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CASE REPORT

Anesthetic Implications in Holt Oram Patients Undergoing Cardiac Procedures. Two Case Reports

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Background Holt-Oram syndrome (HOS) is a rare genetic disorder, first described in 1960, It is characterized by upper limb deformities, congenital heart defects, and conduction abnormalities, posing significant anesthetic management challenges. This report presents two cases of HOS undergoing atrial septal defect (ASD) closure—one through a transcatheter approach and the other via surgical repair.

Case Presentation **Case 1:** A 6-year-old female, weighing 18kg, diagnosed with HOS. The patient underwent transcatheter ASD closure under deep sedation with dexmedetomidine and ketamine. Anesthesia was maintained without complications, and the procedure was completed successfully, with the patient recovering well in the cardiac care unit.
Case 2: A 3.5-year-old female, weighing 17.5kg, with HOS, presented with bilateral upper limb anomalies (absent distal radius, short humerus, absent thumb) and ASD. The patient underwent surgical ASD closure under general anesthesia. The procedure was uneventful, and the patient was successfully weaned from cardiopulmonary bypass, with no residual ASD or arrhythmias observed postoperatively.

Discussion Anesthetic management of HOS requires thorough preoperative assessment, including evaluation of conduction anomalies, chest wall deformities, and airway considerations. Malignant hyperthermia, though reported in isolated cases, was not observed in our patients, despite exposure to potential triggers. Arrhythmias, a common concern in HOS, were anticipated but not exacerbated during the procedures. Special attention was given to femoral arterial cannulation in the surgical case due to upper limb anomalies. The use of TEE was minimized in the transcatheter case to reduce the need for general anesthesia.

Conclusions Anesthetic management in HOS patients undergoing cardiac procedures necessitates a multidisciplinary approach with vigilant monitoring for potential complications. While the risk of arrhythmias and malignant hyperthermia requires careful consideration, our cases suggest that with appropriate planning, these patients can undergo complex cardiac procedures safely.

Keywords ASD closure, Holt-Oram, Skeletal deformity.
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INTRODUCTION

Holt Oram syndrome (HOS) was first described in 1960. Despite having low incidence (1:100,000) [1] it exerts several challenges as regard its anesthetic management including difficult airway, increased risk of arrythmias, restrictive lung disease, difficult cannulation and increase susceptibility to certain anesthetics. Holt-Oram syndrome

is characterized by upper limb deformities, congenital cardiac anomalies, cardiac conduction problems and positive family history [2].

We will discuss two cases of Holt Oram syndrome both were for ASD closure. The first case had transcatheter approach via device and the second case underwent open surgical repair.

Holt Oram syndrome is also known as Heart Hand syndrome, Gladstone and Sybert established a scoring system to assess severity of this syndrome based on cardiac and skeletal affection [3].

However, other systems could be affected like restrictive lung diseases, renal injury, stroke and vascular anomalies [4].

Case Description:

First case was a 6-years-old female patient weighing 18kg diagnosed with Holt Oram syndrome based on skeletal deformity, cardiac finding and family history of mother having also upper limb anomalies and grandfather with upper limb bony deformity and history of conduction anomalies and pacemaker insertion. Features of upper limb anomalies were syndactyly between thumb and index fingers and absent distal radius (Figure 1). While cardiac findings in transthoracic echocardiography (TTE) included atrial septal defect (ASD) secundum 7.3*6mm with left to right shunt.

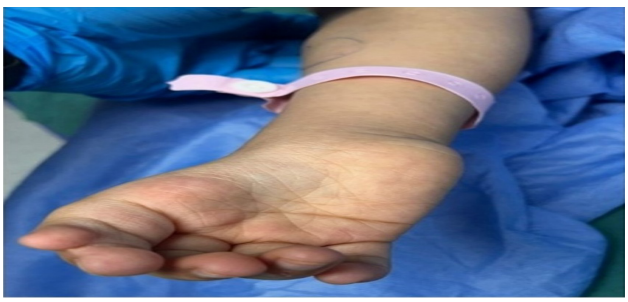

Figure 1: Syndactyly involving the thumb and index finger, along with other upper limb bony deformities, in a patient undergoing transcatheter atrial septal defect closure with a device, which highlights a key clinical feature of Holt-Oram syndrome. The diagnosis is further supported by the presence of associated cardiac anomalies and a positive family history.

Patient was planned for transcatheter ASD closure via device. Anesthetic plan was for deep sedation, 35mg ketamine was administered intravenously (IV) in previously inserted peripheral cannula and then basic monitor attached including noninvasive blood pressure (NIBP), pulse oximeter, electrocardiography (ECG) and micro stream capnography via nasal cannula. Her basal vitals were Heart rate 75bpm, Blood pressure 110/70mmHg and arterial oxygen saturation (SPO2) 100%.

Dexmedetomidine IV loading dose (1mcg/kg over 10 minutes) started while the monitor was being placed. Afterward, we continued maintenance on 1.5mic/kg/hour.

Nasal cannula applied on 2 liters of oxygen and ETCO2 was monitored through micro stream capnography attached to it.

Random blood sugar checked, and it was 126g/dl, so ringer IV fluid infusion started with total amount 100ml throughout the procedure, Temperature probe was placed.

Right groin was sterilized with alcohol and local anesthesia with lidocaine 40mg administered subcutaneously at site of sheath insertion. After catheter insertion through femoral vein heparin was given at dose of (100IU/kg).

During device deployment 10mg propofol was administered. Procedure went uneventful. Following device deployment, confirmation of device site and excluding residual flow was done through TTE, after sheath removal Compression at site of insertion was done for 5 minutes Patient was then transferred to CCU still sedated Patient took around 20 minutes to become fully conscious oral feeding was then started after one hour.

