THE TREACHER COLLINS SYNDROME

By A. G. D. MARAN (Edinburgh)

"All cases are unique, and very similar to others."
T. S. Eliot, *The Cocktail Party*.

FROM Edinburgh in 1889, Berry, an ophthalmologist, described the cases of a mother and daughter with colobomata in the unusual site of the lower lid. The drawing of the daughter showed her to have sloping eyes, micrognathia and an abnormal prolongation of the hair line on to the cheek—the tightly bound hair and bonnet covered her ears leaving us in doubt as to whether a malformation existed. Eleven years later Treacher Collins described a boy of 8 and a man of 32 with lower lid colobomata, antimongoloid eyes and an unusual want of prominence of the malar bones, the drawing of the boy showing micrognathia and normal external ears. It was Franceschetti in 1949 who first gathered together all the main features to form the syndrome we recognize today and he was also the first to claim it as a syndrome. In the United States and Great Britain it is associated with the name of Treacher Collins but continental writers prefer to call it Franceschetti's Syndrome. A welter of cases exists however with the same thing described variously as mandibulo-facial dysostosis, the first arch syndrome, bilateral facial agenesia and multiple facial abnormalities.

It is rather odd to describe a patient as having the Treacher Collins Syndrome when he has few of the characteristics described by Treacher Collins, but to argue about what Treacher Collins himself described is really just saying what happened at a certain historical point in medicine. The question one must ask is whether certain pathological or clinical features occur together frequently enough for one to justifiably recognize a syndrome.

There is a close relationship between this syndrome and other craniofacial abnormalities which are, in general, cursed with an unsatisfactory nomenclature and a conglomeration of misleading eponyms, and it is sometimes difficult to define the borderline. Franchescetti himself complicates the issue further by allowing for a wide variation in the signs by his classification.

Like all so called rare diseases this syndrome is not nearly so rare as reports would have one believe as a search of the available literature has

https://doi.org/10.1017/S0022215100061922 Published online by Cambridge University Press

revealed 85 documented cases with another 30 or so mentioned as existing. The nine cases described here have been seen over a period of 2 months.

Case Reports

Case I: Mrs. G.B. (29) presented the complete picture of the syndrome. The eyes sloped downwards (antimongoloid) and there was a coloboma at the junction of the lateral third and medial two-thirds of the lower lid; eyelashes were missing from the medial two-thirds of the lower lid. On both sides the upper portion of the external ear was missing and while the left meatus was narrow and oblique upwards, the right meatus was absent. There was distinct flattening of the malar bones and maxillae with the resultant high short palate, poorly formed alveolar ridge and also a bifid uvula. The mandible was underdeveloped. The result of the poor facial development was that the nose was a prominent feature and false hypertelorism existed.

Audiogram showed a 65 db. loss in both ears with normal bone conduction. Radiographs showed defective development of the posterior portion of the zygomatic process on both sides. The condyle of the mandible was mis-shapen on both sides and displaced backwards so that the middle ear cleft was markedly narrowed in the antero-posterior direction. The shadows of the ossicles were clearly seen.

Case II: Miss C.B. (4), daughter of Case I, was again a complete case showing antimongoloid eyes, slight ectropion, colobomata and absent lashes on the medial two-thirds of the lower lid. The facial bones were poorly developed with resultant flattening, and a high arched short palate with a bifid uvula. There was micrognathia and malocclusion. Both external ears were abnormal with deformed helices but while both meati were narrow and oblique upwards the tympanic membranes looked normal.

Audiogram showed a conductive loss of 40 db. in the left ear and 25 db. in the right. Radiographs were not done.

Case III: Master T.B. (2), son of Case I, who was typical. He had antimongoloid eyes, ectropion, colobomata and absent lashes on the medial two-thirds of the lower lid. There was a depression at the malar eminence and the maxillae were flattened with a high short palate. Micrognathia resulted in malocclusion. Both helices were slightly abnormal and the meati were pin hole and upward sloping, the drums not being visible.

Audiograms and radiographs were not done but voice tests with play technique showed a hearing loss in both ears.

