MANDIBULO-FACIAL DYSOSTOSIS*

BY
ALLAN H. BRIGGS
Lincoln

The first recorded cases of this comparatively rare congenital anomaly were reported by Berry (1889), but it was not until 11 years later that Collins (1900) recognized that the condition occurred as a syndrome. Further cases in Great Britain have been described by Mann and Kilner (1943) and Johnstone (1943). The clinical picture varies considerably from case to case and atypical, incomplete and unilateral forms may occur. It has been described under various names, in England as "Treacher Collins' Syndrome", and on the continent of Europe as the "Syndrome of Franceschetti".

The original cases described by Berry (1889) comprised a mother, her brother, and her daughter, who all shewed obliquity of the palpebral fissures, colobomata of the eyelids, and defective development of the chin (micrognathia); these are now classified as abortive forms of the full syndrome.

Collins (1900) and Tyrrell (1903) described in addition flattening of the cheeks due to hypoplasia of the malar bones. McMullen (1920) described a unilateral case, and Pires de Lima and Monteiro (1923) first described the fully developed syndrome. Numerous case reports followed—Isakowitz (1927), Waardenburg (1932, 1934), Hermans (1936), Van Lint and Hennebert (1936), Kazanjian (1936), McEnery and Brennemann (1937), Debusmann (1940), Sanvenero-Rosselli (1940, 1948), Mann and Kilner (1943), Johnstone (1943), Franceschetti and Zwahlen (1944), Leopold, Mahoney and Price (1945), Schachter (1947), Brohm and Kluska (1947), Holm (1948), Brégeat and Naud (1949), Waardenburg (1948), Navis (1948), Straith and Lewis (1949), Labourcarie and Gayral (1949), O'Connor and Conway (1949), Halberg and Paunessa (1949), and Streiff (1950).

The fully developed syndrome consists of certain associated congenital and familial deformities of the ears, malar bones, lips, chin, and lower eyelids. The general similarity in appearance of the affected patients is very striking, although the number and extent of the deformities (which are usually bilateral) vary considerably. The appearance is unmistakable and the face has a characteristic fish-like aspect. The main features (see Table of selected cases) are:

1. An anti-mongoloid obliquity of the palpebral fissures, sometimes associated with colobomata of the outer portion of the lids (usually the lower lids).

* Received for publication October 23, 1952.
### TABLE

**SUMMARY OF SELECTED CASES OF TREACHER**

<table>
<thead>
<tr>
<th>Author</th>
<th>Date</th>
<th>Malar Eminence</th>
<th>Lower Orbital Margin</th>
<th>Palpebral Fissure</th>
<th>Axis of Eyeball</th>
<th>Lower Eyelids</th>
</tr>
</thead>
<tbody>
<tr>
<td>Treacher Collins</td>
<td>(1) 1900</td>
<td>Flattened</td>
<td>Deficient</td>
<td>Oblique (downward laterally)</td>
<td>?</td>
<td>Bilateral notches</td>
</tr>
<tr>
<td></td>
<td>(2) 1900</td>
<td>Flattened</td>
<td>Deficient</td>
<td>?</td>
<td>?</td>
<td>?</td>
</tr>
<tr>
<td>Mann and Kilner</td>
<td>(1) 1943</td>
<td>Flattened</td>
<td>Deficient</td>
<td>Oblique</td>
<td>Normal</td>
<td>Thin and atrophic</td>
</tr>
<tr>
<td></td>
<td>(2) 1943</td>
<td>Flattened</td>
<td>Deficient</td>
<td>Oblique</td>
<td>Normal</td>
<td>Bilateral notches</td>
</tr>
<tr>
<td>Straith and Lewis</td>
<td>(3) 1949</td>
<td>Flattened</td>
<td>Deficient</td>
<td>Oblique</td>
<td>Normal</td>
<td>Bilateral notches</td>
</tr>
<tr>
<td></td>
<td>(4) 1949</td>
<td>Flattened</td>
<td>Deficient</td>
<td>Oblique</td>
<td>Oblique</td>
<td>Bilateral notches</td>
</tr>
<tr>
<td></td>
<td>(5) 1949</td>
<td>Flattened</td>
<td>Deficient</td>
<td>Oblique</td>
<td>Normal</td>
<td>Normal</td>
</tr>
<tr>
<td>Present Author</td>
<td>1953</td>
<td>Flattened</td>
<td>Deficient</td>
<td>Slightly oblique</td>
<td>Normal</td>
<td>Normal</td>
</tr>
</tbody>
</table>

* Two cases reported by Berry (1889) had only the

(2) Hypoplasia of the facial bones, especially the malar bones, the zygomatic arch and the mandible, resulting in flattening of the cheeks and recession of the chin (micrognathia).

