THE TREACHER COLLINS-FRANCESCHETTI SYNDROME

By M. SPENCER HARRISON (Lincoln)

CASES showing a similar clinical picture of congenital notching of the outer third of each lower lid and deficient malar bones were described by Berry in 1888 and in 1900 E. Treacher Collins drew attention to the condition as a syndrome. Since then Mann, Kilner and Johnstone (1943) have described cases in this country and Franceschetti (1944-45-49) published a description of the syndrome under the name "mandibulo-facial dysostosis" which describes a much broader series of deformities than those recounted by Treacher Collins. Continental authors now refer to the syndrome of Franceschetti (Holm, 1948; Bregeat, 1949; Gayral, 1950) or of Franceschetti-Zwahlen (Brom, 1947). Cases of this condition have also been presented by Isakowitz (1927), Pires de Lima (1930), Waardenburg (1934-49), Kazanjaan (1936), Hermans (1936), Lint (1937), McEnery (1937), Schacter (1947), Navis (1948), Savenero Roselli (1948), Straith (1949), Hallberg (1949), O'Connor (1950), Harrison (1951), Briggs (1952), McNeil (1953) and Szlazak (1953).

The syndrome consists of certain associated congenital and familial deformities of the ears, malar bones, lips, chin and lower eyelids. The similarity between the patients is very striking though the number and degree of the deformities, which are usually bilateral, varies considerably. From the literature it is thus difficult to conclude what constitutes the fundamentals of the syndrome and what are associated deformities, but it is generally agreed that the findings may be placed under a series of headings.

Deformities of the ears. Variations in shape and size of auricle and external auditory meatus, the auricles often tending to be placed lower than normal and to point backwards. At times microtia and meatal atresia. Auricular fistulae. Malformation of middle and inner ear producing conductive or perceptive deafness.

Deformities of the malar bones and zygomatic arch. The bones may be flattened with bony defects below and lateral to the external canthi.

Deformities of lips and mouth. Cleft lip and palate, enlarged upper lip, macrostomia, high palate, malocclusion of teeth and choanal atresia.

Deformities of chin. Hypoplasia of mandible producing micrognathia.

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Deformities of orbits. Palpebral fissures sloping downward and outwards. Notching of lower lids, which, in their medial portions are atrophic and without lashes and without meibomian glands. Absent puncta and obstruction of nasolachrymal ducts. Also at times subconjunctival lipomata, slight heterochromia, absence of certain eye movements and retraction of the globe.

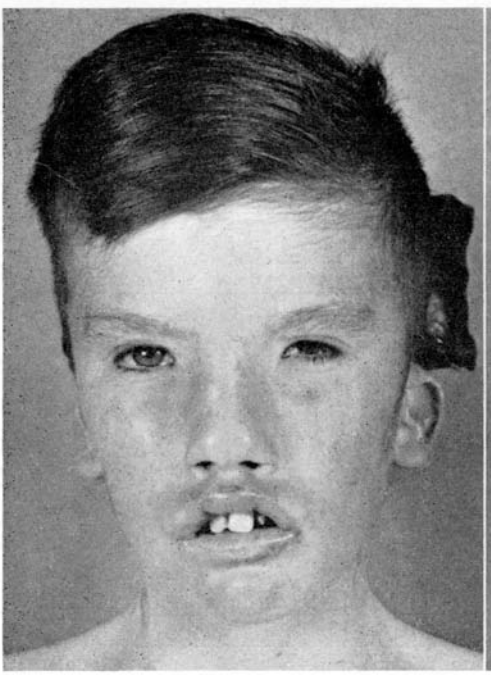


FIG. 1.

R.C., 9 years. Flattened infra-orbital margins, Small receding chin. Lymphangioma of lip.

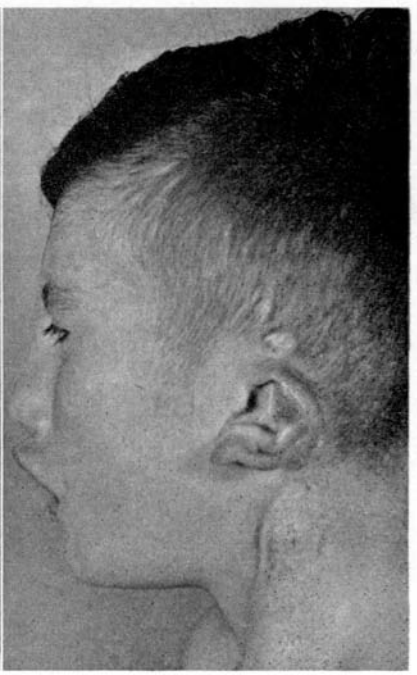


FIG. 2.

