ORIGINAL RESEARCH PAPER
Volume - 12 | Issue - 08 | August - 2023 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

ANAESTHETIC MANAGEMENT OF THE PATIENT WITH BERRY-TREACHER COLLINS AND GOLDENHAR SYNDROME IN PERCUTANEOUS NEPHROLITHOTOMY SURGERY

Anaesthesiology

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ABSTRACT

The Berry-Treacher Collins and Goldenhar syndrome is an autosomal dominant rare craniofacial disorder characterized by bilateral hypoplasia of facial structure and periorbital and ocular and adnexal anomalies, antimongoloid features, palpebral fissures, malar hypoplasia, mandibular hypoplasia, malformation of auricle and pinna, coloboma of an eyelid, conductive deafness, cleft palate and other systemic involvement. The congenital deformity of unilateral anotia and ipsilateral Bell's palsy with other systemic involvement is an uncommon finding of this syndrome. This study aims to discuss the anaesthetic management of 13 years old boy who underwent surgery of percutaneous nephrolithotomy under general anaesthesia for right renal calculi, having narrow and shared airways and facial deformity.

KEYWORDS

Berry-Treacher collins syndrome, Goldenhar syndrome, shared airways, anotia, Bell's palsy, mandibular hypoplasia, percutaneous nephrolithotomy

INTRODUCTION:

Berry-Treacher Collins syndrome and Goldenhar syndrome is an autosomal dominant disorder with variable expression [(1)]. The craniofacial deformity resulted from interference in the development of first and second brachial arches [(1)]. Bell's palsy is a neurological condition caused by upper motor neuron disorder mostly caused by congenital or inherited defects [(1)]. Bell's palsy of developmental origin is found in conjunction with other anomalies including that of the pinna, external auditory canal, laryngeal nerve [(1,2)]. Perinatal trauma, intrauterine distress could be acquired cause of bell's palsy [(2)]. Complete absence of pinna also known as anotia is a rare congenital malformation which may often include partial pinna closure or complete absence of an ear canal [(1)]. The significant difference between the two syndromes is Goldenhar syndrome shares the same Bell's characteristic as Berry-Treacher Collins syndrome but is usually unilateral and asymmetrical in earlier while it is bilateral and symmetrical in later [(1,2)].

Majority of the cases are thought to be derived from new mutation because there is absence of family history of diseases [(3)]. It is suspected to be ribosomopathy which are a genetic abnormality caused by impaired ribosome biogenesis and function resulting in a specific clinical sphenotype [(3)].

Case History:

13 years old boy presented to urology Outpatient Department with complains of nausea, vomiting and pain in the right flank region.

On examination:- slurring of speech, dribbling of saliva, poor oral hygiene, complete absence of the right-sided ear. The child has minimum eyelashes at the lower lids, and hypoplasia of facial bones, especially malar bones, the patient also has mandibular hypoplasia, retrodisplacement of the base of the tongue into the oropharynx, resulting in the obliteration of the airway. The patient was a known case of congenital bell's palsy.

Investigation: -

Ultrasonography: Suggestive of 20 mm renal calculi (partial staghorn calculi) on right side.

***Hearing assessment*:** Right sided conductive hearing loss identified.

***Airway assessment*:** The patient has a narrow airway and a history of snoring. The mouth opening was reduced with Mallampati 3 which anticipates a difficult airway.

***Dental assessment*:** The dental assessment of the patient revealed slurred speech, dribbling of saliva and poor oral hygiene.

The Patient general physical examination revealed respiratory and cardiovascular parameters within normal limits. No murmur and palpable thrill noted.

CT report done in past showed that the bone and cartilage of the right external ear were atretic. The right middle ear ossicle was not seen. The right middle ear cavity was hypoplastic and showed hypodense soft tissue in it with the presence of bony erosion.

13 -year- old patient with Goldenhar syndrome along with narrow airways, and facial deformity anticipates delayed recovery and shared airways would be under very high risk when subjected to anaesthesia.

Figure A & B: Physical characteristics of bell's palsy with unilateral anotia

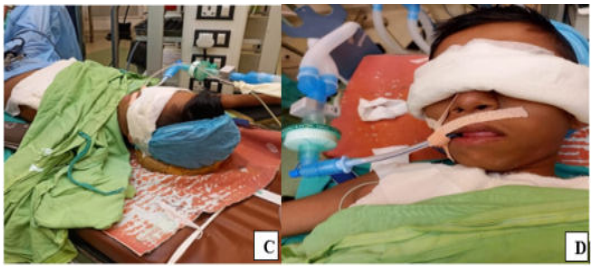

Figure C & D: Intraoperative intubation of patient with difficult airway

Management:

The patient underwent percutaneous nephrolithotomy patient was taken to operation theatre after informed consent and comprehended his parent with complete procedure and benefit and possible risk factors associated with the surgery and added risk due to congenital anomalies. The patient was advised not to eat food or water 8hours before surgery (Nil by mouth). Regular monitors were attached i.e., pulse oximeter, electrocardiography, blood pressure cuff, intravenous access secured with 20-gauge needle, calculated fluid was given considering nil by mouth period. The patient primarily given premedication i.e., injection Midazolam 1 mg, injection Glycopyrrolate 0.2mg, injection Fentanyl 40 mcg. The patient was primarily induced with inhalation agent sevoflurane and inducing agent injection propofol 60mg and the muscle relaxant of choice injection atracurium 15 mg was also administered, after 3 minutes of

the bag and mask ventilation, the airway was secured with 4.5 number cuffed endotracheal tube through direct laryngoscopy technique (cormack lehane grade 2). Throat packing was done to prevent further aspiration, and intraoperatively patient was haemodynamically stable. At the end of surgery, the neuromuscular blocker was reversed adequately by administering neostigmine 1.5 mg and glycopyrrolate 200mcg. Patient was shifted to post anaesthesia care unit after complete reversal and extubation performed after thorough suction. Complete motor power was present in all four limbs, adequate respiration attempt was present. The Patient was closely monitored for about 2 hours in the recovery room.

DISCUSSION:

Managing patients with this syndrome requires a comprehensive preoperative evaluation and anaesthetic management [6].

Availability of different airway devices, with effective communication among the anaesthetist and operating surgeon overcomes the risk of difficult airway[6].

CONCLUSION:

Berry- Treacher Collins and Goldenhar syndrome though a rare entity but associated with difficult airway with facial anomalies. In such a case thorough evaluation and meticulous measures multidisciplinary planning need to be performed before, during and after the surgery to prevent mortality and morbidity. This syndrome represents a significant challenge to anaesthesia providers.

Consent: Written informed consent for publication of the clinical details and clinical images was taken from patient's parents**.**

Conflict of interest: No conflict of interest.

Grant information: No grant involved.

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