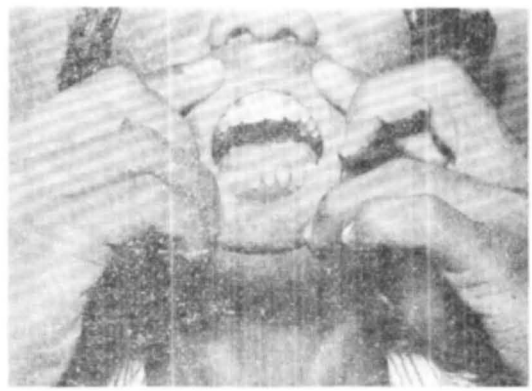

Fig. 2. Diffuse white patches and plaques on labial mucosa.

From the Department of Dermatology and Venereology, Medical College Hospital, Kottayam-686 008, India.

Both eyes showed foamy, gelatinous plaques overlying the hyperaemic bulbar conjunctivae and these resembled pinguenculae (Fig. 3). The vision in both eyes was normal. Fundus examination did not reveal any abnormality. Examination of the mother and younger brother showed lesions in the mouth and eyes (Fig. 4), similar to those described for the reported case.

Routine laboratory tests on blood, urine and stools were normal. Blood VDRL test was negative. Fungus was not seen in KOH preparations of scrapings taken from the mucosal lesions. Histopathological study of the biopsy specimens taken from the buccal mucosa and bulbar conjunctiva showed increased thickening of epidermis with hydropic degeneration of the cells of stratum spinosum (Figs. 5 and 6) and benign dyskeratosis characterized by hyaline eosinophilic cells and 'Cell-within a cell' pattern.

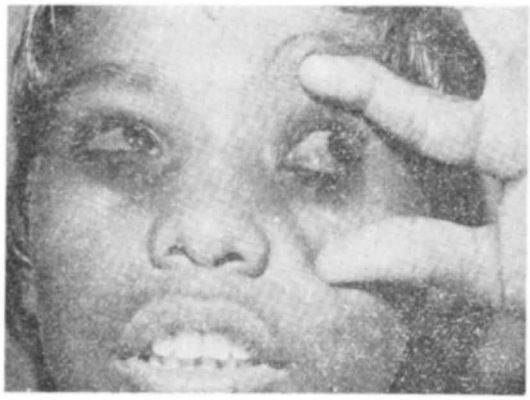

Fig. 3. Foamy gelatinous plaques overlying the hyperaemic bulbar conjunctiva.

Fig. 4. The hyperaemic bulbar conjunctiva in the younger brother of the patient.

Comments

Hereditary benign intra-epithelial dyskeratosis is an extremely rare disease. More important

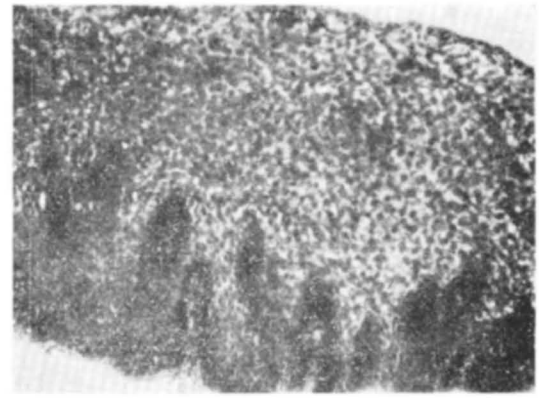

Fig. 5. Histopathology of oral lesion. There is no horny layer. Epithelium is acanthotic and shows extensive hydropic degeneration and dyskeratosis of the cells of the stratum spinosum (X 40).

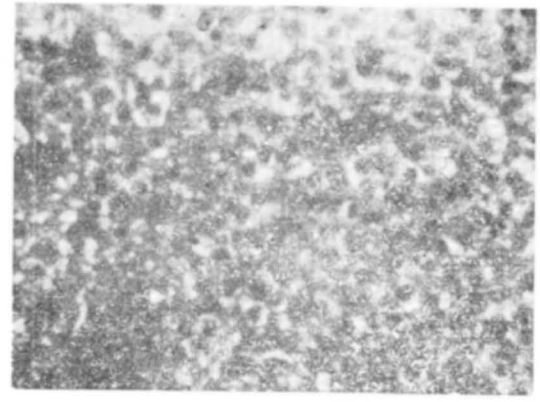

Fig. 6. Hydropic degeneration of the cells of stratum spinosum and benign dyskeratosis characterized by hyaline eosinophilic cells and 'Cell within a cell' pattern (X 400).

for the clinician is to differentiate this condition from the common disorders affecting the mucosa of the oral cavity and conjunctiva. The mucous membrane lesion of the oral cavity in hereditary benign intra-epithelial dyskeratosis may resemble white sponge nevus of Cannon, clinically and histopathologically. But absence of anal and vaginal lesions and presence of conjunctival lesions are characteristic of hereditary benign intra-epithelial dyskeratosis.[3] Eosinophilic dyskeratotic cells and 'cell within a cell' pattern also help in their differentiation. Softness of the involved area, lack of induration and the superficial character of the oral lesion help to differentiate this condition from leucoplakia. The ocular lesions may resemble pingueculae. About a quarter of the affected persons give a history of a flare of ocular lesions in early spring manifested by hyperaemia, photophobia and increased lacrimation. But in spite of extensive conjunctival lesions, our patients remained asymptomatic.

The cause of hereditary benign intra-epithelial dyskeratosis is unknown. It is transmitted as an autosomal dominant trait with a high degree of penetrance. In our case, 3 members in a family—mother and her two children, were affected. Various anomalies reported in association with this disorder include hare lip, cleft palate, scrotal tongue, ankyloglossia, torus mandibularis, polydactyly and congenital heart disease.[3] But our patient had none of these or other defects. Since the anomaly is not associated with neoplasia or premature death from other causes, treatment is either symptomatic or not indicated.

References

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