

Case Report

Oromandibular limb hypogenesis syndrome: a singular variation of a rare syndrome: a case report

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Abstract – Background: A positive prenatal history of maternal fever has been found to express as a range of defects in infants including limb reduction, central nervous system defects, facial dysmorphogenesis and foetal death. Oromandibular limb hypogenesis syndrome is a rare spectrum of congenital disorders characterised by malformations of the tongue, mandible, maxilla and limbs. They present as sporadic cases with extremely low incidence and seldom occur with associated syngnathia. Syngnathia can manifest as fibrous, bony or in combination. **Observation:** Here we represent a rare case of Oromandibular limb hypogenesis syndrome type IV E with combination of syngnathia, cleft palate, retrognathia and hypoglossia-hypodactylomelia. An early surgical release of syngnathia was undertaken on having features of failure to thrive, high risk of aspiration and related complications due to enteral feeding. **Conclusion:** Delaying the surgical procedure could result in growth restriction and progressive ankylosis of the Temporomandibular joint (TMJ). It is essential to document the singular variant of Oromandibular limb hypogenesis syndrome (OLHS) to the medical literature.

Introduction

Congenital defects of the orofacial region are often found with asymmetric hypoplastic deformities of distal extremities. Oromandibular limb hypogenesis syndrome (OLHS) is a rare spectrum of congenital disorders that represent a group of uncommon and overlapping conditions. Only around 50 cases of OLHS have been reported on reviewing the world literature [1]. The syndrome is characterised by malformations of the tongue, mandible, maxilla and limbs. The etiology still remains unknown but a greater acceptance is to the environmental factors over genetic factors. The first case was reported by Rosenthal [2] in 1932 and these syndromes were classified by Hall [3] in 1971 on the basis of different characteristics and their representing combinations (Tab. I).

Observation

Our patient, three-month old male infant, a third child of non-consanguineous married couple, full term, normal vaginal delivery, body weight 2.8 kg was referred to our tertiary care facility having complaint of inability to open his mouth since birth. There was a positive history of antenatal fever in the first

trimester. The antenatal care was uneventful with normal scan at 5th and 9th month. Shortly after delivery the baby was not able to cry. Lack of mouth opening was observed during oral suction. There was no respiratory distress. The patient had difficulty in oral intake since birth and hence was supported with expressed breast milk feeding via nasogastric tube.

The family history revealed first pregnancy of the mother underwent midterm abortion due to anencephaly and neural tube defect. The second child from normal delivery is a 3 years old female, alive and healthy. The present age of the father is 29 years and mother 26 years.

Physical examination revealed abnormality of right hand and foot. Right hand thumb was well developed but the distal phalanges of remaining digits were hypoplastic with syndactyly having web like appearance. The right foot was hypoplastic with absent metatarsals and phalanges (Fig. 1). External genitalia were normal. Findings of maxillofacial region were severe retrognathia of the mandible with lip trap and distortion of lower left lip with loss of vestibular depth. There was no mouth opening or jaw thrust and we were unable to insinuate fingers in the mouth due to fusion of the upper and lower jaws in the midline which extended more towards the left side. No cleft defect was observed as the intraoral examination could not be carried out. The patient was evaluated by neonatologist, pediatric cardiologist, ophthalmologist and ENT specialist to

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Table I. Hall’s classification of oromandibular limb hypogenesis syndrome.

Type I	A	Hypoglossia
	B	Aglossia
Type II	A	Hypoglossia-hypodactylia
	B	Hypoglossia-hypomelia
	C	Hypoglossia-hypodactylomelia
Type III	A	Glossopalatine ankylosis
	B	With hypoglossia
	C	With hypoglossia-hypodactylia
	D	With hypoglossia-hypomelia
	E	With hypoglossia-hypodactylomelia
Type IV	A	Intraoral bands and fusion
	B	With hypoglossia
	C	With hypoglossia-hypodactylia
	D	With hypoglossia-hypomelia
	E	With hypoglossia-hypodactylomelia
Type V	A	Hanhart syndrome
	B	Charlie M syndrome
	C	Pierre Robin syndrome
	D	Moebius syndrome
	E	Amniotic band syndrome

rule out any systemic abnormality. Genetic counselling regarding the recurrence risk of the syndrome in relation to patient’s sibling and offspring was carried out [4].