The above three cases are shown together in Fig. 1.

Case IV: Baby G.B. (1 week), second son of Case I, showed antimongoloid eyes with absent lashes on the lower lid, colobomata and some

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FIG. 1.
Mother, daughter and son, Cases I-III.

ectropion. There was distinct flattening of the cheek showing poor development of the malar bones, a high short palate and micrognathia. Both helices were slightly deformed and the meati were narrow and oblique upwards (Fig. 2).

Case V: Mr. G.C. (18), brother of Case I, showed exactly the same facial and ophthalmological features as the others. There was a slight malformation of the external ear and the tympanic membranes were not seen due to the narrow oblique meati. Rinne was bilaterally negative and a whispered voice was heard only at the ear on both sides. Radiographs showed the malar bones to be rudimentary and the zygomatic process of the temporal bone to be very small. The maxillae were poorly developed, the orbital floor almost non-existent and the antra were small. The condyles of the mandible were malformed and more posteriorly placed than normal and the coronoid processes were everted. There was malposition and overcrowding of the teeth. The middle ear and ossicles were present. He also had hydronephrosis which was operated on (Fig. 3).

Cases VI and VII: These were the 25-year-old brother and father of Case V. They showed the same features as Case V together with deafness but refused to be photographed or submit to any further investigation.

From hearsay, the paternal grandfather was also affected.

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FIG. 2.
Second son of Case I.



FIG. 3.
Brother of Case I.

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Fig. 4 shows the pedigree of this family.

Case VIII: Miss S.M. (10) was an atypical case. She lacked the eye deformity but had flattening of the malar bones and cheek, a high arched short palate, micrognathia and malocclusion. The tongue-shaped process of hair extending on to the cheek sometimes described is well seen in Fig. 5. Both external ears were malformed, the meati sloped upwards and there was a conductive deafness of 50 db. in both ears.

Radiographs showed the posterior end of the zygomatic arch on both sides to be underdeveloped and tomograms showed the ossicles clearly on both sides with the left middle ear larger than normal for a child of 10 and the right middle ear abnormally small.

Dr. A. B. Smith performed a left tympanotomy and found a deep pocket extending superiorly to the suprameatal triangle. The malleus and stapes were normal but the long process of the incus was shortened, the gap being completed by a thin strand of fibrous tissue. The drum was laid on the stapes and the residual air bone gap was 10 db.

There was apparent hypertelorism but although the fingers of both hands were abnormally long and hyperextensile she had none of the other characteristics of Marfan's Syndrome.

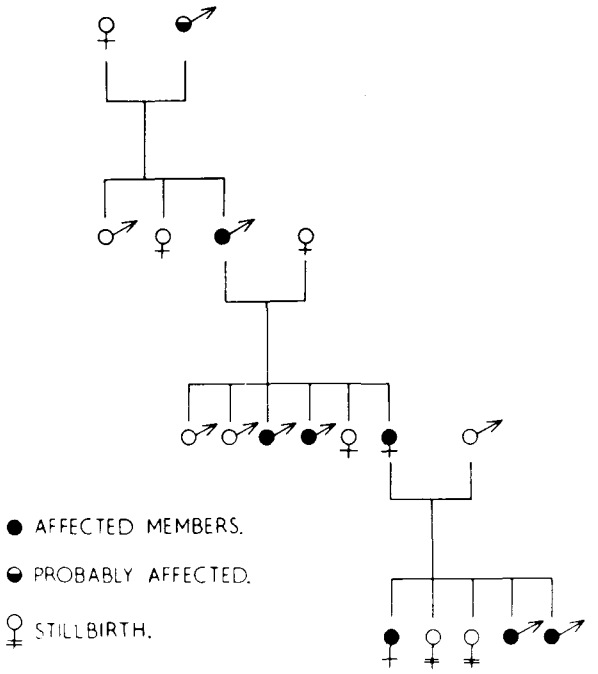


Fig. 4.
Pedigree of Case I.

