OCULODENTODIGITAL DYSPLASIA WITH CUTANEOUS KERATOTIC PAPULES

A Case Report

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Summary

A case of oculodentodigital dysplasia is described with some unusual findings like cutaneous keratotic papules and Duane's retraction syndrome, which have not been documented earlier.

KEY WORDS: Oculodentodigital dysplasia, Cutaneous keratotic papules, Duane's retraction syndrome. Camptodactyly.

Oculodentodigital dysplasia is a very rare hereditary syndrome which consists of ophthalmic and digital anomalies with enamel hypoplasia[1]. The present case is being reported because in addition to anomalies of eyes, digits and enamel it manifested cutaneous lesions which have not been described earlier. Further, the eye changes were those of Duane's retraction syndrome rather than the usual microphthalmia.

Case Report

A five year old female child of nonconsanguinous parentage presented to the skin clinic with asymptomatic yellowish white spots on hands of three years' duration.

On examination the child was found to have flexion deformity of little fingers and toes which on questioning was reported to have been noted at the time of birth. She also had squint and the signs of Duane's retraction syndrome type B (Complete abolition of abduction, normal adduction and slight retraction of the eye ball) in the left eye (Fig. 1). Dental examination revealed imperfect formation of enamel and dentine in the upper and lower incisors and molars. Right upper first molar showed carious pulpal exposure.

Cutaneous examination showed multiple firm keratotic slightly pale papules on the medial aspect of dorsa of both hands extending on to the back of the little and ring fingers (Fig. 2). The lesions were more on the right hand than on the left and varied in size from 1 - 2 mm diameter. Similar lesions present on dorso-lateral aspects of feet were less prominent.

Child was active and of average intelligence. Systemic examination did not reveal any abnormality.

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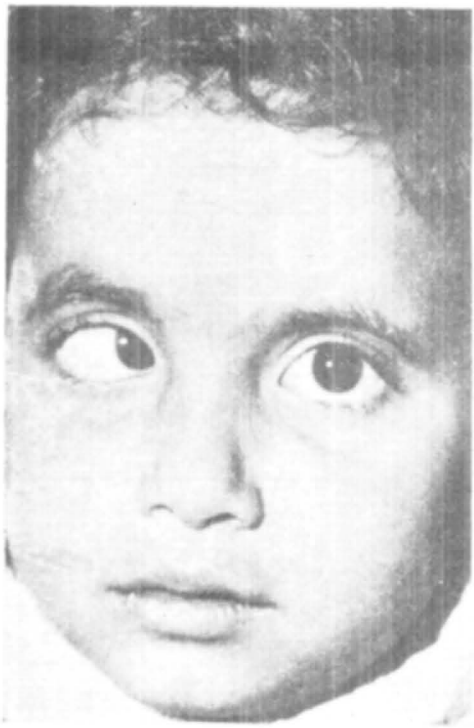

Fig. 1
Eye changes of Duane's Retraction syndrome

One of the lesions from the hand was biopsied. Haematoxylin and eosin section showed marked hyperkeratosis to the extent that the thickness of the keratin layer was greater than the thickness of the rest of the epidermis (Fig. 3). Stratum malpighii was acanthotic while granular layer showed hypergranulosis. Dermis did not show nay change.

Discussion

Gillespie[2] reported a case of this hereditary syndrome which had bilateral microphthalmos, hypotrichosis, microdontia, anodontia, camptodactyly, syndactyly, absence of phalanges and small alae nasi. Eidelman et al[3] described two cases of this syndrome and differentiated it from a closely allied entity, orodigitofacial dysostosis. In his two cases of a brother and a sister, in addition to digital, ocular, dental and nose anomalies described previously there was ocular hypertelorism, prominent epicanthal folds, bilateral conductive deafness and cleft palate. In the four cases described by Solomon et al[4] hypertrophy of peripheral zone of iris was an additional feature.

In our patient there were skin lesions which did not conform to lesions of any known dermatosis. It is reasonable to assume that the cutaneous lesions are related to the syndrome although the nature or pathogenesis of the cutaneous lesions is not clear.

Eye changes in this patient were entirely different from those that were

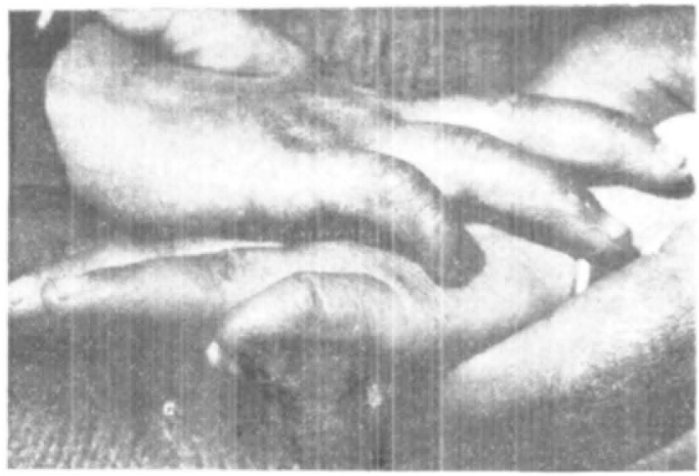

Fig. 2
Showing Camptodactyly and Keratotic Lesions

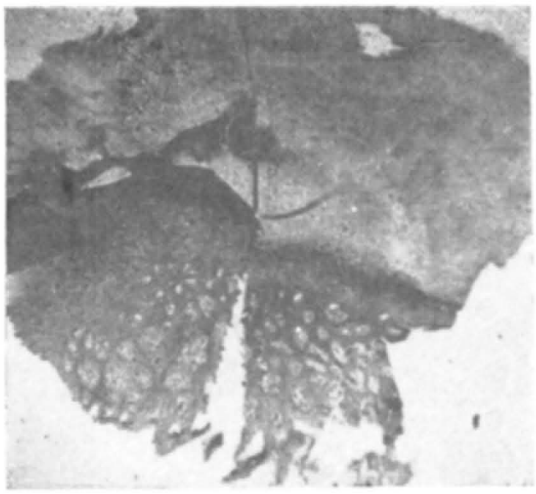

Fig. 3
Showing gross hyperkeratosis and hypergranulosis

earlier reported with oculodentodigital dysplasia. One of the hypothesis on aetiopathogenesis of Duane's retraction syndrome is replacement of one of the extraocular muscle by fibrous tissue bands[5]. Camptodactyly also seems to be due to connective tissue abnormality. Presence of these two features in this case suggests a defect of connective tissue in this syndrome.

Anomalies of nose like defective alae formation, syndactyly or hearing impairment were not present in our case.

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