

An Unusual Case of Congenital Syngnathia

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Abstract

Congenital syngnathia is a rare anomaly characterised by the presence of soft tissue (synechia) or bony adhesions (synostosis) between the maxilla and mandible. A wide spectrum and range of severity have been documented in case reports. Primarily it manifests in the inability to open the mouth; impacting on mandibular growth, nutrition, speech and management of the airway. The bony adhesions can be classified as partial or complete, as well as syndromic and non-syndromic. The aetiology of congenital syngnathia remains unknown. We describe a growth restricted, premature infant diagnosed postnatal with multiple congenital anomalies including syngnathia, craniosynostosis, ventriculomegaly, microcephaly, bilateral cataracts, facial dysmorphism, small kidneys, hypoplastic prepuce of the genitalia and bilateral syndactyly of toes 2, 3 and 4. The rare finding of syngnathia with the associated findings in a premature male infant weighing 1065 g at 31 weeks of corrected gestational age is the first to be reported.

Abbreviations:

TM-temporomandibular joint; BMP-bone morphogenetic proteins

Introduction

Congenital syngnathia is a rare anomaly characterised by the presence of soft tissue (synechia) or bony adhesions (synostosis) between the maxilla and mandible [1]. A wide spectrum and range of severity have been documented in case reports. Primarily it manifests in the neonatal period with an inability to open the mouth; impacting on mandibular growth, nutrition, speech and management of the airway [2]. The bony adhesions can be classified as partial or complete, as well as syndromic and non-syndromic [2]. The aetiology of congenital syngnathia remains unknown [2]. The rare finding of syngnathia with the associated findings in a premature male infant weighing 1065 g at 31 weeks of corrected gestational age is the first to be reported. Presenting case reports of syngnathia remain important to grow an evidence base of knowledge and experience of a rare and often severe anomaly.

Case Report

A growth restricted, premature male infant weighing 1065 g was born by emergency caesarean section for severe maternal pre-eclampsia at 31 weeks of corrected gestational age. No other significant pregnancy complications were noted and the mother denied any illness, trauma or drug use in the antenatal period. The parents were not related and their medical history was non-contributory. This was their 2nd child; a 3 year old daughter had no abnormalities.

The infant required brief resuscitation at delivery and was transferred to the neonatal intensive care unit for nasal CPAP. Examination revealed that the lips could be separated but the maxillary and the mandibular gingivae were completely fused anteriorly except for a small opening to the right of the midline and

the buccal mucosa could not be retracted away from the alveolar gingivae (Figure 1).



Figure 1: Inability to open the mouth. Maxillary and the mandibular gingivae fused except for a small opening to the right of the midline.

Infant had micrognathia and retrognathia of the mandible (Figure 2). Further examination revealed microcephaly (< 3rd centile), dense bilateral cataracts, low anterior hairline, low set and posteriorly rotated ears, short palpebral fissures, bilateral syndactyly of toes 2-4 and a hypoplastic prepuce of the genitalia.

The infant was dependent on high flow nasal cannula to maintain the airway and soon developed signs of feed intolerance and gastroesophageal reflux which resulted in frequent and prolonged apnoea.

A multidisciplinary team including neonatologists, otolaryngologists, geneticists, maxillofacial surgeons, a paediatric palliative care clinician and a social worker counselled the parents at numerous meetings. The team and the parents agreed on the initiation of palliative care rather than embarking on corrective surgeries, given the uncertainty of the prognosis and the potential for further suffering and negative impact on the quality of life of the infant. The infant died a few weeks later.

Discussion

Congenital syngnathia is rare and often causes a severe craniofacial malformation [3]. However since the first description of syngnathia by Burket in 1936 our knowledge of this condition is limited to a few case reports. Inconsistent nomenclature to describe congenital maxillomandibular fusion further complicates literature review of this rare phenomenon [4].

The cause remains undetermined but it is thought to have an embryological basis, with several hypothesis including persistence of the buccopharyngeal membrane, amniotic constriction bands, presence of an aberrant stapedius artery, premature loss of embryonic neural crest cells, alveolar fusion predisposed by failure of tongue protrusion or depressed foetal swallowing reflexes, environmental insults, or drug exposure such as meclozine and large doses of vitamin A [5]. Recently He et al., suggested mutations in the bone morphogenetic proteins (BMP) signalling pathway as aberrant BMP4-mediated signalling in cranial neural crest cells of mice led to congenital bony syngnathia [3].



Figure 2: Micrognathia and retrognathia of the mandible.



