doi: 10.1111/j.1600-0714.2007.00570.x

J Oral Pathol Med (2007) 36: 555–7

www.blackwellmunksgaard.com/jopm

CASE REPORT

Oromandibular limb hypogenesis syndrome, type IIA, hypoglossia–hypodactylia: a case report

Puneet Wadhwani[1], Shadab Mohammad[2], Ruchi Sahu[1]

[1]*Saraswati Dental College, Oral and Maxillofacial Surgery;* [2]*UP King George's University of Dental Sciences, Oral and Maxillofacial Surgery, Lucknow, Uttar Pradesh, India*

A patient with oromandibular limb hypogenesis syndrome, type IIA hypoglossia–hypodactylia, is reported. The features essential for the diagnosis of this congenital defect include a reduction in tongue size (microglossia), micrognathic mandible and limb abnormalities of varying severity. Parents and the other sibling were normal. Past medical history for exposure of the mother to drugs during pregnancy was positive.
J Oral Pathol Med (2007) **36:** 555–7

Keywords: oromandibular limb hypogenesis; syndrome

Case report

A 15-year-old male patient reported to the Department of Oral Diagnosis, UP King George's University of Dental Sciences, Lucknow, with a chief complaint of difficulty in swallowing and unclear speech since birth.

On evaluation both the parents and a younger sibling, were found to be normal; another sibling, died 24 days after birth, the cause of which was not known. At the second month of pregnancy, the mother had suffered from high fever and was treated by a local doctor with some medication. The doctor was unaware of the pregnancy, as the parents did not disclose it.

On general examination, the patient presented as a normal, healthy, well-oriented young boy with slightly altered speech.

On oral examination, the patient had a convex profile with micrognathic mandible and relative macrognathic maxilla (Fig. 1).

Intra oral examination revealed, a high arched palate (Fig. 2a) with proclined upper anteriors and labially placed lateral incisors, a constricted V-shaped mandibular arch with congenitally missing central incisor and lateral incisors, and missing right canine (Fig. 2b). Microglossia with partial ankylosis was also seen.

On radiological examination, impacted mandibular right canine and second molars, bilaterally, were seen in the orthopantomogram (OPG) (Fig. 3)

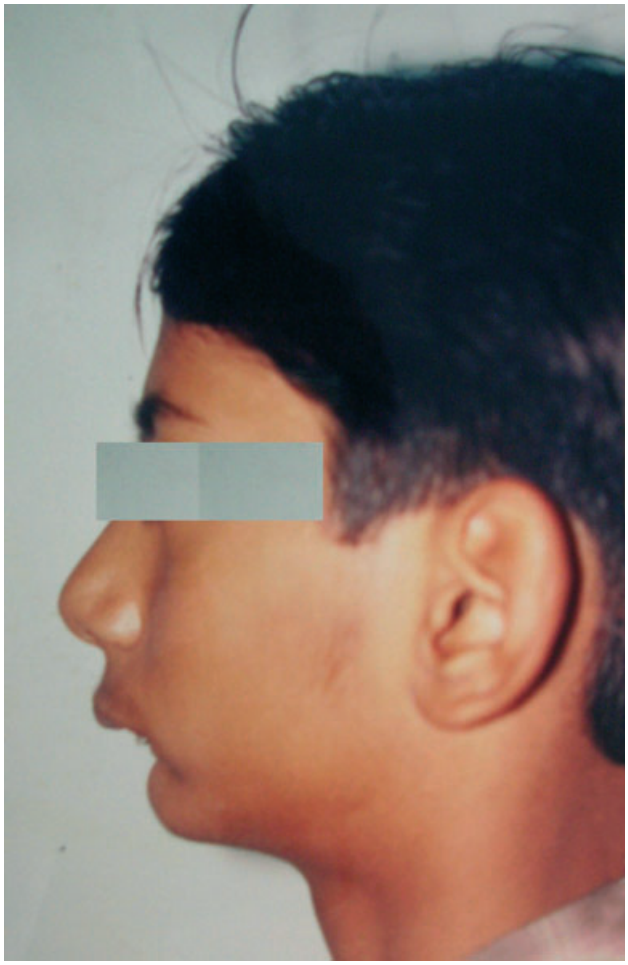

Figure 1 Extra oral profile view.

