www.jpnim.com Open Access eISSN: 2281-0692
Journal of Pediatric and Neonatal Individualized Medicine 2018;7(1):e070119
doi: 10.7363/070119
Received: 2017 Aug 25; revised: 2017 Oct 30; rerevised: 2017 Nov 13; accepted: 2017 Nov 28; published online: 2018 Feb 06

Case report

Early treatment of congenital syngnathia. A case report

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Abstract

Congenital syngnathia is a rare malformation. The synechia or synostosis of the maxilla and the mandible may be partial or complete and also be associated with other malformations. The syngnathia can be developed between the upper and lower alveolar ridges with a wide spectrum and range of severity documented in case reports. The authors report a newborn with complete syngnathia and discuss the management during the first days of life. Our goal is to help attending physicians understand anatomic and functional implications of syngnathia, and become familiar with strategies used in the early caring of a newborn with congenital syngnathia. Literature reveals results of different patients with poorly documented follow-up and different management. The priority is a safe airway and early nutrition with human milk to avoid the risk of aspiration pneumonia, malnutrition, and poor growth.

Keywords

Mouth abnormalities, maxilla abnormalities, hypoplastic mandible, congenital maxillomandibular fusion, congenital syngnathia, synostosis, perinatal management.

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How to cite

Duro EA, Benitez SM, Barreto Paternina AdC. Early treatment of congenital syngnathia. A case report. J Pediatr Neonat Individual Med. 2018;7(1):e070119. doi: 10.7363/070119.

Introduction

Congenital syngnathia is a very rare malformation. When a newborn infant after birth cannot open the mouth due to maxillary and mandibular fusion, attending physicians need to do something. In the front, there are open lips, but the examination of the intraoral space cannot be made. If the child is breathing well (the first priority in the management of any infant born without an opening mouth is to secure the airway, said Dawson [1] in 1997 when he proposed the first classification of congenital bony syngnathia), then physicians need to detect other congenital anomalies (as facial malformations, facial hemiatrophy, genital and limbs anomalies) and provide adequate nutrition with a big challenge: how? There are no standard protocols, but early measures are necessary to improve proper nutrition for the baby.

If other malformations as cleft lip, cleft palate, mucous cysts of the lower lips, popliteal pterygium, digital and genitals anomalies are present, the differential diagnoses must be made with popliteal pterygium syndrome (PPS) (OMIM 119500); and if the baby only has pits or sinuses in the lower lip, and/or cleft lip and/or cleft palate, the diagnosis could be Van der Woude syndrome (VWS) (OMIM 119300), an autosomal dominantly inherited disorder. Both syndromes are in relation to a mutation in the gene encoding interferon regulatory factor-6 on chromosome 1q32-q41 [2, 3].

But isolated congenital syngnathia with synechia of the gums, partial or complete, fibrous or bony, between the upper and lower alveolar ridges, without others malformation is another phenotype with unknown molecular basis (OMIM 119550). ORPHA presumes an autosomal dominant with variable expressivity inherited manner with a prevalence < 1/1,000,000. It is a very rare condition, with no more than sixty cases described in the literature since Burket's report in 1936 marked the difference with the acquired forms [4]. No gold standard exists for making the diagnosis.

Some authors have proposed aetiological causes for congenital syngnathia as abnormal development of the stapedial artery, intrauterine oral pressures or swallowing defects, abnormal early loss of the neural crest cells, hypervitaminosis A or teratogenic exposure [5].

Recently, in mice mutant genetic model of syngnathia researchers demonstrated that *FOXC1* gene in relation to fibroblast growth factor 8 gene (*FGF8*) is required for normal development of jaw formation [6]. The *FOXC1* gene on chromosome 6p25.3 provides instructions for making a protein that regulates the activity of other genes as a transcription factor and plays a critical role in early development, particularly in the formation of structures in the embryo. The *FGF8* gene on chromosome 10q24.32 provides instructions for making a protein called fibroblast growth factor 8 a growth factor involved in regulation of cell growth and maturation. Other researchers suggested mutations in bone morphogenic proteins as aberrant BMP4 in the cranial neural crest of mice led to congenital syngnathia [7].

Laster proposed a classification for congenital bony syngnathia into 4 types from a simple bony fusion of the alveolar ridge to a complex of mandibulozygomatic bony fusion with temporomandibular joint ankylosis [5] (**Tab. 1**).

Case report

A female infant was born at 40 weeks of gestation by normal vaginal delivery from a 26-year-old mother. She was the third child of a nonconsanguineous couple. There was no significant maternal or family history, and the pregnancy was uncomplicated with regular prenatal care before 14 weeks of gestation without problems or teratogenic exposure. Two antenatal ultrasound scans did not reveal anomalies and serologic tests for intrauterine infection were negatives.

