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Case Report

NON-SYNDROMIC CONGENITAL HYPOGLOSSIA WITH HEMIMANDIBULAR HYPOPLASIA - A RARE CASE REPORT

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ABSTRACT

Hypoglossia concomitant with hemimandibular hypoplasia is a rarely reported entity. The congenital anomaly of hypoglossia is uncommon and fewer than 50 cases have been reported in literature so far whereas unilateral mandibular hypoplasia is found to occur in one in 3500 live births. This article focuses on the nonsyndromic category wherein we report a case of unilateral mandibular hypoplasia and rudimentary tongue in a 6-month-old boy baby with presence of mild asymmetry towards the right side of the mandible. The objective of this presentation is to highlight the rarity, possible etiology, manifestations and diagnostic approach of the nonsyndromic group.

Key words: Hypoglossia, Hemimandible, Hypoplasia, Nonsyndromic, Rare entity.

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INTRODUCTION

Hypoglossia or Aglossia is a rare congenital anomaly, manifested by the occurrence of a small or rudimentary tongue. The orofacial features associated with hypoglossia are recessed or hypoplastic mandible [1]. There have been less than 50 cases of hypoglossia reported so far [2-4]. On the other hand mandibular hypoplasia (MH) alone is a craniofacial anomaly which occurs one in 3500 live births [5]. This case report throws light on two different manifestations occurring together in an infant.

CASE REPORT

A 6-month-old male baby was brought to the outpatient department by his mother with complaint of absence of tongue [Figure 1]. On obtaining detailed history it was revealed that baby was born at full term via cesarian section in a local hospital, with 3 ½ kg weight to a 27 year old mother and 34 year old father at gravida 2 para 2: Immediately after delivery the baby was shifted to a private tertiary care hospital for MRI evaluation by neonatologist to rule out any other anomalies. The cry of the baby was normal after birth. Since the baby had difficulty in suckling mother's milk, as per the advice of the neonatologist the baby was fed with formula milk using a feeding bottle with a bigger sized nipple.

The baby was a second child and his five year old elder sister was apparently normal. The parents were unrelated; the family history was otherwise unremarkable with no consanguinity and the mother had no history of drug intake or trauma during her pregnancy.

Intraoral examination revealed that the baby had a small-sized mandible towards the right side and a small 0.8cm to 1cm rounded bud like protuberance evident in the floor of the mouth. The baby was able to close and open

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his mouth normally; there was no evidence of natal or neonatal teeth present. The baby responded well to syllables. At present baby is fed with ceralac, formula milk and mashed semisolid soft food stuffs till date. There was no associated ear, limb or cardiac anomalies present.

Singh and Barlett's classification

Chart 1.

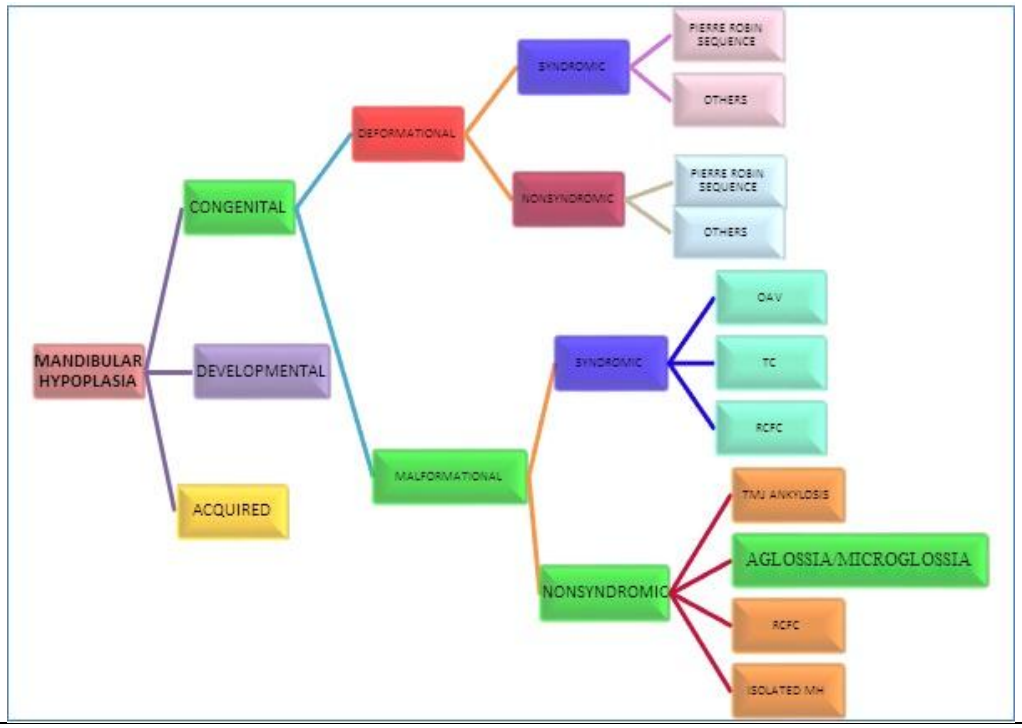


Table 1. Judith .G. Hall's Classification: Syndromes of Oromandibular and Limb Hypogenesis[4]

