Isolated aglossia congenita: A rare case of oromandibular limb hypogenesis syndrome type I B

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ABSTRACT
Aglossia congenita (AC), congenital total absence of the tongue, is a very rare midline developmental anomaly, hypothesized to be associated with vascular disruption between the fourth and eighth week of gestation. It was classified by Hall (1971) as part of oromandibular limb hypogenesis syndrome (OLHS) type I B. Most of the cases reported with OLHS are actually hypoglossia with limb abnormalities whereas isolated aglossia is an extremely rare entity. A case of isolated AC is presented in a 28-year-old Indian male. He had long narrow face, tapering chin, low set ears, and microstomia. Intraorally, he had narrow palatal vault, constricted oropharyngeal isthmus, oligodontia, and maxillo-mandibular hypoplasia. Interestingly, the patient showed a median palatal groove, which has not been reported before. He also had an unusual acquired adaptive mechanism to compensate for aglossia. This report presents the manifestations of this rare syndrome, its complications, differential diagnosis, and rehabilitation strategies.

Key words: Adaptation, anomaly, developmental, palate, rehabilitation, tongue

INTRODUCTION
Aglossia congenita (AC), congenital absence of the tongue, is a rare and unusual midline developmental anomaly usually associated with craniofacial and limb defects and has overlapping features with other syndromes. All these syndromes are now believed to belong to a family of oromandibular limb hypogenesis syndrome (OLHS). The incidence of OLHS is very low (1/175,000 live births) and most cases are actually hypoglossia rather than true aglossia.[1] Isolated aglossia without limb defects is even more rare. This is a rare case of isolated AC, unusual median palatal grooving, and remarkable functional adaptability in a young Indian male patient.

CASE REPORT
A 28-year-old Indian male reported with the chief complaint of missing lower anterior teeth and wanted prosthetic replacement for the same. He was well oriented. He spoke normally and clearly. He gave a history of missing deciduous mandibular incisors and inability to speak until 5 years of age. He was born from a non-consanguineous marriage, and his parents and three siblings were normal with no craniofacial and skeletal abnormalities. The birth was normal but postnatal period was associated with sucking and feeding problems. The mother gave a history of intake of some medication during pregnancy in the first trimester.

He was of medium height with a long narrow face, pointy tapering chin, saddle nose, broad nasal alae, low set ears, fused eyebrows, thin lips, and microstomia. There was no facial palsy, hearing defects or difficulty in swallowing solids and liquids. The taste perception was not compromised. There were no abnormalities of limbs or nails or other skeletal defects.

Intraoral examination revealed a smooth mucosa of the floor of the mouth and complete absence of tongue. The uvula was long, hypertrophic, and projected into the pharyngeal space. The oro-pharyngeal isthmus was constricted with absence of the palatoglossal arches [Figure 1]. The permanent mandibular incisors (32,31,41,42), maxillary lateral incisors (12,22), second premolars (15,25,35,45), and mandibular third molars (38,48) were missing. The mandibular arch was narrow and collapsed anteriorly. The palate was also narrow and constricted with an unusual median groove [Figure 2]. There
was no oronasal/oroantral communication. The periodontal status was poor and he had fixed prosthesis in some quadrants.

Interestingly, any attempt to perform functions related to the tongue would cause the mucosa of the floor of the mouth to balloon out mimicking the absent tongue [Figure 3]. The patient showed excellent adaptability and could perform all the functions of tongue. He had not undergone any specialist speech therapy but had taught himself to speak clearly with repeated practice and personal effort (even lingual consonants). He was employed and faced no difficulty in social adjustment.

The radiographs did not show any impacted teeth or evidence of bony cleft in the palate. There was inter‑dental bone loss and radicular cyst in association with left mandibular first molar (36) [Figure 4]. The pharyngeal air space appeared narrow and the gonial angle was increased. The anterior mandibular alveolus was resorbed and lingually collapsed [Figure 5]. The chest radiograph, electrocardiogram (ECG), and abdominal ultrasound were normal. Routine hematological and thyroid function tests were normal. Karyotype was of normal male (46, XY).

