

AGLOSSIA CONGENITA

REPORT OF A CASE OF THE CONDITION COMBINED WITH OTHER
CONGENITAL MALFORMATIONS

ROBERT ROSENTHAL, M.D.

ST. PAUL

Congenital absence of the tongue is extremely rare; only a few cases have been reported in living children. Usually the tongue has been found to be absent in cases of gross underdevelopment or maldevelopment of the first visceral arches, especially in cases of congenital agnathia or micrognathia.¹

To understand the etiology one has to know some facts about the development of the tongue. The tongue has its origin in the pharynx and grows forward into the floor of the mouth. It begins with a swelling on the floor of the pharynx between the first and second visceral arches, the tuberculum impar, which is present before the embryonal age of 4 weeks. The part formed from it is only a small part of the tongue, that just in front of the foramen caecum. The anterior portion develops from the so-called lateral tongue swellings of the first or mandibular arch, from which arch the mandible also develops (the right and left lingual buds appear in the fifth week). These two swellings later fuse in front of and with the tuberculum impar and grow forward to form the greater part of the tongue. This fusion is completed after the seventh week, as in the seventh week the tip of the tongue is still bifid. The posterior part, the radix linguae, originates from a medial swelling, the cupola, at the base of the second and third arches, posteriorly to the tuberculum impar; this part is the one back of the v-shaped row of the papillae circumvallatae. The union of the anterior and posterior parts is later marked by the sulcus terminalis. The muscles are probably derived from the mesenchyma of the floor of the mouth.²

Underdevelopment and total absence of the tongue are due to nondevelopment of one or more of the aforementioned parts of the embryonal tongue. Incomplete fusion is a cause of deformities, the

1. Aschoff: Missbildung der Zunge bei Agnathie, Verhandl. d. deutsch. path. Gesellsch., 1901, p. 176. Swoboda, N.: Micrognathia, Allg. Wien. med. Ztg., 1904, p. 94.

2. Keith, Arthur: Human Embryology and Morphology, ed. 4, London, E. Arnold, 1921. Dodds, G. S.: The Essentials of Human Embryology, New York, John Wiley & Sons, 1929. Shumway, Waldo: Introduction to Vertebrate Embryology, New York, John Wiley & Sons, 1930.

most common of which is the bifid tongue, or lingua bipartita, due to the lack of fusion of the lateral tongue swellings. In Kettner's case a broad band was found at birth running across the mouth between the tongue and the palate, and he expressed the belief that in his case amniotic amputation should be considered.

Therapy is of no avail. Children with this condition usually learn to eat slowly, but later they become quite adept in using whatever rudiments they have in their mouths and, in the case of solid food, helping with the finger to push it along toward the pharynx. Artificial tongues did not prove practicable, even in adults. The speech, which is very poor in most cases during the first few years, may improve considerably when the child learns to use other muscles in substitution for the missing tongue.

After a very careful search of the literature I was able to collect only three cases. Many of the reported cases of "absence of the tongue," even in children, proved to be aglossia due to loss of the tongue during life, usually due to gangrene following smallpox.

The first case is that reported by de Jussieu.³ He described a case in a 15 year old girl who had only a tiny wartlike elevation in place of a tongue; certain rudiments of musculature were present, as some contractions could be felt. The girl talked quite plainly, although the author did not go into detail as to which letters could not be properly pronounced. The sense of taste was not impaired, and the patient was able to chew with some difficulty. In the absence of the tongue she used the finger to push the food between the teeth. She swallowed fluids fairly well, but she had to be careful to take only small amounts at a time, and she had to bend forward. Nothing is said as to whether the child had been able to nurse at the breast. In the more recent literature this seems to be the only case referred to as an authentic case of congenital aglossia.⁴

3. de Jussieu, M.: Observation sur la manière dont une fille sans langue s'acquitte des fonctions qui dépendent de cet organe, Hist. Acad. roy. d. sc., Par. 1718, 1719, mém. 6 to 14. (This article was republished in several European periodicals, the last time as late as 1770.) Meyer, M. W., in his article: Ueber die angeborenen Fehler der Zunge und die dadurch bedingte Hinderung des Saugens (J. f. Kinderkr. 13:328, 1849) stated that the Portuguese physician, Dr. Somarive, published a description of the same case in a special pamphlet.

4. Gould, G. M., and Pyle, W. L.: Anomalies and Curiosities of Medicine, Philadelphia, P. Blakiston's Son & Company, 1897. Birnbaum, R.: A Clinical Manual of Malformations and Congenital Diseases of the Foetus, translated by G. Blacker, London, J. & A. Churchill, 1912. Birnbaum said that Tillmans had reported a case (of congenital absence of the tongue) from the French literature. This is without doubt the case of de Jussieu. Thomas B. Cooley, in Abt, I. A.: Pediatrics, Philadelphia, W. B. Saunders Company, 1925: "Aglossia: There is at least one authentic case of congenital absence of the tongue, that of Jussieu."

