Acquired Corneal Dystrophy in a Case of Dysgenesis of the Anterior Segment of the Eye, Oligodontia and Metacarpal Dysplasia (dysplasia oculodentodigitalis?)

H.E.

Henkes

Eye Hospital, Rotterdam Medical School, Rotterdam (The Netherlands)

Author's address: Prof. Dr. Harold E. Henkes, Eye Hospital, Rotterdam Medical School, Rotterdam (The Netherlands)

Summary

A 16-year-old girl suffering from an acute corneal dystrophy due to spontaneous (?) rupture of Descemet's membrane in one eye, caused problems of differential diagnosis between the abortive form of Rieger's dysgenesis with dental and skeletal anomalies, and the dysplasia oculodentodigitalis.

Both eyes showed iris stroma hypoplasia, anomalies in the chamber angle and a markedly bluish scleral band, together with glaucoma. Microcornea and dysplasia marginalis posterior were absent. Apart from the 3rd molares, anodontia vera of 10 permanent elements was demonstrated. Maxillar hypoplasia and mandibular hyperplasia were present. Hypoplasia of metacarpalia IV and V of both hands could be demonstrated without, however, camptodactyly or syndactyly being present.

The extended corneal dystrophy greatly improved following extensive diathermic coagulation of the cornea.

(Will be published in extenso in the British Journal of Ophthalmology.)

Discussion

Waardenburg: In an area as small as the anterior chamber of the eye one has the choice of the following hereditary anomalies:

1. Hypoplasia of the iris, as also atypical coloboma, as a "forme fruste" of aniridia.

224

Henkes

Hypoplasia of the iris as such, or with localized iris defects of the sphincter muscle, the stroma or the pigmentary layer.

Peters' syndrome with endothelial defect.

Rieger's syndrome with microcornea (common) and slit pupil (less common).

Oculo-dento-digital syndrome with microphthalmos, hypotelorism, hypoplasia of the median part of the face, particularly of the maxilla, finger and/or toe anomalies.

4 and 5 are often associated with oligodontia.

In my opinion, we must choose in this case between 4 and 5. The absence of microphthalmos and median facial hypoplasia make Rieger's malformation rather more likely. The decision is made difficult by the fact that, in the many publications over Rieger's anomalies, photographs of the patients full face and in profile are hardly ever given, so that we do not know whether 4 and 5 are due to the same gene or not.

You consider that the endothelium and Descemet's membrane must be assumed to be of poor quality. You have mentioned cases in which this is associated with other anomalies in the anterior chamber and disposition to glaucoma. Endothelial hyperplasia appears to play a part in progressive essential iris atrophy (Rochat and Mulder, Pau, a.o.).

De Haas asks Waardenburg: Do you classify the conditions described under 2 and 4 in the same or different classes?

Waardenburg: So far I have separated them

although they do occur together in very large pedigrees such as those of Falls and Gedda. The future will show if there are arguments in favour of combining them

and if the same gene may be considered to be responsible. An example of 2 has been described by Leffertstca. All the malformations 1–5 may lead to the development of glaucoma.