The MRI of head and neck showed hypoplasia of nasal cavity, paranasal sinuses, oral cavity and jaw bones. There were two bony septi seen extending from mandible to maxilla in the midline and towards left side (Fig. 2). Anteriorly the tongue appeared to be partially fused with the palate. Communication of oral cavity with left nasal cavity was suggestive of palatal cleft. There were bilateral punctate ossification present in the spheno-frontal and coronal suture area along with mild flattening of skull bones, orbital floor and mandibular fossae. Temporomandibular joint (TMJ) spaces, mandibular condylar head, ramus and body were normal. However, fusion of both the half of the mandible at midline was more acute. The brain tissue and airway appeared to be normal. The findings of CT scan corroborated to the MRI. The hematological and biochemical parameters were within the normal limit. On clinico-radiological evaluation he was diagnosed as a case of Oromandibular limb hypogenesis syndrome Type IV E.

As patient was having features of failure to thrive and was at high risk of aspiration and related complications due to enteral feeding, it was decided to intervene at the earliest. Delaying the procedure may result in growth restriction and progressive ankylosis of the TMJ. The case was discussed with



Fig. 1. (a) Frontal profile of patient. (b) Frontal facial profile. (c) Hypodactylomelia of right foot with absent metatarsals and phalanges. (d,e) Morphological features showing anonychia of the distal phalanges and hypoplasia of the distal hand (Right).



Fig. 2. Coronal scan showing bony septi seen extending from mandible to maxilla in the midline and left side.



Fig. 4. Immediate post-operative mouth opening.

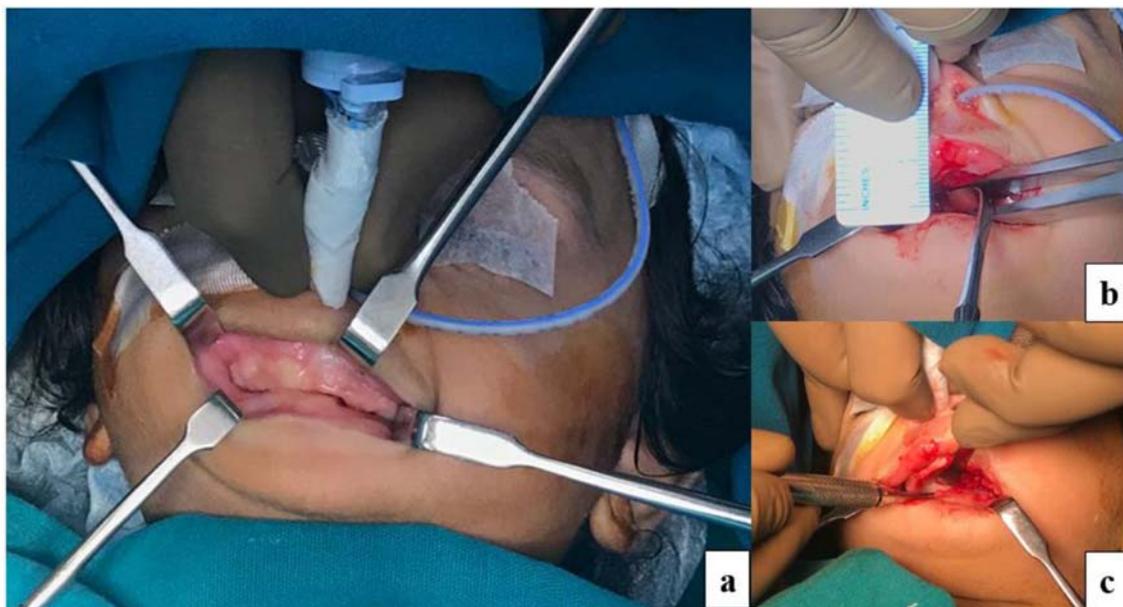


Fig. 3. Intraoperative release of syngnathia and synechia.