Table 1. Laster classification of syngnathia [5].

Type	Category	Clinical features
Type 1a	Simple anterior syngnathia	Bony fusion of alveolar ridge **without** other congenital deformity in the head and neck
Type 1b	Complex anterior syngnathia	Bony fusion of alveolar ridge **with** other congenital deformity in the head and neck
Type 2a	Simple zygomatic-mandibular syngnathia	Bony fusion of the mandible to the zygomatic complex causing **only** mandibular micrognathia
Type 2b	Complex zygomatic-mandibular syngnathia	Bony fusion of the mandible to the zygomatic complex and associated **with** cleft or temporomandibular joint ankylosis

At birth, the patient cried and Apgar scores were 8 and 9 at 1 minute and 5 minutes, respectively, but quickly developed a respiratory distress with grunts. The infant was in good status, was active and had pinkish skin color with adequate SpO_2 but a full-layer adhesion was present on upper and lower gingiva (**Fig. 1**). She also had micrognathia with retrognathia and low seat ears and two preauricular skin tags.

When systemic examination revealed no other anomalies in the rest of the body, the newborn infant was transferred to the neonatal intensive care unit and started oxygen supplementation with a low flow nasal cannula, obtaining a good response. She had a birth weight of 3,030 g, a length of 50 cm, and a head circumference of 34 cm; all the three parameters were appropriate for gestational age.

At two hours after birth, we performed a chest X-ray which was normal; abdominal and transfontanellar ultrasonography showed no abnormalities. With the impossibility to breastfeed, a percutaneously inserted central catheter (PICC) was placed for total parenteral nutrition in the lower extremity of the baby.

With oxygen supplement a Computed Tomography (CT) scan was done which showed bony fusion between the two alveolar ridges; a small oral cavity contained a rudimentary tongue. Brain appeared normal (**Fig. 2**).

Maxillofacial CT with 3D reconstruction revealed an extensive fusion of the mandible with the maxilla and zygoma. The mandibular rami were completely fused to the maxillary tuberosities and zygoma, with ankylosis of the temporomandibular joint (**Fig. 3**). We classified the deformity as a Type 2b according to the Laster classification.

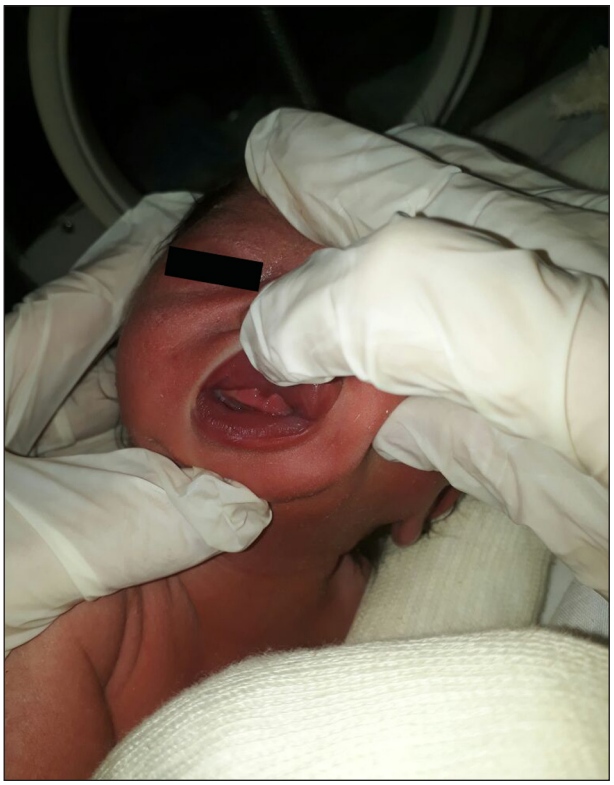

Figure 1. A full-layer adhesion was present on upper and lower gingiva.

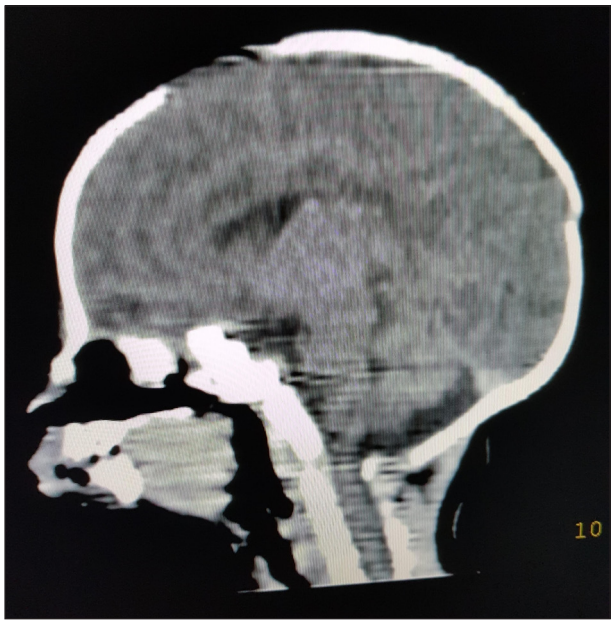

Figure 2. Computed Tomography (CT): bony fusion between the two alveolar ridges, a small oral cavity contained a rudimentary tongue, brain appeared normal.

