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), Sanvenere-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

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

* 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.

COLLINS SYNDROME REPORTED IN THE LITERATURE*

Eyelashes (Medial & Lower Eyelid)	External Ear	External Auditory Meatus	Hearing	Maxilla	Mandible	Other Abnormalities
?	Normal	Normal	Normal	Narrow	Receding chin	?
?	Normal	Normal	Normal	?	?	?
Bilaterally absent	Normal	Normal	Deaf	High, narrow Prominent front teeth	Receding chin	None
Collection of lashes at distance to lateral commisure	Bilaterally deformed	Absent	Deaf	High, narrow	Receding chin	Cleft palate
Absent medial & right lower lid	Slightly deformed bilaterally	Deficiency of helix laterally	Partially deaf	High, narrow Prominent front teeth	Receding chin	Long second metatar- sal bone
Normal	Bilaterally deformed	Absent	Deaf	High, narrow Prominent front teeth	Receding chin	Long second metatar- sal bone Flat parieto-occipital skull
Absent right, sparse left	Bilaterally deformed	Unilaterally absent	Unilateral deafness	Prominent upper front teeth	Receding chin	Long second metatar- sal bone Flat, parieto-occipital skull Hypermotility in metatarso-phalan- geal joints
Bilaterally absent	Bilaterally deformed	Normal	Normal	High, narrow Prominent front teeth	Receding chin	Long second metatar- sal bone Hypermotility in metatarso-phalan- geal joints
Bilaterally absent	Bilaterally deformed	Normal	Normal	High, narrow Prominent teeth	Receding chin	As above
Present	Bilaterally deformed	Very narrow overhanging superficially	Unilateral partial deafness	High, narrow Prominent front teeth	Receding chin	Webbed second and third digits of both feet Lacrimal puncta pre- sent

deficient lower eyelids and are not included.

- (d) Flattened parieto-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.

	6	321	13	6
Present teeth are these:	<hr/>			
	6*	21	12	6*

Upper incisors very prominent with narrow maxillary arch. X rays of teeth show retained roots with apical abscess $\sqrt{7}$. Retained root $\overline{77}$. 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 curetted. 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.

FIG. 1.—Condition before treatment, August 4, 1950, full face.

FIG. 2.—Condition before treatment, August 4, 1950, left lateral view.

FIG. 3.—Condition before treatment, August 4, 1950, right lateral view.

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

- BERRY, G. A. (1889). *Roy. Lond. ophthal. Hosp. Rep.*, **12**, 255.
 BRÉGEAT, P., and NAUD, G. (1949). *Arch. Ophthal.*, **9**, 427.
 BROHM, F., and KLUSKA, V. (1947). *Lékařské listy*, **2**, 329.
 COLLINS, E. TREACHER (1900). *Trans. ophthal. Soc. U.K.*, **20**, 190.
 DEBUSMANN (1940). *Arch. Kinderheilk.*, **120**, 133.
 FRANCESCHETTI, A., and ZWAHLEN, P. (1944). *Bull. Schweiz. Akad. med. Wiss.*, **1**, 60.
 GAYRAL, BRU, and PAINTANDRE (1950). *J. Radiol. Electrol.*, **31**, 97.
 HALBERG, G. P., and PAUNESSA, J. M. (1949). *British Journal of Ophthalmology*, **33**, 709.
 HERMANS, R. (1936). *Bull. Soc. belge Ophthal.*, **73**, 60.
 HOLM, E. (1948). *Ugeskr. Laeg.*, **110**, 1366.
 ISAKOWITZ, J. (1927). *Klin. Mbl. Augenheilk.*, **78**, 509.
 JOHNSTONE, I. L. (1943). *British Journal of Ophthalmology*, **27**, 21.
 KAZANJIAN, V. H. (1936). *Int. J. Orthod.*, **22**, 259.
 LABOUCARIE and GAYRAL (1949). *Rev. Oto-neuro-ophthal.*, **21**, 318.
 LEOPOLD, I. H., MAHONEY, J. F., and PRICE, M. L. (1945). *Arch. Ophthal.*, Chicago, **34**, 210.
 VAN LINT, A., and HENNEBERT, P. (1936). *Bull. Soc. belge Ophthal.*, **73**, 51.
 MANN, I., and KILNER, T. P. (1943). *British Journal of Ophthalmology*, **27**, 13.
 MCENERY, E. T., and BRENNEMAN, N. (1937). *J. Pediat.*, **11**, 468.
 McMULLEN, W. H. (1920). *Proc. roy. Soc. Med. Sect. O.*, **13**, 85.
 NAVIS, H. (1948). *Ned. T. Geneesk.*, **92**, 740.
 O'CONNOR, G. B., and CONWAY, M. E. (1950). *Plast. reconstr. Surg.*, **5**, 419.
 PIRES DE LIMA, J. A., and MONTEIRO, H. B. (1923). *Arq. Anat. Antrop.*, **8**, 185.
 SANVENERO-ROSSELLI, G. (1940). *Plast. chir.*, **1**, 184.
 ——— (1948). *Deutsch. Zahn. Z.*, **3**, 816.
 SCHACHTER, M. (1947). *Ann. Paediat.*, **169**, 345.
 STRAITH, C. L., and LEWIS, J. R. (1949). *Plast. reconstr. Surg.*, **4**, 204.
 STREIFF, E. B. (1950). *Ophthalmologica, Basel*, **120**, 79.
 TYRRELL, F. A. C. (1903). *Trans. ophthal. Soc. U.K.*, **23**, 263.
 WAARDENBURG, P. J. (1932). "Das menschliche Auge und seine Erbanlagen", p. 51. Nijhoff, The Hague.
 ——— (1934). *Klin. Mbl. Augenheilk.*, **92**, 29.
 ——— (1948). *Ned. T. Geneesk.*, **92**, 3455.
 WOLFF, E. (1934). *Arch. Anat. Strasbourg*, **18**, 229.

ADDITIONAL BIBLIOGRAPHY

- DUKE-ELDER, S. (1952). "Text-book of Ophthalmology", vol. 5, p. 4720. Kimpton, London.
 FRANCESCHETTI, A., BROCHER, J. E. W., and KLEIN, D. (1949). *Ophthalmologica, Basel*, **11**, 796.
 ——— and KLEIN, D. (1949). *Acta ophthal., Kbh.*, **27**, 143.
 ——— and VALERIO, M. (1945). *Confin. neurol., Basel*, **6**, 255.
 HALLERMANN, W. (1948). *Klin. Mbl. Augenheilk.*, **113**, 315.
 PIRES DE LIMA, J. A. (1930). *Ann. Anat. path. méd-chir.*, **7**, 377.