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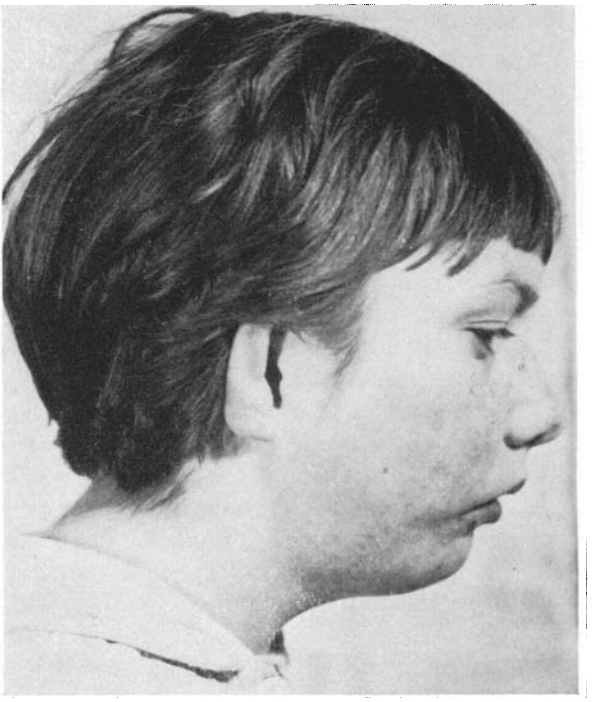


FIG. 5.
Case VIII showing tongue-shaped extension of hair to cheek.

This case showed a very close resemblance to her mother, who, having none of the real characteristics of the syndrome, has a conductive deafness of 40 db. in her right ear with normal tomograms.

Case IX: Miss W.H. (12) is either an atypical case or else one of those cases who does not quite fit in to any syndrome. The axis of the eyes was slightly off the horizontal and although no colobomata existed there were no lashes in the medial third of the lower lids. The right lacrimal duct was absent, a divergent squint existed and there was hypertelorism. There was some flattening of the cheek and the palate was short and cleft. The mandible was underdeveloped with resulting malocclusion and a peculiar feature of the teeth was that the upper central incisors were fused. Posterior rhinoscopy showed a completely bifid posterior end of the septum although anterior rhinoscopy was normal. Although the external ears were normally formed they were rather low set and the meati were sloping obliquely upwards and so narrow that no drum could be seen. The left foot had a talipes equino-varus deformity and the hands and feet were webbed.

A conductive deafness of 60 db. in the right ear and 45 db. in the left ear existed. Radiographs were interesting as they showed a defective

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development of the tympanic plate on both sides; no external meatus could be seen on the right side and it was deficient on the left, a bony plate being present at the inner end. The ossicles on both sides were seen normally on tomography and the middle ears were normally developed. The cleft palate was well seen and the bony vomer was fused to the right side of the palatal split (Fig. 6).

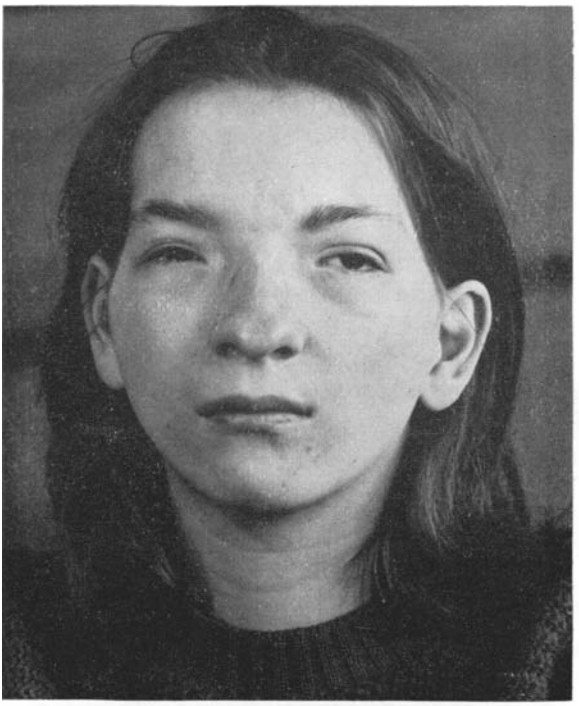


FIG. 6.
Case IX—many atypical features.

