ORIGINAL ARTICLE

AGLOSSIA: A RARE CASE REPORT AND REVIEW

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ABSTRACT

Aglossia is a rare malformation resulting in the partial development or complete absence of the tongue. It is commonly associated with craniofacial and limb defects (Oromandibular Limb Hypogenesis Syndrome).The etiology is unknown and various theories have been proposed for this. The objective of presenting the case report and literature review is to discuss the clinical presentation, causative factors and possible management strategies of this rare malformation.

Key Words: *Aglossia, Microglossia and Hypoglossia.*

INTRODUCTION

The term Aglossia implies "the absence of tongue". It is a rare malformation attributed to the failed embryogenesis of developmental swellings during fourth to eighth intrauterine weeks.[1] Microglossia and Hypoglossia refer to the presence of "a very small or rudimentary tongue".[2] However, all the three terms have been used in co-occurrence in literature.[3]

Most cases of aglossia in literature have been described in correlation with other congenital or developmental defects. Limb defects are the most common, particularly Adactylia or Hypodactylia in hands or feet or both (with or without syndactyly).[4] Aglossia-Adactylia sequence has been mentioned in literature.[5] In 1971; Hall classified these entities under the title of Oromandibular Limb Hypogenesis Syndrome, which has a very low incidence.[6] (Table A). Syndromes associated with Aglossia include Moebius, Hanhart, Pierre Robin, Charlie M, Pfaundler-Hurler, Orofacial-digital, Amniotic band and Glossopalantine-Ankylosis Syndrome.Mental stature is usually normal in such cases, but a case in conjunction with mental retardation has also been reported.[7] Deafness has also been highlighted in some cases.[8,9] Situs Inversus and more recently Hypothyroidism has also been observed with this malformation.[10,11]

Cranio-facial abnormalities reported in combination with this defect include cleft lip and palate, defects of the eye-lid, asymmetry of the face and cranial-nerve palsies.[12] Aglossia appears to be congenital and sporadic in nature but it may be acquired due to surgery needed in case of malignancy. Till date, no genetic mutations or chromosomal aberrations leading to aglossia have been identified.[13] Isolated aglossia, without any systemic deficiency, as reported here, is a rare entity, which has only been mentioned by a few. A review of the previous cases of this defect indicates that the most common clinical findings in such patients include maxillo-mandibular hypoplasia, microstomia, micrognathia, hypertrophy of salivary glands and mylohyoid muscle, palatal-grooving and a conspious uvula. Oligodontia is a frequent finding and most cases presented with congenitally missing mandibular incisors.[13]

As per the classification an noted by Hall, this

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case report of patient falls in the category Type-IB. (Table A)

TABLE A: CLASSIFICATION OF OROMANDIBULAR LIMB HYPO GENESIS SYNDROME

Type I	A:	Hypoglossia
	B:	Aglossia
Type II	A:	Hypoglossia-hypodactylia
	B:	Hypoglossia-hypomelia
	C:	Hypoglossia-hypodactylomelia
Type III	A:	Glossopalantine ankylosis (Ankylossum superius syndrome)
	B:	With hypoglossia
	C:	With hypoglossia-hypodactylia
	D:	With hypoglossia-hypomelia
	E:	With hypoglossia-hypodactylomelia
Type IV	A:	Introral bands and fusion
	B:	With hypoglossia
	C:	With hypoglossia-hypodactylia
	D:	With hypoglossia-hypomelia
	E:	With hypoglossia-hypodactylomelia
Type V	A:	The hanhart syndrome
	B:	Charlie M syndrome
	C:	Pierre- robin syndrome
	D:	Moebius syndrome
	E:	Amniotic band syndrome

CASE REPORT

A 40 -year old female patient reported to the Prosthodontics Department, Islamabad Dental Hospital, with the presenting complaint of ill-fitting removable partial dentures, owing to which she had difficulty in mastication. She was well-oriented and spoke normally. No mental deficiency was observed.

External physical appearance was normal and no deformity was observed. Her complexion was fair. She had very thin lips and a slightly prominent chin. Intra-oral examination revealed the hollow space, which would otherwise have been occupied by the tongue .The absent tongue was replaced by a small, inverse U-shaped mucous membrane projection on the floor of the mouth, near the oropharyngeal isthmus (Fig 1). Upon examination, it was found to be hard, firm and slightly mobile.

