CLINICAL VIGNETTE

Goldenhar Syndrome in a Pediatric Patient Undergoing ENT Surgery and Its Anesthetic Considerations

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Introduction

Goldenhar syndrome (GS) refers to a congenital disorder characterized primarily by a wide range of craniofacial abnormalities. Incidence has been cited from 1 in 5,600 to 1 in 150,000.[1] Common craniofacial manifestations of GS include asymmetrical mandibular hypoplasia and auricular deficits. Additionally, extra-craniofacial abnormalities are seen frequently and most often involve the skeletal and circulatory systems.[2–5] GS has also been referenced, in the literature as craniofacial microsomia (CFM), hemifacial microsomia, and oculoauriculovertebral spectrum to name a few examples.[1,2] Although GS is seen as a specific variant of CFM with features most notable for epibulbar dermoids and vertebral abnormalities, there are no clear diagnostic criteria and patients are clinically indistinguishable from those with other forms of CFM.[6]

Patients with GS often require surgical intervention either for functional or aesthetic reasons, with many requiring a multistage treatment plan involving successive surgeries early in their lives.[2,7] Given the extensive surgical requirement and unique anatomic differences in this patient population, there are many potential anesthetic considerations. We present a case of a patient with GS who underwent general anesthesia for a mastoidectomy at our institution.

Case

A 7-year-old male with GS presented for a scheduled mastoidectomy for acute-on-chronic left ear mastoiditis. Pertinent medical history included left microtia, hearing loss, mandibular hypoplasia, scoliosis, and possible obstructive sleep apnea. The patient had a history of bilateral epibulbar cysts and periauricular tags that were excised in the past, as well as pulmonary stenosis that had previously resolved. He otherwise met developmental milestones and had an adequate exercise capacity.

On the day of surgery, the patient's vital signs were within normal limits. Physical exam was notable for multiple craniofacial abnormalities including left ear hypoplasia, blind pouch in both left and right external auditory canals, and left eye medial deviation. Preoperative airway exam demonstrated adequate mouth opening, a Mallampati class II airway, and full cervical neck range of motion. Cardiac and respiratory examination were unremarkable.

Prior to surgery, the patient was premedicated with oral midazolam. After arrival to the operating room, the patient was placed on standard anesthesia monitors. Inhalation induction was achieved with a combination of nitrous oxide and sevoflurane. There was no difficulty with mask induction and ventilation. An 18-gauge peripheral IV was placed in the left hand after inhalational induction. Propofol and fentanyl were given via the peripheral IV to deepen the anesthesia prior to intubation. A grade 1 Cormack-Lehane view was obtained under direct laryngoscopy with a Macintosh size 2 blade, and a 5.5 cm cuffed endotracheal tube (ETT) was successfully inserted and secured. Anesthesia was maintained with sevoflurane for the duration of the 3-hour case. The patient's vital signs remained stable throughout surgery. He was extubated awake at the end of the surgery and was transferred to the post-anesthesia care unit (PACU) for further monitoring. The patient recovered well in the PACU and was later discharged home with his parents.

Discussion

Patients with Goldenhar syndrome have complex co-morbidities and a number of anatomic and functional changes. Therefore, there are several considerations to providing safe and effective anesthesia care in the peri-operative setting.

<u>Airway and Skeletal:</u>

GS patients are at an increased risk of difficulty with ventilation. Mandibular deficiency has been noted in upwards of 89% of patients with CFM.[2] Although our patient had left-sided mandibular hypoplasia noted on a past CT scan, it was not severe enough to affect mask induction and ventilation. Another challenge of ventilation is the risk of upper airway obstruction considering the high incidence of mandibular retrusion and facial muscle hypoplasia.[2] Although awake intubation has been performed successfully in patients with GS,[8] this may not be suitable for the younger pediatric population, which comprises the majority of GS surgical patients. Thus, anesthesiologists should be adequately prepared for possible difficult mask ventilation and any potential difficult intubation in these patients.

