DYSCEPHALY WITH CONGENITAL CATARACT*†

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FRANÇOIS (1958) described a new syndrome with an analysis of 21 cases from the literature and one of his own; its main characteristics were dyscephaly with "bird face", dental anomalies, dwarfism, hypotrichosis, cutaneous atrophy, microphthalmos, and congenital cataract. Falls and Schull (1960) reported another six cases (one of which had been described by Moehlig (1946) and referred to by François (1958) as Case 5); they called the condition the "Hallermann-Streiff syndrome", because Hallermann (1948) and Streiff (1950) had distinguished it from mandibulo-facial dysostosis of the type described by Franceschetti and Zwaan (1944). Gillespie (1964) reported two cases of a syndrome named "dysplasia-oculo-dentodigitali" by Meyer-Schwickerath, Gruterich, and Weyers (1957), which was similar except that

FIG. 1.—X ray of skull of Case 1, showing gaping sagittal suture.

FIG. 2.—Lateral view, showing gaping sagittal suture and aplasia of maxilla and mandible.

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the patients had no cataract but showed syndactyly, hypoplasia of the middle phalanx of the fifth finger, and missing phalanges in the feet.

Thus, since Audry (1893) described the first case (François, 1958), less than thirty others have been reported.

It is the purpose of this paper to present two more examples, both with unusual additional features.

Case Reports

Both these patients came to the Ein-Shams University Hospital with extreme visual loss. Their families were normal and they showed no abnormality of any of the bodily systems apart from those tabulated below. The important physical features are illustrated in Figs 1 to 9.

TABLE
CLINICAL PARTICULARS OF TWO CASES OF DYSCEPHALY

Case No.	1	2
Sex	Male	Male
Age (yrs)	20	15
Visual Acuity { R L	Hand movements Counting fingers	} Counting fingers
Family History	Negative	Negative
Dyscephaly	Gaping sagittal suture (Figs 1 and 2) Glabella depressed Bird face (Figs 3 and 4)	Gaping sagittal suture Glabella flat Bird face (Figs 8 and 9)
Teeth	Upper incisors prominent and separated	Upper incisors prominent
Dwarfism	Height 157 cm. Weight 39.5 kg.	Small for his age
Hair	Small deficient areas on scalp (Figs 3 and 4)	Deficient along sagittal suture
Skin	Facial acne vulgaris and dimpling of chin	Normal
Eyes	Microphthalmos	Both corneal diameters 9 mm.
	Cataract { R L	Left corneal diameter 8 mm.
	Other abnormalities	} Residual after needling
	Blue sclera R concomitant convergent squint Irregular nystagmus Left fundus marked tessellation and choroidal scar temporal to a normal disc	Blue sclera L concomitant convergent squint Horizontal nystagmus
Other Defects	S-shaped clavicles (Figs 5, 6, 7)	—

Discussion

Besides the regular characteristics of this new syndrome, Case 1 also had S-shaped clavicles and both had a marked gaping of the sagittal suture. Little can be added to the description given by François (1958). The condition is sporadic, and perhaps

FIG. 3.—Profile of Case 1, showing characteristic "bird face".

FIG. 4.—Back view of head of Case 1, showing hypotrichosis.

FIG. 5.—Front view of Case 1, showing strabismus and S-shaped deformity of both clavicles.

FIG. 6.—Lateral view of Case 1, showing deformity of right clavicle.

FIG. 7.—X ray showing S-shaped deformity of the clavicles.

FIG. 8.—Front view of Case 2, showing characteristic "bird face".

FIG. 9.—Side view of Case 2, showing characteristic "bird face".

due to a gene mutation caused by an intra-uterine viral infection during the early months of pregnancy, too slight to be noticed by the mother.

Although Hallermann (1948) and Streiff (1950) differentiated this syndrome from the mandibulo-facial dysostosis of Franceschetti and Zwaalen (1944), it was François (1958) who reviewed the literature, collected and analysed most of the reported cases, and differentiated the condition from other ectodermal dysplasias.

Summary

Two examples are described of a syndrome characterized by dyscephaly with "bird face", dental anomalies, dwarfism, hypotrichosis, microphthalmos, and congenital cataract.

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