The palate was high-arched, constricted and V-shaped with medial grooving (Fig 2). The uvula was conspious. Mandibular and maxillary alveolar ridges were severely resorbed. In case of mandibular ridge, the resorption at the posterior section was greater.

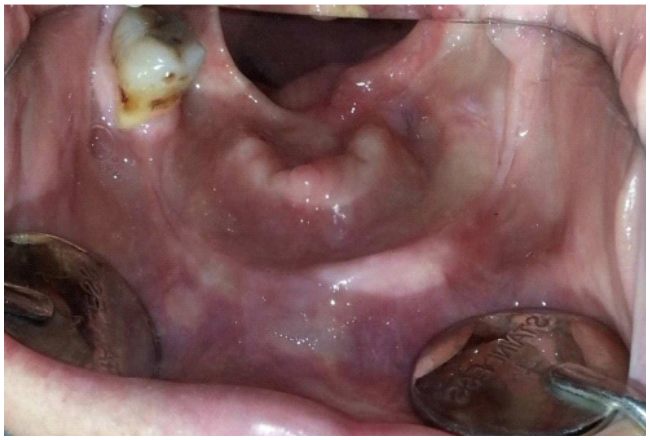

Fig 1: Intra-oral view of the floor of the mouth (Lingual view) 94x63mm

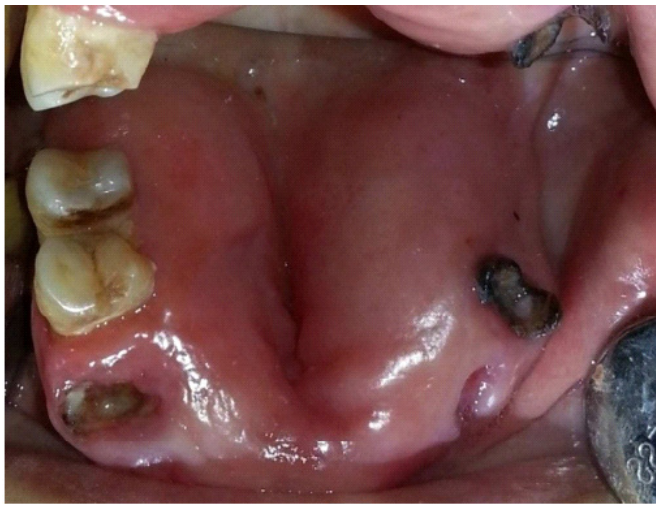

Fig 2: Intra -oral view, Maxillary arch with palatal defect (Palatal view), 74x57mm

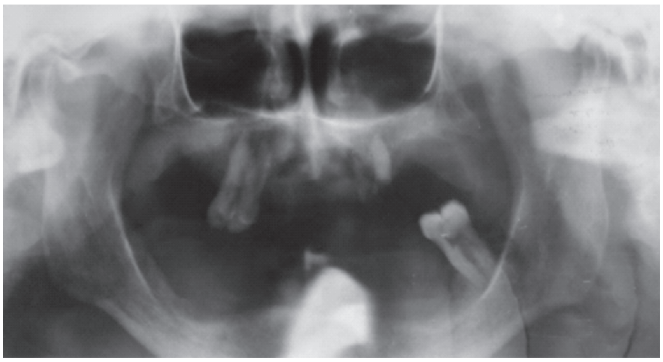

Fig 3: Radiographic view 132x72mm

There was no deviation and the opening and closing of mouth was within the normal range. No oro-antral communication was apparent.

History was taken to evaluate her adaptability and efficacy related to functions of the tongue. No history of trauma, intake of drugs and / or radiation exposure during pregnancy was reported by the patient. The family history was not relevant. She had a normal

birth but she gave an account of the difficulties in swallowing and sucking that she had to face later. She had been wearing dentures since twelve years of age and maintained that she had lost her tongue during childhood. According to the patient, during her early childhood, she had a disease in which tiny nodules appeared on her facial skin and tongue, after which the tongue shed. For as long as she could remember, she has been living with this malformation. She was unable to speak till six years of age. However, at the present age, her speech was surprisingly clear and understandable and slurred only during the pronunciation of the lingual consonants. The aglossia was probably compensated by the smooth floor of the mouth and the slight elevation of the floor during swallowing and speech. Taste perception, according to the patient, was normal. No associated abnormalities of the limbs or nails were noted. No hearing defects were present and there were no signs of facial palsy.

