AGLOSSIA CONGENITA

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CINERADIOGRAPHIC FINDINGS

BY

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Aglossia may be either congenital or acquired. Only congenital aglossia is considered in this paper. The principal features of the reported cases are summarized in the table.

In 1932 Rosenthal recorded a case of aglossia, and in a review of the literature found three previously reported cases. He cites de Jussieu who, in 1718, reported a Portuguese girl of 15, who in the floor of her mouth had a tiny wart-like elevation which was muscular and could be moved. She could talk easily and clearly in French except for certain sounds. Chewing was difficult and she used her fingers to move the food in her mouth. Fluids were swallowed easily if she leaned forward and only took small amounts; solids were more difficult. The sense of taste was unimpaired.

Spiller (1816) described a child with a shallow elevation across the floor of the mouth, in the region of the root of the tongue, which contracted on crying. The soft palate was absent except for two lateral rudiments.

Kettner (1907) reported a boy of 4 years who had a small triangular mass where the body of the tongue normally arises. In the floor of the mouth there were also two antero-posterior ridges which could be made almost to fill the space between the teeth. The palate was completely cleft and there was no sign of the band between the tongue and palate which had been divided at birth. There were also severe abnormalities of the hands and feet. The right foot consisted of a heel and a soft ridge replacing the metatarsus. The left foot had only a rudimentary heel. The index and middle fingers of both hands were absent and the other fingers were deformed except for the right fifth finger. The boy pronounced his letters well except for d and t.

Rosenthal’s case was a 3-year-old girl, the third child of healthy parents, with no family history of congenital abnormalities. Pregnancy and delivery had been normal. The face had a retruding lower jaw, a narrow chin and a rudimentary median cleft of the lower lip. The tongue consisted of a small notched median rudiment in the anterior part of the floor of the mouth. A hypertrophic ridge of mucous membrane extended along the lower edge of the left mandible, covering the left parotid duct. A hypertrophic sublingual ridge was capable of some movement and simulated part of the tongue. The tonsils were large and divided into upper and lower lobes. These came together on crying and apparently on swallowing, almost closing off the oral cavity posteriorly. The teeth were irregular and some were missing. The child had no difficulty in talking and in making herself understood, although speech was not clear. There was a moderate and continuous drooling and she found it difficult to swallow solids without the help of the fingers. The right hand was represented by a thumb which consisted of two bones; all the carpals were smaller than normal. A rudimentary fifth metacarpal was present. The wrist, third, fourth and fifth fingers of the left hand were normal but the thumb was underdeveloped and the index finger had a rudimentary second and no terminal phalanx. The right foot had a normal calcaneus and talus, but all the other bones were lacking except for a rudimentary phalanx of the first toe. The left foot was normal. The child walked normally and used her hands well.

Watkin (1925) reported a girl with a narrow face and small lower jaw. The anterior two-thirds of the tongue were missing but there was a hypertrophied sublingual gland. The posterior one-third was rudimentary. The permanent dentition was not complete in either jaw. The viscera, including dextrocardia, were transposed. Speech was good,
the t and th sounds were normal, but she could not 'hiss' as in c, s and x.

Sinclair and McKay (1945) published a post-mortem report of a female child of 4 weeks. The facial features were normal except for an upper median hare-lip. The anterior two-thirds of the tongue was missing and a bifid posterior stump projected from the floor of the mouth. The hard and soft palates were represented by ridges on the medial aspects of the upper jaw. The teeth, larynx and nerves were normal. The first cervical vertebra was compressed and there was a small heart. All the fingers were short and webbed, as were three toes of each foot.

Farrington (1947) reported a man of 23 years with a sharp narrow and receding lower jaw. There was no tongue. At birth a fibrous band had passed between the hard palate and the floor of the mouth but it had been divided and no sign of it remained. The submandibular and sublingual glands were hypertrophied and the sublingual ridges were prominent and appeared hypertrophied. The palate was normal. The deciduous dentition had been normal, but the permanent teeth were irregular and decayed early. He talked fairly normally but pronounced q as in 'to'. Chewing and swallowing were performed satisfactorily, the fingers being used. The sense of taste was normal.