Clinical Features

Franceschetti (1949) classified the syndrome into complete, incomplete, abortive, unilateral and atypical. The features are as follows:

Complete form

(*a*) The palpebral fissures are oblique and slope downwards and laterally (antimongoloid) with a coloboma in the outer portion of the lower lids which are S-shaped due to the notch at the junction of the medial two-thirds and lateral third. There are no lashes in the medial two-thirds of the lower lid and meibomian glands are absent. The morphology of the antimongoloid axis can be racial, a variation within the population or the result of some malformation. Having eliminated the former

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two, one is left with a completely absent malar bone or one demonstrating the merest vestige. If this obliquity is to be associated with the syndrome there must be ample evidence that the malar bone which forms the outer and lower quadrant of the orbit is missing or defective.

(*b*) All bones arising from the maxillary and mandibular processes show some disturbance of growth potential. The absent malar has already been noted but its absence does not prevent the complete encirclement of the orbit by bone. The maxilla presents a defective pattern of growth and the palate can be high and narrow or cleft. The fronto-nasal elements which are unaffected in the syndrome tend to dominate the face and stand out in great contrast to the reduced maxillary process components and the nose tends to be a prominent feature. The mandible shows changes that range from severe degrees of micrognathism to jaws with a more normal form. The condyloid process is the most affected part and the reduction in length of the mandible depends on the extent to which the condyloid cartilage is affected. The maxillary and mandibular defect result in malocclusion of the teeth.

(*c*) Malformed external and middle ears—this will be dealt with separately later.

(*d*) Blind fistulae between the angles of the mouth and ear.

(*e*) Atypical hair growth in the form of a tongue-shaped process of hair line extending towards the cheek occasionally exists. This is a feature not seen in any other malformation of the cranio-facial structures.

Incomplete form

The appearance of the palpebral fissure and lower lids is characteristic and there is under-development of the malar bones and mandible. External ear form is normal but hearing is impaired. In this group the syndrome is present but the development is less extensive and less striking.

Abortive form

Only the eyelid deformities are present. Franceschetti (1949) commented that it was difficult to decide whether some abortive cases represent a pathological stigma, the so-called "forme fruste" of the syndrome.

Unilateral form

The abnormalities of whatever degree are on one side only: this according to Greer Walker (1961) does not exist and these are cases of intra-uterine facial necrosis.

Atypical form

This includes incomplete forms of the syndrome in which one of the

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principal characteristics is missing while other abnormalities such as microphthalmus which do not belong to the syndrome may be present.

Using the above classification we have found that the literature consists of 50 per cent. complete forms, 18 per cent. incomplete forms, 12 per cent. abortive, 4 per cent. unilateral and 16 per cent. atypical. Because of the possible relationship of the Treacher Collins Syndrome to other syndromes both clinically and genetically and since it is difficult to classify most non-complete cases it is felt that this classification serves only to cloud the issue and is artificial in its horizons. The main abnormalities are those of the eye, jaw, maxillary processes and ear, and one must only judge whether there are enough of these present to warrant its description as a Treacher Collins Syndrome.

Associated Anomalies

As with any congenital defect other anomalies seemingly unrelated to the main one are often seen and often missed. A great variety have been described in relation to the Treacher Collins Syndrome.

Most commonly described are abnormalities of the extremities such as bilateral aplasia of the thumb and radio-ulnar synostosis (Franceschetti and Klein, 1949), club foot (Kumar and Sood, 1962; Granrud, 1953, and Case IX), abnormally long second metatarsals (Straith and Lewis, 1949), bilateral partial syndactyly of the toes (Brégeat and Naud, 1949), webbing of the digits of the feet and hands (Ronconi, 1962; Harrison, 1957; and Case IX), klinodactyly (Herberts, 1962) and hyperextensile metatarso- and carpo-phalangeal joints (Straith and Lewis, 1949, and Case VIII).