Multidisciplinary approach towards treatment was adopted. The faulty prosthesis was removed. Tooth 36 was extracted and the associated radicular cyst was enucleated. Teeth 18, 17, 16, 24, 34, and 44 were endodontically treated. The patient is undergoing transverse maxillary arch expansion at present to be followed by anterior mandibular transverse expansion and mandibular ridge augmentation with autologous rib graft. Implant supported prosthetic rehabilitation is being considered.

DISCUSSION

AC was first described in the eighteenth century by de Jussieu (1718/1719). The association of aglossia with anomalies of limbs was noted by Kettner (1907) and the first report of Aglossia – Adactylia syndrome was reported by Rosenthal (1932). Aglossia/hypoglossia has been reported with numerous other developmental abnormalities and systemic
manifestations, the most common being craniofacial and limb defects [Table 1].[1-4]

Hall’s classification of OLHS (1971), dividing the spectrum of manifestations into five types, is widely accepted [Table 2].[5]

AC associated with microstomia, oligodontia, median palatal groove, and maxillo-mandibular hypoplasia without any limb defects was seen in this case. According to Hall’s classification, this case would represent OLHS type I B.[6]

Embryologically, aglossia could be explained by the failure of development of the two lingual swellings and the tuberculum impar but the cause for this is not known.[8] Several theories have been proposed like role of intrauterine factors and membranous strands produced by rupture of amnion in early pregnancy causing constriction or amputation of limbs and oral anomalies due to ingestion of these strands during oral development.[11] Recently, heart and neural crest derivatives expressed protein 2 (Hand 2), a transcription factor has been found to play a role in tongue morphogenesis and limb and branchial arch development in experimental animal studies and may be associated with OLHS.[9] Most of the cases are sporadic but a few intra-familial cases led to the hypothesis of mutation in an autosomal recessive gene. No genetic mutations or chromosomal abnormalities have been identified till date. It does not have any sex or racial predilection. Maternal febrile illness, hypothyroidism, and cytomegalovirus infection have also been implicated as etiological factors. Teratogenic etiology due to ingestion of drugs and herbs (Perovskia abrotanoidis) by mother during gestation could play a role and should be avoided.[11] Drugs ingested by the mother may be an etiological factor in this case although the type of drug could not be determined. It is now believed to be caused by heat-induced vascular disruption around the fourth embryonic week. Chorionic villous sampling when performed before 10 weeks of amenorrhea has been associated with this disorder giving further support to the disruptive vascular hypothesis.[7]

Aglossia may be associated with a number of syndromes, some of which are part of OLHS and which have to be excluded before a final diagnosis. Moebius syndrome would also have chest wall abnormalities and paralysis of VI and VII cranial nerves, while Hanhart syndrome would be associated with hearing loss. Glossopalatine ankylosis syndrome would also have cleft palate and an intraoral band attaching tongue to palate or maxillary alveolar ridge. Splenogonadal fusion syndrome would be associated with spleen that is abnormally connected to gonads along with cardiac defects, pulmonary hypoplasia, and anorectal atresia. Charlie M syndrome would typically be also associated with urinary obstruction, pulmonary hypoplasia, and cleft palate. Otocephaly is a lethal condition associated with agnathia, synotia, and holoprosencephaly. Amniotic band syndrome would be associated with craniofacial clefts and clubfoot deformities. The diagnostic work up would, therefore, include a thorough extra- and intra-oral examination along with referral to ENT, Medicine, Cardiology, Urology, and Gastroenterology departments. Cranial nerve examination, taste testing, and audiometry should be carried out to rule out any neurological functional defects. An ultrasound of abdomen may be followed by endoscopy if any defects are detected. ECG may be advised to rule out cardiac involvement. Radiographic examination should include the jaws (Orthopantomograph (OPG), lateral cephalogram), chest, and limbs. Three-dimensional imaging (Cone beam computed tomography (CBCT)) can be used to better visualize the craniofacial skeletal defects. Thyroid function tests may be done to rule out hypothyroidism.