A secure airway with an endotracheal intubation is an important consideration for airway management and protection in patients with GS. The anesthesiologist should plan accordingly and

consider various instrumental adjuncts and alternatives to routine direct laryngoscopy (DL) as approximately 40% of intubations were noted to be difficult in one study.[9] Although there have been cases of successful intraoperative ventilation solely with supraglottic airways,[10,11] the benefits should be weighed against the risk of aspiration especially with the types of corrective oral and maxillofacial surgeries that this patient population will likely receive early in their lifetime. Supraglottic airway devices have been useful mediums to facilitate endotracheal intubation after unsuccessful intubation with DL in patients with GS.[12,13] Nasal intubation has been successfully utilized especially when the anatomy of the oropharyngeal airway makes it difficult for an oral intubation.[14,15] Video laryngoscopy[16] and flexible scope intubation[17] are also uniquely valuable for patients with GS due to the prevalence of oropharyngeal and vertebral abnormalities.

Given that vertebral malformations are the most common extra-craniofacial abnormality observed in GS patients,[2,5] the cervical neck range of motion should be thoroughly assessed in all GS patients. Particularly, the cervical spine has been affected in GS, with torticollis one of the most common conditions.[3]

Additionally, obstructive sleep apnea (OSA) has been noted in up to 17% of patients with CFM.[18] In our case, the patient could have undiagnosed OSA which was suspected because of an apneic event in a previous post-operative period. Thus, GS patients should be meticulously monitored until they are adequately alert and comfortably breathing on room air in the post-anesthesia care unit (PACU).

Although rare, lower airway malformations can be associated with GS.[3] A case report discussed airway management of a GS patient with tracheoesophageal fistula. The ETT had to be placed distal to the fistula to prevent insufflation of the stomach.[19] Pulmonary malformations in these patients may not manifest in symptoms, nor are they definitively seen on routine imaging. Another case report discussed failure to ventilate after apparent successful endotracheal intubation that led to subsequent discovery of a tracheal bronchus in the GS patient.[20] Although pulmonary malformations are one of the more rare extra-craniofacial abnormalities of GS, they should be considered in the differential diagnosis for abnormal ventilation in this patient population.

Of note, most procedures to improve the airway are performed within 6 months of birth in GS patients; however, airway management may also become more challenging with age.[3] Therefore, even if there were no complications with securing the airway in a GS patient in the past, the anesthesiologist should still be carefully prepared for the possibility of a difficult airway for future procedures.

Cardiac:

Cardiac defects are a common extra-craniofacial abnormality in GS patients, with an incidence of up to one third reported in one study.[2] Cardiology consultation is essential in the neonatal period, with tetralogy of Fallot and ventricular septal defects being some of the most common cardiovascular malformations observed in this population.[21,22] Cardiovascular defects should be routinely followed and considered for pre-operative evaluation. In our case, the patient had a peripheral pulmonary stenosis diagnosed in the past but was no longer observed during the more recent cardiac evaluation. When preparing for the anesthetic management of GS patients with suspected cardiovascular abnormalities, the myocardial depressant effects of commonly used anesthetic agents should be considered and vasoactive medications and additional hemodynamic monitoring should be readily available.

Conclusion

Although GS is a rare congenital disorder, patients with this condition will present early and often to the anesthesiologist. The wide phenotype and lack of clear diagnostic criteria of this syndrome underscores the importance of a thorough pre-operative evaluation. We have presented the anesthetic management of a 7-year-old child with GS. Although his operative course was uncomplicated, there are many anesthetic considerations, especially focused on airway management, that had to be cautiously considered and prepared. Anesthesiology providers should prudently review these potential challenges when caring for patients with GS or other forms of craniofacial microsomia.