There are cranio-facial anomalies such as facial asymmetry (Pires de Lima and Montiero, 1923; Schachter, 1947; and van Lint and Hennebert, 1936), internal hydrocephalus (Franceschetti and Klein, 1949), ankylosis of the temporo-mandibular joint (Brégeat and Naud, 1949), absence of the mastoid process (Franceschetti and Klein, 1949; and Ronconi, 1962), absent parotid gland (McKenzie and Craig, 1955), mental deficiency (Schachter, 1947; and Granrud, 1953), and lymphangioma of the upper lip (Harrison, 1957). To this group can be added the eye anomalies of heterochromia (Franceschetti and Klein, 1949), microphthalmia (Ronconi, 1962), double row of lashes on the lower lid (Halberg and Paunessa, 1949), webbed skin in the external canthus (Snyder, 1956), absent lacrimal ducts (O'Connor and Conway, 1950, and Case IX), and a divergent squint (Case IX). The dental anomalies of anomalous implantation of teeth (Halberg and Paunessa, 1949, and Case V) and twin incisors (Case IX) have been described. Choanal atresia (McNeil and Wynter Wedderburn, 1953) and a bifid vomer (Case IX) may occur in the nose.

An asymmetrical thorax (Schachter, 1947; Hermans, 1936; and Ronconi, 1962), sternal deformities (Mann, 1943) and fusion of the verte-

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brae (Franceschetti and Klein, 1949; and Ronconi, 1962) have been described.

There are recorded three cases each of hydronephrosis (Livingstone, 1962, and Case V) and undescended testes (Brégeat and Naud, 1949; Hermans, 1936; and Schachter, 1947), but perhaps one of the most significant features is that the two cases which have been brought to autopsy by Fujino *et al.* (1961) and Granrud (1953) both had a patent foramen ovale; Fujino's case also had a patent ductus arteriosus, a bicornuate uterus and oblong soft ovaries.

In view of all this one cannot agree too strongly with Livingstone (1962) who stated that the possibility of multiple lesions was so great that a full pediatric examination should be done in all cases.

Otological Defects

Although these cases usually present to the otologist as deafness we must remember that many more attend the ophthalmologist or plastic surgeon. This is probably why only 48 per cent. of cases in the literature are described as being deaf. This cannot wholly be ascribed to the disinterest of the ophthalmologist and plastic surgeon to the problems of hearing as a good number of cases are noted as having normal hearing.

All manner of abnormalities of the external ear have been reported varying from the normal, through minor abnormalities of the helices to complete microtia. The meatus may be normal or absent but is quite often narrowed and sloping upwards (Holborow, 1961) with a deep pocket superiorly (Herberts, 1962, and Case VIII). The drum is usually normal but has been described as perforated (Aubry *et al.*, 1953), or replaced by bone (Herberts, 1962; Lockhart, 1929; and Case IX).

Harrison (1957) found no attic and a small antrum while Lockhart (1929) in his dissection found absence of both attic and antrum. On one side he found no ossicles and a large tensor tympani muscle and on the other he found "an attempted malleus like a prehistoric bone axe, the head almost immovably applied to the roof of the cavity" and no other ossicles. This sounds like fusion of the malleus and incus which may be attached to the anterior wall as described by Livingstone (1958, 1959). Herberts (1962) described one case with the head of the malleus missing, the incus represented by a shapeless bone and what appeared to be the remains of the stapes lacking in both crura and plate, and a second case in which the handle of the malleus was missing, the malformed head and incus remaining and a mobile normal stapes. Holborow (1961) described a brother and sister both with an absent incus. If the syndrome were derived solely from the first arch one would expect deformities of the malleus and incus, but Plester (1961) and Keerl (1962), describing the same case show a drawing of a columella stapes fixed in the oval window by a malformation

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of the footplate and a small bony bridge joining it to the facial canal, while McKenzie's case (1955) showed an absent incus and stapes.

Harrison (1957) mentions an abnormal placement of the lateral canal and Livingstone (1958) mentioned that in three fenestrations the canal was found in only one case. In Herberts' (1962) first case the labyrinth mass was missing in the upward and forward direction and corresponding to the posterior canal there was a raised portion of labyrinth bone. In his second case the facial canal was devoid of a bony covering but this is the only recorded abnormality of the facial nerve.

Anatomy

The clinical signs can be illustrated better by a study of the anatomy and two cases have been fully dissected. Professor Lockhart of Aberdeen in 1929 dissected a case of a man of 75 years without realizing that it was a Treacher Collins Syndrome (Fig. 7).

Both zygomata were absent apart from a small posterior root and the malars were rudimentary. There was no articular eminence, the inferior aspect of the squamous temporal bone was restricted and the glenoid fossa was shallow. The plate of the squamous temporal bone was small and it was completed by numerous sutural bones. The floor of the orbit sloped downwards and forwards with no fossa posterior to the infra-orbital margin. The frontal process of the maxilla was large. The condyle of the mandible was not convex and elongated transversely but was small, flat and circular with a well formed articular disc. The coronoid notch was small as it was encroached on by a very large coronoid process which was markedly everted. The upper jaw had a curious gothic arch appearance and both pterygoid plates were badly developed. A striking feature was the massive development and fusion of the temporalis and masseter muscles. The absence of the zygoma allowed the origin of the temporalis to extend very low on the squama and it was inserted to both sides of the coronoid process.

The findings of McKenzie and Craig (1955), also from Aberdeen, were almost exactly the same.

Radiology

The main radiological abnormalities according to Campbell (1954) are non-fusion of the zygomatic arches, absence of the bodies of the malar bones, ossification of each malar bone from two centres with non-fusion of the frontal process, agenesis of the palatine bones, cleft palate and agenesis of the mandible. In a review of 64 cases, Stovin *et al.* (1960) found that only 24 had been X-rayed and of these 17 had defective malar bones, nine had hypoplastic maxillae and the surprisingly high number of six had inner ear abnormalities. In 1959 Livingstone stated that X-rays were of little value to show whether the middle ear was

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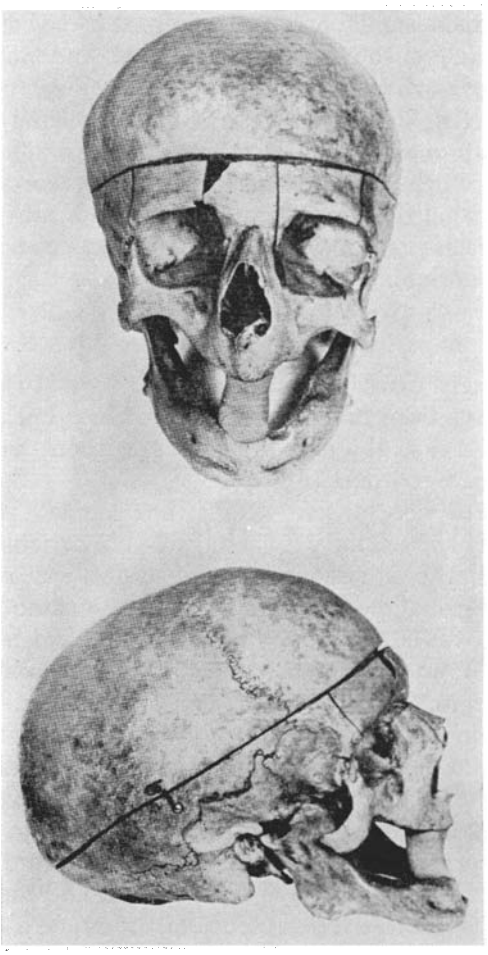


FIG. 7.
Professor Lockhart's case.
(From D. G. Walker: *Malformations of the Face*. Edinburgh: Livingstone.)

present or not but this can hardly be held now in view of the recent advances in tomography of the middle ear.

Pathogenesis

No useful purpose can be served by discussing at length all the suggestions with regard to the cause of the Treacher Collins Syndrome since so much of the true understanding of congenital malformations is only beginning to come to light. It may be that eventually the simple morphological explanation will be replaced by concrete evidence of defects in organization, differentiation or migration.

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The Treacher Collins Syndrome

Treacher Collins (1900) mentioned that the mother in his paper had had a fright during pregnancy from a rat which had something wrong with its eyes, and Mann and Kilner (1943) noted a similar upset caused by a dog. Mall (1917) noted that normal embryos were influenced by environment and suggested that poor nutrition caused a faulty implantation of the ovum in the uterus while Keith (1940) blamed placental circulatory failure. Granrud (1953) presented five cases and blamed the cause respectively on influenza and ante-partum hæmorrhage, rubella, diphtheria, attempted abortion and hydramnios, and Kibel (1962) mentioned varicella as a cause. Herberts (1962) gives a comprehensive list of various factors which have been known to cause fœtal abnormality.

McKenzie (1955) reviewed the embryology of the first visceral arch with particular reference to the vascular supply during the first 2 months. He stressed the hazardous existence of this region from the third to the fifth week dependent as it is on the relay of blood via three successive vessels each of which in turn takes over the dominant supply—the remains of the first aortic arch, the stapedial artery with its branches and the external carotid artery. These early vascular changes causing absolute or relative malnutrition to the highly active embryonic tissues "may result in complete chaos in the subsequent development of the first visceral arch and its derivatives such as the inhibition of growth in the maxilla, mandible, or ear, or, if the conditions at the time happen to be particularly favourable, no perceptible abnormality".

Ida Mann (1943) stated that the syndrome was due to retardation of differentiation of the maxillary mesoderm at and after the 50 mm. stage because in her case the boy was deaf and there was evidence of malformation of the middle ear cleft. This points to a delay or arrest at or before the end of the second month when differentiation of the middle ear is proceeding. The position of the eyes was normal showing that upgrowth of the visceral wedge of mesoderm in the temporal region was normal until the third month (50 mm. stage). Arrest towards the end of the second month is compatible with the presence of teeth in the upper jaw since the primary dental lamina begins to appear early in the sixth week before the rudiments of the lashes so that an arrest a little later might not inhibit the formation of the teeth.

This explanation does not however complete the whole picture of the Treacher Collins Syndrome. As Harrison (1951) points out, the syndrome involves not only the maxillary process but also the mandibular and hyoid arches to no small degree as is seen by the occurrence of cleft lip and palate and stapedial abnormalities. The timing of events is important as different injuries which occur at identically the same time in development can result in the same defect, whereas identical injuries occurring at different stages can result in different defects (Stockard, 1921). For example, if the labyrinth is affected it would seem probable that the

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causal mechanism of the deformation arises at an earlier stage of embryonic development since the foundations of the labyrinth are laid and developed earlier (first and second embryonic month) than those of the external ear and meatus (third embryonic month).

Hereditary Factor

There are so many geneologies recorded that one must assume that this syndrome is an independent genotypic entity which follows the irregular dominant form of transmission—Berry (1889), Pires de Lima and Montiero (1923), Isakowitz (1927), Debusmann (1940), Leopold *et al.* (1945), Schachter (1947), Sanvenero-Roselli (1948), Waardenberg (1948), Straith and Lewis (1949), Harrison (1957), Wildervanck (1960), and Holborow (1961). Leopold *et al.* (1945) described a transmission of a homologous picture in three generations, while Brohm and Kluska (1947) and Harrison (1957) showed intra-familial variability with increasing severity in three generations. Debusmann (1940) showed a dominant mode of transmission in three generations of increasing severity with all three cases in the third generation dying. In our series Case I had two stillbirths (third generation) but we were unable to find out if these were affected or not.