The second case appeared about one hundred years later, and was reported by Dr. Spiller, a physician of Pennsylvania, in the early part of the nineteenth century.⁵ This patient showed in place of the tongue a shallow elevation across the mouth in the region of the root of the tongue. When the child cried or when this region was touched a contraction occurred, and this rudiment appeared like a ridge. There was an almost entire absence of the soft palate, except for two lateral rudiments. The infant had considerable trouble in nursing; one could see the cheeks work like a bellows. The fact that the milk frequently regurgitated through the nose was probably due to the defect of the palate.

The third case, which occurred rather recently, was reported by Kettner.⁶ It was perhaps not a case of aglossia in the strictest sense, but the defect was so great that the case can be included in this group. Kettner described a boy who showed practically complete absence of the tongue; there was only a small mass, the size of a quarter, near the pharynx. This mass was triangular and pointed in front, at the site where normally the body of the tongue should start. There were two parallel ridges on the floor of the mouth extending anteroposteriorly, which showed good mobility; they could be stretched so as to almost fill the space between the teeth. They represented the sublingual ridges, and no doubt contained rudiments of the styloglossus and the hyoglossus. They took on the functions of the tongue, pressed the food between the teeth and made a furrow during swallowing to allow fluids to flow toward the pharynx. This child was able to talk, and he could pronounce all the vowels and most of the consonants, especially the ones that do not depend on the tongue for pronunciation. Besides this anomaly there were a complete cleft of the palate and severe anomalies of both feet: of the right foot only the heel and a soft ridge where the metatarsus should be were preserved, and the left foot was entirely absent except for a trace of the heel. There were also anomalies of the hands: both index and middle fingers were absent, and all the other fingers were deformed in some way, with the exception of the right little finger.

I wish to add another case to this small group. The case herein reported is similar to that of Kettner on account of the presence of multiple deformities besides the aglossia.

5. Quoted from Meyer, M. W. (footnote 3, second reference). Meyer presented a complete account of the case. He gives as reference the "Philadelphia Medical Examiner of the year 1816." This is evidently incorrect as this journal did not appear until 1838 and its indexes fail to show such an article by Dr. Spiller.

6. Kettner: Kongenitaler Zungendefekt, Deutsche med. Wchnschr. 23:532, 1907.

REPORT OF A CASE

The patient was a girl, 3 years and 3 months old, whose parents were well. There were no congenital anomalies on either side of the family, and the two older brothers of the patient were perfectly normal. The mother's pregnancy and delivery were normal. The child was unable to nurse at the breast, but had to be fed with a long, soft nipple. The patient was rather small, and a fairly marked retrusion of the lower jaw combined with a narrow chin gave the face a peculiar

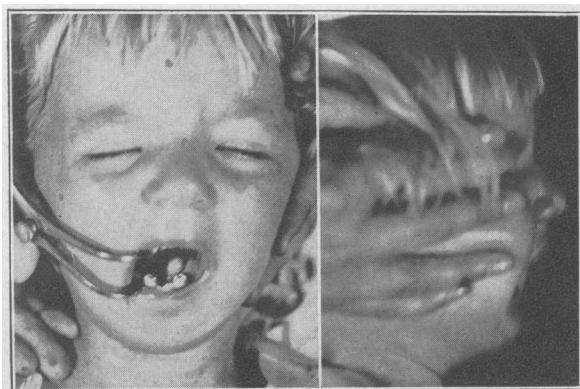


Fig. 1.—On the left, a front view of the patient is shown. Note the hypertrophic left sublingual ridge simulating a rudimentary tongue. The real tongue rudiment is not visible. Note the absence of lower incisors. On the right, a lateral view is shown. Note the retrusion of the lower jaw.

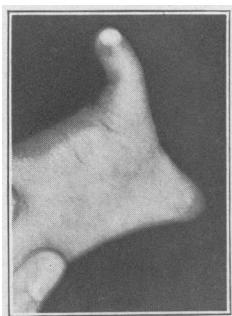


Fig. 2.—Photograph of the right hand.

appearance (fig. 1). The child presented multiple deformities of the mouth and the extremities.

The right hand (fig. 2) had only one finger, which appeared to be the thumb. A roentgenogram revealed that the distal epiphyses of the forearm were normal; the ossification centers of the os magnum and os unciforme were much smaller than normal, as were also those of the semilunar and scaphoid bones. There was a rudimentary fifth metacarpal. Two bones in the thumb represented either a rudimentary metacarpal and a rudimentary first phalanx or two rudimentary phalanges.

The left hand (fig. 3) showed a perfectly normal wrist. The last three fingers were normal; the thumb showed a first phalanx that was broader and shorter than normal and also some underdevelopment of the end-phalanx. The index finger had a rudimentary second phalanx; the end-phalanx was absent.