REFERENCES

1. **Gabbett MT**. The oculoauriculovertebral spectrum: Refining the estimate of birth prevalence. *J Pediatr Genet*. 2012 Jun;1(2):71-7. doi: 10.3233/PGE-2012-014. PMID: 27625806; PMCID: PMC5020924.
2. **Brandstetter KA, Patel KG**. Craniofacial Microsomia. *Facial Plast Surg Clin North Am*. 2016 Nov;24(4):495-515. doi: 10.1016/j.fsc.2016.06.006. PMID: 27712817.
3. **Bogusiak K, Puch A, Arkuszewski P**. Goldenhar syndrome: current perspectives. *World J Pediatr*. 2017 Oct;13(5):405-415. doi: 10.1007/s12519-017-0048-z. Epub 2017 Jun 15. PMID: 28623555.
4. **Horgan JE, Padwa BL, LaBrie RA, Mulliken JB**. OMENS-Plus: analysis of craniofacial and extracraniofacial anomalies in hemifacial microsomia. *Cleft Palate Craniofac J*. 1995 Sep;32(5):405-12. doi: 10.1597/1545-1569_1995_032_0405_opaoca_2.3.co_2. PMID: 7578205.
5. **Renkema RW, Caron CJJM, Pauws E, Wolvius EB, Schipper JAM, Rooijers W, Dunaway DJ, Forrest CR, Padwa BL, Koudstaal MJ**. Extracraniofacial anomalies in craniofacial microsomia: retrospective analysis of 991 patients. *Int J Oral Maxillofac Surg*. 2019 Sep;48(9):1169-1176. doi: 10.1016/j.ijom.2019.01.031. Epub 2019 Mar 13. PMID: 30878275.
6. **Tuin J, Tahiri Y, Paliga JT, Taylor JA, Bartlett SP**. Distinguishing Goldenhar Syndrome from Craniofacial Microsomia. *J Craniofac Surg*. 2015 Sep;26(6):1887-92. doi: 10.1097/SCS.0000000000002017. PMID: 26267577.