The marked variability in the degree of the manifestation observed in the reported cases of the Treacher Collins Syndrome which makes it necessary to classify them into a number of different categories points to an unstable gene of variable expression, penetrance and specificity. Expressivity is (*a*) the activity of the gene itself which depends on the quantity of some substance produced by it in initiating the reaction for which it is responsible as well as, (*b*) its activity in relation to the rest of the genes and the environment (Waddington, 1939). Penetrance is a measure of the frequency with which the characteristic features of a gene appear in the carrier while its specificity refers to the kind of effect rather than to the amount. Picó (1961) notes that if accessory abnormalities occur then one is dealing with a pleiotropic gene. McKenzie (1958) presumes that the gene is responsible for maldevelopment or even suppression of the stapedial artery while the compensatory processes (anastomoses) will provide the degrees of penetrance, although the other factors must be sought to explain the variability of the compensatory reactions. The expressivity and specificity of the gene on the other hand depend on the details and timing of the maladjustments occurring among the vessels concerned.

Wildervanck (1961) states that each child of a parent with the Treacher Collins Syndrome has a 50 : 50 chance of having a congenital lesion but only one in 14 of having deafness with complete meatal atresia. This is borne out in our geneology (Fig. 4) and also that of Debusmann (1950).

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It is interesting to note the distinct familial resemblance between Case VIII and her mother who does not have the Treacher Collins Syndrome. This is similar to the family described by Harrison (1957).

We examined the chromosomes of seven of our cases by a modified method of Moorhead (1960) with negative results and this agrees with the negative findings of Livingstone (1962) who examined skin biopsies.

Relationship to Other Syndromes

The Treacher Collins Syndrome, already allowed a variability of expression by Franceschetti's classification, has also been related to various other cranio-facial syndromes such as cranio-cleido-dysostosis (Brohm and Kluska, 1947), Klippel Feil syndrome (Franceschetti and Klein, 1949), Crouzon's syndrome (Clerc and Deumier, 1958), familial primary hypoplasia of the orbital margin (Urrets-Zavalia, 1955), congenital ectropion and distichiasis (Picó, 1961), Francois' syndrome (Ronconi, 1962), intra-uterine facial necrosis (Harrison, 1951) and acrocephalic syndactyly of Apert (Picó, 1961). McKenzie (1958) described a group of syndromes arising from abnormal development of the first visceral arch including the Treacher Collins Syndrome, the Pierre Robin Syndrome, mandibular dysostosis, hypertelorism, cleft lip and palate, deformities of the external and middle ear, and congenital deaf mutism. He suggested that all the anomalies are one hereditary syndrome caused by a dominant gene or groups of genes varying in expression. While no one would deny that most of these are due to maldevelopment of the first arch, to make any further conclusions in the light of present genetic knowledge is not warranted. Stark and Saunders (1962) argued along the same lines and concluded that to lump together the main deformities of the Treacher Collins Syndrome as a single entity was no more justified than grouping together as a syndrome the separate anomalies of cleft lip and palate even though they commonly co-exist.

Greer Walker (1961) summed this up by saying that general agreement was needed about defining the various malformations and the specific lesions should be separated from the non-specific ones that present no steadfast clinical manifestation. These specific malformations are frequently grouped with a heterogenous collection of cases under the belief that all are part and parcel of the same syndrome. This has happened with the Treacher Collins Syndrome and, as he says, the appearance is classical, but in spite of this it has been incorporated into other syndromes. The cases included under the wider terminology vary considerably in their clinical appearance and etiology.

To conclude, may we be allowed to tamper with the quotation from T. S. Eliot and say, "All syndromes are unique, and very similar to others."

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Summary

Seven typical and two atypical cases of the Treacher Collins syndrome are reported. The clinical features, associated anomalies and otological defects are reviewed. Some points in the anatomy and radiological study are presented. With regard to the pathogenesis it can be stated that until we are in a position to study the chromosome more minutely, McKenzie's vascular theory is the most likely. The genetic upset is evaluated and the probable false relationship to other cranio-facial anomalies is pointed out.

Acknowledgements

Thanks are due to Dr. A. B. Smith, Dr. J. P. Stewart and Dr. J. Forfar for permission to report on cases under their charge. Acknowledgements are also due to Dr. A. Bain for carrying out the chromosome studies and to Dr. A. Large, the family doctor, for his help in following up cases V, VI and VII. The help of Miss A. Barker was invaluable in tracing the references.

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