Correspondence: Puneet Wadhwani, MDS (Oral and Maxillofacial Surgery), 3/233, Vishal Khand – III, Gomtinagar, Lucknow 226010, Uttar Pradesh, India. Tel: +91 5222392571, +91 9415408088, Fax: +91 5222815392, E-mail: drpuneetwadhwani@hotmail.com
Accepted for publication February 27, 2007

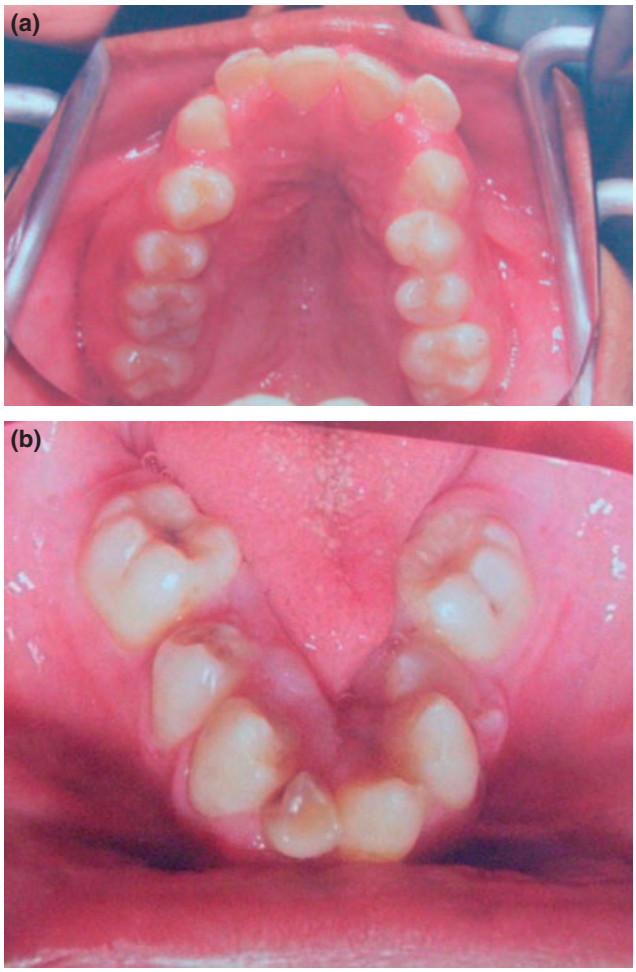

Figure 2 (a) Maxillary arch, a high arched palate. (b) Mandibular arch, constricted, V-shaped, congenitally missing central incisor, lateral incisors, missing right canine and microglossia with partial ankylosis.

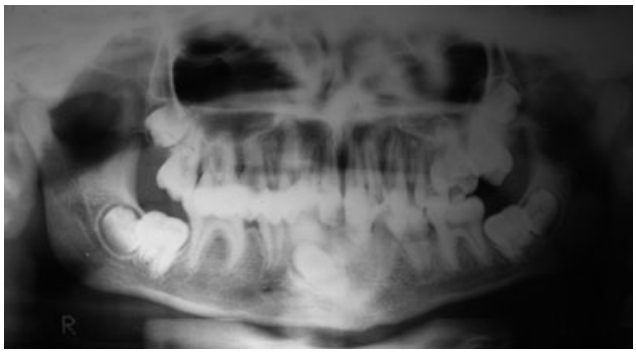

Figure 3 Orthopantomogram (OPG), impacted mandibular right canine and second molars, bilaterally.

