Kufa Journal for Nursing Sciences
Open Access Full Text Article

Publisher
University of Kufa, Faculty of Nursing

Congenital Syngnathia as a Part of Oro-Mandibular Limb Hypogenesis Spectrum: Case Report

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Abstract:
Syngnathia (congenital union of the maxilla and mandible) is uncommon and can range in severity from mucosal bands (synechiae) to complete bone fusion (synostosis). Most of them appear as unilateral, incomplete fusion. Bony fusion (syngnathia) without any other systemic disease or congenital abnormality is extremely uncommon. Oro-mandibular limb hypogenesis syndrome, which includes syngnathia, is an extremely rare disorder characterized by variable degrees of congenital abnormalities involving the tongue, jaw, and limbs.

Keywords: congenital syngnathia, congenital synechiae, oro-mandibular limb Hypogenesis syndrome, congenital maxillomandibular fusion

INTRODUCTION

Congenital fusion of the jaws is a rare condition that was first reported by Burket (1936) [(1)], that is characterized by the limitation of mouth opening at birth. It is caused by a fusion of the maxilla or zygoma with the mandible. The severity of fusion varies From mucosal bands (synechiae) to complete bone fusion (syngnathia) [(2)]. Fusion can happen in the midline, laterally, and unilaterally or bilaterally. A unilateral fibrous fusion is seen in the majority of described cases [(3)].

A etiological explanations for congenital syngnathia have been hypothesized by some writers as faulty stapedial artery development, intrauterine oral pressures or swallowing abnormalities, abnormal early loss of neural crest cells, hypervitaminosis A, or teratogenic exposure [(4)].

Recently, some researchers demonstrated congenital syngnathia as a result of a mutation on FOXC1 gene on chromosome 6p25.3, FGF8 gene on chromosome 10q24.32 plays a critical role in early development, particularly in the formation of

structures in the embryo and involved in the regulation of cell growth and maturation [5].

Oro-mandibular limb hypogenesis syndrome (OLHS), Popliteal Pterygium syndrome, Nager syndrome, Aglossia–Adactylia syndrome, Van der Woude syndrome, and Hanhart syndrome are all associated with syngnathia [6, 7]. Syngnathia within the scope of OLHS; is an extremely rare condition and is characterized by varying degrees of congenital malformation involving the tongue, maxilla, and mandible, as well as reductive limb anomalies [8]. Chicarilli and Polayes [9] divide the oro-mandibular limb hypogenesis syndrome (OLHS) into three categories based on the primary abnormalities of micrognathia, microglossia, and dysgnathia (ankylosis).

Case presentation

A 38-days-old male child of Asiatic origin from Iraq presented to our department of Oral and Maxillofacial Surgery Al-Wasity Teaching Hospital with a history of inability to open his mouth from the first day of birth. The patient presented without any health problems, nor was any family history of congenital anomalies. Clinical examination revealed facial asymmetry, mandibular micrognathia with class 2 skeletal relationship, microstomia, with a complete bony fusion of maxilla and mandibular alveolus. In addition, the soft tissues of the alveolar segments were completely fused. The patient had a nasogastric tube for feeding. Additionally, there were upper and lower limb anomalies (partial amelia) (Fig 1). All usual laboratory tests revealed normal results, as did an abdominal ultrasound and two-dimensional echocardiography.

A computed tomography (CT) of the facial scan revealed a completely bony fusion between the mandibular and the maxilla alveolus, along with mandibular hypoplasia without temporomandibular joint ankylosis (Fig 2). The child was diagnosed with oro-mandibular limb hypogenesis syndrome in the first 3 weeks after birth by a pediatric specialist following the genetic test.

Surgical procedure

The primary goal of our surgical intervention was to improve functions, esthetic and prevent secondary developmental deformity.

The patient was prepared for surgery, the sterile preparation and draping were carried out in the same conventional manner and tracheostomy was done to secure the airway (Fig 3). The first step was an infiltration of local anesthesia then, direct mucosal incisions at the area of upper and lower jaw union to expose the boney fusion, and then fine Osteotome was used to separate the mandible from the maxilla. Finally, the mouth gag is used to achieve a satisfactory mouth opening of about 15 mm (Fig 4). Bleeding was minimal and hemostasis was easily achieved. The healing of oral mucosa by secondary intention without the need for suturing. A splint was performed intraoperatively by used wax as impression material, and then cold cure acrylic was used to fabricated the splint with hooks extended extra orally to stabilized the splint. the splint was trimmed and adapted over the bare maxilla to prevent recurrence of bony fusion (Fig 5). microglossia and cleft palate was identified when the mouth opened. The patient was scheduled for cleft palate repair at age 9-18 months. A splint was removed after 2 weeks. A follow up continuous for 13 months postoperatively and showed maintained mouth opening (Fig 6).

