

# Derموchondral corneal dystrophy (of François)

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## Abstract

**Derموchondral corneal dystrophy (of François) has been reported rarely in the literature. It consists of a triad of findings characterised by the development of skin nodules, acquired deformities of the extremities, and a corneal dystrophy. The corneal dystrophy is central and superficial with whitish subepithelial opacities. We present two brothers who display previously unreported ocular findings. Specifically, they developed confluent opacification of their central corneas with anterior stromal involvement, and peculiar anterior cortical cataracts. These findings should be added to the spectrum of findings seen in this rare disorder.**

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Derموchondral corneal dystrophy is a rare familial disease initially described by François in 1949.<sup>1,2</sup> It consists of a triad of abnormalities affecting the bones of the extremities, the skin, and the cornea. We describe previously unreported ocular manifestations in a pair of brothers whose dermatologic findings were previously investigated.<sup>3</sup>

## Case reports

Two Mexican brothers, aged 20 and 22<sup>1/2</sup>, with a history of derموchondral corneal dystrophy were referred for evaluation. Both had been born by spontaneous vaginal delivery and were healthy without abnormalities at birth. Four siblings are healthy and there is no history of consanguinity. As early as 9 months of age they began to develop nodules on their hands. Limb abnormalities and corneal findings were seen as early as age 3. The corneal opacities did not affect vision at that time. Histopathology of the skin lesions revealed 'spongiocytes' without staining by special lipid or mucopolysaccharide stains.<sup>3</sup> They have experienced a gradual decrease in

visual acuity over the last several years. However, there is no history of ocular inflammatory episodes, and the patients have no history of redness, pain, or photophobia.

On ocular examination, the best visual acuity of the elder brother was 20/100 with both eyes and 20/80 in the younger brother. Their corneas showed numerous superficial and central corneal opacities which were white with irregular and blurred edges. Most opacities were subepithelial; some caused an elevation of the overlying epithelium. There was no breakdown of the corneal epithelium and there was no staining with fluorescein. The intervening cornea showed subepithelial and anterior stromal haziness (Fig 1). Gonioscopy revealed normal angle structures. Anterior cortical cataracts in a stellate pattern were evident (Fig 2). The rest of the examination was unremarkable.

## Discussion

Derموchondral corneal dystrophy is a rare disorder with only nine cases having been reported in the literature.<sup>1-7</sup> Consanguinity was demonstrated in one case suggesting that transmission may be autosomal recessive.<sup>4</sup> This syndrome consists of a triad of findings which affects the bones of the extremities, the skin, and the cornea. There is an abnormal ossification of cartilage leading to abnormal skeletal development which is limited to the hands and feet. This leads to subluxations and retractions of tendons which gives rise to the deformities. The skin nodules look similar to xanthomas and involve the dorsal surface of joints of fingers, posterior surface of elbows, nose, and external ear (Figs 3, 4). Although the nodules appear to be xanthomatous they do not stain with lipid stains and probably represent a proliferation of anomalous fibroblasts.<sup>7</sup>

The ocular syndrome consists of a superficial and central corneal dystrophy with opacities of varying dimensions under the epithelium. The

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Figure 1 Slit-lamp photograph of the cornea. The large white opacities are subepithelial. There is haziness of the anterior third of the stroma.

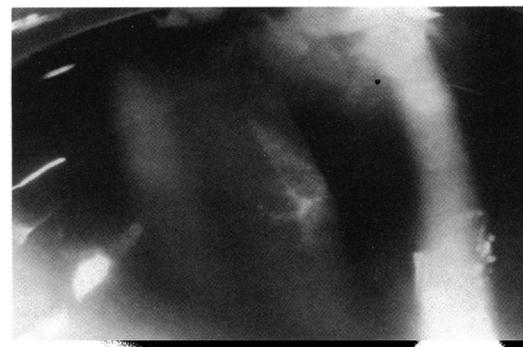


Figure 2 Slit-lamp photograph of the crystalline lens. There is a stellate anterior cortical cataract visible.

Figure 3 (A) Dermal nodules on the elbows in dermochondral corneal dystrophy. (B) Higher magnification of elbow nodules.



Fig 3A



Fig 3B

opacities are white with irregular margins. In large opacities, there is elevation of the epithelial surface with an anterior convex wall, a posterior concave wall, and an optically empty space between them. The periphery of the cornea, the parenchyma, and the endothelium appear normal. Histological examination of François' cases showed an abnormal epithelium with Bowman's membrane being poorly defined and containing some histiocytes. The stromal tissue had scanty cells and cavities were evident.

The natural history of this syndrome is not

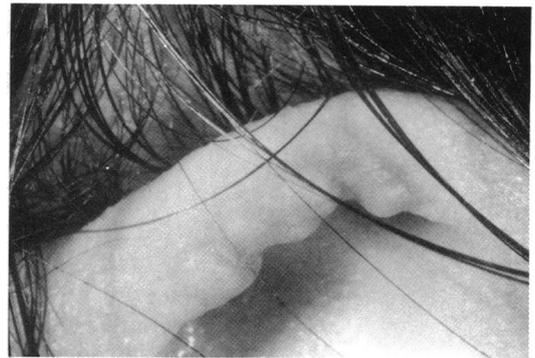


Figure 4 Nodules on the outer part of the ear in dermochondral corneal dystrophy.

clearly defined. François observed that his patients developed hand deformities between age 1 and 2, foot and skin abnormalities by age 5, and ocular abnormalities between age 8 and 9. Another case report suggested that the onset may develop later in life (age 33) in association with pregnancy and may also have gingival and conjunctival abnormalities. Analysis of collagen urinary metabolites in this case suggested there was a hyperproduction of type III collagen.<sup>7</sup>

By contrast, our patients had developed skin lesions in infancy, and limb and corneal abnormalities as early as age 3. Moreover, our patients developed anterior corneal stromal opacities, and cataracts which were previously unreported. These observations suggest that the ocular manifestations of this syndrome can be progressive and will probably require therapeutic intervention in the future.

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