Type I A) Hypoglossia B) Aglossia	**Type IV** A) Intraoral Bands and Fusion B) With Hypoglossia C) With Hypoglossia-Hypodactyly D) With Hypoglossia-Hypomelia E) With Hypoglossia -Hypodactylomelia
Type II A) Hypoglossia-Hypodactyly B) Hypoglossia-Hypomelia (Peromelia) C) Hypoglossia-Hypodactylomelia	**Type V** A) Hanhart Syndrome B) Charlie M. Syndrome C) Pierre Robin Syndrome D) Moebius Syndrome E) Amniotic Band Syndrome
Type III A) GlossopalatineAnkylosis B) With Hypoglossia C) With Hypoglossia-Hypodactyly D) With Hypoglossia-Hypomelia E) With Hypoglossia-Hypodactylomelia	

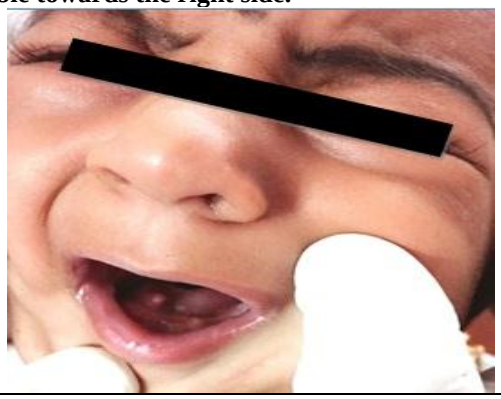
Table 2. Modified Hall's classification proposed by various authors [8,20]

Modified Hall's classification	Modified subtypes	Clinical Features	References
	Type I B.1	Isolated aglossia	Salles F et al, GuptaS
	Type I B.2	Aglossia with adactyli	Nevin NC et al, Purohit et al
	Type I B.3	Aglossia with hypodactyli (mental	Preis et al, Kantapura and

Type I B (Aglossia)		retardation, cardiac defect, anodontia, hypothyroidism)	Tanpaiboon
	Type I B.4	Aglossia with rudimentary ear (deafness)	Higashi and Edo,

Fig 1. Profile photograph showing hemimandibular hypoplasia of right side.

Fig 2. Intraoral photographs of the baby with a small bud like projection of the tongue and hypoplastic mandible towards the right side.



DISCUSSION

Congenital aglossia/hypoglossia:

Congenital aglossia/hypoglossia was first reported in 1718 by Antoine de Jussieu, renowned French botanist and physician, in his study, "Observation About How a Girl Born Without the Tongue was Able To Perform All Functions That Depend On That Organ [6]. It was Rosenthal, in 1932, who first described a true case of aglossia-adactylia, with cleft of the lower lip, absence of the maxillary incisors, and only a small rudimentary tongue. The case reported by Rosenthal was the fifth reported case in the 2 centuries since de Jussieu's report [7].

In 1971, Judith.G.Hall classified these entities into 5 different groups, all under the generic term Syndromes of Oromandibular and Limb Hypogenesis (Table I) [4].

Isolated case reports describe congenital hypoplasia or complete absence of the mandible and tongue, often associated with other deficits of the first branchial arch derivatives or the limbs .These cases have been associated with high mortality due to respiratory distress and at times due inability to suckle mother's milk .We report a case of an infant with a rare and possibly unique anomaly with severe hypoplasia of the right side of the mandible which many authors used the term 'agnathia'. Similarly aglossia is often used to refer to both the total absence of anterior two third and posterior one third of the tongue or the hypoplasia of either thirds of the tongue. Most cases of aglossia and hypoglossia reported in the literature were associated with syndromes like Moebius, Pierre Robin, and Hanhart in which there were limb deformities, cleft palate, deafness and situs inversus respectively [8,9,10,11&12].

Various etiologies like traumatisation of the fetal cells due to intrinsic and extrinsic factors were believed to be the cause behind these anomalies. Deficient migration of neural crest cells into the ventral part of the first branchial arch during the fourth week of embryogenesis is the most likely mechanism underlying the defects in the infant reported here. Vascular insufficiency in the territory of the distal maxillary artery may contribute to the arrest in the development or be the primary cause of aglossia [13].

