



ISSN: 0975-833X

## CASE REPORT

### ORO MANDIBUAR LIMB HYPOGENESIS SYNDROME TYPE II C ... A RARE CASE REPORT

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#### ARTICLE INFO

##### Article History:

Received 21<sup>st</sup> July, 2015

Received in revised form

19<sup>th</sup> August, 2015

Accepted 30<sup>th</sup> September, 2015

Published online 20<sup>th</sup> October, 2015

##### Key words:

Hypoglossia,  
Hypomelia,  
Micrognathia.

#### ABSTRACT

Hypoglossia –Hypomelia is rare congenital anomaly affecting the tongue and limbs. Hall in 1971 classified it under a complex group of disorders called oro Mandibular limb hypogenesis Syndrome. It is an extremely rare condition with around 40 cases reported in world literature. The etiology of the syndrome is unknown. Some type of intrauterine trauma is the most widely accepted etiology. The characteristic features of the syndrome are hypoglossia, limb anomalies of variable degrees and micrognathia of the mandible. It is characterized by failure of development of intraoral region and distal extremities. It is congenital and there seems to be no sex predilection. We hereby report a case of 15 years old girl patient presenting with retruded mandible, hypoglossia and limb anomaly (hypomelia) Her parents and other siblings were normal. Positive prenatal history of maternal hyperthermia was obtained suspecting it to be cause of Oro Mandibular limb hypogenesis

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**Citation:** Dr. Sanjay Kumar Sinha, Dr. Shailesh Chandra Gupta and Dr. Brajesh Kumar, 2015. "Oro mandibular limb hypogenesis syndrome type ii c ... A rare case report", *International Journal of Current Research*, 7, (10), 21354-21356.

## INTRODUCTION

Oromandibular-limb hypogenesis syndromes (OLHS) represent a group of rare conditions characterized by congenital malformations involving multiple sites such as the tongue, mandible, and limbs. (Brockmann *et al.*, 2009) In 1971, Hall classified OLHS into 5 major types and according to this, the case report presented here falls under type II C, which is hypoglossia-hypodactylomelia syndrome. (Rasool *et al.*, 2009; Jang *et al.*, 1997) The hypoglossia- hypodactyly syndrome, the Moebius syndrome, the Hanhart syndrome, the Charlie M syndrome and OMLH are possibly variants of the same condition, and it is often difficult to define the phenotypic boundaries between them. (Hanhart, 1950; Kaissi *et al.*, 2005) There is considerable overlap between these syndromes gathered under the term OLHS, with a marked variability of face and limb anomalies as well as other additional malformations. (Brockmann *et al.*, 2009) Limb deficiencies are major congenital mal-formations and can result from a number of etiologi-cal factors. (Hall, 1971) Heat- induced vascular disruption has been considered as one of the etiological factors for these syndromes. (Superneau and Wertelecki, 1985) Apart from this, teratogenic etiology has also been implicated. (Hanhart, 1950) The genetic origin of these syndromes is uncertain. (Chicarilli and Polayes, 1985)

However, most of the cases are sporadic. (Hall, 1971) Forty-seven cases of hypoglossia-hypodactylia (Type I A) syndrome have been reported before 1990. (Bagnulo *et al.*, 1999) The radiographic features consisted of retruded mandible in the lateral cephalograms, multiple missing teeth in orthopantomogram and incomplete development of limb since it is suggestive of this syndrome.

#### Case History

A 15 year old female patient reported to our clinic with the chief complaint of her aesthetics. Patient also complained of small jaw, irregular teeth and inability to move her tongue in either direction. Patient had no difficulty in swallowing or mastication. She was the third child of her parents. Her other siblings and parents are normal. There was no history of similar findings in the family.

#### On general examination

Patient was well-oriented on time and space. Her upper limb was not well formed (Hypomelia) (Fig.1). On extraoral examination, Patient exhibited mandibular micrognathia (Fig. 2), protruded upper anteriors (Fig.3), retruded chin, incompetent lips, deep mentolabial sulcus, hyperactive mentalis muscle and considerable lip trap.