the intensivist and pediatric anesthesiologist. The patient was placed supine and administered general anaesthesia through fiberoptic guided nasal intubation. The release of the bony syngnathia along the occlusion plane and surgical transection of synechia (posterior right, left) were pursued using osteotomes and electrocautery. Achieving a mouth opening of 18 mm and release of fusion of the tongue from the palate, the cleft palate and microglossia was appreciated (Fig. 3). Bilateral condyles palpated endaurally and mandibular movements were confirmed intraoperatively. The patient was transferred to neonatal ICU and extubated uneventfully after 24 hours. Nasogastric tube was removed on post-operative day

5 and oral feed was commenced (Fig 4). The child was discharged on 7th post-operative day and parents were advised monthly follow up. 4 months post-surgery the oral feeding of the patient is sustained with achieving timely growth milestones and weight gained to 5.6 kg (Fig. 5).

Discussion

OLHS represents a spectrum of disorders affecting the orofacial region and the limbs. Having overlapping features, these syndromes have been classified for ease of diagnosis. Our case is OLHS Type IV E: syngnathia with hypoglossia-hypodactylomelia,



Fig. 5. 4 months post-operative mouth opening.

has a heterogenous etiology. There was a positive prenatal history of maternal fever. Evidence of maternal hyperthermia causing OLHS has been well documented [1,5]. Maternal fever at/above 102 °F (38.9 °C) between 4 and 14 weeks of pregnancy results in a range of defects including limb reduction, central nervous system (CNS) defects, facial dysmorphogenesis, and foetal death. It is pathogenesis points towards heat induced vascular disruption of the embryo. The severity, duration and timing of the maternal fever, relates to the nature of the anomalies present. CNS defects appear to be the most common consequence of gestational hyperthermia. Yet, patients with OLHS are often born with normal intelligence [5]. However, more research is required to be undertaken for association of maternal hyperthermia and OLHS.

Micrognathia, hypoglossia and limb anomalies are three characteristic features of the syndrome [6]. In our case, hypoglossia was diagnosed during the surgery due to the

presence of syngnathia. The extent of limb deformities varies from syndactyly (incomplete separation of the fingers) to amelia (complete loss of the limb). Severe micrognathia is observed in all the cases of OLHS, due to the osseous defect in the mandibular midline region and occasionally involving the fusion of the maxillae as was present in our case.

Syngnathia (“syn”=fusion and “gnathos”=jaw) means fusion of jaws. This fusion could be fibrous, bony or a combination of both. It is a rare craniofacial disorder seen in infants, association with a number of syndromes, varying in severity from a single mucosal band (synechia) to complete bony fusion (synostosis). Till date 118 [7] cases have been documented with the first case reported in 1936 by Burket *et al.* [8] which was associated with congenital bony temporomandibular ankylosis, facial hemiatrophy and Horner’s syndrome. Congenital syngnathia primarily affects mouth opening causing difficulty in feeding and respiration. Only 8 combination cases of syngnathia and synechia present together have been reported [7].

This syndrome is diagnosed at birth. Despite the tongue being deficient, majority of cases improve oral functions of speech and swallowing with time with the help of other muscles substituting for tongue. Speech therapy improves phonetics and surgery is recommended on presence of severe hypoplasia. Prosthetic rehabilitation of limbs is desired for improvement in locomotion. All these treatment procedures undertaken in turn will uplift the quality of life.

Authors’ contribution

Nandakishore Sahoo: Conceptualization; Ravinder S. Semi: Methodology; Praneet K. Rana: Writing original draft; Nandakishore Sahoo: Visualization; Lakshmi shetty: Investigation; Nandakishore Sahoo: Supervision; Praneet K Rana: Writing-Reviewing and Editing.

Conflict of interests

The authors declare that there is no conflict of interest.

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Informed consent

Written informed consent was obtained from parents.

Ethical approval

Ethical approval was not required.

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