(3) Malformation of the external ears, sometimes involving the middle and internal ears with consequent impairment of hearing. The auricles are placed lower than the normal position, and point backwards. Auricular fistulae may be present.

(4) Deformities of the lips and mouth: the lip and palate may be cleft; the palate tends to be high and arched, with malocclusion of the teeth, and the upper lip may be enlarged with macrostomia.

(5) The chief associated deformities are:

- (a) Blind fistulae between the angles of the mouth and the ears.
- (b) Tongue-shaped projections of the hair line on to the cheeks.
- (c) Atrophy of the medial portions of the lower lids, with absence of the lashes and Meibomian glands over the affected area. The puncta may be absent; the naso-lacrimal ducts may be obstructed.
## MANDIBULO-FACIAL DYOSTOSIS

### COLLINS SYNDROME REPORTED IN THE LITERATURE*

<table>
<thead>
<tr>
<th>Eyelashes (Medial $&amp;$ Lower Eyelid)</th>
<th>External Ear</th>
<th>External Auditory Meatus</th>
<th>Hearing</th>
<th>Maxilla</th>
<th>Mandible</th>
<th>Other Abnormalities</th>
</tr>
</thead>
<tbody>
<tr>
<td>?</td>
<td>Normal</td>
<td>Normal</td>
<td>Narrow</td>
<td>?</td>
<td>Receding chin</td>
<td>?</td>
</tr>
<tr>
<td>?</td>
<td>Normal</td>
<td>Normal</td>
<td>Normal</td>
<td>?</td>
<td>?</td>
<td>?</td>
</tr>
<tr>
<td>Bilaterally absent</td>
<td>Normal</td>
<td>Normal</td>
<td>Deaf</td>
<td>High, narrow</td>
<td>Receding chin</td>
<td>None</td>
</tr>
<tr>
<td>Collection of lashes at distance to lateral commissure</td>
<td>Bilaterally deformed</td>
<td>Absent</td>
<td>Deaf</td>
<td>High, narrow</td>
<td>Receding chin</td>
<td>Cleft palate</td>
</tr>
<tr>
<td>Absent medial $&amp;$ right lower lid</td>
<td>Slightly deformed bilaterally</td>
<td>Deficiency of helix laterally</td>
<td>Partially deaf</td>
<td>High, narrow</td>
<td>Receding chin</td>
<td>Long second metatarsal bone</td>
</tr>
<tr>
<td>Normal</td>
<td>Bilaterally deformed</td>
<td>Absent</td>
<td>Deaf</td>
<td>High, narrow</td>
<td>Receding chin</td>
<td>Long second metatarsal bone Flat pariетo-occipital skull</td>
</tr>
<tr>
<td>Absent right, sparse left</td>
<td>Bilaterally deformed</td>
<td>Unilaterally absent</td>
<td>Unilateral deafness</td>
<td>Prominent upper front teeth</td>
<td>Receding chin</td>
<td>Long second metatarsal bone Flat, pariетo-occipital skull Hypermotility in metatarso-phalangeal joints</td>
</tr>
<tr>
<td>Bilaterally absent</td>
<td>Bilaterally deformed</td>
<td>Normal</td>
<td>Normal</td>
<td>High, narrow</td>
<td>Receding chin</td>
<td>Long second metatarsal bone Hypermotility in metatarso-phalangeal joints</td>
</tr>
<tr>
<td>Bilaterally absent</td>
<td>Bilaterally deformed</td>
<td>Normal</td>
<td>Normal</td>
<td>High, narrow</td>
<td>Receding chin</td>
<td>As above</td>
</tr>
<tr>
<td>Present</td>
<td>Bilaterally deformed</td>
<td>Very narrow overhanging superficially</td>
<td>Unilateral partial deafness</td>
<td>High, narrow</td>
<td>Receding chin</td>
<td>Webbed second and third digits of both feet Lacrimal puncta present</td>
</tr>
</tbody>
</table>

deficient lower eyelids and are not included.

(d) Flattened pariетo-occipital bones.

(e) Nasal deformities.

(f) Skeletal deformities, such as long second metatarsal bones, club foot, and hypermotility of the metatarso-phalangeal joints.

