DEFICIENCY OF THE MALAR BONES WITH DEFECT OF THE LOWER LIDS*

BY

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*With Notes of a Similar Case, Treatment and
Suggestions.* By T. POMFRET KILNER

THE subject of this communication is a boy aged 11 years, sent to me by Major Peet. He was brought to the out-patient department by his step-mother with the request that something be done about his appearance as everyone laughed at him and thought him mentally deficient, which she considered was not the case. As soon as

A

B

FIG. 1.

A. The case described by Treacher Collins

B. The present case

he was seen the resemblance to the classic case reported by Treacher Collins in 1900 (Collins; Case with symmetrical congenital notches in outer part of each lower lid and defective development of the malar bones. *Trans. Ophthal. Soc., U.K.*, 1900,

* Read at the Oxford Ophthalmological Congress, 1942.

Vol. XX; p. 90) was noted, and we have no doubt that the condition is the same. Fig. 1 shows Treacher Collins' case and the present one side by side in approximately the same attitude.

The main points seen on superficial examination are:—

1. *Eyelids and Eyes.*—The upper lids are normal, but droop downwards at the outer ends owing to the defect of the lower lids,

FIG. 2.

The present case with the lids held up in position

so that a false appearance of squint is given in some positions since the eyes are not in the centres of the palpebral apertures. There is no ocular defect whatever, the eyes being emmetropic, visual acuity 6/5 R. and L., with perfect binocular vision and stereopsis and an amplitude of fusion of 50° at least. Fig. 2, in which the lids are held in position and the eyes are seen to be straight, brings this out. The lower lids are peculiar. The outer 2-3 mm. and the outer canthus are normal but the rest of the lid is thin, atrophic

looking, and shows no marginal differentiation, lashes, Meibomian glands and intermarginal strip being absent. The lower bony margin of the orbit is defective and the cheek depressed. The fibres of the orbicularis are absent at the inner canthus and along the lower lid. A small bony boss can be felt under the skin at the anterior end of the zygoma.

2. *Ear*.—The external ears are well formed but there is deafness, probably due to a malformation of the middle and possibly inner ear.

3. *Upper Jaw*.—This is narrow and there are abnormalities of spacing of the teeth.

Radiographs were taken of the skull in various positions by Mr. Kemp, who reports as follows:—

Radiographs show defective development of all the facial bones. In particular, the malar bones are very small and structurally malformed. The following are the principal deformities:—

The orbital cavities seem to be oval in shape. Their roofs are well formed but are inclined sharply downwards and outwards. The floors seem shallower than normal. The infra-orbital edges are much narrower than normal, especially at their outer margins where they are formed by the malar bones. The malar processes of the frontal bones and the orbital processes of the malar bones are much thinner than normal, and on the left structurally incomplete. Both maxillae are very small. The hard palate is small and foreshortened but comparatively well formed. The teeth are crowded together, and there is a moderate degree of superior protrusion of the incisors and canines. The nasal bones are well formed. Both frontal sinuses are well formed. The ethmoidal cells are normal but the maxillary antra are very small. The mandible is small and its body is bowed upwards to accommodate the teeth. The front teeth are crowded together and there is a moderate degree of inferior protrusion to meet the comparable deformity in the upper jaw. The premolar and molar teeth show mal-alignment. The vault of the skull seems normal in size and shape. Both mastoid processes are very small. The tympanic antra are not formed and there are no cells in the mastoids. The cochlea on both sides does not seem to be formed properly, for only a single coil can be made out. The bony labyrinths seem to be normal. The sternum shows malformations of the body. No other deformities are known to be present in the skeleton.

Discussion of Case

This centres round two points of interest, namely the embryological meaning of the defect and the possible lines of treatment.

The embryological interest of the case is great as it would

appear to throw further light on the still obscure parts played by the visceral and paraxial mesoderm respectively in the formation of the face. The general lines of development in this region are of course well known. The paraxial mesoderm surrounding the central nervous system extends in the form of enveloping capsules around the sense organs (olfactory, optic and otic) while the visceral mesoderm advances secondarily in the form of the maxillary process of the first visceral arch (mandibular) to produce the supporting skeletal and outer coverings necessitated by the great increase in width of the brain in the higher vertebrates. Thus both phylogenetically and ontogenetically we find a steady increase in the size and importance of the maxillary process. It forms in the first place the wedge of tissue which, extending upwards behind the laterally placed eyes to form the temporal bone and muscle, is mechanically responsible for the swinging round of the eyes from the lateral to the forward position required in binocular vision. In the second place it is responsible for the maxilla and malar bones, and thirdly, its superficial extensions account for the lower lid, the vestibule of the nose and the whole moulding of the side of the face. Put another way, one can say that the area formed of maxillary (visceral) mesoderm is that supplied by the second division of the Vth cranial nerve, while the paraxial part of the face is supplied by the first division, and the mandibular (visceral again) by the third division.

The interest lies in the mapping of the exact lines of junction on the adult face. In the present case it is obvious that there is no defect in the paraxial mesoderm. The upper lids, upper orbital margins, sclerae and extrinsic ocular muscles are all normal. The position of the eyes is normal, showing that the upgrowth of the visceral wedge in the temporal region was normal until nearly the third month (50 mm. stage or earlier) by which time the angle between the optic stalks is already 60° (the adult angle). The differentiation in this wedge is however defective as the boy is somewhat deaf, and there is evidence of malformation of the middle ear. This points to a delay or arrest at or before the end of the second month, at which time active differentiation of the middle ear should be proceeding.

The extension of the soft tissues of the maxillary process over the face to form the lower lids also appears to have occurred but their differentiation has been arrested. The first rudiments of lashes and Meibomian glands in the lower lid appear at the 50-60 mm. stage, and in this case they are absent, the lid fold being present but still in the undifferentiated condition seen during the second month. An arrest towards the end of the second month is also compatible with the presence of teeth in the upper jaw, since the primary dental lamina begins early in the sixth

week, long, before the rudiments of eyelashes, so that an arrest a little later might not inhibit the formation of teeth.

The presence of an intermarginal strip with normal lashes and glands at the outer end of the lower lid would seem to point to the formation of this part as well as the outer canthus itself, from the paraxial mesoderm of the upper lid fold. If this is so, then the sensory supply of the outer 2-3 mm. of the lower lid margin should be from the first division of the Vth cranial nerve. This does not appear to be known with certainty, but a patient in whom the 2nd division was recently injected by Mr. Pennybacker showed definite anaesthesia of the lower lid to within 2 mm. of the outer canthus only. A dental surgeon has also assured us that when using a novocaine block in the upper jaw, the lower lid is usually anaesthetic in its inner 3/4 at least, but never quite to the outer canthus. This overlap, or lack of complete co-incidence of nerve supply and apparent line of division, is repeated again at the vestibule of the nose, where the maxillary mesoderm, carrying its 2nd division of the Vth, overlaps this time the paraxial part

FIG. 3.

Figure from Buchanan's Anatomy showing the formation of the face, c. 16mm. stage. The extending visceral mesoderm is darkened.

of the nostril supplied by the 1st division. A case reported by me showed very clearly how in failure of the paraxial portion of the nostril the vestibule of the nose was nevertheless present. (Mann; Developmental abnormalities of the eye. Camb. Univ. Press, 1937). A diagram (Fig. 3) from Frazer's Manual of Embryology (London; Baillière, Tindall & Cox, 1931), also indicates this arrangement of the maxillary mesoderm, the falling away of the process from the outer canthus, and its overlap at the nostril being indicated at the 16 mm. stage.

Embryologically therefore, we would put this case as one of retardation of differentiation of maxillary mesoderm at and after the 50 mm. stage, and consider that it offers further evidence for the supply of the outer 2-3 mm. of the lower lid by the 1st division of the Vth cranial nerve.

It is rare and few records of similar cases exist. The earliest which seems to be in any way comparable was reported in 1888 by G. A. Berry (*Ophthalm. Hosp. Reports*, Vol. XII, p. 255, 1888). Berry describes a mother and daughter with congenital notches in the lower lids and gives a drawing of them. The notches were in the same position but there is practically no flattening of the cheek shown. Also lashes are shown in the sketch all along the lower lids, though thinner and finer internal to the notch. Nothing is said about the malar bone. Both the patients have receding chins as in the present case. In the mother the condition was bilateral, in the daughter on the right side only. The mother had also had a hare lip. The twin brother of the girl with the right-sided notch also had a hare lip. The mother's brother had a notch in the lid but was not examined. Here we have a definitely hereditary condition associated with hare lip in the same pedigree (and in one case in the same patient). Hare lip itself is now known to be due to defective development of the maxillary process, so that the co-existence of hare lip and congenital notches in the lower lids would appear to implicate the visceral mesoderm with an even greater certainty.

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Notes by Mr. T. Pomfret Kilner

Photographs of the case described above were sent to me by Major Peet with a request for advice on treatment.

I immediately recognised that I had under treatment at the time a case falling into the same category and it is at Miss Mann's

invitation that I present a description of the boy whose photographs appear in the illustration No. 4.

This child was brought to see me in 1937 at the age of fourteen months. He was the second child of parents of 35 and 38, the previous child, a girl, being then six years old and without congenital abnormality. There was no family history of congenital defects. The child was three weeks premature and the mother

FIG. 4.

Mr. T. Pomfret Kilner's case, before and after operation.

gave a history of having been frightened by a dog when three months pregnant.

My advice was sought chiefly for treatment of a *cleft soft palate*, but notes were made of the following abnormalities:—

Ears.—Both ears crumpled forwards and with very poor development of the pinna: no external auditory meatus present on either side.

The case was seen by my E.N.T. colleague, Mr. Bateman, who advised against any attempt to reconstruct a meatus, the prospects of hearing being so very poor.

Eyes.—Palpebral fissures slope downwards at their outer ends. Peculiar angulation of lower eyelids with small bushy collections of eyelashes at junction of outer and middle thirds.

Mother says eyelids were “practically absent” at birth. Deep depressions in infra-orbital regions.

The palate defect was successfully repaired at 14 months, May 31, 1937.

When the child was almost six years old, work was commenced on the ears and eyelids. The ear and eyelid of the right side were treated at the same sitting on February 21, 1942.

The ear was freed anteriorly so that it could be folded back into relatively normal position and the raw surface thus produced was covered by a Thiersch skin graft applied on a mould of dental composition.

A triangular skin flap was transposed from upper to lower eyelid across the outer canthus with the object of raising the outer angle of the palpebral fissure, giving some support to the lower eyelid and providing better protection for the eye. Three weeks later precisely similar procedures were executed on the ear and eyelid of the left side.

There remain deep depressions in the outer parts of the infra-orbital margins and the original treatment prescription includes the filling out of these by cartilage implants. This should provide improved contour in these regions and assist in restoring natural level to the lower eyelid margins, but if, owing to lack of muscle action, the lower eyelids still droop, thin laminae of cartilage will be introduced to provide support.