Anesthetic Considerations in a Pediatric Patient with Patau Syndrome

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INTRODUCTION

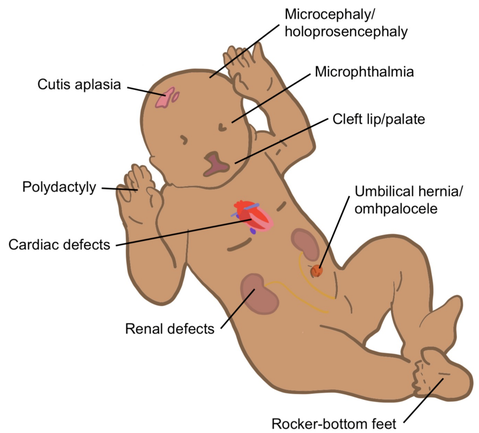
- Patau syndrome, or Trisomy 13, is a chromosomal disorder first described in 1960[1]. It is the third most common autosomal trisomy disorder with an estimated prevalence of 8 to 15 per 100,000 live births[2,3].
- A majority of prenatal diagnoses are terminated or spontaneously lost, with 20% of cases resulting in live births[4]. Median life expectancy is 5 to 12 days and over 90% of infants die within the first year, with a majority of deaths occurring in the first month of life[2,5,6].
- Patau syndrome phenotype is characterized by multiple craniofacial and congenital organ malformations, particularly involving the cardiac and central nervous systems[7]. These anatomic and functional changes present potential challenges to anesthetic management.
- We present a case of a patient with Patau Syndrome who underwent monitored anesthesia care (MAC) for a magnetic resonance imaging (MRI) of the lumbar spine.

CASE PRESENTATION

- A 3-year-old female child presented for repeat lumbar spine imaging in preparation for possible neurosurgical intervention. Her past medical history included a prenatal diagnosis of Trisomy 13, Lennox Gastaut Syndrome, global developmental delay, sacrococcygeal teratoma with tethered cord, optic nerve atrophy, bicuspid aortic valve, patent ductus arteriosus (PDA), atrial septal defect (ASD), pulmonic valve dysplasia, suspected obstructive sleep apnea (OSA), urogenic bladder complicated by multidrug resistant urinary tract infections, and failure to thrive. The patient was born at full term via cesarean section.
- Prior to the start of the case, oral midazolam was given for anxiolysis. In preparation for the procedure, the American Society of Anesthesiology's standard monitors (EKG, non-invasive blood pressure, pulse oximetry, capnography) were placed, and a peripheral intravenous (IV) access was obtained with a 24-gauge catheter in the left hand. The patient was positioned supine, head neutral, and all pressure points padded. Propofol was titrated to the patient's level of sedation. The patient maintained spontaneous respirations throughout the procedure with 6 L/min of supplemental oxygen via face mask.
- The 41-minute procedure proceeded uneventfully, and the patient was then transported to the post-anesthesia care unit (PACU) with continued oxygen supplementation at 4 L/min via face mask. She continued to do well in the PACU and was discharged later the same day.

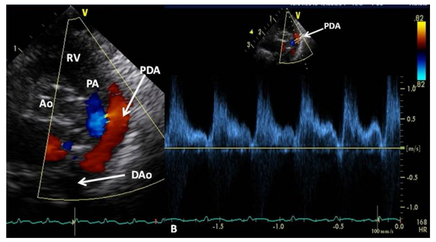
DISCUSSION

Given the breadth of possible congenital abnormalities associated with Patau Syndrome, anesthetic management for these patients emphasizes diligent preoperative planning and intraoperative vigilance. Anesthetic goals include seizure prevention, maintenance of hemodynamic stability, and prevention of and monitoring for apnea.

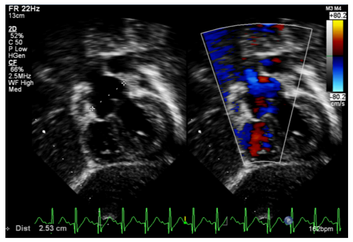


Representative illustration of characteristic features of Patau Syndrome (Trisomy 13). Adapted from Osmosis ©.

- **Neurologic** Abnormal intrauterine cerebral development is characteristic and results in severe cognitive impairment; serious developmental delay is reported in all patients surviving beyond the first year of life[7]. Seizure disorders are present in 50% of patients[7].
- **Cardiac** Congenital heart disease (CHD) is present in 80% of patients with Patau syndrome, with most cases having multiple non-cyanotic lesions including PDA, ASD, and ventricular septal defect (VSD)[7,8]. Less commonly, dextrocardia, Tetralogy of Fallot and transposition of the great arteries may be seen[7]. Most notably, uncorrected cardiac abnormalities may not be adequately diagnosed or optimized due to poor life expectancy.



Representative transthoracic echo of parasternal short axis view with Doppler demonstrating patent ductus arteriosus (PDA) with left-to-right shunt and high Doppler flow velocity. Suggestive of normal/low pulmonary artery pressure. Ao, aorta; D, diastolic; DAo, descending aorta; RV, right ventricle; S, systolic[15].



Patient's transthoracic echo demonstrating aortic root dilation to 25 mm at end diastole.

DISCUSSION (continued)

- **Respiratory** A number of craniofacial abnormalities commonly reported in Patau Syndrome are associated with difficult ventilation and intubation and they include short neck, micrognathia, high arched cleft lip and palate and nasal malformation[8].
- **Renal** Renal involvement, such as renal agenesis, hydronephrosis, and polycystic kidneys, is present in 60% of cases[8].
- **Musculoskeletal** Joint contractures and subluxation may develop with age and may present additional challenges to direct laryngoscopy, intravenous access, positioning, and transportation of these patients.
- **Hematologic** Long-standing hypoxemia due to CHD is associated with secondary erythrocytosis. This both increases thrombosis risk due to hyperviscosity and stasis and bleeding risk due to coagulation profile derangements[9].

CONCLUSION

- Despite high mortality rates, cases of long-term survival past the first decade have been reported[10-12]. One-year survival rates have increased from 3% to 5-20% in recent years[2,5,6,13].
- Although these estimates may be influenced by less severe genetic mosaicism and increased termination of pregnancies with major malformations, these reports have led to more physicians transitioning from palliative care to more aggressive inpatient management[2,14].
- One year survival after first surgery increases to 70% in those with Patau syndrome; therefore, a deeper understanding of anesthetic management and its potential challenges in these patients benefit both the anesthesiologist and the patient[2].

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