Discussion

According to many studies in the literature, a congenital fusion of the upper and lower jaws is a rare condition. the diagnosis of syngnathia is usually confirmed clinically and radio graphically via CT scan showing bony fusion with or without temporomandibular joint involvement. A prenatal diagnosis of syngnathia may now be possible by 3D

ultrasonography [10]. Laster et al classified congenital bony syngnathia into four categories in 2001, ranging from a simple bony fusion of the alveolar ridge to a complex of mandibulozygomatic bony fusion with temporomandibular joint ankylosis [4] (Tab. 1).

The type of syngnathia presented in our case report is classified as type 1b, also fits within the spectrum of oro- mandibular limb hypogenesis syndrome, a syndrome characterized by congenital abnormalities affecting various areas such as the tongue, mandible and limbs. Hall classified OLHS into five categories in 1971 (Tab.2) [11]. According to this classification, our case report presented here falls under type IIC, which is a hypoglossia-hypodactylomelia syndrome.

Embryo genic development of the face and limbs occurs between the 4th& 8th weeks of gestation so both areas can susceptible to teratogenic stimuli. Many studies on this specific topic suggest that abnormal development of the tongue and the mandible causes prolonged interalveolar contact, which may result in either osseous or fibrous synechiae or both [12]. The goal for early surgical intervention is important to ensure airway patency, enable feeding, swallowing and mastication. So, the surgical treatment is essential to allowed jaw mobilization and prevent temporomandibular joint (TMJ) ankylosis. Although there are no standard protocols for congenital syngnathia treatment, surgical osteotomy to separate the maxillomandibular fusion is the workhorse procedure along with methods to prevent refusion [13].

Conclusions

Syngnathia is a rare congenital abnormality that can result in high developmental morbidity and even mortality, due to both airway difficulties and an inability to take food. The main treatment for syngnathia is osteotomy, accompanied by methods such as internal distractors, and soft tissue or plastic implants, to prevent ankylosis, and bony contact. As syngnathia fits within the spectrum of OLHS, multidisciplinary treatment is necessary in order to provide satisfactory rehabilitation.

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TABLES AND FIGURES:

Figure 1: preoperative photograph of the patient reveal upper and lower limb anomalies