The tongue plays an important role in suckling, speech, mastication, swallowing, taste perception, and development of jaws and occlusion. It also affects jaw development, facial aesthetics, and hence psychological, sexual, and social development. AC is associated with difficulty in performing these functions, especially suckling after birth, and has mostly resulted in death of the newborn within 3 days.[9] Taste perceptions are not severely compromised, probably due to presence of taste buds in the mucosa of the floor of the mouth.[10] Speech development is delayed and slurred, and lingual consonants are most affected. In this case, the patient’s speech was near normal and gave no indication that there was aglossia. This remarkable adaptation is made possible by the hypertrophy of the muscles of the floor of the mouth, especially the mylohyoid, due to constant stimulation during swallowing from early life. This helps the patient in elevating the floor of the mouth to touch the palate, which then mimics the absent tongue and helps in phonation, mastication, and swallowing. The hypertrophied uvula helps in closing the oropharyngeal opening and forces the air through the nasal passage making the articulation of nasalized sounds possible.[9] The adaptive mechanisms developed by these patients to compensate for absence of the tongue can be used in rehabilitation of patients who have acquired aglossia due to other causes like surgery.
Table 1: Manifestations reported in association with aglossia/hypoglossia[^4^]

| Craniofacial          | Oral/Dental                  | Limb defects                                                                 | CVS^*                        | CNS^†                      | Visceral                | Functional                      | Endocrinial         |
|-----------------------|------------------------------|------------------------------------------------------------------------------|-------------------------------|----------------------------|--------------------------|---------------------------------|---------------------|
| Microcephaly          | Anodontia/Hypodontia         | Upper limb defects usually distal to humerus and lower limb defects distal to femur (Unilateral/Bilateral) | Hypertension                  | Mental retardation         | Situs inversus totalis     | Speech difficulties (slurred speech/ Inability to pronounce lingual consonants) | Hypothyroidism       |
| Facial and Jaw asymmetry | Congenital absence of mandibular incisor teeth | Sydactyly                                                                   | Ischemic heart disease        | Unilateral/Bilateral 6th and 7th nerve palsies | Jejunal atresia          | Difficulty in feeding/ Swallowing (neonatal period) |                     |
| Micrognathia          | Abnormal teeth               | Bony fusion of phalanges                                                    | Systolic murmur               | Esophageal atresia         | Conductive deafness       |                                 |                     |
| Retruded chin         | Protuded upper anterior teeth | Congenital absence of parts of lower limb (Hemimelia)                       | Dextrocardia                  | Hypoplastic epiglottis     |                          |                                 |                     |
| Convex profile        | Bilaterally impacted teeth   | Congenital absence of parts of upper limb (Peromelia)                       | Ventricular septal defect     | Gastrochisis               |                          |                                 |                     |
| Prognathic mandible   | Malocclusion                 | Nail dysplasia                                                               | Chest deformity              | Choanal atresia/ Stenosis  |                          |                                 |                     |
| Thinning of condylar heads | Bony/ Cartilagenous fusion of jaws (syngnathia) |                                 | Pulmonary hypoplasia          |                           |                          |                                 |                     |
| Increased gonial angle | Collapse of mandibular arch  | Cleft palate                                                                 |                               |                           |                          |                                 |                     |
| Absence of mandibular rami |                          | Fusion of alveolar mucosa with labial mucosa (synechiae)                     |                               |                           |                          |                                 |                     |
| Microstomia           |                              | Intraoral band                                                               |                               |                           |                          |                                 |                     |
| Incompetent lips      | Agenesis of lower lip        |                                                                           |                               |                           |                          |                                 |                     |
| Median notch/ Cleft in upper/ lower lip | Ventromedial auricular position (melotia) |                               |                               |                           |                          |                                 |                     |
| Epidermoid cyst of eyelid |                          |                                                                           |                               |                           |                          |                                 |                     |

[^4^]: CVS: Cardiovascular system. CNS: Central nervous system
The movement of the developing tongue plays a role in causing the elevation of the palatal shelves from vertical to horizontal position to form a continuous hard secondary palate during the 8th intrauterine week. The palatal grooving in this patient could be due to overgrowth of lateral palatine ridges due to absence of tongue during secondary palate development.

