

Non-surgical treatment of upper airway obstruction in oculoauriculovertebral dysplasia: a case report

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SUMMARY A non-surgical technique for the treatment of upper airway obstruction in oculoauriculovertebral dysplasia using an intra-oral orthopaedic appliance is described, which resulted in respiratory and feeding problems being solved without side-effects. This non-invasive management might also be of major benefit in the treatment of airway obstruction associated with Pierre Robin sequence, mandibular micrognathia in other craniofacial anomalies, or obstructive sleep apnoea.

Introduction

The presence of retrognathia and glossoptosis which leads to respiratory problems is normally seen in 'Pierre Robin sequence'. The term 'glossoptosis' was first introduced by Robin (1934), to describe the tendency of the tongue to fall back into the respiratory space, causing pharyngeal obstruction in infants with cleft palate.

Newborns who develop severe upper airway obstruction require emergency treatment or they become exposed to prolonged hypoxia, risking permanent brain damage, cor pulmonale, congestive heart failure, pulmonary oedema, or right-sided heart hypertrophy (Hoffman *et al.*, 1963). Furthermore, feeding problems are found to be common, with frequent regurgitation, swallowing of air, and aspiration resulting in failure to thrive and possible pneumonia.

A number of conservative approaches and surgical procedures have been developed to bring the mandible forwards and prevent the tongue from falling back: these include downward positioning of the infant (Robin, 1934), nasopharyngeal intubation (Routledge, 1960; Stern *et al.*, 1972; Wada *et al.*, 1983), glossopexy (Douglas, 1946), perimandibular fixation and extension (Kiskadden and Dietrich, 1953), and tracheostomy.

Despite controversial discussion of the appropriate treatment procedure, tracheostomy has been considered as the last resort because of the high rate of complications (bronchopneumonia, need for constant tracheostomy and negative influence on speech development), and other methods with lower rates of short- and long-term complications have been employed.

Hotz and Gnoinski (1982) reported the successful use of orthopaedic appliances in the treatment of upper airway obstruction in patients with Pierre Robin sequence. Based on the hypothesis that the backward and upward tilting of the tongue causes mandibular displacement and respiratory distress, the aim of the procedure was to correct the posture of the tongue by means of intra-oral orthopaedic appliances. Upper plates with long posterior extensions were inserted, obturating the whole length of the velar cleft, so that the tongue was mechanically held forward.

Respiratory distress has also been described in cases of oculoauriculovertebral dysplasia (Burstein *et al.*, 1995), a deformity with an extreme variability in terms of characteristics (Gorlin *et al.*, 1963, 1990; Rollnick *et al.*, 1987; Cohen *et al.*, 1989). The mandible is involved and, depending on the skeletal anatomy of the ramus and temporomandibular joint (TMJ), the mandibular

deformity is classified into three types (Pruzansky, 1969; Kaban *et al.*, 1981). Type III-deformity represents the most severe anomaly with complete absence of the ramus, glenoid fossa and TMJ, which can result in a retropositioning of the mandible to the abnormal side and a falling back of the tongue creating upper airway obstruction.

A new conservative approach undertaken to treat respiratory distress in an infant with oculoauriculovertebral dysplasia is reported. A modification of the upper plate described by Hotz and Gnoinski (1982) resulted in the successful treatment of the upper airway obstruction.

Case presentation

A girl, the first baby of a healthy mother, 29 years of age, and a father of 43, was born after an uncomplicated full-term pregnancy. At birth, her body weight was 3100 g. The parents were non-consanguineous, and the family did not have any history of facial or other congenital anomalies.

The patient exhibited the phenotypic characteristics of oculoauriculovertebral dysplasia: marked facial asymmetry with complete absence of the right mandibular ramus, glenoid fossa, and zygomatic arch; macrostomia; corneal opacity,



Figure 1 Extra-oral photograph of the newborn patient with oculoauriculovertebral dysplasia demonstrating severe retrognathia of the mandible on the right side.

and aphakia of the right eye; bilateral microtia; preauricular tags; malformations of the right middle and internal ear associated with agenesis of the ossicular chain; fusion of ribs; butterfly vertebrae; ventricular septal defect; patent ductus arteriosus; and pulmonary artery anomalies (Figure 1).

The complete absence of the right ramus resulted in a falling back of the mandible to the abnormal side (Figure 2). Subsequently, the

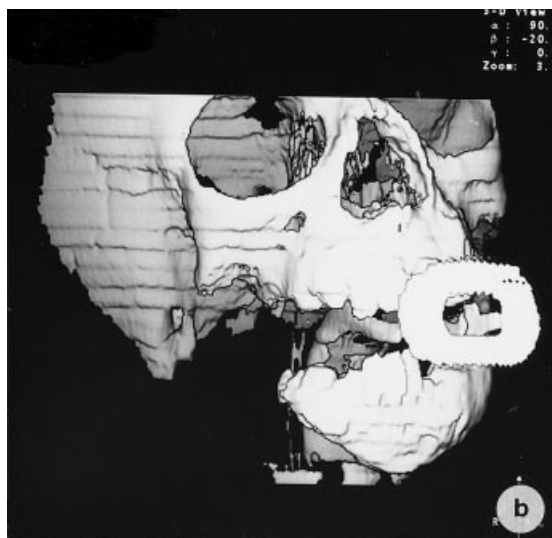


Figure 2 (a,b) Three-dimensional reconstruction of the CT scans. There is a marked facial asymmetry with complete absence of the right mandibular ramus, glenoid fossa, and zygomatic arch.

marked unilateral retrognathia caused glossoptosis leading to severe airway obstruction characterized by stridor, cyanosis, and failure to thrive. Apnoea was noted in both waking and sleeping phases, and the O_2 saturation values frequently decreased below 80 per cent. During obstructive episodes, mandibular manipulation was successful in moving the shortened side of the mandible forward. Through this intervention, the base of the tongue simultaneously advanced out of the pharynx.