Fitzwilliam (1927) quotes de Jussieu and Kettner’s cases and the following, but without giving references:

‘Blumenbach saw a person without a tongue and whose sense of taste was perfect.

Moncorvo saw a case with complete congenital absence of the tongue.

Filho saw a 10-day-old male with a badly developed jaw and a small elevation in the floor of the mouth representing the tongue. The tonsils and soft palate were missing and ossification of the head defective. The baby could not feed properly.

Griffiths saw a premature male in whom there was a gross deformity of the lower jaw, absent tongue and cleft hard palate.

Maygrier and Haller saw a patient in whom the tongue was represented by a small mass in the floor of the mouth. Other deformities were present.’

Case History

J.W., a boy, aged 5 weeks, was admitted to hospital because he had not been taking his feeds well and had lost weight.

The mother, a primipara, lost a little weight at the beginning of the pregnancy and a mild toxaemia developed during the last two months, but there were no infectious diseases.

Labour was uneventful and resulted in the birth of a full-term baby (5 lb. 12 oz.) who cried lustily. The only abnormality noticed at the time was webbing of the fingers of the left hand (Fig. 1). When breast feeding was started on the third day it was found that the baby did not feed properly and an abnormality of the tongue was then noticed. Bottle feeding was begun but the baby still had difficulty in feeding, requiring an hour to take 3 oz. He thrived and put on weight steadily until the week before admission when he fed poorly and lost some weight.

Examination on admission showed a fairly well nourished baby with a receding lower jaw (Fig. 2). The vestibular mucosa was directly continuous with that of the floor of the mouth as the anterior alveolar margin of the mandibles was depressed. The mucosa of the floor of the mouth was filiform and appeared to consist
of normal tongue mucosa, but there was no tongue (Fig. 3). Laterally this mucosa was raised into two ridges which ran alongside and parallel with the medial borders of the mandibles. These folds probably contained muscle as they moved when the baby cried. In the midline of the posterior part of the floor of the mouth, and visible only on crying, there was a mobile pyramidal projection about \( \frac{1}{2} \) cm. in height. The hard and soft palates, uvula, tonsils and larynx were normal. The mouth was infected with *Oidium albicans*.

The only other abnormality was syndactyly of the left hand. The left thumb was normal, the second and third digits were completely joined, the third and fourth were webbed to the proximal interphalangeal joint and the fourth and fifth were webbed to the distal interphalangeal joint.

![Image](image_url)

**Fig. 3.—J.W., aged 4 months, showing the lateral folds in the floor of the mouth and the depressed alveolar margins of the mandibles anteriorly.**

The baby fed slowly from a bottle using a soft teat with a large hole, but swallowed without choking or coughing and there was no nasal regurgitation. He breathed and cried normally.

Radiographs showed three tooth buds in each mandible and a normal bony structure of the hands and feet. There was no transposition of the viscera or dextrocardia. The oral infection with *Oidium albicans* responded to treatment with gentian violet and the baby started to gain weight.

After discharge from hospital the baby continued to thrive and his rate of feeding improved. At 3 months he was feeding normally. The baby is now 10 months old and has had no further feeding trouble. He has no teeth, but takes most minced and sieved foods when fed with a spoon or cup; he appears to discriminate taste. Food which is stuck to the hard palate is dislodged with the fingers. He says ‘Mum’ and ‘Dad’. The sub-mandibular gland now appears a little hypertrophied and the sublingual ridges are well marked.

**Cineradiographic Investigations**

Cineradiographs were taken at 25 frames per second with an image intensifier. Three films each of four seconds’ duration were obtained.