Ptosis auriculae and microtia. Scar of supra-auricular fistula. Scarring below ear.

Associated deformities. Flattened parieto-occipital bones. Nasal deformities. Deformities of the extremities such as long second metatarsal bones and hypermotility of metatarso-phalangeal joints. Family history of deafness and deaf mutism as well as other hereditary disorders.

The hereditary character of the syndrome has been established by genealogies (Debusmann, 1940; Leopold, 1945, and Brohm, 1947). The 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 amongst the causative factors. The etiology of the condition is clearly genetic and defective ossification of the bones of the face, derived from the visceral mesoderm, provides the

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primary lesion. The defect must presumably date from the seventh week of fœtal life. Similar anomalies have been produced in chicks by X-ray irradiation during development (Wolff, 1934).

Franceschetti and Klein (1954) have described the syndrome in association with the Klippel-Feil syndrome.

Illustrative case histories follow:



FIG. 3.

Grandmother, parent and child. Showing syndrome progressively becoming more marked.

R.C. 9 YEARS (Figs. 1 and 2)

This intelligent boy with satisfactory articulation was making normal progress at school. There was no family history of facial or other congenital abnormalities. Deficiency of both malar bones was obvious with noticeable flattening in the region of the infra-orbital margins. Cilia were present throughout the lower lids, but both lachrymal ducts were blocked. The left eye was amblyopic. The chin was small and receding. The palate was high and narrow and the nasal bridge was broadened. Scarring was present in relation to the upper third of each sternomastoid muscle. The upper lip contained an extensive lymphangioma. The second and third digits of both feet were webbed. Scars of supra-auricular fistulae were present on both sides, the right one still discharging. Both auricles were small (microtia) and placed caudally to the normal (ptosis auriculae) and rotated, pointing backwards and upwards and compressed from above downwards. Both external auditory meatuses were narrow and the drumheads appeared irregular. A little conductive hearing loss was present which was

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diagnosed as the result of otitis media earlier in life. Bilateral dacryocystorhinostomies were carried out with satisfactory results.

M.C. (Figs. 3, 4 and 5)

Male child aged 2 months, first seen on 5.8.54 was referred owing to upper respiratory obstruction thought by family doctor to be associated with recession of the lower jaw. Examination showed typical mandibulo-facial dysostosis with congenital laryngeal stridor. The auricles were rudimentary and almost sym-



FIG. 4.
M.C., aged 2 months. Sloping palpebral fissures. Notching of lid.



FIG. 5.
Rudimentary auricle with absence of external auditory meatus.

metrical. No external auditory meatus could be found on either side. The child appeared intelligent and could hear different notes when sounded loudly in both ears

G.C. (Fig. 6)

Child's mother aged 32 years. Appearance similar to that of child, though features of the condition did not appear so marked. The auricles were similar and there were no external auditory meatuses.

Hearing	Right	Left	
Rinne	negative	negative	} 512
A.B.C.	very little fall		

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FIG. 6.

Mother of child (in Figs. 4 and 5). Very similar condition of auricle and meatus.

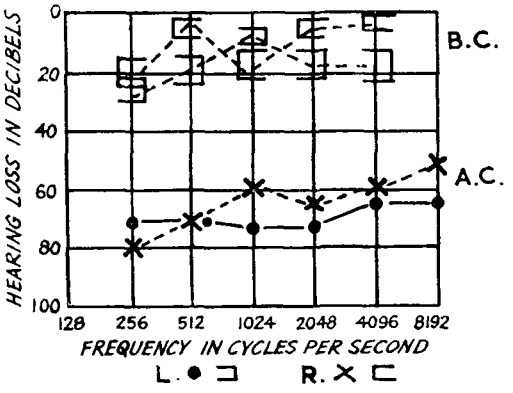


FIG. 7.

Audiogram A.61. 5.8.54. Pre-operative air and bone conduction curves.

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X-rays of the petrous bones showed absence of bony meatuses, the middle ears were present, but further details were difficult to interpret.

