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 Zwahlen (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

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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

<table>
<thead>
<tr>
<th>Case No.</th>
<th>1</th>
<th>2</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sex</td>
<td>Male</td>
<td>Male</td>
</tr>
<tr>
<td>Age (yrs)</td>
<td>20</td>
<td>15</td>
</tr>
<tr>
<td>Visual Acuity</td>
<td>R: Hand movements Counting fingers</td>
<td>L: Counting fingers</td>
</tr>
<tr>
<td>Family History</td>
<td>Negative</td>
<td>Negative</td>
</tr>
<tr>
<td>Dyscephaly</td>
<td>Gaping sagittal suture (Figs 1 and 2) Glabella depressed Bird face (Figs 3 and 4)</td>
<td>Gaping sagittal suture Glabella flat Bird face (Figs 8 and 9)</td>
</tr>
<tr>
<td>Teeth</td>
<td>Upper incisors prominent and separated</td>
<td>Upper incisors prominent</td>
</tr>
<tr>
<td>Dwarfism</td>
<td>Height 157 cm. Weight 39.5 kg.</td>
<td>Small for his age</td>
</tr>
<tr>
<td>Hair</td>
<td>Small deficient areas on scalp (Figs 3 and 4)</td>
<td>Deficient along sagittal suture</td>
</tr>
<tr>
<td>Skin</td>
<td>Facial acne vulgaris and dimpling of chin</td>
<td>Normal</td>
</tr>
<tr>
<td>Microphthalmos</td>
<td>Both corneal diameters 9 mm.</td>
<td>Left corneal diameter 8 mm.</td>
</tr>
<tr>
<td>Cataract</td>
<td>R: Residual after repeated needling Aphakic</td>
<td>L: Residual after needling</td>
</tr>
<tr>
<td>Other abnormalities</td>
<td>Blue sclera R: concomitant convergent squint</td>
<td>Blue sclera L: concomitant convergent squint</td>
</tr>
<tr>
<td>Other defects</td>
<td>S-shaped clavicles (Figs 5, 6, 7)</td>
<td>—</td>
</tr>
</tbody>
</table>

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.
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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 Zwahlen (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.

REFERENCES

Dyscephaly with congenital cataract.

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