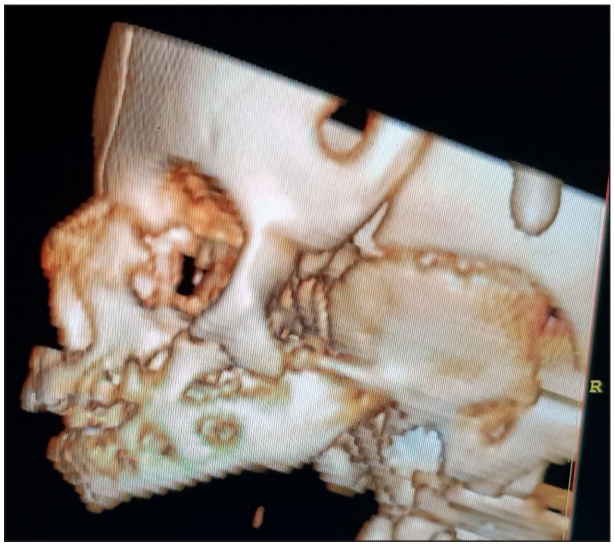

Figure 3. The mandibular rami were completely fused to the maxillary tuberosities and zygoma, with ankylosis of the temporomandibular joint.

The girl was referred to the level IV National Pediatric Hospital [8] at three days of age with the diagnosis of Type 2b congenital syngnathia and jaundice. Despite the girl's confused prognosis, parents received emotional care by a multidisciplinary team consisting of midwife, psychologist, and nurse who counseled parents with frankness and with accurate medical information about the possible infant evolution.

Discussion

Congenital syngnathia is rare, but physicians need to be alert in front of this severe craniofacial malformation. An absence of standardized treatment protocols and the paucity of long-term outcomes data make treatment a daunting and uncertain process. Congenital maxillomandibular fusion further complicates when the newborn baby starts with respiratory distress. Room air will often not be sufficient for these babies and the use of a pulse oximetry is mandatory as appropriate oxygen-air blenders. SpO_2 should rise gradually over 85% 10 minutes after birth.

Our patient had a good response to oxygen supplementation with a low flow nasal cannula, but as we don't have a small size fiberscope to make nasal fiber optic intubation, which is the technique of choice for a complete syngnathia, we prepared for the emergency requirements: a surgical tracheostomy [9].

Tracheotomy is a definitive procedure for airway management for neonates whose condition fails to respond to other measures. It is associated with frequent and serious adverse effects, complications, and even death [10].

Difficulties in providing an adequate feeding are the main causes of undernutrition in newborn infants with syngnathia. It is important to be alert about feeding and growing issues, aspiration, and/or gastroesophageal reflux, which may negatively affect the baby. It is useful to consider the energy needs of infants with congenital syngnathia, who need to grow and gain weight. Close nutritional follow-up by a nutritionist or a physician with expertise in nutrition is necessary for detecting early growth failure and optimizing child nutritional needs [11].

The possibility of starting feeding with a nasogastric tube was contemplated, but the respiratory distress needed free nostrils. If the choice is a nasogastric probe, attending physicians will consider the possibility of gastroesophageal reflux with the risk of aspiration and the mandatory use of human breast milk.

Although the early need for gastrostomy tube placement is another option for feeding issues and avoiding weight loss, in congenital syngnathia this surgical intervention has not been universally accepted.

Conclusions

Congenital syngnathia is a disorder present at birth requiring early management measures. Literature reveals results of different patients with poorly documented follow-up and different management. The priority is a safe airway and early nutrition with human milk to avoid the risk of aspiration pneumonia, malnutrition, and poor growth. Difficulties in providing an adequate feeding are the main cause of undernutrition in newborn infants with syngnathia. When respiratory assistance at birth is finished, the priority is the best feeding for better growth. If a congenital syngnathia is present, aspiration is always a risk. A feeding tube will help the child get proper nutrition until the risk of aspiration improves, and, if surgery is delayed, a gastrostomy tube placement is a proper option.

Declaration of interest

The Authors declared that no competing interest exists.

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