



## Lingual Agenesis: A Case Report and Review of Literature

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### Abstract

Aglossia is a rare condition characterized by the complete absence of the tongue. Its etiology is still unknown. The underlying pathophysiology involves disruption of the development of the lateral lingual swellings and tuberculum impar during the second month of gestation. In this case report, a 26-year old African American female with aglossia presented to the Detroit Mercy Oral Surgery Clinic. The patient presented for extraction of tooth #28, which was located within a fused bony plate at the floor of the mandible. This study presents a case of aglossia, as well as a critical review of aglossia in current literature.

### Introduction

Aglossia is a rare condition characterized by the complete absence of the tongue. The exact etiology of aglossia is still unknown. Possible etiologic factors during embryogenesis include maternal febrile illness, drug ingestion, hypothyroidism, and cytomegalovirus infection. Heat-induced vascular disruption in the fourth embryonic week and chronic villous sampling performed before 10 weeks of amenorrhea, also called the disruptive vascular hypothesis, may be another possible cause of aglossia [1,2]. The underlying pathophysiology involves disruption of the normal embryonic development of the lateral lingual swellings and tuberculum impar during the second month of gestation [3]. Most cases of aglossia are associated with other congenital limb malformations and craniofacial abnormalities, such as hypodactyly, adactyly, cleft palate, Pierre Robin sequence, Hanhart syndrome, Moebius syndrome, and facial nerve palsy. Animal studies have shown that the transcription factor Hand2 plays a role in tongue morphogenesis and the development of the limb and branchial arch [2,4]. Situs inversus and thyroid dysfunction are also described to occur with aglossia. An isolated defect limited to the absence of the tongue alone is extremely rare and only a few cases have been described in current literature. Normal tongue development is vital for many functions, such as suckling, swallowing, chewing, and speech. At birth, the tongue is essential for nourishment and the suckling reflex. Newborns with aglossia seldom survive more than three days [3]. In addition, the tongue is associated with the normal growth and position of the mandible, hyoid bone, and teeth. Due to the lack of muscular pressure of the tongue, the establishment of occlusion and the development of the dental arch shape are altered [5]. Multiple cases in literature have described hypertrophy of the floor of the mouth, exaggerated lip movements, constriction of the oropharynx, and hypertrophy of the uvula [6]. Other orofacial alterations reported are the absence of deciduous mandibular teeth and the crowding of permanent mandibular teeth [7,8]. Mandibular hypoplasia, micrognathia, macrosomia, and Angle's class II malocclusion are also features associated with aglossia [9]. In regards to phonation, most patients have intelligible speech; however, speech impediments are common. The hypertrophic mylohyoid, hyoid bone, and tongue base act as a pseudo-tongue that creates a seal with the posterior palate. The creation of the palatal seal allows for speech and swallowing functions.

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### Materials and Methods

A systematic PubMed and Medline search was conducted using the key words aglossia, lingual agenesis, and aglossia adactyly and aglossia congenita. Age at presentation, gender, race, comorbidities, limb abnormalities, speech deficits, clinical characteristics, rendered treatment, and treatment outcome were collected, tabulated, and subjected to analysis (Table 1). A 26 year old African American female presented to our Oral Surgery clinic for the extraction of tooth #28 (Figure 1). Medical history revealed asthma, sleep apnea, and sickle cell trait. Patient stated that she used a feeding tube to eat until the age of 10 and a tracheal tube until the age of 7 to assist with breathing. No limb abnormalities were reported. Intraorally, the patient presented with lingual agenesis, a fused mandible, and bony plate in the floor of the mouth, uncharacteristic of typical aglossia cases. Her mandibular teeth are displaced lingually and tooth #28 is located in the center of the bony plate. A panoramic radiograph was taken (Figure 2). Tooth #28 showed extensive decay and diagnosed



**Figure 1:** Clinical visualization of tooth #28. Mandibular teeth are lingually tipped. Tooth #28 located at the center of the bony plate of the mandible.



**Figure 2:** Panoramic radiograph showing fused mandible and bony plate at the center of the mandible. Tooth #28 is located at the center of the bony plate.



**Figure 3:** Reflection of the periodontium during the extraction of tooth #28.

with symptomatic apical periodontitis. Patient was anesthetized and tooth #28 was removed using straight elevator (Figure 3). To close the defect created following extraction, a lateral repositioned flap was advanced, a collagen membrane was placed, and secured by four (3-0) Chromic Gut sutures.