The right foot (figs. 4 and 5) had a normal ankle; the calcaneus and talus also appeared normal. The other tarsal bones, however, were absent, as were also all of the metatarsal bones. There was only one toe, the large one; in this toe only one bone was present, apparently a rudimentary first phalanx. The left foot was perfectly normal. The child was not incapacitated by the aforementioned deformities; she walked like a perfectly normal child, and used both hands with great dexterity.

The face was deformed, owing to a marked retrusion of the lower jaw, a slight protrusion of the upper jaw (intermaxillary region) and a narrow chin, which gave the face a tendency toward a so-called "bird face" appearance, which is

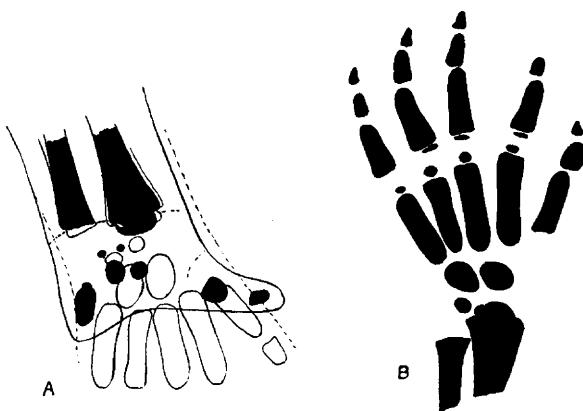


Fig. 3.—*A*, graphic representation of roentgenogram of patient's right hand (in black) superimposed on a drawing of a normal wrist. *B*, graphic representation of roentgenogram of patient's left hand.

usually due to an underdevelopment of the lower jaw (fig. 1). There was a rudimentary cleft in the lower lip (median notch). All of the incisors in the lower jaw were missing; the space between the left and the right half of the mandible was very narrow (fig. 1). A roentgenogram revealed an unerupted tooth in the anterior portion of the mandible, but it was difficult to ascertain whether it was a retained incisor or a permanent canine or bicuspid. The first molars and the permanent upper teeth were plainly visible in the roentgenogram.

The tongue was practically absent. There was only a tiny rudiment where the *musculus genioglossus* normally inserts on the lower jaw, a small notched bud. A fairly large fold extending along the lower edge of the left mandible was formed by a hypertrophic fold of mucous membrane covering the left duct of Steno, a hypertrophic sublingual ridge (fig. 1); it simulated part of the tongue. The fold was capable of certain motions. The tonsils were very large, and were divided into an upper and a lower lobe. On crying and, apparently, on swallowing, the tonsils

approximated and practically closed the posterior part of the floor of the mouth. The epiglottis was not flat, as usual, but folded.

The speech was not very clear, but one could understand the patient quite well; she talked much and seemed to have very little difficulty in articulating the words. The child had some difficulty in the swallowing of solid food, and frequently assisted in this act by pushing the food toward the pharynx with the finger; the food then entered the esophagus without any trouble. The anomaly of the epiglottis did not interfere with the function. There was a moderate continuous dripping of saliva, which was aggravated during colds.

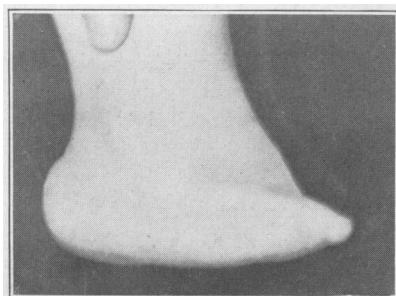


Fig. 4.—Photograph of patient's right foot.

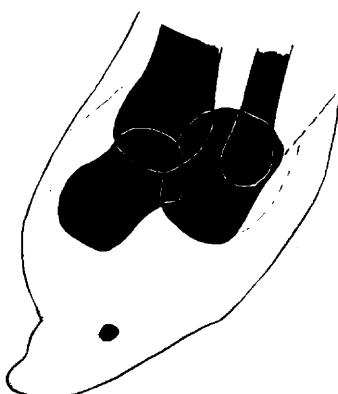


Fig. 5.—Graphic representation of roentgenogram of patient's right foot.

SUMMARY AND COMMENT

In the case reported, there was almost complete absence of the tongue; the notched bud in front apparently represented a rudiment of the lateral tongue swellings. There was no evidence of the part normally formed from the tuberculum impar or of the radix linguae. The non-development of the anterior part of the tongue and the underdevelopment of the mandible occur together, as both originate from the same visceral arch. In cases of agnathia the tongue—at least the anterior part—is always absent. The lower parts of the tonsils help to form a part

of the posterior floor of the mouth. The absence of the lower incisors and the consecutive narrowing of the floor space make it easier for the left hypertrophic sublingual ridge to form part of the floor of the mouth and to aid in the act of mastication, especially as this ridge seems to contain muscle fibers, which make some voluntary motions possible. Teratologically, one has to assume that in the case reported the damage must have occurred at a very early age in order to produce anomalies in so many and in such widely separated organs.