

Available online at [www.sciencedirect.com](http://www.sciencedirect.com)

# Resuscitation Plus

journal homepage: [www.journals.elsevier.com/resuscitation-plus](http://www.journals.elsevier.com/resuscitation-plus)

## Case report

# Initial resuscitation of a neonate with congenital maxillomandibular syngnathia: A case report and literature review



Samantha J. DeMarsh<sup>a,\*</sup>, Zubin S. Shah<sup>b</sup>, Ahmed F. Osman<sup>b</sup>

<sup>a</sup> Department of Pediatrics, Nationwide Children's Hospital, 700 Children's Drive, Columbus, OH 43205, USA

<sup>b</sup> Division of Neonatology, The Ohio State University College of Medicine and Nationwide Children's Hospital, 410 W 10<sup>th</sup> Ave, Columbus, OH 43210, USA

## Abstract

We report a full-term male neonate found to have undiagnosed syngnathia requiring extensive resuscitation at birth followed by urgent tracheostomy. We conducted a systematic literature review to study the presentation, resuscitation methods, and outcomes of neonates with congenital syngnathia. Of the 174 cases reported to date, 91 had initial resuscitation data available. Extensive resuscitation was required in 16 of these 91 infants (18%). This ranged from nasal intubation to emergent tracheostomy. These neonates are potentially higher risk deliveries for which methods in addition to those recommended by the American Heart Association neonatal resuscitation guidelines may be needed.

**Keywords:** Case report, Congenital, Syngnathia, Synechia, Resuscitation, Neonate

## Introduction

Syngnathia is the presence of bony (synostotic) or fibrous (synechial) adhesions that create a fixed fusion between the maxilla and mandible with potential involvement of the zygoma,<sup>1–3</sup> tuberosity, hard palate, and/or temporal bone.<sup>1</sup> This is a rare congenital anomaly in neonates with fewer than 175 cases reported in the literature to date.<sup>1–9</sup> These neonates may require extensive resuscitation at birth.<sup>4</sup> It has been reported that up to 37% of these infants can require nasotracheal intubation and up to 32% can require tracheostomy prior to surgical management of the syngnathia.<sup>2</sup> Here we present the case of a full-term neonate with undiagnosed syngnathia who presented with respiratory depression requiring extensive resuscitation at birth. Consent for publication was obtained from both parents, and exemption from ethics committee review was obtained from our Institutional Review Board. Additionally, we summarize previously

reported birth presentations and resuscitation requirements that may provide anticipatory guidance for future cases.

## Case presentation

The neonatal team was called to attend a cesarean section delivery of a 30-year-old primigravid mother at 37 weeks gestation. While fetal heart tracings were reassuring, cesarean delivery was indicated for abnormal uterine artery doppler. The pregnancy was complicated by polyhydramnios, intrauterine growth restriction, and multiple abnormal ultrasound findings including micrognathia, abnormal shape of both hands, micropenis, ventricular septal defect, and hyperechoic kidneys. Amniocentesis with genetic testing resulted as 46, XY with normal single nucleotide polymorphism microarray. Maternal serologies were negative for human immunodeficiency virus, hepatitis B, and syphilis, and she was rubella immune. The parents were not

**Abbreviations:** CPAP, continuous positive airway pressure; CT, computed tomography; EXIT, ex utero intrapartum treatment; FiO<sub>2</sub>, fraction of inspiratory oxygen; IPPV, intermittent positive pressure ventilation; NICU, neonatal intensive care unit; TMJ, temporomandibular joint.

\* Corresponding author at: Pediatric Residency Program, Nationwide Children's Hospital, 700 Children's Dr., Columbus, OH 43205, USA.

E-mail addresses: [Samantha.demارش@nationwidechildrens.org](mailto:Samantha.demارش@nationwidechildrens.org) (S.J. DeMarsh), [Zubin.shah@nationwidechildrens.org](mailto:Zubin.shah@nationwidechildrens.org) (Z.S. Shah),

[ahmed.osman@nationwidechildrens.org](mailto:ahmed.osman@nationwidechildrens.org) (A.F. Osman).

<http://dx.doi.org/10.1016/j.resplu.2021.100080>

Received 8 November 2020; Received in revised form 1 January 2021; Accepted 8 January 2021

2666-5204/© 2021 The Authors. Published by Elsevier B.V. This is an open access article under the CC BY-NC-ND license (<http://creativecommons.org/licenses/by-nc-nd/4.0/>).

