

Goldenhar Syndrome: Airway and Anesthetic Management - A Case Report

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ABSTRACT

A 10-year-old female child presented to ophthalmic outpatient department with history of decreased vision and painless swelling of left eye since birth. Clinical examination of left eye revealed an infero-lateral limbal dermoid with normal fundoscopic examination. There were multiple preauricular appendages in front of the left ear. Both ears were normal in all other respects. The X-ray of cervical spine showed fused third to fifth cervical vertebrae with loss of intervertebral disc but clinical examination found no neurological deficits. X-ray of occipital-mental view found hypoplasia of left maxilla with normally appearing mandible. Clinical examination of the airway revealed restricted head and neck movements with Mallampatti class II which reliably predicted difficult laryngoscopy and intubation.

The history, clinical examination of vertebral, auricular and ocular regions, and radiological evaluation confirmed her to be a case of Goldenhar syndrome. A difficult airway continues to be a major cause of anesthesia-related morbidity and mortality and fiberoptic intubation of the spontaneously breathing patients is the technique of choice for elective management of a difficult airway. Therefore, fiberoptic intubation under sedation was planned for the surgery. The intraoperative and postoperative period was uneventful. Lack of anesthesia-related and surgical complications encouraged us to present the advantage of fiberoptic intubation under sedation for successful management of predicted difficult airway.

Key words: Goldenhar syndrome; Oculo-Auriculo-Vertebral spectrum (OAV); Craniofacial deformity; Fiberoptic intubation

Conflicting Interest:
None declared

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CASE REPORT

Goldenhar syndrome, also known as oculo-auriculo-vertebral dysplasia, is a rare congenital disorder. This syndrome with malformations of face, eyes and ears was first recorded by the nineteenth-century German physician Carl Ferdinand von Arlt (1812-1887) but was first described by Dr. Maurice Goldenhar in 1952 [1]. The syndrome occurs randomly, with no apparent cause. Most cases of Goldenhar syndrome are not inherited, however few families show unclear inheritance pattern. The male to female ratio is 2:1 and its occurrence is estimated from 1 in every 3,000 to 1 in every 5,000 live births. This syndrome mostly involves face, ears, eyes and vertebrae and is characterized by a wide spectrum of symptoms and physical features that range from mild to severe and is often associated

with difficult intubation due to characteristic asymmetrical malformations of the face. Usually children with Goldenhar syndrome have normal life span [1-4].

CASE HISTORY

A 10-year-old female child presented to the ophthalmic outpatient department of our hospital with history of decreased vision and painless swelling of the left eye since birth. On ophthalmic examination, visual acuity in the right eye was 6/6 but best corrected visual acuity in the left eye was 6/60 which did not improve with refraction. There was inferolateral limbal dermoid of about 5x5 mm size with 30 to 35 degree divergent squint of left eye (Figure 1). Fundoscopic examination and intraocular pressure were normal in both eyes but extra-

ocular movements of the left eye were restricted. The physical examination showed facial asymmetry due to under development of left maxilla and normally appearing mandible. Airway evaluation showed Mallampatti class II with restricted movements of head and neck, which suggested possibility of difficult laryngoscopy and intubation. On ear examination, the child had multiple preauricular skin tags in front of left ear, lying in line joining tragus and angle of mouth (Figure 2). The external ears were normal in shape and size. Her hearing was normal. Cardiovascular and central nervous system examinations were normal with no other associated congenital anomaly. Lateral view of the cervical spine X-ray showed fused third to fifth cervical vertebral bodies with loss of intervertebral disc (Figure 3). X-ray chest, skull and computed tomography of head were normal. Haemogram, bleeding time, and clotting time were within normal limits. Fenigold and Baun criteria were used to diagnose her as a case of Goldenhar syndrome as she had pre-auricular appendages on left side, infero-lateral limbal dermoid of left eye and vertebral anomalies.

Airway and Anesthetic Technique

In preanesthetic assessment, she was evaluated for airway by mouth opening, Mallampatti classification which was MP-II, thyromental distance, and neck movement. Nasal patency was checked for both nostrils. The fiberoptic intubation technique under sedation was selected for airway management during surgery. A written informed parental consent was taken after discussing risks and management of difficult airway with parents and ophthalmic surgeon. Local anesthetic, lignocaine sensitivity test was done. The 'difficult airway cart' with percutaneous tracheostomy set, was kept ready. The intravenous infusion of ringer lactate was started at 6 to 8 mL kg⁻¹ and standard monitors for heart rate, systemic arterial blood pressure, pulse oximetry and oxygen saturation (SpO₂), electrocardiography (ECG), capnography and temperature were attached. The topical anesthesia was achieved by nasal packing with lignocaine 4% and xylometazoline 0.1% nasal drops, instilled in both nostrils 15 minutes prior to procedure. The pharynx was sprayed with 4 to 6 puffs of lignocaine 10% aerosol. She was premedicated with intravenous glycopyrrolate (.01mg kg⁻¹), fentanyl (1µg kg⁻¹) and midazolam (.05mg kg⁻¹). After preoxygenation with 100% oxygen for three minutes, fiberoptic intubation (Pentax -PMS, FI 10P2, Pentax Corporation, Medical instrument division, Japan) was performed under sedation, by nasal route with 5.5 mm portex cuffed endotracheal tube. During procedure the sedation was supplemented with fentanyl. The correct positioning of tracheal tube was confirmed with capnograph and was firmly secured after confirming the equal bilateral air entry by auscultation. Anesthesia was induced with intravenous propofol (1%) in a dose of 2 mg kg⁻¹,

