Hereditary Benign Intra-Epithelial Dyskeratosis (HBID) (Witkop-Von Sallman Syndrome)

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Abstract:
Hereditary Benign Intraepithelial Dyskeratosis (HBID) is a rare autosomal dominant recurring disorder of the oral and ocular mucosa first described in 1960 among Haliwa Native American Indians in North Carolina. A few cases have been reported also in other parts of the United States, South America and Europe. This report describes a case with clinical and histopathological features of HBID.

Case Report:
A 15-year-old Egyptian white female born in Qatar was referred to the Dermatology Department for evaluation of recurrent asymptomatic white oral lesions since early neonatal life. The pediatric history recorded recurrent attacks of apparent oral thrush and chronic oral candidiasis with unsatisfactory responses to both topical and systemic antifungal treatment. Apart from the oral lesions and recurrent eye irritation, redness and lacrimation, the patient denied any systemic complaint or family history of a similar condition.

On examination the oral lesions were white rough diffuse and folded plaques on the dorsal aspect and sides of the tongue and buccal mucosa (Figures 1A, 1B, 1C, 1D). The ocular lesions, diagnosed as allergic conjunctivitis, were dilated vessels in both conjunctivae. The patient was otherwise healthy with no other mucous membrane lesions, and the skin, hair and nails showed no abnormalities. HBID, white sponge disease, and other conditions causing developmental oral leukoderma in the mouth were considered in the differential diagnosis.

Routine clinical chemistry and hormonal (thyroid, parathyroid and adrenal gland) profiles and humoral and cellular immunity were within normal ranges. A 4 mm punch biopsy specimen of a tongue lesion stained with

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Figure 1D

hematoxylin and eosin and Periodic Acid Schiff (PAS) showed increased epithelial thickness and inflamed hyperplastic mucosa (Figure 2). Numerous large vacuolated cells and dyskeratotic cells were present mainly in the superficial part of the epidermis (Figure 3), with some appearing to be "engulfed" by the surrounding cells in a "cell-within-cell" pattern (Figure 4). Surface candidal pseudohyphae and spores were seen occasionally in PAS stained sections (Figure 5).

Discussion:

HBID is a rare hereditary disease first described by Witkop et al and by Von Sallmann and Paton (1,2) in 1960 in a triracial isolate (white-black-Indian) in North Carolina, USA. In 1981, more cases were detected in other American states in descendants of North Carolinian ancestors. (3) HBID has also been reported in other parts of USA, Europe and South America. (4-10) HBID affects oral and ocular mucosa. The onset is usually at birth or early childhood with persistent waxing and waning. Oral lesions are usually asymptomatic, may vary in extent and are typically bilateral. Most oral lesions go unrecognized until examined. (1) Ocular lesions in HBID also begin early in life. Eye lesions are typically characterized by the development of white granular to gelatinous triangular, perilimbal and bulbar conjunctival plaques associated with dilated conjunctival vessels which are responsible for its most striking feature "red eye". The lesions are typically bilateral and may encircle or extend over the cornea. (2,8)

All this was true for our patient who had been followed since birth as a case of oral thrush in the beginning and thereafter as chronic oral candidiasis and allergic conjunctivitis; the ocular lesions consisted of prominent dilated conjunctival vessels and she suffered frequently...
from eye irritation, redness, lacrimation, foreign body sensation, and occasional photophobia.

Characteristic histological findings in samples of oral and eye lesions include an increase of the epithelial thickness with numerous dyskeratotic cells in the superficial layers and cells appearing to be engulfed by normal cells giving the so called "cell-within-cell" pattern. Infiltrations of chronic inflammatory cells are present beneath the intact epithelial basement membrane as in our patient (Figures 2, 3, 4) plus occasional Candida pseudohyphae and spores found in the superficial epithelium (Figure 5). The presence of Candida albicans in HBID section was previously reported as an accidental finding during an ultrastructural study.\(^\text{(11)}\)

HBID is rare autosomal dominant condition with incomplete penetrance.\(^\text{(1)}\) It is associated with chromosome 4 anomalies that have been localized recently to chromosome 4q35.\(^\text{(12)}\) Other etiologic possibilities such as mutation and influence of environmental factors need to be considered.\(^\text{(13)}\) Genetic analysis could not be carried out in our patient. Her negative family history may be explained by incomplete penetrance of the trait or by the possibility of a new mutation but, we are satisfied that the case that we describe in this Egyptian girl fulfills the criteria for a diagnosis of HBID.

References: