A CASE OF AGNATHIA, OR CONGENITAL ABSENCE OF THE LOWER JAW.

By BERTRAM M. H. ROGERS, M.D., B.Ch., B.A. (Oxon.), *Physician and
Pathologist to the Hospital for Women and Children, Bristol.*

(PLATE XVII.)

THE word agnathia is used, according to the "New Sydenham Society's Lexicon of Medicine," to denote malformation from arrest of development, in which one or both jaws are defective. This work also says that the mouth may be absent or closed posteriorly, and that when this occurs there is commonly defective development of the upper jaw, the palate, and vomer. Occasionally the two temporal bones are brought into proximity, or are in contact, and the ears are approximated.

Though, no doubt, all these conditions may be found, the word itself cannot be meant to define anything more than the absence of the jaw; and I have made use of the word, therefore, to describe a specimen in which this condition exists, though there are other interesting and uncommon developmental deficiencies that I shall include in the description.

The mother, Mrs. T., was a patient of my colleague, Dr. Lees, who kindly brought the specimen to add to the collection at the Children's Hospital. She had given birth to four living children, three boys and a girl, and had had three miscarriages, the last one about three years ago. From that time her catamenia were regular till July 1896, when they ceased, and calculating from this date she expected her confinement in May 1897. As I shall point out later there must have been some mistake, for the skiagram shows that the foetus was about 6 months old. There is nothing peculiar to record about this last pregnancy, except that she suffered more severely than usual from nausea, and that the foetal movements were stronger than those she had experienced on former occasions. She is a healthy woman, with no organic disease, and has had no serious illness, except acute rheumatism eighteen months ago.

At my request Dr. Lees inquired whether the parents could explain the abnormality of the foetus, but he was unable to obtain any history of maternal impressions, or of malformations in the family.

The four children are all normally formed, and, as far as the father knew, the fœtuses in the miscarriages were so too. The father, who is a butcher by trade, remarked, when he saw the fœtus, that it looked as if its throat had been cut—a postnatal paternal impression.

The labour was normal and easy from the small size of the fœtus, but the placenta was as large as that found at term, and Dr. Lees informs me that he is absolutely certain that the child took several breaths. As I have not dissected the specimen, I have not had the opportunity of seeing how inflated the lungs are, but for reasons stated below I believe that the respirations took place through the nose and not the mouth.

In order to see whether there was any bony formation to represent the lower jaw, I got Mr. Tyack of University College, Bristol, to take a skiagram for me, which is shown in Fig. 1. From this reproduction it can be seen that there is nothing to represent the inferior maxilla in the mass of tissue between the mouth and the hyomandibular clefts.

The illustration (Fig. 2) shows very well the general appearance of the fœtus, and though the shadows are rather dark, the absence of chin, persistent hyomandibular cleft with the misplaced ears, are well seen. It will be noticed, too, that instead of a horizontal there is a vertical triangular slit for a mouth.

The fœtus is $13\frac{1}{2}$ in. long, and weighs 2 lb. 4 oz. This is about the normal length and weight of a fœtus of 6 months, which age I conclude it to be, from the fact that the only one centre of ossification seen in the tarsus, that of the os calcis. There probably must have been some mistake in the mother's calculations, for had it been a month older a centre of ossification would be visible in the astragalus.

When handed over to me, the mouth of the fœtus was glued up with dried mucus, and the edges almost touched, but handling the specimen to examine the interior of the mouth has somewhat stretched it, and made it into a triangular opening with the apex directed upwards. Inside can be seen the gums of the upper jaw and the hard palate, but I can find no tongue, nor can I pass a probe into the pharynx.

Just above the hyoid bone, which can be distinctly felt and seen, are two small openings, half an inch apart. These openings lead into small sacs, a quarter of an inch deep, and by careful probing I am unable to find any opening, such as is described to occur sometimes, leading into the œsophagus or pharynx.¹ The direction that the probe takes is backwards, downwards, and inwards.

Outside these clefts are the two ears, placed almost horizontally instead of in the usual vertical position. They are both perfectly formed, but no external auditory meatus exists.

¹ Vogel, "Diseases of Children," translated by Raphael, p. 24.

Besides these deformities the foetus has double talipes varus.

Taking the various points of interest in the order above stated:—

1. *The mouth.*—At first sight I was unable to account for the peculiar shape. It certainly was not a hare lip, from its central position, and how to account for its being vertical instead of horizontal

FIG. 1.

FIG. 2.

puzzled me not a little. But Professor Fawcett of University College, Bristol, has suggested to me that it might be explained in the following way. The mouth really is properly formed, but from the want of being stretched, so to speak, by the absence of the inferior maxilla, it has become vertical instead of being pulled out into a horizontal position. This explanation appears to me to be a very feasible one.

I have stated above, that the alveolar arch with the gums and hard palate are properly formed, but that I can find no communication between the cavity of the mouth and the pharynx; nor can I find anything to represent the tongue.

In the normally developing foetus there is till the ninth week a lining of epiblast drawn across the back of the buccal cavity, at which time it begins to disappear, and remains only visible in the full-term foetus as the pillars of the fauces. This tissue does not seem to have split, and ceased to grow, but has continued to block up the passage. I mentioned before that, when the specimen was first brought to me, the sides of the mouth were in close proximity and glued together with dried mucus; and as Dr. Lees assures me that the child certainly breathed several times, I conclude it did so through the nose. The child never cried, and I am unable to pass a probe down its throat through the mouth. Why this dividing membrane has not disappeared I cannot explain, except that the cause of other abnormalities may have acted here as elsewhere.

The absence of the tongue may be accounted for, I think, in this way. First, there was no arch formed by the lower jaw to develop in, and, as the tissues were not expanded, there was no room for it to grow in. Secondly, the tongue is developed from the first three visceral arches—the mandibular, the hyoid, and the thyro-hyoid, but mainly from the first of the three named. And, in consequence, as I shall show later, of a failure of union between the mandibular and the hyoidean, the lower two arches have not been able to supply their part to its formation. These two reasons may be sufficient to account for the absence of tongue, or of its being present in so rudimentary a state, that through the small aperture I am unable to see it without dissection.

2. *The hyomandibular cleft.*—Just above the hyoid bone, which can be seen on the specimen but not in the illustration, as well as distinctly felt, are situated the two openings, as described above.

The distance between these two clefts is half an inch, that between the basis of the ear is seventh-eighths of an inch, while from the tip of the one ear to the other is $1\frac{1}{2}$ in.

It may be as well to describe the formation of the lower jaw and neck from the branchial or visceral arches, for without a clear understanding the malformation cannot be well appreciated.

The branchial or visceral arches are four or, according to some authorities, five in number, and assist in the formation of the lower and lateral portions of the face, the hyoid apparatus, the auricles, and tympano-Eustachian passages, and part of the neck. The first arch is known as the mandibular, and takes part, as already stated, in the formation of the lower jaw and partially of the tongue. It contains the cartilage known as Meckel's. Appearing at a very early stage of foetal life, and remaining visible to the sixth or seventh

month, this cartilage is connected with the development of the inferior maxilla, the malleus, and the internal lateral ligament of the jaw. By the union of the two mandibular arches, the lower jaw and the space below, reaching nearly to the hyoid bones, are completed.

The second arch is known as the hyoid, the space between it and the first being known as the hyomandibular cleft. In this arch are developed the cartilage which subsequently becomes the incus, the tympano-hyal and stylo-hyal centres of ossification (which become later known as the styloid process of the temporal bone), the stylo-hyoid ligament and the lesser wings of the hyoid bone.

The remaining three arches are of less importance in the present case. The third or thyro-hyoid forms from its cartilage the great wings and body of the hyoid bone the thyroid cartilage, and is closely connected with the formation of the larynx.

The other two arches need not be considered, as they take no part in the malformations under observation.

3. *The misplaced ears.*—Both of them are perfectly formed, as regards the pinna, but there is no external auditory meatus. They are placed horizontally, and in about the position that one would expect to find the angle of the lower jaw. The upper border, which in a fully formed child should be on a level with the eye, is barely as high as the lower border of the mouth.

This completes the description of the foetus and its abnormalities, except the talipes, which is such a common condition, though possibly still unsatisfactorily explained, that no reference need be made to it here.

As is usual in these monsters the developmental faults are multiple. I have not been able to find any lengthy description of one similar to that which has come into my hands, and so cannot say whether the persistent clefts are usually associated with agnathia or not. Coats¹ says: Cyclopia sometimes occurs with it, and Dr. Ballantyne informs me, in writing, that one which came under his observation had that condition.

It is not, perhaps, a very hopeful speculation, but one cannot help wondering how far one malformation is connected with another. Why, for instance, should there be a persistent hyomandibular cleft when there is no lower jaw? Or, again, why the fault should be a double one? Why should Meckel's cartilage be absent on both sides? In the commonest developmental failure, hare lip, the single variety is more frequently seen, but in agnathia and anencephalic conditions the absence of bone is always on both sides.

Then, again, what is the cause of these arrests in development? Many reasons have, I know, been assigned, but we do not really seem to have got beyond Tol's statement, that there is some irregular impregnation of the ovum, which, after all, does not land us much further.

¹ "Manual of Pathology," 3rd edition, p. 51.

When I showed the specimen at the meeting of the Bath and Bristol Branch of the British Medical Association, it was suggested that it was not the hyomandibular cleft that was patent, but one lower down, perhaps between the hyoid and the third arch. The hyoidean arch takes part in the formation of the thyroid cartilage, so that, if the cleft was at the lower border of this arch, it would be near the thyroid cartilage and not above the hyoid bone, where the junction of the mandibular and hyoid arches takes place. Further, the ears are generally developed at the dorsal end of the hyomandibular cleft, and they are just outside the cleft in my specimen.

Hamilton¹ gives an illustration of a case, but the ears are placed much higher than in my specimen, and the foetus, being placed in profile, does not show whether there are any persistent clefts or not, or the shape of the mouth.

These cases are so rare that the above description may prove of interest. I may possibly dissect the foetus at a later date, and shall then be able to ascertain for certain the condition of the mouth and the internal parts of the ears.

¹ "Textbook of Pathology," vol. ii. pt. 2, p. 919.