Scholars Journal of Applied Medical Sciences (SJAMS)
Abbreviated Key Title: Sch. J. App. Med. Sci.
ISSN 2320-6691 (Online)
ISSN 2347-954X (Print)

A Unit of Scholars Academic and Scientific Society, India
www.saspublishers.com

Anesthesiology

Anesthesia Approach to a Patient with Patau Syndrome

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Case Report

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Article History
Received: 16.02.2018
Accepted: 26.02.2018
Published: 30.03.2018

DOI:
10.36347/sjams.2018.v06i03.046

Abstract: Trisomy 13 syndrome (Patau Syndrome) was originally found cytogenetically in 1960 while its clinical phenotype was described later. It is a congenital genetic disorder caused by an extra copy of 13 Chromosome. Cardinal findings of Trisomy 13 syndrome are motor and mental retardation, microcephaly, microphthalmia, holoproencephaly, hypotelorism, cleft palate and lip, cardiaovascular, genitourinary and ocular malformations. In Patau syndrome, difficulty of intubation due to craniofacial deformities and cardiac anomalies occurring in 80% of the cases make anesthesia management more challenging. We aimed to present anesthesia management in a patient who was diagnosed with Patau syndrome and was planned to undergo operation for inguinal hernia.
Keywords: Patau Syndrome, anesthesia management, central apnea.

INTRODUCTION

Trisomy 13 syndrome (Patau Syndrome) was originally found cytogenetically by Patau *et al*. [1] in 1960 while its clinical phenotype was described by Smith *et al*. [2]. It is a congenital genetic disorder caused by an extra copy of 13 Chromosome and presenting with multiple craniofacial, cardiac, neurological and renal anomalies. The frequency of this syndrome is between 1:3000 to 1:29 000 live births [3,4]. Trisomy 13 is the third most common autosomal Trisomy at birth, with Trisomy 21, followed by Trisomy 18, occurring more frequently [5,6].

Cardinal findings of Trisomy 13 syndrome are motor and mental retardation, microcephaly, microphthalmia, holoproencephaly, hypotelorism, cleft palate and lip, cardiaovascular, genitourinary and ocular malformations. Microphthalmia, cleft palate and lip and polydactly triad is characteristic for the syndrome [3]. 80% the cases has congenital heart defects such as atrial septal defect, ventricular septal defect, patent ductus arteriosus and dextrocardia. Central apnea, capillary hemangioma, omphalocel, hernia, polycystic kidneys, micropenis and hyperthrophy in clitoris may also occur [7,8]. In addition, inguinal and umbilical hernias are common, and gall bladder stones, malrotation and Meckel diverticula has also been reported. Microscopic pancreatic dysplasia is a special anomaly reported in these patients [9].

CASE REPORT

A 1 year old patients who weighted 8 kg and was diagnosed with Patau syndrome was planned to undergo operation for inguinal hernia. He had ASD, facial anatomic changes (micrognathia, large forehead) polydactyly and pectus carinatum. He had previously undergone operation for, cleft palate and lip, omphalocel and cataract. The patients was monitorized with pulse oximeter, ECG and non invasive pressure. Preparation was made for probable difficult airway. Anesthesia induction was performed with %8 sevoflurane inhalation. Under anesthesia, venous access was obtained and 2 mg/kg propofol was administered. 1,5 no laryngeal mask was put on the patient. After induction, anesthesia maintenance was made with %2.5 Sevoflurane and %40 O_2/ air mixture. Isotonic sodium chlorur and %5 dextrose solution was infused, calculating fluid deficit. The patients was extubated without encountering any problems.

DISCUSSION

The most common cranial imaging finding is holoproencephaly, which leads to severe mental retardation [6]. Chromosome analysis is required for definitive diagnosis. It may be complete, partial or mosaic type. Phenotype and survival rate rests on underlying genotype. In this syndrome, in most of the fetuses, spontanous abortion or still birth occurs. Intrauterine growth retardation occurs and they have low birth weight. Multiple congenital anomalies in patients with Patau syndrome are not compatible with life, with approximately 50% of the cases dying within 1 month and 90% within 1 year. However, mosaic type cases may have more favorable course. In the literature,

a 12 year old case has been reported [10]. We had no information regarding the caryotype of our patient, but his being older than one year suggests mosaic type. Our case had ASD, micrognathia, large forehead, polydactly and pectus carinatum.