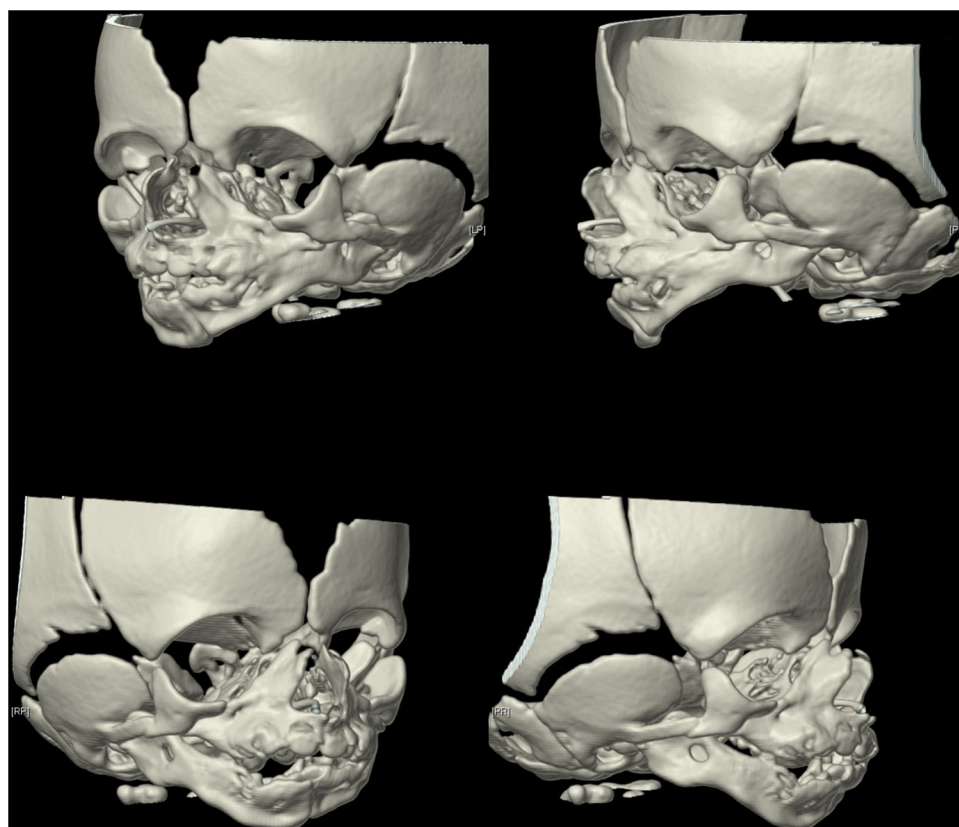


**Fig. 1 – Visualization of maxillo-mandibular fusion with presence of jaw syngnathia.**

consanguineous, and mother had no known exposure to teratogenic agents.

At delivery, the baby was limp and apneic. He was immediately brought to the radiant warmer, dried, stimulated, and placed in “sniffing” position for nasal bulb suctioning. Oral suctioning was attempted but unsuccessful as the mouth was found to be fused (Fig. 1). His 1-minute heart rate was 80 beats per minute, and he continued to have poor respiratory effort. Intermittent positive pressure ventilation (IPPV) was administered using a T-piece resuscitator via face mask with a maximum fraction of inspiratory oxygen ( $\text{FiO}_2$ ) concentration of 60%. Effective ventilation was achieved with rising heart rate, adequate chest rise, and improving muscle tone. While a nasopharyngeal airway was considered, it was

not required as the infant’s condition improved. He established spontaneous regular breathing by 8 min of life. Support was changed to continuous positive airway pressure of +6  $\text{cmH}_2\text{O}$  and  $\text{FiO}_2$  was gradually weaned to 30%. Apgar scores of 2 and 7 were assigned for 1 and 5 min of life, respectively. Initial glucose was 63  $\text{mg/dL}$  and cord blood gases showed no significant metabolic acidosis or hypercarbia. Birth weight was 1840 g (<3<sup>rd</sup> percentile), length was 40.9 cm (<3<sup>rd</sup> percentile), and head circumference was 29.5 cm (<3<sup>rd</sup> percentile). Physical examination was significant for large open anterior and posterior fontanelles with splayed sagittal suture, micrognathia, partial syndactyly and overlapping fingers of both hands, no testicles palpable in the scrotum, and micropenis. He was briefly admitted to the neonatal intensive care unit (NICU) at the delivery hospital prior to



**Fig. 2 – CT reconstruction of skull demonstrating fusion of the left temporomandibular joint (TMJ) and less extensive fusion of the right TMJ.**

transport to a quaternary care children's hospital NICU for further evaluation.

Upon arrival to the level IV NICU, he underwent urgent operative laryngoscopy with fiberoptic intubation followed by tracheostomy and placement of tracheostomy tube. The patient remained stable on ventilator support without significant physiologic derangements. Feeding was initiated via nasogastric tube and advanced towards goal. Computerized tomography (CT) scan on the eighth day of life showed: 1. Fusion of lateral aspects of maxillary and mandibular alveoli, 2. Mandibular hypoplasia, 3. Cleft hard palate and likely soft palate with elevation of tongue into inferior nasal cavity, 4. Midline cleft face with widened inferior metopic suture and presence of a cleft through midline nasal bones, and 5. Bilateral cochlear aperture stenosis and ossicular dysplasia (Fig. 2).