Ombredanne's operation, 11.10.55. Post-auricular incision. There was no bony meatus at all, but a depression in front of the markedly deformed auricle had a fibrous attachment to the bone of the anterior part of the mastoid process. The latter was smaller than usual in both dimensions. The bone was very sclerotic and the lateral sinus was very far forward, part of it being exposed. The dura of the middle fossa was rather low. As the deeper part of the mastoid was approached with a dental burr the ivory hard bone was a little softer near what might have been the site of the antrum and retro-facial cells, but no other anatomical details could be made out. There was no attic and no bony dome of the lateral semicircular canal. The facial nerve was discovered in its lower part and followed upwards and the middle ear opened and the rudimentary ossicles seen. The semicircular canal was found some distance from the facial nerve in a mass of very hard bone and a wide fenestration opening was made in the one stage as it was impossible to find the canal without opening it. The membranous canal appeared healthy and a large flap of skin from below and behind the ear was turned into the mastoid cavity and over the fenestration area and packed in position with sponges soaked in chloromycetin solution. The remainder of the cavity was lined with a Thiersch graft. Figs. 8 and 9.

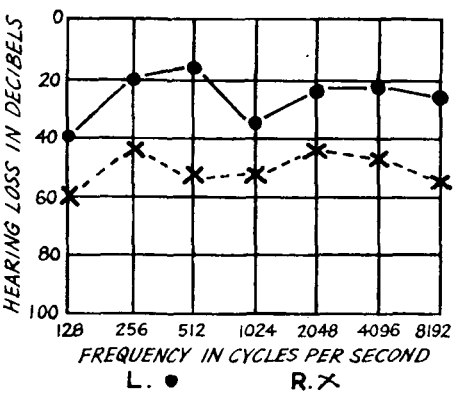


Fig. 8.
Post-operative audiogram. A.61. 10.11.55.

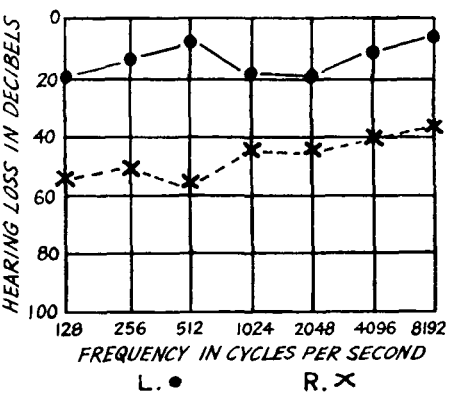


Fig. 9.
Post-operative audiogram. A.61. 16.12.56.

The grandmother's features are similar in some respects to those of her daughter and grandson and there can be little doubt that the early features of mandibulo-facial dysostosis are present in her case.

A.M.H. 8 Months (Figs. 10 and 11). Kindly sent to me by Mr. S. W. Allinson.

The seventh child in a family in which all others were normal. Both the auricles and the facial appearance closely resembled those of the male child, M.C. illustrated in Figs. 3, 4 and 5. The auricles were rudimentary and the external meatuses absent. The malar bones showed the characteristic depression

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so that the palpebral fissures sloped downwards and outwards. There was notching of the lower lids, a considerable area of which were without eyelashes. Associated deformities of other parts of the body were not discovered.

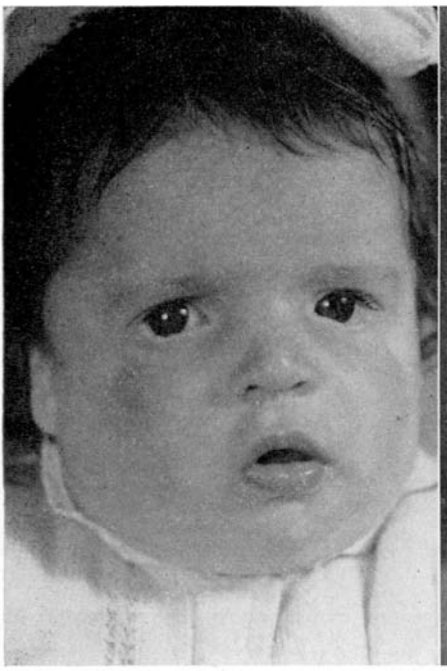


FIG. 10.

Lower lids lacking in lashes and notched. Palpebral fissures sloping. Malar depression.



FIG. 11.

Rudimentary auricle with absent meatus.

Summary

A hereditary cervico-oculo-facial dysmorphia which appears to represent a morphological entity is described with five illustrative cases. This condition may occur in association with many other hereditary disorders. The appearance of the patient is often misleading for the mental condition is usually normal and if the hearing is reasonably good the child does well at school. The combination of the facial appearance together with deaf-mutism must often have caused the unfortunate child to be considered an imbecile, little attempt being made to teach it language.

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