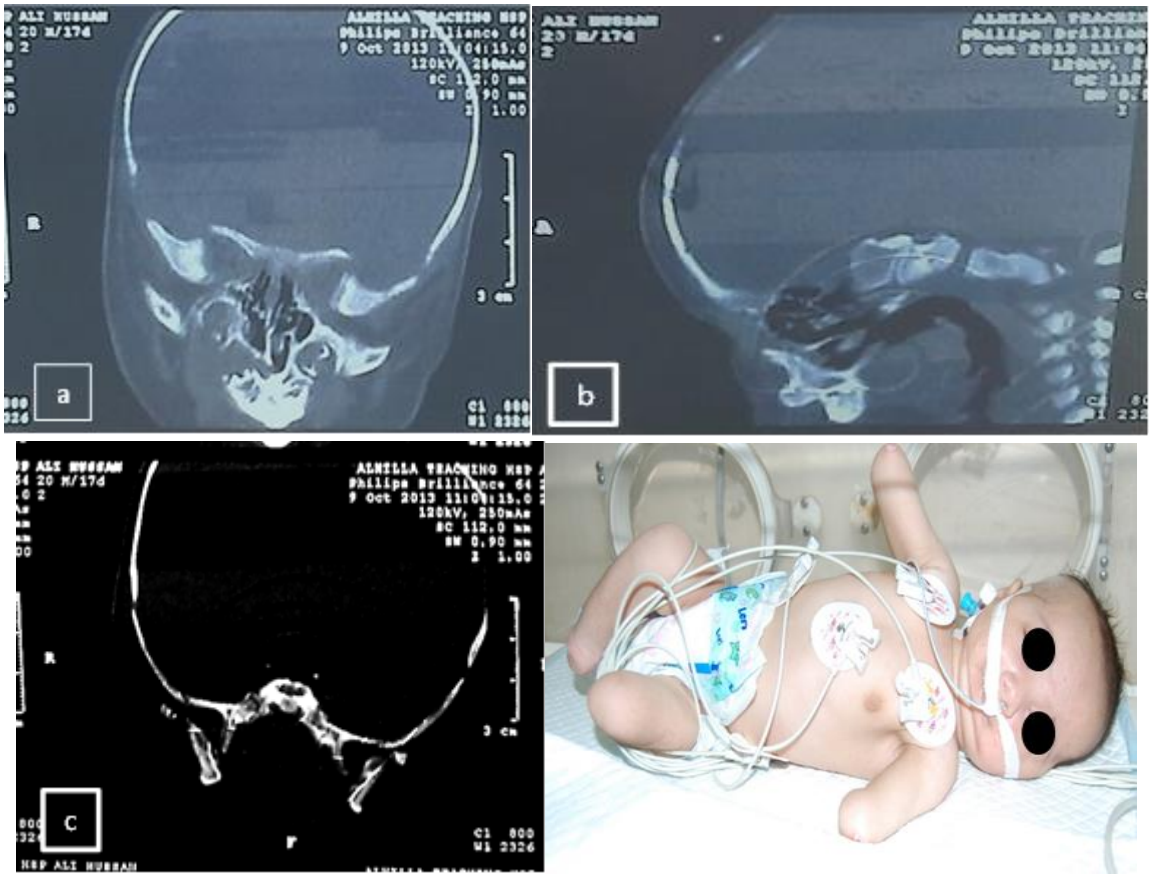

Figure 2: a. CT scan coronal view revealed maxillomandibular fusion (syngnathia), b. CT scan sagittal view revealed maxillomandibular fusion (syngnathia), c. coronal view with the normal temporomandibular joint.