7. **Pluijmers BI, Caron CJJM, van de Lande LS, Schaal S, Mathijssen IM, Wolvius EB, Bulstrode N, Evans RD, Padwa BL, Koudstaal MJ, Dunaway DJ**. Surgical Correction of Craniofacial Microsomia: Evaluation of Interventions in 565 Patients at Three Major Craniofacial Units. *Plast Reconstr Surg*. 2019 May;143(5):1467-1476. doi: 10.1097/PRS.0000000000005554. PMID: 31033829.
8. **Sahni N, Bhatia N**. Successful management of difficult airway in an adult patient of Goldenhar syndrome. *Saudi J Anaesth*. 2014 Nov;8(Suppl 1):S98-S100. doi: 10.4103/1658-354X.144086. PMID: 25538532; PMCID: PMC4268539.
9. **Hirata M, Miwa T, Ono K, Namatame R, Hiroki K**. [Analysis of difficult tracheal intubation in patients with first and second brachial arch syndrome]. *Masui*. 2001 Sep;50(9):986-90. Japanese. PMID: 11593721.
10. **Khan WA, Salim B, Khan AA, Chughtai S**. Anaesthetic Management in a Child with Goldenhar Syndrome. *J Coll Physicians Surg Pak*. 2017 Mar;27(3):S6-S7. PMID: 28302228.
11. **Sukhupragarn W, Rosenblatt WH**. Airway management in a patient with Goldenhar syndrome: a case report. *J Clin Anesth*. 2008 May;20(3):214-7. doi: 10.1016/j.jclinane. 2007.09.020. PMID: 18502367.
12. **Kim YL, Seo DM, Shim KS, Kim EJ, Lee JH, Lee SG, Ban JS**. Successful tracheal intubation using fiberoptic bronchoscope via an I-gel™ supraglottic airway in a pediatric patient with Goldenhar syndrome -A case report-. *Korean J Anesthesiol*. 2013 Jul;65(1):61-5. doi: 10.4097/kjae.2013.65.1.61. Epub 2013 Jul 19. PMID: 23904941; PMCID: PMC3726849.
13. **Camkiran A, Pirat A, Akovali NV, Arslan G**. Combination of laryngeal mask airway and pediatric Boussignac bougie for difficult tracheal intubation in a newborn with Goldenhar syndrome. *Anesth Analg*. 2012 Sep;115(3):737-8. doi: 10.1213/ANE.0b013e31825d0a18. PMID: 22910640.
14. **Madan R, Trikha A, Venkataraman RK, Batra R, Kalia P**. Goldenhar's syndrome: an analysis of anaesthetic management. A retrospective study of seventeen cases. *Anaesthesia*. 1990 Jan;45(1):49-52. doi: 10.1111/j.1365-2044.1990.tb14505.x. Erratum in: *Anaesthesia* 1990 May;45(5):424. PMID: 2316841.
15. **Kaji J, Higashi M, Sakaguchi Y, Maki J, Kai T, Yamaura K, Hoka S**. [Fiberoptic intubation using two tracheal tubes for a child with Goldenhar syndrome]. *Masui*. 2010 Dec;59(12):1526-8. Japanese. PMID: 21229697.
16. **Milne AD, Dower AM, Hackmann T**. Airway management using the pediatric GlideScope in a child with Goldenhar syndrome and atypical plasma cholinesterase. *Paediatr Anaesth*. 2007 May;17(5):484-7. doi: 10.1111/j.1460-9592.2006.02149.x. PMID: 17474957.
17. **Char DS, Gipp M, Boltz MG, Williams GD**. Case report: airway and concurrent hemodynamic management in a neonate with oculo-auriculo-vertebral (Goldenhar) syndrome, severe cervical scoliosis, interrupted aortic arch, multiple ventricular septal defects, and an unstable cervical spine. *Paediatr Anaesth*. 2012 Sep;22(9):932-4. doi: 10.1111/j.1460-9592.2012.03915.x. PMID: 22834469.
18. **Caron CJJM, Pluijmers BI, Maas BDPJ, Klazen YP, Katz ES, Abel F, van der Schroeff MP, Mathijssen IMJ, Dunaway DJ, Mills C, Gill DS, Bulstrode N, Padwa BL, Wolvius EB, Joosten KFM, Koudstaal MJ**. Obstructive sleep apnoea in craniofacial microsomia: analysis of 755 patients. *Int J Oral Maxillofac Surg*. 2017 Oct;46(10): 1330-1337. doi: 10.1016/j.ijom.2017.05.020. Epub 2017 Jun 19. PMID: 28641899.
19. **Guerrero-Domínguez R, López-Herrera-Rodríguez D, Benítez-Linero I, Ontanilla A**. Anesthetic management for surgery of esophagus atresia in a newborn with Goldenhar's syndrome. *Braz J Anesthesiol*. 2015 Jul-Aug;65(4):298-301. doi: 10.1016/j.bjane.2013.07.011. Epub 2015 Apr 28. PMID: 26123147.
20. **Hasham F, van Helmond N, Sidlow R**. Anaesthesia and orphan disease: Difficult ventilation following intubation in Goldenhar syndrome. *Eur J Anaesthesiol*. 2017 Mar; 34(3):181-183. doi: 10.1097/EJA.0000000000000556. PMID: 28141737.
21. **Choudhury M, Kapoor PM**. Goldenhar syndrome: Cardiac anesthesiologist's perspective. *Ann Card Anaesth*. 2017 Jan;20(Supplement):S61-S66. doi: 10.4103/0971-9784.197802. PMID: 28074825; PMCID: PMC5299831.
22. **Digilio MC, Calzolari F, Capolino R, Toscano A, Sarkozy A, de Zorzi A, Dallapiccola B, Marino B**. Congenital heart defects in patients with oculo-auriculo-vertebral spectrum (Goldenhar syndrome). *Am J Med Genet A*. 2008 Jul 15;146A(14):1815-9. doi: 10.1002/ajmg.a.32407. PMID: 18553555.