sufficient to abolish the eyelash reflex and was maintained with vecuronium .08 mg kg⁻¹, halothane 0.5-1% and nitrous oxide 60% in oxygen on controlled ventilation via pediatric Bain's circuit. The surgical procedure was smooth, uneventful and lasted for 60 minutes. Residual neuromuscular blockade was reversed with neostigmine 0.05mg kg⁻¹ and glycopyrrolate. She was extubated, when fully awake and breathing spontaneously with adequate tidal volume. The postoperative period was also uneventful.

DISCUSSION

Goldenhar syndrome is a rare congenital disorder and consists of ocular, auricular and skeletal anomalies with variable presentation. Although, in most of cases, such malformations affect one side of the body (hemifacial microsomia) but in 10 to 33% of cases, it may be bilateral. Ocular abnormalities include epibulbar dermoids and lipodermoids, coloboma, microphthalmia, palpebral fissures, blepharophimosis, strabismus, vision defects including diplopia of various degrees and/or other eye abnormalities and are seen in 60% of cases. Amongst ocular features, epibulbar dermoids are the commonest (75%) and are classically located in the inferio-temporal quadrant. Among auricular afflictions, preauricular skin tags and accessory are common. Hearing defect of various degrees from near normal to severe hearing loss (conductive type) may occur. Involvement of axial skeleton (vertebrae and ribs) has been observed in 24% of the patients. The spina bifida is the commonest and least severe of all anomalies. Craniofacial abnormalities may include malar hypoplasia, maxillary, mandibular and temporal hypoplasia, macrostomia, cleft lip and/or palate. Many affected individuals may have additional skeletal, neurological, cardiac, pulmonary, renal, and/or gastrointestinal abnormalities including feeding difficulty [2-4]. Feingold and Baun listed criteria for Goldenhar syndrome, of which at least two are required for the diagnosis of the syndrome [5]. These included eye abnormalities such as lipoma, lipo-dermoid, epibulbar dermoid, in association with ear, mandible or vertebral anomalies.

Genetic Profile

Goldenhar syndrome is caused by disruption of normal facial development which is formed between eighth and twelfth weeks of intrauterine gestational life of pregnancy. Its etiology may be environmental or due to certain medication taken during pregnancy. Some cases of positive family history suggested autosomal dominant or recessive inheritance. There may be interaction of many genes possibly in combination with environmental factors (multifactorial inheritance) [6].

Our patient showed inferolateral limbal dermoid of left eye with 30-35 degree divergent squint and restricted extra ocular movements on abduction, preauricular skin tags, maxillary hypoplasia of left side and fused bodies of third to fifth cervical vertebrae with loss of intervertebral discs, thus fulfilled requisite criteria (oculo-auriculo-vertebral spectrum) for diagnosis of Goldenhar syndrome (Figure 1-3).

The presence of mandibular abnormalities have 100% sensitivity and 96% specificity for predicting difficult laryngoscopy. As the number of associated craniofacial anomalies of Goldenhar syndrome increases, the risk of difficult intubation also increases [7]. The airway and anesthetic management of the case presented was challenging as the preoperative assessment of her airway revealed the presence of maxillary hypoplasia, fused cervical vertebrae and limited head and neck movements with Mallampatti class II.

Flexible fiberoptic intubation under local anesthesia with sedation is the technique of choice for management of the anticipated difficult airway with restricted mouth opening in the patient undergoing an elective procedure. It is generally regarded as a gold standard method for endotracheal intubation in patients with cervical spine instability or immobility[8, 9] Madan *et al* found that intravenous induction was preferable to the gaseous one [10]. Other alternative method for such patients are

lighted stylet guided intubation under general anesthesia. Tracheostomy should be performed only in emergency or when other options failed [8-10]

Treatment of Goldenhar syndrome is usually confined to surgical intervention that may be necessary to allow the child to develop normally e.g. jaw distractions/bone grafts, ocular dermoid debulking, repairing cleft palate/lip, repairing heart malformation and spinal surgery [11, 12] In our patient, excision of limbal dermoid of left eye was performed successfully under general anaesthesia. Surgery was smooth, uneventful and lasted for approximately 60 minutes.

CONCLUSION

Goldenhar syndrome is a rare congenital disorder of unknown etiology, associated with craniofacial vertebral abnormalities and characterized by a wide spectrum of physical features that vary in range and severity. No single airway test can provide a high index of sensitivity and specificity for prediction of difficult airway in patients of Goldenhar syndrome. The airway and anesthetic management for such patients depend on the type, extent and severity of craniofacial-vertebral anomalies, associated cardiovascular problems and nature of surgery. Awareness of this condition will help in diagnosing more of such cases.



Figure 1: Infero-lateral limbal dermoid of the left eye in the Patient



Figure 2: Pre-auricular skin tags



Figure 3: Fused bodies of cervical vertebrae (C3-C5) with loss of intervertebral discs are visible in this X-ray

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