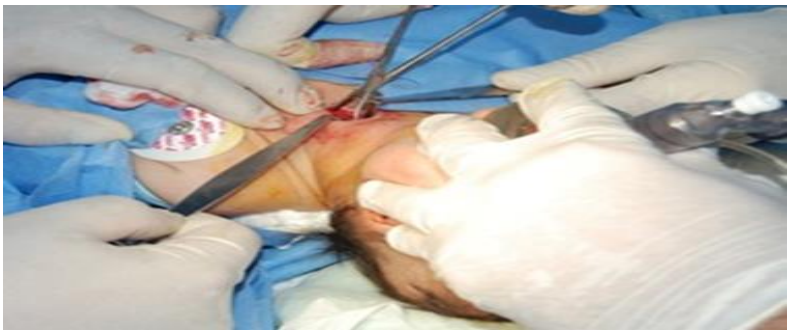

Figure 3: Intraoperatively tracheostomy done

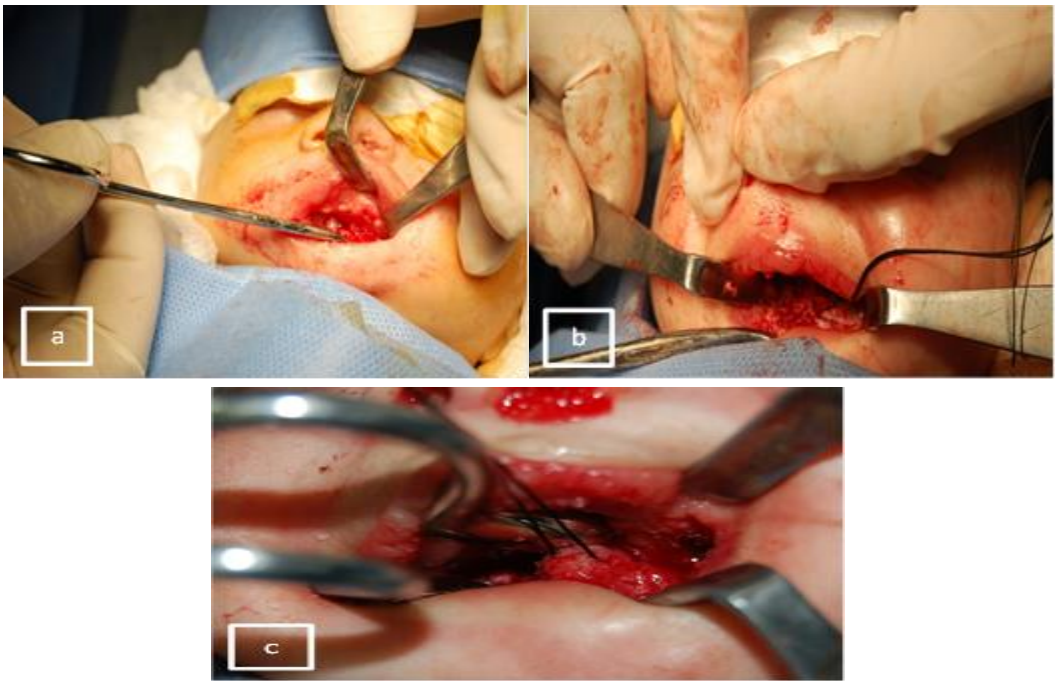

Figure 4: a.soft tissue band separation, b. bony fusion separated by osteotome, c. mouth opening achieved by mouth gauge.

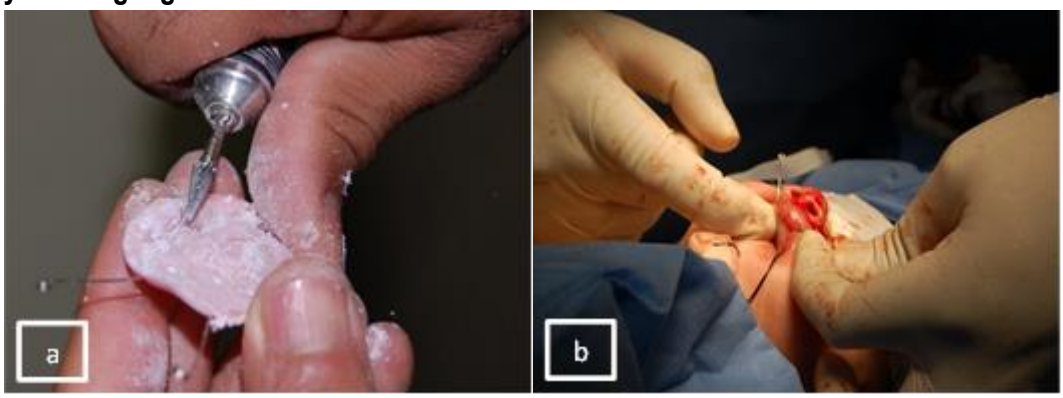

Figure 5: a. splint trimming, b. splint adapted intraorally to avoid recurrent fusion.

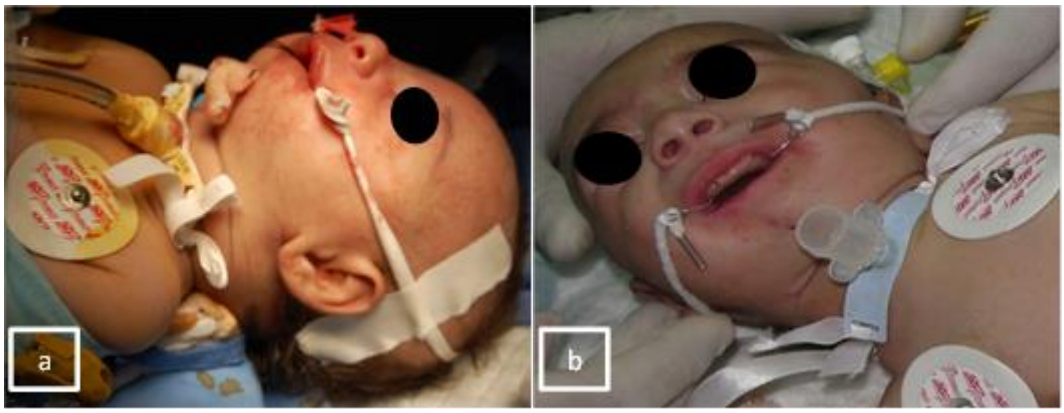

Figure 6: a. immediate postoperatively, b. 2 days postoperatively.

Table 1: Laster classification of syngnathia (4).

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

Table 2: Hall's classification of OLHS (11).

Type -I	**A**	**Hypoglossia**
	B	Aglossia
Type - II	**A**	Hypoglossia - hypodactylia
	B	Hypoglossia - hypomelia
	C	Hypoglossia - hypodactylomelia
Type - III	**A**	Glossopalatine ankylosis (Ankylos-sum superius syndrome)
	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**	The Hanhart syndrome
	B	Charlie M – syndrome
	C	Pierre – Robin syndrome
	D	Mobius syndrome
	E	Amniotic band syndrome