At the first examination at 6 months of age the child was given a mixture of milk and barium suspension in a bottle supplied by the mother. When the bottle was inverted the mixture dripped slowly from the teat. The teat was adjusted in position in the mouth (Fig. 4). Jaw movements began. When the mandible was lowered the teat distended with barium; the lips formed a seal round the base of the teat; the floor of the mouth was raised above the position of rest and applied to the lower surface of the teat, the upper surface of the teat being apposed to the hard palate. The posterior part of the soft palate was thickened and drawn forwards to meet what seemed to be a normal posterior part of the tongue. No free or anterior portion of the tongue was recognized. A small collection of the contrast medium occupied the space between the end of the teat and the soft palate. The larynx and pharynx were air filled and in communication with the nose; they appeared normal. As the lower jaw was raised the floor of the mouth was lifted higher and the teat was narrowed and displaced forwards; the soft palate reverted to the appearance normally seen at rest, then moved backwards and slightly upwards towards the posterior wall. Palato-glossal closure was maintained by elevating the posterior portion of the tongue against the soft palate. These actions resulted in an increase in the size of the pool of contrast medium in the mouth. Two or three repeated jaw movements seemed to be necessary to obtain a sufficiently large bolus. The tongue and soft palate then separated and the bolus flowed into the pharynx. The back of the tongue was then applied to the soft palate from before backwards and the bolus expressed into the pharynx and swallowed. The movement of the bolus through the pharynx seemed to be entirely normal. As the bolus was expressed from between the posterior part of the tongue and the soft palate, the mandible was lowered to allow the teat to re-fill.

At the second examination at 10 months the child was given barium suspension by teaspoon while sitting up (Fig. 5). Cineradiographs showed that fluid flowed off the spoon into the child’s mouth and over the dorsal surface of the tongue into the valleculae, some spilt over the edge of the epiglottis. When a considerable quantity of the barium had
Fig. 4.—A selected series of cineradiographs from an examination of J.W. at 6 months while being fed with barium sulphate and milk from a bottle.

(a) Position of the floor of the mouth at rest.
(b) Diagram of Fig. 4 (c): (a) floor of the mouth, (b) posterior part of the tongue, (c) soft palate.
(c) The teat is in position in the mouth. The soft palate is thickened and drawn forwards to make contact with what seems to be a normal posterior part of the tongue thus establishing palato-glossal closure in the normal manner. The floor of the mouth is raised and apposed to the lower surface of the teat. A small quantity of barium suspension is held close to the end of the teat and there is a thin line of barium in the line of the palato-glossal closure; at a lower level a further small quantity of barium suspension is held between the posterior pillars and the tongue just above the valleculae in front of the tonsils.
(d) A further quantity of the barium has been drawn into the mouth. The teat is slightly narrowed and the tongue and soft palate are parting.
(e) Palato-glossal closure is no longer maintained. Barium spills downwards.
(f) The posterior part of the tongue is raised against the soft palate: a quantity of barium has been displaced into the pharynx. The nasopharynx is closed by apposition between the soft palate and the posterior pharyngeal wall.
(g) The barium is displaced downwards in the pharynx. The soft palate is down and closely apposed to the dorsal surface of the tongue. Nasopharyngeal closure is maintained by forward movement of the upper posterior pharyngeal wall.
(h) Further displacement of the bolus with bowing forwards of the upper posterior pharyngeal wall.
(i) and (j) The bolus passing through the lower pharynx into the oesophagus.
(k) Further displacement of the bolus downwards. Air is entering the pharynx.
(l) The airway is re-established.
FIG. 5.—The child sitting erect taking barium suspension from a spoon. A series of prints from the cineradiographic examination at 10 months.

(a) Barium taken into the mouth from the bowl of the spoon; the floor of the mouth is raised from the position of rest. Palato-glossal closure maintained between the soft palate and the posterior part of the tongue. The barium in the oesophagus is part of a previous bolus.

(b) The head is being extended. Note the position of the chin and compare it with the position shown in Fig. 5 (a). The soft palate and the tongue have parted to allow the barium to flow down to the level of the tonsils; the floor of the mouth and the posterior part of the tongue have been lowered.

(c) and (d). Further extension of the head. Barium held up at the level of the tonsils.

(e), (f) and (g). Barium descending through the pharynx in the normal manner. The back of the tongue has arched backwards (Fig 5 (f)) and the posterior pharyngeal wall has been brought forwards so that the cavity of the upper pharynx is obliterated and the soft palate apposed to the dorsum of the tongue.

(h) Most of the contents of the pharynx expressed into the oesophagus.

(i) A trace of barium remains upon the upper surface of the epiglottis. Pharyngeal cavity obliterated.
collected in the pharynx the posterior portion of the tongue was elevated against the soft palate, cutting off the barium in the pharynx from the contents of the mouth. This action was then continued as a normal swallowing movement, the contents of the pharynx being expressed into the oesophagus. When the airway was re-established, the child tilted the chin a little upwards and parted the tongue and soft palate; the posterior portion of the tongue was moved forwards, its arched dorsal surface being flattened to form a steep slope allowing a further quantity of barium to flow into the valleculae; the soft palate at this stage was straight and not appreciably elevated. The action described above was then repeated. Two more swallowing movements were needed to take the bulk of one teaspoonful. Before the last swallowing movement the child extended the head to the maximum degree, apparently in an attempt to empty the mouth, and as this movement was taking place there was some elevation of the floor of the mouth towards the hard palate; the changes in the contour of the floor of the mouth suggested that there might be some contraction of the fibres of the tongue musculature. When swallowing movements were concluded the residuum left in the forepart of the mouth was considerably greater than the amount which would be left in the normal child. The films taken in the antero-posterior projection showed that the bolus of barium passed into the pharynx over the midline of the dorsal part of the tongue. Another film in the lateral projection, with the child lying on its back, showed that the method of swallowing was similar to that described in the sitting position, except that the child did not have to extend the chin so far in order to clear the mouth.

In normal babies the tip of the tongue is thrust upwards against the upper gum and the forepart of the tongue is applied to the teat from before backwards, so tending to express the contents of the teat and displace the bolus backwards into the mouth (Ardran and Kemp 1955). In this patient the flow of barium from the teat into the mouth appeared to be brought about by three separate factors. First, by drips from the teat under the influence of gravity. Secondly, by suction when the jaw and the floor of the mouth were depressed and thirdly, by compression of the teat by elevation of the floor of the mouth. The last factor was probably the least effective as it was not accompanied by occlusion of the neck of the teat, since contents of the teat would be more readily displaced into the bottle through the wide open neck of the teat rather than onwards through the much smaller opening at the tip of the teat.

The behaviour of the floor of the mouth at the second examination strongly suggests that there are some fibres of the tongue present in this situation, possibly in the two lateral ridges, which have been described clinically.

**Embryology**

According to Baxter (1953) the floor of the mouth in the early embryo consists of six pharyngeal arches separated by pharyngeal grooves. Ventrally the first and second arches meet their fellows in the midline and at the point of fusion of the first (mandibular) arches a small median swelling, the tuberculum impar, appears before the fourth week. A larger midline swelling, the hypobranchial eminence, appears between the third, fourth and fifth arches.

Embryologically the tongue consists of anterior and posterior parts. The anterior part first appears as the tuberculum impar, but at the fifth week elevations of the first arches arise on each side of it. Later, after the seventh week, they fuse with the tuberculum impar and with each other to form the anterior two-thirds of the tongue.

The posterior one-third is formed by the third arch mesoderm, which grows forward as a V-shaped process from the anterior part of the hypobranchial eminence, burying the second arch. The line of fusion of the anterior and posterior parts is V-shaped and is indicated in the adult tongue by the vallate papillae. The posterior part of the hypobranchial eminence forms the epiglottis.

These swellings from the first and third arches form the mucous membrane, mucous glands, lymphoid and connective tissues. The muscle is formed by three or four occipital myotomes which migrate ventrally bringing with them their own nerve supply, the hypoglossal nerve.

**Discussion**

The embryological formation of the tongue is a complex process and in all these patients it appears to have been interrupted at an early stage. The anterior part of the tongue is absent and the posterior part is represented by a rudiment. In one case the posterior median rudiment was bifid, suggesting that it has been formed from the hypobranchial eminence and not from the tuberculum impar. The epiglottis, which is also formed from the hypobranchial eminence has been present in all cases. The failure of formation of the tongue is, therefore, most commonly due to an agenesia of the first mandibular arch. In every reported case the rudiments of the tongue, when present, were mobile and, therefore, contained innervated muscle. Thus the
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AGLOSSIA CONGENITA

occipital somites, and their nerves, migrate normally, but because of the previous agenesis of the first arch they cannot form the normal musculature. This view is confirmed by Sinclair’s case which was dissected and found to have a normal hypoglossal nerve.

Some malformation apart from the aglossia is always found, and may be either associated with the branchial arches or apparently unrelated.

The face is usually sharp and narrow with a receding chin, the appearance being described as ‘bird-like’. This is due to a hypoplasia of the mandible and a narrow mandibular arch. A median hare-lip occurred in two cases, once of the upper and once of the lower lip; a cleft palate was present in four cases, three times complete and once of the soft palate only. The mandibular teeth, especially the permanent dentition, are usually irregular and of poor quality. An intra-oral band, between the floor of the mouth and the palate, was divided at birth in two cases; this was probably the remains of the bucco-pharyngeal membrane.

The other deformities which have been described mainly affect the digits. Two cases had adactyly and one syndactyly; the present case has syndactyly. Transposition of the viscera with dextrocardia occurred once. The frequency with which aglossia is accompanied by digital deformity is striking; its explanation is obscure. Since the tongue completes its development before the hard palate appears it is unlikely that a single noxious influence can be responsible and it is more likely that the association is determined genetically.

The disability caused by the aglossia is not very great. It was found that the baby had to be bottle-fed, using a long soft teat with a large hole. The reason for this was shown by the radiographic investigation. In normal babies the tip of the tongue is thrust upwards against the upper gum, closing the neck of the teat, then the forepart of the tongue is applied from before backwards compressing the teat against the hard palate, thus expressing the contents of the teat into the mouth.

The baby with aglossia is unable to perform these actions and milk enters the mouth under the influence of gravity, through the large hole in the teat. This is assisted by suction and compression of the teat performed by a raising and lowering of the floor of the mouth. Compression cannot be very effective as there is no closure of the teat and the floor of the mouth cannot be brought into apposition with the hard palate. Three patients showed hypertrophy of the structures in the anterior part of the floor of the mouth and this appears to have started in the present case by the tenth month. This hypertrophy increases the effectiveness with which the floor of the mouth can be raised against the hard palate during the compression movement and thus partially compensates for the aglossia. Once the food has left the mouth the posterior part of the tongue functions normally; the bolus is swallowed in the normal manner which explains why there is no nasal regurgitation, choking or coughing.

The sense of taste was normal in the two adult cases and the present case appears to differentiate tastes. These children are slow in learning to talk, but they later speak without difficulty and are easily understood, although there are always some sounds which are not properly formed.

Summary

A case of aglossia congenita in a baby is reported. Twelve previously reported cases are reviewed. The embryological formation of the tongue is considered. Investigation by cineradiography shows the way in which the baby feeds and swallows. No cause for the deformity has been found. The disability caused by aglossia congenita is not very great after the first three months.

We wish to thank Dr. A. C. Kendall for his permission to publish this case, and for his advice during the preparation.

References