The rate of those who undergo operation for trisomy 13 during newborn period is % 23 [3]. It has been stated that patients with trisomy 13 undergo cardiac surgery most frequently, followed by cleft palate and lip operations [11,12]. Our patient had also undergone operations for cleft palate, omphalocel and cataract.

Short-thick neck, cleft palate with high arched palate, along with small mouth and micrognathia are factors making intubation and placement of laryngeal mask more difficult [9]. In the literature, difficulty in intubation and unsuccessful intubation has been reported in a child with multiple anomalies [13]. In addition, in another patient with intubation difficulty, who was about to undergo operation due to cleft palate, as pediatric fiberoptic was not suitable, intubation was carried out by using urologic guide wire [14]. In the present case, since intubation was not necessary in inguinal hernia operation, airway safety was provided easily with LMA. Apnea episodes have been reported in 50% of the patients with Patau syndrome [14]. Due to probability of postoperative apnea, intraoperative opioid and neuromuscular blockers should be titrated and monitorized during use. Opioid was not used in our case owing to the risk of postoperative apnea.

In Patau syndrome, difficulty of intubation due to craniofacial deformities and cardiac anomalies occurring in 80% of the cases make anesthesia management more challenging. It is our suggestion that the difficulties can be overcome by careful preoperative evaluation and being prepared for difficult intubation.

It should be stressed that difficut intubation, cardiac anomalies and central apnea are the main points for which care should be taken by anesthesists.

REFERENCES

1. Patau K, Smith DW, Therman E, Inhorn SL, Wagner HP. Multiple congenital anomaly caused by an extra autosome. Lancet 1960;1:790-3.
2. Jones KI. Chromosomal Abnormality Syndromes. In: Jones KI (ed). Smith’s Recognizable patterns of human malformations. Sixth Edition. Philadelphia; Elsevier Saunders. 2006. 18-21
3. Baty BJ, Blackburn BL, Carey JC. Natural history of trisomy18 and trisomy 13. I: Growth, physical assessment, medi-cal histories, survival, and recurrence risk. Am J Med Genet.1994;49:175---88.
4. Rasmussen SA, Wong LY, Yang Q, May KM, Friedman JM. Population-based analyses of mortality in trisomy 13 and trisomy 18. Pediatrics. 2003 Apr 1;111(4):777-84.
5. Whittle MJ. Structural Fetal Abnormalities. The Total Picture. Archives of Disease in Childhood Fetal and Neonatal edition. 1996 Jul;75(1):F72.
6. Driscoll DA, Mennuti MT. Genetic defects and gene therapy. In:Spitzer AR, editor. Intensive care of the fetus and neonate. St.Louis, MO: Mosby; 1996. p. 164---77.5
7. Tsukada K, Imataka G, Suzumura H, Arisaka O. Better prognosis in newborns with trisomy 13 who received intensive treatments: a retrospective study of 16 patients. Cell biochemistry and biophysics. 2012 Jul 1;63(3):191-8.
8. Plaiasu V, Ochiana D, Motei G, Anca I, Georgescu A. Clinical relevance of cytogenetics to pediatric practice. Postnatal findings of Patau syndrome–Review of 5 cases. Mædica. 2010 Jul;5(3):178.
9. Baum VC, O’Flaherty JE. Anesthesia for genetic, metabolic and dysmorphic syndromes of childhood. Lippincott Williams and Wilkins 2007;374.
10. Fogu G, Maserati E, Cambosu F, Moro MA, Poddie F, Soro G, Bandiera P, Serra G, Tusacciu G, Sanna G, Mazzarello V. Patau syndrome with long survival in a case of unusual mosaic trisomy 13. European journal of medical genetics. 2008 Aug 31;51(4):303-14.
11. Pollard RC, Beasley JM. Anaesthesia for patients with trisomy 13 (Patau's syndrome). Pediatric Anesthesia. 1996 Mar 1;6(2):151-3.
12. Nelson KE, Hexem KR, Feudtner C. Inpatient hospital care of children with trisomy 13 and trisomy 18 in the United States. Pediatrics. 2012 May 1;129(5):869-76.
13. Demir HF, Erakgün A, Çertuğ A. Patau sendromu ön tanılı pediyatrik olguda anestezik yönetim: Olgu sunumu ve literatüre kısa bir bakış. Ege Tıp Dergisi. 2013;52(4).
14. Kamal M, Varghese D, Bhagde J, Singariya G, Simon AM, Singh A. Anesthesia in a child operated for cleft lip associated with Patau's syndrome. Brazilian Journal of Anesthesiology (English Edition). 2016 Apr 20.