The use of a nasopharyngeal tube prescribed for Pierre Robin patients could possibly manage to maintain the anterior position of the tongue. However, the reason for the glossoptosis in our patient was not a deficiency in mandibular growth, as seen in Pierre Robin sequence, but a severe deformity of the mandibular skeleton. The suggested maximum 4-week period for using nasopharyngeal tubes seemed insufficient for a myofunctional forward stabilization of the distorted mandible. The high morbidity risk of nasopharyngeal tubes, when left in place for more than a few weeks, is regarded a significant problem (Sher, 1992).

As an alternative to this procedure the management of upper airway obstruction by means of an orthopaedic appliance was discussed. In our patient, airway obstruction resulted from the posterior displacement of the mandible causing a falling back of the tongue. The idea was that, instead of mechanically pulling the mandible forward, a triggering of a functional process, i.e. in this case the provocation of the vomiting reflex, could be utilized to treat pharyngeal obstruction. In order to avoid the unpleasant vomiting reflex situation the patient would learn to bring and hold the mandible in a forward position. For this reason, an upper plate with a new type of posterior border was designed.

An alginate impression was taken, without sedation, while the hypoplastic side of the mandible was moved forward. From the plaster cast an upper plate with a posterior bow was fabricated (Figure 3). The angulation of the bow was approximately 130 degrees, with a length of 1.5 cm. In order to prevent bruising on the posterior part of the tongue, the wire was covered with acrylic. After the plate was properly adapted,

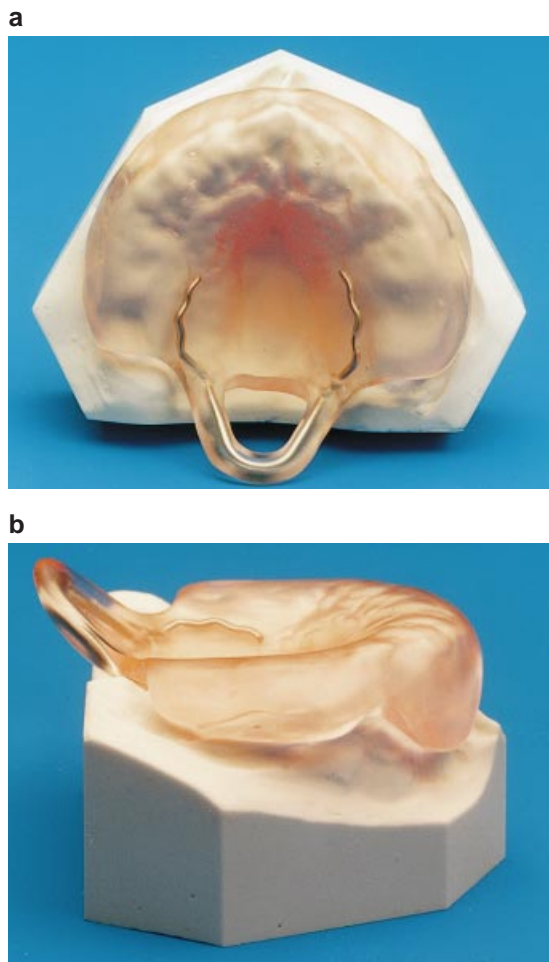


Figure 3 (a) Occlusal and (b) lateral view of the appliance.

the baby wore it permanently. The appliance was cleaned three times a day and no inconvenience was noticed. In order to increase the stability of the plate, an adhesive cream was used.

Within the first 5 days after insertion, the respiratory problems were considerably reduced so that airway obstruction was only occasionally noticed during sleeping. Even then oxygen saturation values did not fall below 80 per cent. No inadequate ventilation occurred whilst the baby was awake. After 1 week the infant was discharged with a pulse-monitoring device to be attached during sleep.

During the following 4 months the oxygen saturation value rarely fell between 80 and

90 per cent. Even when it did, the baby did not need any external help to keep the mandible forward. As a result of positional changes of the mandible and the tongue, the suck-swallow mechanism could be established and tube-feeding was gradually reduced.

It seems that, through the myofunctional stimulation of the plate, the infant learned to hold the mandible forwards in awake phases. After 6 months, the child also learned to cope without the aid of the plate while sleeping.

The use of orthopaedic myofunctional appliances in the treatment of respiratory distress occurring due to skeletal dysplasias seems promising. Similar types of appliances might be beneficial in treating other situations associated with airway obstruction, for example obstructive sleep apnoea.

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