The right hand was severely hypoplastic with unossified carpal bones with a vestigial digit (Fig. 4a); left hand was fully formed and normal. The left foot was severely hypoplastic with unossified tarsal bones (Fig. 4b), right foot was fully formed and normal. No CNS abnormalities or hearing loss were observed.

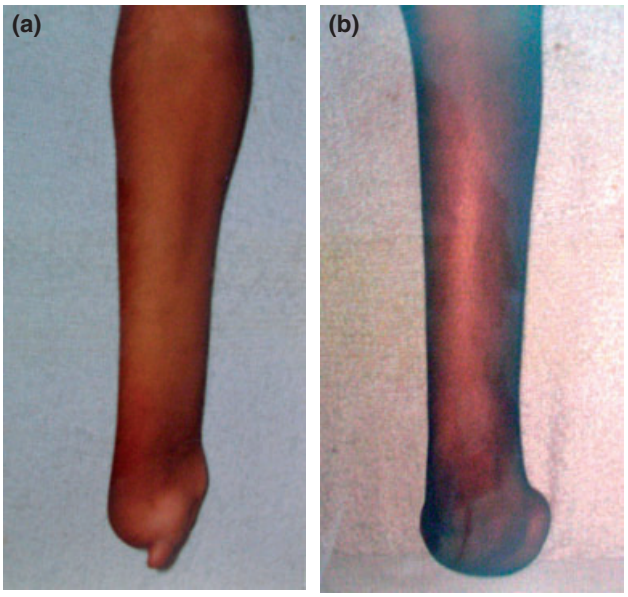

Figure 4 (a) Right hand, severely hypoplastic with unossified carpal bones with a vestigial digit. (b) Left foot, severely hypoplastic with unossified tarsal bones.

Comments

The oromandibular–limb hypogenesis syndrome was first reported by Rosenthal in 1932 as Aglossia Congenita. In 1971, Hall used the term hypoglossia–hypodactylia as part of a classification of syndromes of oromandibular and limb hypogenesis (1).

The features essential for the diagnosis of type IIA, hypoglossia–hypodactylia include (2):

1. reduction in tongue size (microglossia);
2. micrognathic mandible;
3. limb abnormalities of varying severity.

Additionally, a number of other features include, gingival abnormalities and absence of teeth (hypodontia) (3).

Our case presented with the above-mentioned essential features, in accordance with hypoglossia–hypodactylia syndrome, type IIA.

The aetiology of hypoglossia–hypodactylia is unknown. Most cases are sporadic and exhibit extreme variability with regard to limb anomalies (1). However, according to Gorlin, some type of intrauterine trauma is currently the most accepted cause for this syndrome.

Hyperthermia is a proven teratogen in animals and humans (4). Similar findings have also been reported by Martinez-Frias et al. (5). Hyperthermia during pregnancy was reported in our case, for which some medication was given when the patient's mother was 2 months pregnant.

However, there is no evidence that heredity plays a role and there seems to be no sex predilection. Treatment, if any, is long term and multidisciplinary, directed towards improving tongue, lip and mandibular functions (1).

References

1. Alexander R, Friedman JS, Eichen MM, et al. Oromandibular limb hypogenesis syndrome; type IIA, hypoglossia–hypodactylia – report of a case. *Br J Oral Maxillofac Surg* 1992; **30**: 404–6.
2. Lustmann J, Lurie R, Struthers P, et al. The hypoglossia–hypodactylia syndrome: report of 2 cases. *J Oral Maxillofac Surg* 1981; **51**: 403.
3. Gorlin RJ, Cohen MM. *Syndromes of head and neck*, 4th edn. New York: Oxford University Press Inc., 2001; 822–32.
4. Edwards MJ. Hyperthermia as a teratogen a review of experimental studies and their clinical significance. *Teratog Carcinog Mutagen*. 1986; **6**: 563–82.
5. Martinez-Frias ML, Garcia Mazario MJ, Caldas CF, et al. High maternal fever during gestation and severe congenital limb disruptions. *Am J Med Genet* 2001; **98**: 201–3.