Figure 3: Lateral aspect of skull X-ray demonstrating the hypoplastic ascending mandibular rami, particularly on the left where the horizontal ramus of the mandible approximate the maxilla.

To date only 55 cases of congenital maxillomandibular fusion have been reported, 28 of which had no associated congenital or systemic defects [6]. Common associations reported include anomalies of the tongue, palate, limbs, eyes, ears, and vertebral column. No known genetic associations have been made, although syndromes include craniofacial microsomia, Van der Woude syndrome (VWS), popliteal pterygium syndrome (PPS), cleft palate lateral alveolar synechiae syndrome (CPLSS), and Fryns syndrome [7]. VWS is an autosomal dominant disorder characterized primarily by cleft lip or palate and lip pits (85% of cases). VWS is the second most common cause of syndromic clefts, second only to chromosome 22q11 microdeletions. PPS is similar to VWS except for the additional findings of popliteal and oral webs, nail deformities, syndactyly, ankyloblepharon (fusion of eyelids), and genital anomalies [8]. CPLSS findings include cleft palate with multiple cordlike adhesions between the free borders of the palate and lateral parts of the tongue and floor of the mouth and abnormal facial features, such as short palpebral fissures, beaked nose, small mouth with protruding lower lip, large cheeks, and short chin. The major findings in Fryns syndrome include coarse face, small eyes, cloudy cornea, cleft of the soft palate, hypoplasia, and absence of lobulation of both lungs, defects of the diaphragm, digitalization of the thumbs, and distal limb deformities [8]. The clinical features in our infant have never been reported in the literature and did not meet the criteria of any of the above syndromes.

Congenital syngnathia, with or without syndromic associations is a complex disorder and management requires a multidisciplinary team. Literature review reveals varied patient outcomes, poorly documented long term follow-up in post-surgical survivors and differences in management strategies (early versus delayed intervention). Early intervention prioritises securing of the airway; however a high recurrence of re-fusion exists requiring multiple surgeries [9]. Delayed intervention places the patient at risk of asphyxia, aspiration pneumonia, malnutrition and poor growth of the facial bones [9].

Conclusion

A lack of standardised treatment protocols and the paucity of long term outcomes data make prognostication a daunting and uncertain process. It therefore remains an important exercise to continue presenting case reports of syngnathia using a universal nomenclature and classification system to grow an evidence base of knowledge and experience to facilitate the development of standard management guidelines and more comprehensive counselling regarding long term functional prognosis.

References

1. Rezende GDC, Pereira AK, Reis ZSN, Parola AR, Bsstos FM, et al. (2013) Prenatal diagnosis of congenital syngnathia by 3D ultrasound and pathological correlation. *J Med Ultrason* 1: 85-87.
2. Said Hamed M, Prasad P (2014) Congenital Fusion of the Mandible and Maxilla- A Case Report from Ajman, United Arab Emirates. *Int J Dent Sci Res* 2: 8-9.
3. He F, Hu X, Xiong W, Li L, Lin L, et al. (2014) Directed Bmp4 expression in neural crest cells generates a genetic model for the rare human bony syngnathia birth defect. *Dev Biol* 391: 170-181.
4. Burket LW (1936) Congenital bony temporomandibular ankylosis and facial hemiatrophy review of the literature and report of a case. *JAMA* 106: 1719-1722.
5. Fallahi HR, Naeini M, Mahmoudi M, Javaherforoosh F (2010) Congenital zygomatic-maxillo-mandibular fusion: a brief case report and review of literature. *Int J Oral Maxillofac Surg* 39: 930-933.
6. Shah J, Desai S (2013) Congenital unilateral complete bony syngnathia with non-formation of TM joint and zygomatic arch disruption, Presenting at 11 years of age. *J Cranio-Maxillary Dis* 2: 59-61.
7. Naikmasur VG, Sattur AP, Joshi SK, Rai A (2010) Congenital syngnathia: case report and review of literature. *Cleft Palate Craniofac J* 47: 654-660.
8. Turksen Z, Ozakpinar HR, Tellioglu AT (2012) A case of syngnathia, cleft palate and hypospadias: an isolated case or syndromic syngnathism? *J Craniomaxillofac Surg* 40: 8-10.
9. Vahidi K, Joe BN, Glenn OA, Barkovich AJ, Filly R, et al. (2007) Prenatal imaging of congenital maxillomandibular fusion: case report and review of the literature. *J Ultrasound Med* 26: 381-389.