At three weeks of life, he underwent operative release of bilateral maxillary and mandibular bony fusion and laparoscopic gastrostomy tube insertion. While inpatient, he received rehabilitation of the temporomandibular joint and optimization of nutrition. He underwent extensive evaluations for neurological, endocrinological, and genetic abnormalities. Whole exome sequencing ultimately revealed a de novo hemizygous single nucleotide deletion in the HDAC8 gene, associated with X-linked Cornelia de Lange syndrome. He was discharged home on tracheostomy tube with cool mist humidification during the day with bilevel positive airway pressure through the night and on full enteral tube feeds.

## Literature review

After consultation with a medical librarian, we conducted a comprehensive literature review to identify the methods of resuscitation at birth for neonates with syngnathia. Articles were retrieved from PubMed database using the following search terms: "congenital syngnathia," "maxillomandibular fusion," "congenital synostosis," and "oral synechiae." We included individual case reports and case series from all years of publication up to May 2020. References and review articles were manually searched for additional case articles not previously included. Results were filtered for those written in English. Cases of acquired syngnathia were excluded. Full texts of all articles meeting inclusion criteria were thoroughly reviewed for data extraction (Supplemental data). We extracted reports of prenatal ultrasound findings, gestational age at birth, birth weight, patient clinical condition at birth including Apgar scores, the resuscitation methods utilized (if any), and survival outcome.

## Results

Our search yielded 132 articles reporting a total of 174 unique cases. Clinical reports ranged in publication dates from 1936 to 2020, with most case reports being published within the past three decades. We extracted perinatal data and resuscitation details when available and calculated median values with interquartile ranges (IQR) where appropriate. Most cases were born at term (38 weeks, IQR 37–40,  $n = 40$ ) with a median birthweight of 2800 g (IQR 2500–3280,  $n = 81$ ). The median Apgar score at 1 and 5 min were 8 (IQR 6–9,  $n = 29$ ) and 9 (IQR 8–9,  $n = 29$ ), respectively. The descriptive condition at birth was reported in 91 (52%) of the 174 cases with extensive resuscitation required for 16 patients (Table 1). These patients tended to have lower

**Table 1 – Most extensive resuscitation method required for patients with congenital syngnathia.**

Extensive birth resuscitation required (N = 16)	n/N (%)
Face mask	2/16 (12.5%)
Nasal intubation	3/16 (18.8%)
Oropharyngeal airway	1/16 (6.3%)
Emergent tracheostomy	7/16 (43.8%)
Not detailed	3/16 (18.8%)

gestational age (37 weeks, IQR 36–39,  $n = 10$ ) and weight (2500 g, 1700–3000,  $n = 13$ ) at birth.

## Discussion

It is known that 4–10% of all infants will require resuscitation beyond the initial steps of warming, drying, and stimulating.<sup>6</sup> For syngnathia cases with reported conditions at birth, 18% required extensive resuscitation. Reports without reported condition at birth may indicate patients brought to medical attention later in life, although details were extremely limited in some cases. Publication bias may underestimate the need for resuscitation in these patients, as demise soon after delivery may have gone underreported. The nature of resuscitation of neonates with syngnathia was often unconventional with almost half of the patients that received extensive resuscitation requiring emergent tracheostomy placement ( $n = 7$ ). The remainder were successfully resuscitated with face mask or advanced airway. The most frequently utilized airway (other than surgical tracheostomy) was nasal intubation. Each of these patients survived their respective resuscitation. Only one patient was reported to have died on day four of life.

Prenatal ultrasound results were provided 14 (8%) of the 174 cases. The most common abnormalities visualized were micrognathia and polyhydramnios (Table 2). Micrognathia was reported in over a third of prenatal ultrasounds ( $n = 5$ ) when results were available. Polyhydramnios was reported in over a quarter of ultrasounds compared to a detection rate of 0.2–1.6% in the general population.<sup>7</sup> A prenatal sonographic detection rate of 1 in 1500 of pregnancies in the general population has been reported, but a majority may not be clinically significant postnatally.<sup>1,8</sup> Consensus guidelines to standardize the prenatal diagnosis of micrognathia using inferior facial angle and jaw index have been published.<sup>9</sup> However, the use of such guidelines was not reported in these patients. Fetal stomach fluid volume and movement of fluid in the fetal trachea with diaphragmatic

**Table 2 – Abnormal prenatal ultrasound findings in patients with congenital syngnathia.**

Findings on prenatal ultrasound (N = 14)	n/N (%)
Micrognathia	5/14 (35.7%)
Polyhydramnios	4/14 (28.6%)
Both micrognathia and polyhydramnios	2/14 (14.3%)
Additional abnormalities	6/14 (42.9%)

movement were also not reported. Use of such standardized measurements can assist in prognostic evaluation, decisions to pursue advanced imaging, and planning for postnatal management.