Intra oral examination revealed the presence of Maxillary right first and second premolars and Mandibular left second molar. Broken dental roots of left and right maxillary canine and left maxillary premolar were present. The teeth present, according to FDI notation were 14, 15 and 37 and broken down roots (BDRs) were 13, 23 and 24.

Panoramic radiograph was obtained. No impacted teeth or bony cleft in the palate was visible in the radiograph (Fig 3). It exhibited signs of poor periodontal support and reduced alveolar bone height. The patient was not willing for any treatment for aglossia so only denture treatment was advised and replaced. Informed Verbal Consent was taken from the patient regarding this case report and photographs.

DISCUSSION

The most mobile and adept organ of the human body is the tongue. It plays an important role in suckling, swallowing, mastication and speech. It contributes in the development and the upcoming shape of the jaws as well as establishment of occlusion and prevention of the malocclusion of dentition. It also has the special property of taste sensation associated to it.[12]

During the fourth to eighth intrauterine weeks, the anterior portion of the tongue is formed by the two lateral lingual swellings and one medial swelling; "the tuberculum impar." All of these are derivatives of the first pharyngeal arch. The appearance of a second medial swelling known as the "Copula or hypobranchial eminence" follows. It is the derivative of the second, third and fourth pharyngeal arch. Innervation of the tongue can be explained on the basis of its development; the body of the tongue is supplied by the Trigeminal nerve (CN V)-Mandibular branch whereas the root is supplied by the Glossopharyngeal (CN IX) and Vagus (CN X) nerves. The Facial Nerve (CN VII).-Chorda tympani branch is responsible for the taste innervation.[15] Failed embryogenesis during this time period contributes to aglossia. Musculature of tongue, innervated by the Hypoglossal Nerve (CN XII), except for the palatoglossus (which is supplied by the Vagus nerve) is formed by the Occipital somites. The Occipital somites migrate normally, but because of agenesis of first arch, they are unable to form musculature in cases of aglossia.[13,14]

The etiology of aglossia is unknown. Internal and external factors during fetal development may play a role in the pathogenesis of this malformation. Various theories have been proposed. One theory suggests intake of drugs such as Meclizine, Chlorpromazine and Benzamine hydrochloride[14] Another proposed mechanism of fetal insult is radiation exposure during the first month of pregnancy. Membranous strands may be produced by the amnion rupture during pregnancy which may adhere to structures and cause constriction. Vascular disruption of Stapedial artery, which supplies the first pharyngeal arch, may be responsible for this defect. Genetic theories of multifactorial inheritance are suggested on the basis of two incidents of consanguinity.[2] According to a study, Neural crest derivatives have expressed a transcription factor which has been found to play a role in tongue morphogenesis.[13] Cytomegalovirus has also been implicated as an etiological factor.[13]

The very first case of congenital lingual hypoglossia, reported in literature dates back to 1719 by de Jussieu.[3] Suckling reflex is one of the primary modes of nourishment for infants. The Portuguese mother mentioned in the de Jussieu report, that she used to enlarge the opening of the nursing bottle and milk was poured directly into the infant's throat.[3] The earliest association of tongue and limb anomalies, occurring in conjunction, was reported by Kettner in 1907.[3] A true case of Aglossia-Adactylia was first documented by Rosenthal in 1932.[3] Isolated Congenital Aglossia was reported by Kumar et al in a four-year old Indian female child.[7] Rasool et al also reported a similar finding in a six-year old Indian male child.[12] The earliest case of Isolated Aglossia in an adult was reported in 1949 in Washington; a 22-year old Chinese man reported with the complaint of pain in lower anterior teeth. Upon clinical examination, no tongue or any rudimentary structure resembling a tongue was found in the oral cavity.[16]

Gradual hypertrophy of the muscles of floor of mouth is a common finding in most cases of aglossia, especially of the Mylohyoid muscle, possibly due to constant stimulation during swallowing. The hypertrophy helps in compressing tissues against hard palate during swallowing.[13,14] The hypertrophied uvula, on the other

hand, helps in closing the oropharyngeal opening. In this way, the articulation of nasal sounds is made possible as air is forced through the nasal passage.[13] The medial palatal grooving is possibly due to the overgrowth of the lateral palatine ridges. The movement of the developing tongue plays a role in causing elevation of the palatal shelves from vertical to horizontal position during secondary palate development, in the eighth intrauterine week. Therefore, the absence of tongue, affects this change of position.[13]