## Results

Although aglossia is normally incompatible with life, many cases have been reported and described in literature. Jussieu first described congenital aglossia in 1718, and Rosenthal was the first to describe aglossia's correlation with adactylia in 1932. Our review of literature consisted of 19 case reports of patients with either aglossia or hypoglossia. A 52.6% (10/19) of patients were male and 47.4% (9/19) of patients were female. Most patients in the reviewed literature had some type of limb abnormality or craniofacial malformation. A 47.4% (9/19) of patients presented with limb abnormalities, such as hypodactyly, syndactyly, adactyly, or hypomelia. A 10.5% (2/19) presented with cleft palate and 5.2% (1/19) presented with Moebius Syndrome. Situs inversus was found in 15.8% (3/19) of the cases reviewed. Uncharacteristically, 31.6% (6/19) of patients did not

have an associated limb or craniofacial abnormality. A 36.8% (7/19) of patients suffered from a speech impediment. Hypertension, hypothyroidism, and patent ductus arteriosus were comorbidities associated with singular cases of aglossia. 26.3% (5/19) of patients were of Asian descent and 10.5% (2/19) of patients were of Caucasian descent; however, 63.2% (12/19) of cases did not report the patient's race. Orthopedic distraction osteogenesis was used to treat patients in 21.1% (4/19) of cases, only one of which provided additional speech and hearing therapy. There was no reported treatment in 42.1% (8/19) of cases.

## Discussion

The exact etiology of lingual agenesis is still unknown; however, it is frequently associated with certain limb and craniofacial malformations. In 63.2% of the reviewed cases, patients with aglossia presented with a limb abnormality or craniofacial defect. Similar to the patient presented in our case report, 31.2% of studies did not observe patient limb or craniofacial abnormalities. Our case report also describes mandibular fusion and a bony plate at the floor of the mouth. Characteristically, the reviewed cases reported hypertrophy of the floor of the mouth, acting as a pseudo-tongue to assist with speech and swallowing.

Orofacial alterations due to the absence of the tongue ultimately affect psychological, physical and social development; therefore, professionals in the areas of nutrition, psychology, speech, general dentistry, orthodontics, maxillofacial surgery, and implantology are needed. At birth, the tongue is essential for nourishment and the suckling reflex. Newborns with aglossia seldom survive more than 3 days. Adjusting the way the child feeds allows for proper nourishment and growth for newborns and infants [3].

Orthodontic and surgical treatment are used to correct the malocclusion and orthognathic constriction caused by aglossia. A 21.1% of the reviewed cases utilized orthopedic extraction osteogenesis to expand the dental arch. Rapid maxillary expansion, mandibular expansion, and distraction osteogenesis, followed by orthodontic treatment early in life have been successfully utilized to align crowded teeth, provide functional occlusion, and produce proper esthetics. In some cases, however, these appliances were unsuccessful because they altered the patient's naturally modified swallowing and chewing functions. Because older patients have shown functional adaptation to most tongue functions, dental arch expansion and surgical reconstruction of the tongue is not a preferred treatment [3,7].

Delayed and slurred speeches were the most common problems during the development of patients with aglossia. Although speech therapy was conducted in only one of the reviewed cases, speech therapy is the suggested treatment option. In patients with aglossia, the hyoid bone and the mandible strongly correlate with the movements of the pseudo-tongue during speech. Due to the restricted range of motion of the pseudo-tongue, the mylohyoid and tongue base are more dependent on the mandible and hyoid for range of motion during speech. Exaggerated movements of the hyoid and mandible provide compensatory movements for speech in the absence of a tongue. Over time, adjusting to the altered range of motion through speech therapy can allow for speech that is more comprehensible [2,6-22].

## Conclusion

The complete absence of the tongue is an uncommon occurrence.

Table 1: Data Collection.