Given this data, we suggest the following approach for resuscitating infants with obstructive airway anomalies to be used in conjunction with the American Heart Association Neonatal Resuscitation Guidelines.<sup>6</sup> Each step below should be trialed in stepwise fashion.

- 1 Consult with an appropriately skilled anesthetist and pediatric otolaryngologist for delivery planning, including consideration of ex utero intrapartum treatment (EXIT) procedures in select cases.
- 2 Reposition as needed with trial of prone or side-lying position if there is significant retrognathia.
- 3 Provide IPPV using face mask.
- 4 Use oropharyngeal airway or laryngeal mask airway if able. Otherwise, insert a nasopharyngeal airway.
- 5 Attempt blind nasotracheal intubation while contacting on-call otolaryngologist for fiberoptic intubation (if available).
- 6 Utilize emergent tracheostomy as last option.

## Conclusion

Findings of both micro-retrognathia and polyhydramnios on prenatal ultrasound may serve as the first indication of obstructive airway malformations, including the rare diagnosis of congenital syngnathia. While most patients with syngnathia may not need significant resuscitative efforts, some may require extensive measures and unconventional approaches for facilitating adequate ventilation. The approach to resuscitation of these patients should prioritize multidisciplinary delivery planning, airway clearance, appropriate positioning, and providing IPPV via facemask before considering an advanced airway.

## Contributor's statement

Dr. DeMarsh performed literature review, collected data from individual case reports, analyzed data, drafted the initial manuscript, reviewed and revised the final manuscript.

Dr. Shah analyzed data, drafted the initial manuscript, reviewed and critically revised the final manuscript for important intellectual content.

Dr. Osman conceptualized the study, performed literature review, reviewed and critically revised the final manuscript for important intellectual content.

All authors approved the final manuscript as submitted and agree to be accountable for aspects of the work.

## Declaration of interests

None.

## Funding

This research did not receive any specific grant from funding agencies in the public, commercial, or not-for-profit sectors. No sponsors were involved in the study design, collection, analysis, or interpretation of data, in the writing of the report, or the decision to submit for publication.

## Acknowledgments

Thank you to the Ohio State University Division of Neonatology and Nationwide Children's Pediatric Surgery and Otolaryngology departments for their exquisite care of this patient and clinical expertise. Additional thanks to the medical library staff for their efforts in locating the articles reviewed.

## Appendix A. Supplementary data

Supplementary material related to this article can be found, in the online version, at doi:<https://doi.org/10.1016/j.resplu.2021.100080>.

## REFERENCES

1. Kumar V, Rattan V, Rai S. Congenital maxillomandibular syngnathia: review of literature and proposed new classification system. *J Maxillofac Oral Surg* 2019.
2. Olusanya AA, Akadiri OA. Congenital syngnathia: review of clinical profile and proposal of a new classification-based management protocol. *Int J Oral Max Surg* 2020;49:505–14.
3. Trigg DJ, Mau IT, Rosbe KW. Complete bony syngnathia: report of a case and review. *Arch Otolaryngol Head Neck Surg* 2007;133:187–90.
4. Rao S, Oak S, Wagh M, Kulkarni B. Congenital midline palatomandibular bony fusion with a mandibular cleft and a bifid tongue. *Br J Plast Surg* 1997;50:139–41.
5. Ferre C, Callaghan W, Olson C, Sharma A, Barfield W. Effects of maternal age and age-specific preterm birth rates on overall preterm birth rates—United States, 2007 and 2014. *MMWR Morb Mortal Wkly Rep* 2016;65:1181–4, doi:<http://dx.doi.org/10.15585/mmwr.mm6543a1>.
6. Weiner GM, Zaichkin J, Kattwinkel J, American Academy of Pediatrics, American Heart Association. Textbook of neonatal resuscitation. 7th edition Elk Grove Village, IL: American Academy of Pediatrics; 2016.
7. Hamza A, Herr D, Solomayer EF, Meyberg-Solomayer G. Polyhydramnios: causes, diagnosis and therapy. *Geburtshilfe Frauenheilkd* 2013;73:1241–6.
8. The Fetal Medicine Foundation. Micrognathia. <https://fetalmedicine.org/education/fetal-abnormalities/face/micrognathia>. Accessed 23 September 2020.
9. Puricelli MD, Rahbar R, Allen GC, et al. International Pediatric Otolaryngology Group (IPOG): consensus recommendations on the prenatal and perinatal management of anticipated airway obstruction. *Int J Pediatr Otorhinolaryngol* 2020;138:110281, doi:<http://dx.doi.org/10.1016/j.ijporl.2020.110281>.