Another fact to be taken into consideration is that mandible is the derivative of the same pharyngeal arch. The Malocclusion of teeth and mandibular hypoplasia is due to the lack of lingual muscular stimulus.[14] Speech development may be delayed and it is slurred (particularly- the lingual consonants).[13] However, in some documented cases, speech defects are surprisingly relatively minor. Enlarged sublingual mucosa is a common finding in such cases and the muscles of the floor of the mouth exhibit immense adaptability in such cases.[14] The susceptibility towards caries and periodontitis can be attributed to absence of cleansing action of the tongue.[13]

Taste is a subjective sensation and taste perceptions in aglossia patients have been documented to be intact.[18] This can be attributed to the fact that taste buds are present not only on the tongue but also on the floor of mouth, epiglottis, soft palate and mucous membrane projection which is actually a remnant of embryonic buccopharyngeal membrane. [14]

Possible treatments of aglossia sequelae have been discussed in literature. Borez et al used a palatal expander for mandibular expansion but the results were unsuccessful. The patient was reported to have lost the ability of phonation and swallowing.[17] It was assumed that palatal expanders are responsible for blocking the elevation of the hypertrophic floor of the mouth, thereby making both swallowing and speech difficult.[3] Surgical treatment to build up the tip of the tongue has been suggested in literature, but later it was maintained that surgical reconstruction of tongue is not required as mastication, swallowing and taste sensations are usually intact, and improvement of speech has no link with surgical intervention.[8]

A multidisciplinary approach is needed for adequate function and esthetic rehabilitation of the patient. In early stages, speech therapy should be employed to correct the delayed and slurred speech. Orthodontic, orthopedic and surgical measures are required to correct malocclusion and jaw alignment. Modified palatal expanders often worsen the situation.[12] Distraction Osteogenesis following symphyseal osteotomy has also been suggested.[3] A better alternative for mandibular expansion is the use of rapid prototyping models for making custom made tooth and/ or bone-borne distractors. Distraction vectors are then carefully determined.[3] Prosthetic rehabilitation should be considered with the option of fixed or removable prosthesis. Implant-supported prosthesis may be considered if sufficient bone height and width is present. Otherwise in most cases ridge augmentation with bone grafts may be needed accounting to severe resorption.[12] Difficulties in swallowing and pooling of liquids occurs in the floor of mouth due to the absent tongue. Tongue prosthesis has been advocated in literature in 1972, by Moore. This type of prosthesis provides the patient with a foundation for directing food and thereby assisting in swallowing, as well as speech.[20] Prosthodontic treatment plan can be modified according to the patient's needs. It can be fabricated in conjuction with a mandibular partial denture in case of a dentate patient. Tentative contours are developed in wax and retentive framework is extended into the defect. Mandibular two-piece partial denture tongue prosthesis has also been described. One component is based in acrylic and the other in silicone. The rubber tip of silicon can be depressed and helps in maintaining posterior contacts. In edentulous patients; a complete denture in conjunction with the tongue cap is advised. In cases of severe ridge resorption, palatal tongue prosthesis may need to be considered.[19]

On January 14th 2013, two famous cases of the very rare and unusual Isolated Congenital Aglossia were presented in a symposium held in Brazil. Theoretically these patients should have had a very brief life expectancy but in both cases the patients survived on account of the mothers instinct in which the opening of the nursing bottle was enlarged and milk was directly inserted into the infant's throat. Auristela Vena de Silva reported to Salles in the year 1996 at the age of five and now she is 24 years old.[3] For the first time, a person with this malformation had its sequelae treated. The multidisciplinary approach was opted with the collaboration of departments of Oral and Maxillofacial Surgery, Prosthodontics, Orthodontics, Speech Therapy, Radiology and Nutritional Rehabilitation to treat the anatomic, functional, esthetic, nutritional and psychological difficulties. The girl underwent three surgeries in the year 2001, 2002 and 2003 respectively, in which a 30mm expansion was performed in three stages of 10mm.[3] She underwent orthodontic treatment for six years along with speech therapy. The other case is of a girl named Kelly Rogers in California who was referred to Mcmicken in 1986 by an ENT specialist.[18] But their cases differ in one aspect: the patient of Salles underwent surgery to improve her condition where as Mcmicken's patient did not undergo any surgical intervention as Mcmicken thought it could hamper with speech.[18] Both of them have managed to cope with this condition.

Aglossia is a rare and unusual defect. A multidisciplinary attitude for proper functional and esthetic rehabilitation is required to treat its complications, along with sustained care and monitoring.

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5 Ayesha Aslam:	Editing of the article.
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