Functional and aesthetic rehabilitation of patients with aglossia is difficult and requires multidisciplinary and multistep approach. Surgical reconstruction of the tongue is not a priority as the patients show functional adaptation to most functions except speech. Speech therapy is required in early stages as most of the times speech is delayed and slurred. Correction of malocclusion and jaw anomalies would require orthodontic and surgical treatment. Expansion of collapsed mandibular arch and its retention is difficult, and uses of modified palatal expanders often worsen the swallowing and speech function. Distraction osteogenesis following symphyseal osteotomy and use of rapid prototyping models for making custom made (tooth-borne or bone-borne) distractors and determining distraction vector has been found to be an useful alternative for mandibular expansion.

Prosthetic rehabilitation with fixed/removal appliances or implant-supported prosthesis can be considered where sufficient bone height and width is available. In this case, the mandibular alveolar bone height was greatly reduced and ridge augmentation with autologous bone graft would be required before prosthetic consideration.

AC is a rare, potentially fatal developmental defect. Early diagnosis, sustained care, and multidisciplinary approach to management and rehabilitation are required right from birth for survival of the patient.

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Table 2: Halls classification of oromandibular limb hypogenesis syndrome

| Type | A: Hypoglossia-hypodactylia | B: Hypoglossia-hypomelia | C: Hypoglossia-hypodactylomelia |
|------|----------------------------|-------------------------|-------------------------------|
| Type II | A: Hypoglossia-hypodactylia | B: Hypoglossia-hypomelia | C: Hypoglossia-hypodactylomelia |
| Type III | A: Glossopalatine 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-robins syndrome |
|          | D: Moebius syndrome | E: Amniotic band syndrome |

for malignancy and trauma.

Most cases of AC report characteristic facies with lingually collapsed mandibular arch, further exaggerated due to missing teeth, especially the mandibular incisors. This patient also had characteristic facial features and oligodontia, which included the mandibular incisors but the mandibular transverse arch deficiency was only limited to anterior region, probably because of the action of the hypertrophied mylohyoid muscle, which compensated for the absent tongue during mandibular arch development. The acquired adaptive mechanism may have reduced the severity of mandibular transverse arch deficiency in this patient. Low caries activity in maxillary teeth has been reported in AC, but this patient had a high caries activity and periodontal disease leading to early loss of remaining teeth and radicular cyst formation. The susceptibility towards caries and periodontitis could be due to lack of cleansing action of the tongue. Cleft palate has been reported in association with aglossia but a median groove without bony cleft as seen in this case has not been reported before. Erenberg and Nowak concluded that palatal grooving is for survival of the patient.
Announcement

Warm Greetings from Belgaum,

Firstly I would like to extend my heartfelt thanks for IAOMP office bearers for permitting us to conduct regional convention in collaboration with IAOMP.

The regional convention was one day scheduled program which was held on 26-05-2012 was conducted successfully.

The theme of the convention was “Diagnostic cytology in head and neck lesions”. It was being conducted by Department of Oral Pathology and Microbiology of Maratha Mandal’s Nathajirao G. Halgekar Institute of Dental Science and Research in association with IAOMP.

We had a total of 61 registered members, from various dental colleges of Karnataka and Maharashtra contributed to the success of this event. Participants included faculty, post graduate students from Oral Pathology and General Pathology, and also consultants. The convention was inaugurated by President IOAMP Dr. Alka Kale and Smt. Rajashree Halgekar, Chairperson of Maratha Mandal group of institutions. Addressing the gathering Smt. Rajashree Halgekar emphasized the need and importance of Oral Pathologists to be a part of such various regional programmes. Dr Alka Kale appealed all students and staffs of Oral Pathology to take interest in such educational gathering and attend the same. Eminent speakers - Dr Nirmala Jambekar, Dr Wiseman Pinto, Dr. Prakash V. Patil and Dr Ramakant S. Nayak from different prestigious institutions delivered lectures on Oral Cytology and its role in diagnosis. Slide seminars on FNAC of bone and lymph node pathology benefitted the participants and were appreciated.

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