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IRIS DYSGENESIS WITH OTHER ANOMALIES*†

BY

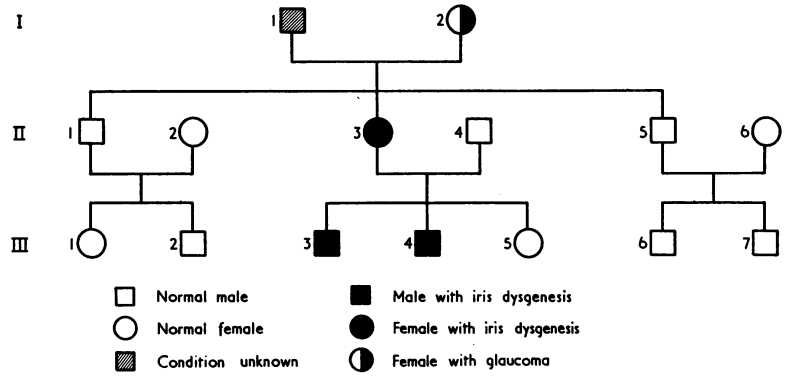
R. A. D. CRAWFORD

Canterbury

THREE members of a family with severe iris defects and dental and other congenital anomalies are described. This association was first reported by Vossius (1883); Rieger (1935) later described defects in the anterior iris layers but not aniridia.

Case Reports (see Family Tree)

I.2.—The grandmother was known to have chronic glaucoma from 1929; there was no record of iris anomaly. When she was seen in 1963 there was no perception of light and the corneae had become degenerate; it was not possible to determine whether the iris had been normal or not.



II.3.—Her daughter was seen first in 1959. There were slit-pupils, shallow anterior chambers with narrow angles, and peripheral anterior synechiae. Correction of myopia gave a visual acuity of 6/12 in the right eye and 6/36 in the left. The intra-ocular pressure was controlled by miotics until 1962, when surgery to the right eye, and the following year to the left, was undertaken. Since then there have been gradually increasing cataracts, and in 1965 a total right retinal detachment occurred. There was gross peripheral retinal degeneration, and surgery for the detachment has not been successful. The left eye retains a visual acuity of 6/24 with field loss. There are no other defects apart from the eyes (see col. pl.).

III.3.—This boy showed partial aniridia in the right eye and complete aniridia in the left (col. pl.). When first seen in 1957, four operations had been performed on each eye to control glaucoma; further surgery was followed by retinal detachment in the left eye, which was lost. The right eye has remained free from pain but has no perception of light.

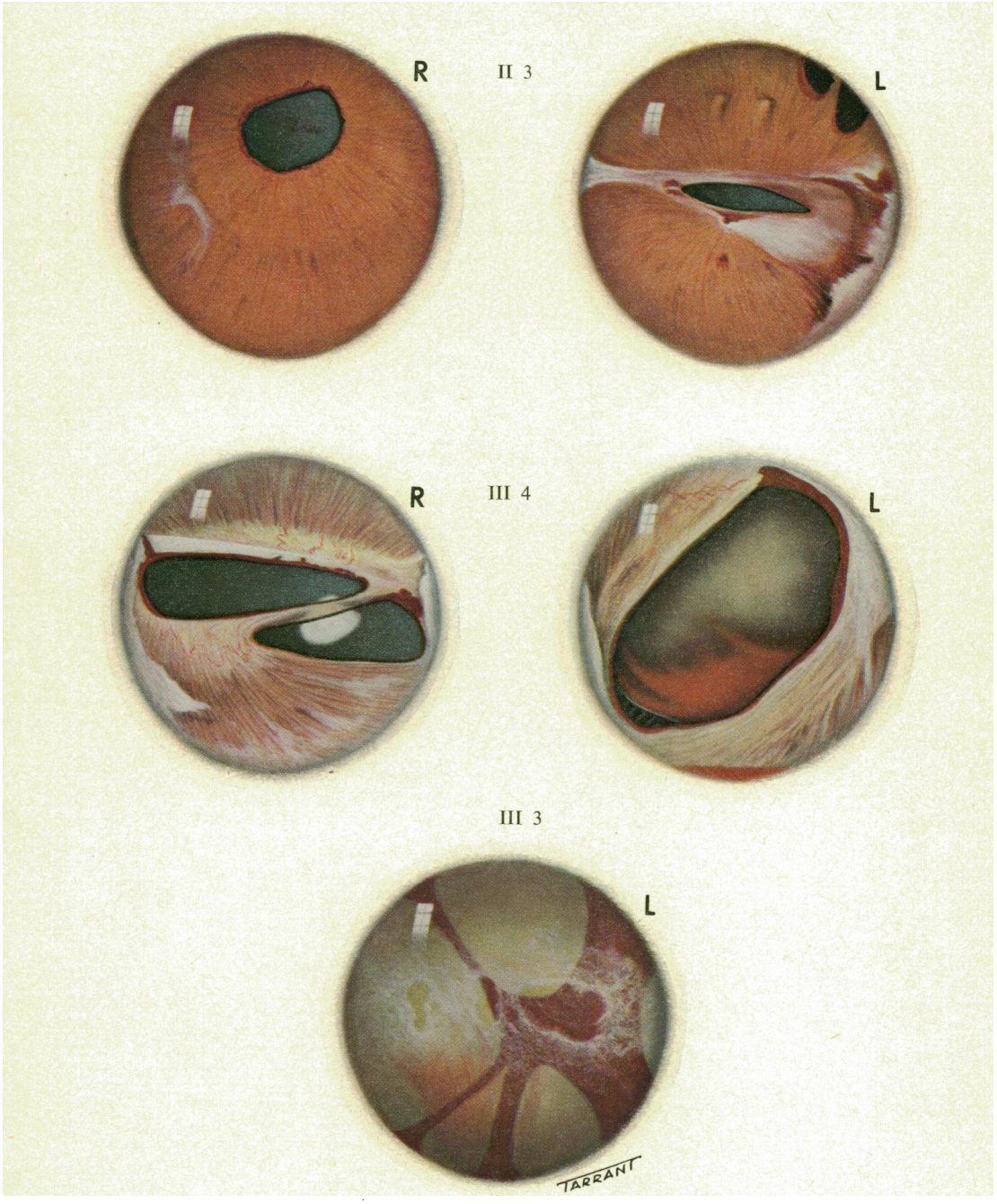
There is a defect of the premaxilla, on which Mr. Denis Glass reports:

"His teeth are very poor and many are missing. The mandible is well developed but the maxilla is small and agenetic. This results in a pseudo-prognathism". Anal stenosis was successfully treated in infancy; there was also an umbilical hernia. He has kyphosis and is of poor physique, but in good general health; chest *x* rays and electrocardiogram being normal. He has low-set bat ears.

III.4.—This boy, seen in 1957, had slit-pupils and a right anterior subcapsular lens opacity (col. pl.). Later glaucoma in the left eye was controlled by surgery until an injury rendered this eye

* Received for publication: February 1, 1966.
† Address for reprints: R. A. D. Crawford, M.D., 80 Gladstone Road, Broadstairs, Kent.

IRIS DYSGENESIS



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blind. The right eye remained quiet with a visual acuity of 6/9 and a full field of vision until April, 1967, when glaucoma appeared. He shows the same dental and ear defects as his brother and also had anal stenosis as an infant. He is small for his age but in good health, and no other defects have been found.

I.1 is no longer alive and no history has been obtainable.

III.5, the sister of the affected boys, is normal.

Chromosome analysis of both parents and all three children is normal. This is in contrast to a family which Mr. M. J. Gilkes has kindly shown me, whose members have similar eye defects, though without the early onset of glaucoma. There is no kinship between the two families, and his cases do show chromosomal abnormalities (personal communication).

Discussion

Vossius (1883) described a girl aged 9 years who had gross iris defects in one eye and virtual aniridia in the other. There were strands of tissue across the anterior chamber, with leucoma adherens, absence of the premaxilla and upper incisors, abnormal lower incisors, and high arching of the palate.

Waardenburg, Franceschetti, and Klein (1961) reviewed the literature. They considered that cases with aniridia might be a separate group from those with anterior iris defect described by Rieger (1935), the error occurring earlier in development. Rieger considered the defect to be entirely ectodermal; his patient showed an anterior polar cataract. Hagedoorn (1937) did not accept Rieger's view, pointing out that it did not explain the aniridia or the dental defects; he believed that persistence of the anterior vitreous led to distortion of the iris and residual tissue in the anterior chamber angle, and proposed the name "Mesostroma anterior persistens".

Falls (1949) described a family with a wide variety of anterior chamber disorders causing glaucoma which was very difficult to control; he considered the condition to be due to a single dominant gene. Lemmingson and Riethe (1958) reported three cases of iris dysgenesis, one with ectopia lentis and two with dental defects. Kittel (1956) found six defective eyes in one family, associated also with skeletal and muscle defects. Gassler and Berthold (1960) noted an ear deformity in one case. Haab (1878) and Schachenmann, Schmid, Fraccaro, Mannini, Tiepolo, Perond, and Sartori (1965) reported anal atresia in two cases.

The only anomaly not previously described is thus the umbilical hernia, which is too common to be important.

Summary

An account is presented of three members of a family showing severe developmental errors in the iris, associated with maldevelopment of the ear and maxilla, umbilical hernia, and anal stenosis. Glaucoma has occurred in all six eyes. The maternal grandmother (I.2) was blind when seen shortly before her death, but it was not possible to establish whether she also showed a congenital anomaly.

I am glad to acknowledge the help of Mr. T. Keith Lyle and Mr. Arthur Lister in tracing and allowing me to quote old records, of Mr. Denis Glass in reporting on the dental state, and of Dr. Peter Hansell and the Medical Illustration Department of the Institute of Ophthalmology in producing the paintings. I am grateful to Mr. M. J. Gilkes for allowing me to see the similar cases in his care.

REFERENCES

FALLS, H. F. (1949). *Amer. J. Ophthal.*, **32**, no. 6, pt II, p. 41.
GASSLER, H., and BERTHOLD, H. (1960). *Klin. Mbl. Augenheilk.*, **136**, 52.
HAAB, O. (1878). *v. Graefes Arch. Ophthal.*, **24**, abt 2, p. 257.
HAGEDOORN, A. (1937). *Arch. Ophthal. (Chicago)*, **17**, 223.
KITTEL, V. (1956). *Klin. Mbl. Augenheilk.*, **129**, 464.
LEMMINGSON, W., and RIETHE, P. (1958). *Ibid.*, **133**, 877.
RIEGER, H. (1935). *v. Graefes Arch. Ophthal.*, **133**, 602.
SCHACHENMANN, G., SCHMID, W., FRACCARO, M., MANNINI, A., TIEPOLO, L., PEROND, G. P., and SARTORI, E. (1965). *Lancet*, **2**, 290.
VOSSIUS, A. (1883). *Klin. Mbl. Augenheilk.*, **21**, 233.
WAARDENBURG, P. J., FRANCESCHETTI, A., and KLEIN, D. (1961). "Genetics and Ophthalmology", vol. 1, pp. 570–575. Royal Van Gorcum, Assen, Netherlands.