The distal portion of the first pharyngeal arch gives origin to the mandibular swelling, from which arises Meckel's cartilage. The mandible forms by intra membranous ossification from around the ventral part of Meckel's cartilage after which it disappears. The presence of normal mandibular rami but mild hypoplasia of the mandibular body presumably reflects premature arrest of mandibular intra membranous ossification. The normal epitympanic part of the malleus and incus implies sparing of the dorsal part of Meckel's cartilage. Absence of the anterior two-thirds of the tongue implies failure of development of the two lateral lingual swellings on either side of the tuberculum impar. Absence of the posterior third of the tongue implies arrested development of the second arch copula or third and fourth arch of hypobranchial eminence. The severe microstomia is most likely a secondary defect, occurring secondary to hypoplastic musculature of the floor of mouth [13&14].

Thornton in 1982 has suggested that rupture of an amnion during early pregnancy may produce membranous strands, which adhere to extremities and may also be responsible for facial abnormalities [15]. Scott suggested that the presence of organized thrombi in fetal vessels maybe responsible for an ischemic event. A vascular accident has also been postulated as a possible causative

factor [16]. Before external carotid artery takes over the blood supply to tongue and mandible, the stapedial artery supplies first brachial arch. Injury to stapedial artery or premature involution can hamper the vascular supply and may be responsible for the orofacial anomalie [17]. Oulis and Thornton 1982 considered radiation therapy received by mother at around first month of pregnancy as an environmental factor that could be contributory [15].

As per a case report published in the year 1963by GM Ardan et al, a six month baby who was diagnosed with congenital aglossia, when re-examined at the age of eight had a portion of the posterior tongue present which has grown with sufficient motility together with the very mobile floor of the mouth to make substitute movements that mimic a normal tongue [18].

Hemimandibular hypoplasia

Studies related these rare groups of nonsyndromic malformations of the mandible were very few. In a retrospective analysis by Bhattacharya S et al in (2015) out of 266 patients with congenital Mandibular Hypoplasia, only 18 turned out to be nonsyndromic with only one having unilateral growth disturbance as in our patient [19]. Clinical findings to make a clear differentiation between the syndromic and the nonsyndromic categories and the subsequent diagnostic procedures, treatment modalities, and prognosis vary accordingly.

According to Singh and Bartlett Classification of mandibular hypoplasia in 2005. The most promising discussion was made, where they illustrated an algorithm to classify mandibular hypoplasia. According to their classification congenital hypoplasias can be either deformational or malformational on the basis of whether extrinsic forces or an intrinsic growth disturbance causes the resultant deformity. These patients can be further classified as having syndromic or nonsyndromic mandibular hypoplasias.

The subset of patients who are with nonsyndromic, malformational mandibular hypoplasia were categorised under four subgroups:

1. Patients with TMJ ankylosis
2. Those with aglossia/microglossia,
3. Those with rare craniofacial clefts (RCFC), and
4. Those with isolated mandibular hypoplasia.

Singh and Bartlett Classification of mandibular hypoplasia [chart1]. Besides the fundamental functions of speech, mastication and swallowing, the tongue also has an important role in the growth of jaws, particularly on that of the mandible and in the prevention of malocclusion. Moreover it affects facial esthetics and therefore aglossia impairs psychological and social development. There have been reports where there is gradual hypertrophy of the mylohyoid muscle which aids in stimulation of swallowing by elevating the floor to touch the palate. [20&21]

As far as speech is concerned marked distortions in the articulation of the phonemes /t/, /d/, /n/, /s/, /z/, /š/,/ž/, and omission of the phonemes /l/ and /r/.Speech may be slurred and having a nasal tone. As per the previous reports by Weingarten and Roth the speech defects are minor in almost all who were old enough to speak, did so intelligibly [22, 23&24]

Our patient did not present with any major difficulty in breathing, although suckling reflex was initially difficult later when a larger sized nipple was used with a feeding bottle the baby learned to suckle normally. The baby responded well to syllables. These kinds of patient should be closely monitored at different milestones of ages as babies grow in unique ways [24].

Hence, they should be evaluated for various orofacial manifestations and treated accordingly.[25&26] High level of compliance is expected from the parent's part for planning a comprehensive care.

CONCLUSION

Treatment of hypoglossia associated with hemimandibular hypoplasia sequelae may involve several conditions that cannot be treated with a single procedure. Facial sequelae of aglossia compromises facial esthetics and severely affect psychological well-being of the patient. The treatment of these sequelae is possible and may improve quality of life of these patients. Surgical correction is usually impractical for patients with hypoglossia. To treat the anatomical, functional, psychological, nutritional and esthetic problems, a multidisciplinary approach is necessary.

STATEMENT OF HUMAN AND ANIMAL RIGHTS

All procedures performed in human participants were in accordance with the ethical standards of the institutional research committee and with the 1964 Helsinki declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

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Nil

CONFLICT OF INTEREST

The authors declare no conflict of interest.

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