Author	Age (Years)	Sex & Race	Comorbidities	Limb Abnormalities	Speech Deficit	Clinical Characteristics	Treatment Rendered	Treatment Outcome
Ardran Fulford [10]	5 weeks	M	None	Syndactyly	-	1. Receded lower jaw 2. Filiform mucosa at floor of mouth 3. Mobile pyramidal projection	Cineradiographic Investigation	Suckling and feeding difficulties
Ardran Beckett [11]	8	M	None	Syndactyly	No	1. No tongue clinically 2. Posterior portion of tongue present	Cineradiographic Investigation	1. Mobile floor of mouth 2. Normal speech and swallowing
Nevin [12]	31	M	1.Hypertension	1. Adactylia 2. Hemimelia 3. Syndactyly	No	1. Abnormally shaped tongue 1/3 <sup>rd</sup> of normal size	-	-
Nevin [12]	21	M	None	1. Adactylia 2. Syndactyly	No	1. Small abnormal tongue 2. Retroclined lower incisors	-	-
Mishima [13]	39 weeks	M	None	1. Hypodactylia 2. Syndactylia	-	1. Hypoplastic tongue 2. Abnormal gingiva 3. Lower lip attached to mandibular gingiva 4. Micrognathia	-	-
Jang [14]	12 days	F	1. Situs Inversus	None	-	1. Absence of tongue 2. Micrognathia	Two year follow up	1. Mild physical underdevelopment 2. Phonation disturbance
Grippaudo [15]	9	M Asian	1.Scoliosis	1. Hypodactylia 2. Adactylia 3. Hemimelia	No	1. Retrusive mandible 2. Edentulous 3. TMJ abnormal 4. High vaulted palate 5. Absence of tongue 6. Fibromucous tissue connecting FOM with palate 7. Micrognathia	-	
Amor [16]	6 months	F Asian	1. Dextrocardia 2. Situs Inversus 3. Patent ductus arteriosis	None	-	1. Microglossia 2. Choanal stenosis 3. Hypoplastic epiglottis 4. Prominent FOM 5. Micrognathia	Follow up	1. Normal growth development 2. Mild gross motor delay
Cho [17]	6	F Asian	1. Situs Inversus 2. Dextrocardia	None	Yes	1. Mandibular retrusion and constriction 2. Complete aglossia 3. Congenitally missing teeth	1. Orthopedic management 2. Bonded hyrax	1. Obtained occlusal harmony 2. No relapse of intercanine width
Kantaputra [18]	13	F Asian	1. Subclinical Hypothyroidism	None	Yes	1. Absent tongue 2. Persistent buccopharyngeal membrane 3. Microsomia 4. Congenitally absent mandibular incisors 5. Collapse of mandibular arch 6. Hypertrophy of FOM 7. High caries	Thyroid function tests	1. Slightly elevated thyroid stimulating hormone 2. Normal free T4 levels
Kumar [5]	4	F	None	None	Yes	1. Congenital absence of tongue 2. Hollow space 3. Raised folds of mucous membranes 4. Constricted pharynx 5. Mild lower anterior crowding	Pathological investigations	None
Salles [3]	14	F White	None	None	Yes	1. Hypertrophic uvula 2. Absent tongue 3. Constricted mandible 4. High palatal vault 5. Absent mandibular incisors 6. Hypertrophic FOM 7. Enhanced taste perception 8. Micrognathia	1. 30mm symphyseal osteogenic distraction using buccal intraoral distractor 2. Speech and hearing therapy	1. Improved speech 2. Improved occlusion and esthetics

Rasool [9]	6	M Asian	None	None	Yes	1. Tongue replaced by small mucous membrane projection near floor of oropharyngeal isthmus 2. Hypoplastic mandible 3. Class 2 occlusion	-	-
Gathwala [19]	Newborn	M	1. Cleft Palate 2. Supernumerary nipples 3. Micropenis 4. Cryptorchidism	Hypodactyly	-	1. Rudimentary tongue 2. Membranous structure with narrow opening communicating to the pharynx 3. Microretrognathia	-	-
Gupta [2]	28	M Asian	None	None	No	1. Missing mandibular incisors 2. Absent tongue 3. Smooth FOM mucosa 4. Hypertrophic uvula & FOM 5. Absent palatoglossus arches 6. Collapsed mandibular arch 7. Narrow palate 8. Microstomia	1.Extraction and enucleation of radicular cyst 2. Transverse maxillary arch expansion 3. Future anterior transverse expansion and mandibular ridge augmentation 4. Future implant prosthetic rehabilitation	-
Cappellette [7]	15	F	1. Glossopalatine ankyloses 2. Difficulty breathing 3. Micrognathia 4. Cleft palate 5. Bifid uvula	Middle finger hypomelia	Yes	1. Persistent buccopharyngeal membrane 2. Maxillary and mandibular hypoplasia 3. Underdeveloped anterior 2/3rds of tongue 4. Abnormal swallow 5. Nasal cavity hypoplasia 6. Maxillary cavity hyperplasia	1. Orthodontic intervention 2. 3 rapid maxillary expanders 3. Mandibular distraction osteogenesis	1. Restored form and function of maxilla and mandible 2. Able to speak, chew, and swallow 3. Incomplete orthodontic phase
McMicken [20]	44	F White	None	None	No	1. Wart-like rudimentary tongue 2. Micrognathia 3. No geniohyoid muscle 4. Hypertrophy of mylohyoid and tongue base	Electropalatography	Able to activate electrodes in anterior and posterior palate during swallowing and speech similar to normal
Bommarito [21]	8	F	None	None	Yes	1. Absent tongue 2. Deep hard palate 3. Atretic maxillary and mandibular dental arch 4. Oblong uvula 5. Absence of mandibular lateral incisors	-	-
Kaissi [22]	9 months	M	1. Moebius Syndrome	Adactylia	-	1. Left facial nerve palsy 2. Bilateral abducent nerve palsy 3. Absent tongue	-	-

When managing aglossia patients, the intricate complications associated with the absence of a tongue call for a multidisciplinary approach. Early management of lingual agenesis is crucial for the development of feeding, swallowing, and speech. Treatment of aglossia and its accompanying characteristics later in life must be elective due to possible disruption of the adaptive function of the pseudo-tongue.

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