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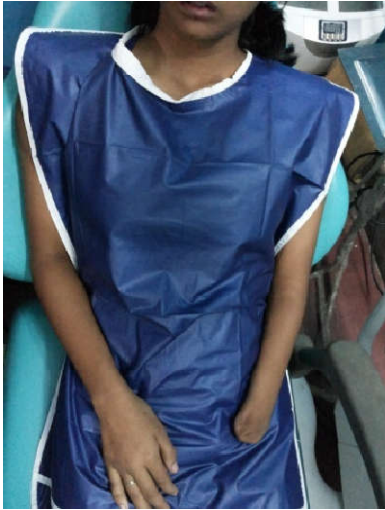


Fig.1. Displaying limb deformity



fig.2 patients side view showing severe micrognathia of mandible

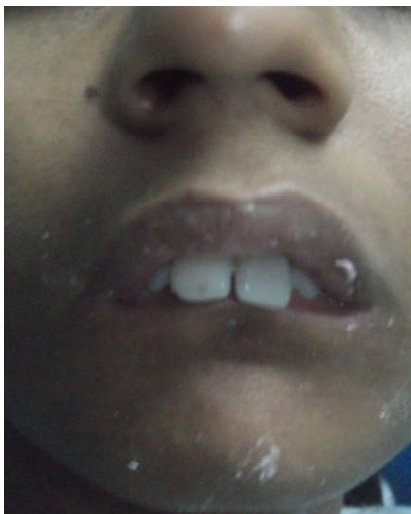


Fig. 3. Front view showing protruded upper anterior teeth



Fig. 4. Patient intraoral view



Fig. 5. Patient lateral Cephalogram showing severe Retrusion of mandible

On intraoral examination, Patient exhibited constricted maxillary and mandibular arches, hypoglossia (Fig.4) and hypodontia. Her upper two lateral incisors along with three lower incisors were found missing. Patient was unable to move her tongue in forward and lateral directions. She was operated twice for ankyloglossia. Still, she was not able to move her tongue anteriorly and laterally. Her Lateral cephalogram x-ray (Fig. 5) reveals severely retruded mandible and protruded maxillary arch. Her OPG (Fig. 6) revealed missing upper lateral incisors and lower multiple incisors. Patient was the youngest out of three children of her parents. Her other siblings are normal. Her mother had frequent bouts of fever during pregnancy. Her mother was engaged in over exertional activities during pregnancy, including tiring travelling. The patient was normal delivered child, no surgery took place.

## DISCUSSION

According to Gorlin, intrauterine trauma is the most widely accepted cause of this syndrome.

Three features essential for the diagnosis of this syndrome are

- (1) Limb anomalies of varying severity.
- (2) Micrognathia of the mandible (or maxilla) in the midline segment;
- (3) Variable reduction in the tongue size (microglossia);



**Fig. 6. Patient orthopantomogram**

Our patient presented with extraoral features, including convex profile, micrognathic mandible with relative maxillary prognathism and intraoral features, including microstomia, hypodontia, hypoglossia, and constricted maxillary and mandibular arches. Our patient also presented with upper anomaly. It was first reported by Rosenthal (Perks *et al.*, 1998) in 1932 as ag-lossia congenita. The OLHS is a rare complex of jaw and limb defects with unknown aetiology. (Wadhvani *et al.*, 2007) There seems to be no sex predilection. (Perks *et al.*, 1998) However, the pro-posed aetiology is heredity, maternal hyperthermia and positive drug history during pregnancy. This syndrome sometimes presents with cranial nerve palsies (sixth and seventh). (Hall, 1971) Hermann *et al.* (Hermann *et al.*, 1976) analysed OLHS cases and found that there was severity of upper limb involvement, especially malformation of the feet, but they did not find cranial nerve palsies and this was significant in differentiating the cases.

The group of patients with cranial nerve palsies included some with limb defects similar to those observed in Hanhart syndrome and others with Poland anomaly; finally, cases with cranial nerve palsies without limb involvement were documented. (Herrmann *et al.*, 1976) No evidence of cranial nerve palsy was observed in the case presented here. Multiple site involvement and the wide range and combination of anomalies make classification difficult. (Alexander *et al.*, 1992) There is overlap and similarity between different syndromic entities among similarities with OLHS, including a long list of syndromes like Moebius syndrome, hypoglossia hypodactylia syndrome, Hanhart syndrome, glossopalatine ankylosis syndrome, limb deficiency, splenogonadal fusion syndrome, and Charlie M syndrome. All are very uncommon except for Moebius syndrome. (Alexander *et al.*, 1992) These groups of syndromes require a long-term and multidisciplinary approach. (Preis *et al.*, 1996) The case reported here is a rare syndrome with multiple site involvement. The treatment includes replacement of upper limb with prostheses, correction of malocclusion. Patient should go under orthodontic treatment to achieve proper aesthetics and masticatory apparatus. Multi disciplinary approach is needed to handle such cases.

## Conclusion

The case presented here rare subtype of oro mandibular limb hypogenesis syndrome with oral manifestations like hypodontia. Hypoglossia, mandibular micrognathia and malalign teeth along with limb anomaly. Almost all cases reported till now seemingly sporadic.

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