The hereditary character of the syndrome has been established by numerous genealogies reported in the literature (Debusmann, 1940; Leopold, Mahoney, and Price, 1945; Brohm and Kluska, 1947). Cases appear to occur more frequently in the lower social and economic grades, and it has been suggested that advancing age in the mother (and possibly deficiencies in her diet) may be among the causative factors. The aetiology of the condition is clearly genetic, and defective ossification of the bones of the face, derived from the visceral mesoderm, provides the primary lesion. The defect must presumably date from about the seventh week of foetal life. Similar anomalies have been produced in chicks by x irradiation during development (Wolff, 1934). Treatment of the defects involves plastic procedures to raise the level of the outer canthus in severe cases. Cartilage implants may be desirable to alter the contour of the face and various forms of plastic operation may be required to remedy the position and shape of the ears.
Case Report

A boy aged 10 was originally presented for treatment with a history of inflammation and discharge from both eyes over a period of several years. His facial appearance had been abnormal from birth, but there was no family history of facial or other congenital abnormalities. He had been noted to be a mouth-breather and his mouth had always been deformed. Articulation was satisfactory. There was some deafness of the right ear but he was making satisfactory progress at school. His general appearance was that of mandibulo-facial dysostosis, although the eyelids did not show bilateral notching. There was deficiency of both malar bones and noticeable flattening in the region of the infra-orbital ridges. Cilia were present throughout the lower lids but on both sides there was lacrimal obstruction, with copious muco-purulent regurgitation from the lacrimal sacs and some associated chronic conjunctivitis. The chin was small and receding and there were bilateral deformities of the external ears, which were caudal to the normal position, pointing backwards and upwards. There was a supra-auricular fistula on the right side (which was still discharging) with a scar in a similar position on the left side, and some scarring down the anterior borders of the sterno-mastoids. The palate was high and narrow: the nose showed a broadened, enlarged bridge: the external auditory meatuses were present but very narrow and overhanging superficially: the upper lip showed the presence of a fairly extensive lymphangioma, with considerable deformity (see Figs 1, 2, and 3).

Eyes.—Apart from the condition of the lacrimal apparatus and the associated conjunctivitis, both eyes were normal externally, the pupils equal and active, the media clear and the fundi normal. The left eye was, however, amblyopic. Vision in the right eye was $-0.75$ D sphere with $+2.50$ D cylinder, axis $80^\circ = 6/9$, and in the left $-2.25$ D sphere with $+2.50$ D cylinder, axis $95^\circ = 6/24$.

Teeth.—A dental surgeon reported as follows:

Congenital deformity of the face: many temporary teeth missing.

<table>
<thead>
<tr>
<th>Present teeth are these:</th>
<th>6</th>
<th>321</th>
<th>13</th>
<th>6</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>6*</td>
<td>21</td>
<td>12</td>
<td>6*</td>
</tr>
</tbody>
</table>

Upper incisors very prominent with narrow maxillary arch. X-rays of teeth show retained roots with apical abscess $\sim 17$. Retained root $\sim 77$. Teeth marked* are septic roots.

Body.—The second and third digits of both feet were found to be webbed. No other skeletal deformities were found.

Treatment.—A series of operations was carried out. A dacryocystorhinostomy was performed on the right side and a week later the lymphangioma of the upper lip was subjected to surgical diathermy. A dacryocystorhinostomy was performed on the left side. Eleven months later a further wedge of tissue was excised from the inner portion of the upper lip, and later still a plastic operation was performed on the right ear by transposition of skin flaps, free mobilization and advancement of the auricle to a higher point of attachment to the skull. The preauricular fistula on the right side was found to be very extensive, the deep portion of it containing a little thick pus. This area was explored down to a narrow opening into the bone above the ear, which was curedtted. Later a similar plastic procedure was carried out upon the left ear, with exploration of the corresponding fistula, which did not, however, reach the bone. Substantial improvement in the patient’s condition and appearance resulted.
Summary

A case of mandibulo-facial dysostosis is described with records of similar cases collected from the literature.

The condition is due to delayed or defective development of the mesoderm of the maxillary process about the end of the second month of foetal life.

Multiple congenital defects are described in association with this syndrome.

The variable nature and extent of the defects renders routine treatment difficult. Each case has to be treated on its individual merits.
I wish to record my sincere thanks to Mr. M. E. Spencer-Harrison for his co-operation in performing the ear, nose and throat operations mentioned above and for his assistance in the preparation of this paper.

My thanks are also due to Mr. Stacey for the preparation of the clinical photographs and to Miss Bryden and Miss Gower for secretarial help.

REFERENCES


DEBUSMANN (1940). *Arch. Kinderheilk.*, 120, 133.


ADDITIONAL BIBLIOGRAPHY


Mandibulo-Facial Dysostosis

Allan H. Briggs

Br J Ophthalmol 1953 37: 171-176
doi: 10.1136/bjo.37.3.171

Updated information and services can be found at:
http://bjo.bmj.com/content/37/3/171.citation

These include:

Email alerting service
Receive free email alerts when new articles cite this article. Sign up in the box at the top right corner of the online article.

Notes

To request permissions go to:
http://group.bmj.com/group/rights-licensing/permissions

To order reprints go to:
http://journals.bmj.com/cgi/reprintform

To subscribe to BMJ go to:
http://group.bmj.com/subscribe/