Second case was a female patient 3.5 years old 17.5kg diagnosed as Holt Oram through cardiac anomaly in form of secundum ASD, skeletal anomaly in form of bilateral absent distal radius, bilateral short humerus and absent left thumb also there was syndactyly between right thumb and right index (Figure 2). Multi slice CT revealed mild Pectus Excavatum secondary to sternal body hypoplasia. Patient underwent surgical closure of ASD.

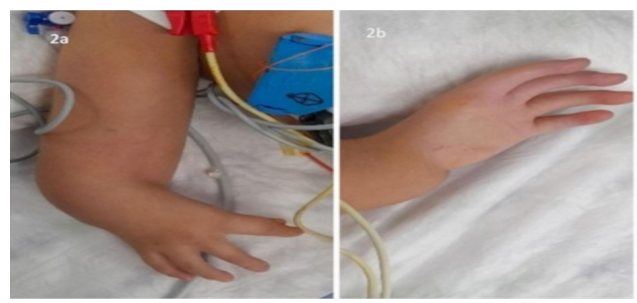

Figure 2: (A) Syndactyly between the thumb and index finger of the right hand. (B) Absence of the thumb on the left hand in the same patient, who underwent open surgical closure of a secundum atrial septal defect. The presence of upper limb bony deformities associated with a congenital cardiac defect raises suspicion for Holt-Oram syndrome. Evaluation for conduction anomalies and a positive family history are essential for confirming the diagnosis.

In operating room 1mg Midazolam was given as sedation in previously inserted peripheral cannula 24G. Full monitor attached in form of ECG, pulse oximetry, and NIBP. Then induction of general anesthesia via Propofol 2.5mg/kg, Fentanyl 2mcg/kg and cisatracurium 0.2mg/kg. Cuffed Endotracheal tube of internal diameter (ID) 5mm was inserted easily. A right femoral arterial line was inserted with 3Fr. Catheter ultrasound guided. Trans-esophageal echocardiography (TEE) probe was inserted, and another Anesthetist was charged to monitor the process of insertion the central venous line (CVL), 5fr CVL was placed in right internal jugular vein guided by both ultrasound and TEE.

Intraoperative monitoring also included central venous pressure (CVP) measurement, temperature, end-tidal carbon dioxide (EtCO2), and transesophageal echocardiography (TEE). Anesthesia maintained with sevoflurane (1%) in 50% oxygen in air mixture, cisatracurium infusion 2mcg/kg/min, and fentanyl infusion 2mcg/kg/hour.

Surgeons were notified to minimize cardiac manipulations for fear of arrhythmias. Closure of secundum atrial septal defect done on cardiopulmonary bypass via patch. On weaning from bypass, A full TEE study was done demonstrating no residual Atrial septal defect, normal functioning tricuspid valve. Smooth weaning from bypass, no inotropes or vasopressors needed, patient was then transferred to Cardiac Surgery Intensive Care Unit intubated ventilated and sedated.

DISCUSSION

Diagnosis of Holt Oram was done on these cases based on clinical criteria in form of skeletal deformity in upper limb, Atrial septal defect and positive family history. Genetic testing couldn't be done due to its cost and unavailability in our center.

Preoperative assessment is a challenge we have to rule out conduction anomalies via ECG assessment, CXR to exclude chest wall deformity and restrictive patterns, proper airway assessment and preparing for difficult intubation. Association between Holt-Oram syndrome and malignant hyperthermia (MH) is not established, Holt Oram is still a rare syndrome several case reports admit that they delivered general anesthesia with triggers to MH safely to Holt Oram cases [4-6] however in a single case report for 2-month child undergoing cardiac surgery malignant hyperthermia like symptoms occurred [7]. SO, after searching for evidence of association, decision was taken to receive general anesthesia in second case with close monitoring of ETCO2, heart rate and body temperature and confirming availability of dantrolene and cooling blanket before starting. The case went uneventful which suggest that solid association between Holt Oram and malignant hyperthermia is not proven and still questionable.

Holt Oram cases usually associated with higher risk of arrhythmias, we expected higher incidence during cardiac procedures due to direct cardiac manipulation and right atrial cannulation during open cases and wire triggered arrhythmias during transcatheter procedures. Surprisingly no exaggerated arrhythmias were noted than the usual incidence. Before starting the second case extra precautions were taken in form of applying external adhesive pads for immediate cardioversion if needed and central venous line insertion was monitored via TEE and ultrasound to avoid excessive advancement of CVL wire which may trigger arrhythmia.

As regard invasive blood pressure monitoring in second case, Femoral artery cannulation was preferred as upper limb anomalies associated with HOS will increase difficulty of radial artery cannulation as it will be more mobile as there is hypoplasia or absent radial bone to fix radial artery against it.

Second Case had pectus excavatum which is usually associated with reduction in lung capacities according to severity of chest wall deformity, however our patient showed normal resistance and compliance on mechanical ventilation with tidal volume 8ml/kg and respiratory rate 22 breath per minute and PEEP 3 CmH_2O

Also, routine precautions for ASD cases including avoidance of air emboli and guarding against infective endocarditis should be implicated.

In Conclusion Anesthetic management in HOS patients undergoing cardiac procedures necessitates a multidisciplinary approach with vigilant monitoring for potential complications. While the risk of arrhythmias and malignant hyperthermia requires careful consideration, our cases suggest that with appropriate planning, these patients can undergo complex cardiac procedures safely.

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PATIENT CONSENT

Written informed consent was obtained from legal guardian for publication of this case and any accompanying images.

CONFLICT OF INTERESTS

There are no conflicts of interest.

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