Case Report

Indian Journal of Anesthesia and Analgesia
Volume 9 Number 6 November-December 2022
DOI: http://dx.doi.org/10.21088/ijaa.2349.8471.9622.3

Anesthetic Management in a Case of Rett Syndrome: A Case Report

Anita R Chhabra[1], Nishigandha G Sawant[2], Vijay L Shetty[3], Atul M Ganatra[4]

How to cite this article:
Anita R Chhabra, Nishigandha G Sawant, Vijay L Shetty, et. al./Anesthetic Management in a Case of Rett Syndrome: A Case Report/Indian J Anesth Analg. 2022;9(6)295-298.

Abstract

Rett syndrome (RS) is a progressive neurological disorder that almost exclusively affects females. It is characterized by severe mental retardation following a period of normal development during childhood. Classical features include autism, stereotypical hand movements, seizures, microcephaly and abnormal respiratory control. Anesthetic challenges in patients with RS include difficult airway, seizure management, positioning, increased sensitivity to sedatives, autonomic dysfunction, anesthesia triggered episodes of apnoea, hypoxemia and sudden death. Here, we describe the successful anesthetic management of a 13-year-old patient with RS exhibiting most of these features who underwent laparoscopic SOS open ovarian cystectomy and hysterectomy.

Keywords: Rett Syndrome, seizures, anesthesia

Key Messages: The estimated prevalence of Rett syndrome is 25% among the population of severely retarded females and are very likely to present for some procedure before a practicing anesthesiologist. Here we have discussed its pathophysiology, anesthetic implications and successful management in a child posted for gynaecological procedure under general anesthesia with a supportive multi-specialty team work.

INTRODUCTION

Rett syndrome (RS) is a progressive neurological disorder that almost exclusively affects females.[1] Incidence is 0.5 in 10,000 female births, being rare in boys.[2] It is characterized by severe mental retardation following a period of normal development during childhood. It is only second to Down syndrome as a cause of mental retardation in females.[3] Classical features include autism, stereotypical hand movements, seizures, microcephaly and abnormal respiratory control. Patients with RS are extremely sensitive to sedatives and exhibit slow recovery from anesthesia.[4] Anesthesia challenges include difficult airway, seizures management, positioning due to musculoskeletal involvement, anesthesia triggered apnoea episodes, hypoxemia and sudden death.[5,6] Here we discuss some of the anesthetic challenges we overcame in the successful management of a

Author's Affiliation: [1,2]Senior Consultant, [3]Head, Department of Anesthesia, [4]Senior Consultant, Department of Obstetrics and Gynecology, Fortis Hospital, Mumbai 400080, Maharashtra, India.

Corresponding Author: Anita R Chhabra, Senior Consultant, Department of Anesthesia, Fortis Hospital, Mumbai 400080, Maharashtra, India.

E-mail: canita24@yahoo.com

Received on: 05.09.2022

Accepted on: 10.10.2022

Rett child.

CASE REPORT

A 13-year-old, 30 kg female, a known case of Rett syndrome had presented in emergency with pain abdomen. Following acute pain management and further investigations she was diagnosed as having ruptured left ovarian cyst and was scheduled by the gynecologists' team for laparoscopic SOS open ovarian cystectomy and hysterectomy. Birth history revealed that she was delivered normally at full term following an uncomplicated antenatal period of the mother. Her developmental milestones were delayed till 13 months of age. Subsequent course of illness revealed history of regression of milestones, history of seizures on irregular treatment with sodium valproate, difficulty in swallowing solid food, vomiting, repetitive hand movements, frequent upper respiratory tract infections and frequent hospitalizations till the age of 8 years Paediatricians took care of her peri-operative management and reinstituted sodium valproate (10 mg/kg/day) therapy.

The pre-anesthesia check-up further revealed that she had frail habitus, was afebrile and hypertonic. Her head circumference was small at 45 cm, which is less than 3rd percentile. Airway was normal. Mild thoracic scoliosis was present. The extremities were cool, grade 1 clubbing was noted while cyanosis was absent. Her vitals revealed heart rate of 102/min, blood pressure (BP) – 100/66 mmHg and oxygen saturation – 99% on room air. On auscultation, the heart-sounds and chest were normal.

Preoperative laboratory values were within normal range. The ECG and chest X-ray were unremarkable. Her CT abdomen was performed uneventfully a day prior to the surgery under intravenous sedation using midazolam, 1 mg and titrated dose of propofol.

The patient had her last seizure episode on the morning of laparoscopic surgery which was managed by a loading dose of levipil, 20 mg/kg over 15 minutes.

The patient was calm on arrival to operation theatre. A 22 G peripheral intravenous line was in-situ with ringer lactate on flow. Patient had received aspiration prophylaxis, antacid pantoprazole 30 mg and antiemetic, ondansetron 3 mg intravenously preoperatively in the ward. In OT, standard monitors (ECG, non-invasive BP, pulse oximeter) were attached and modified rapid-sequence induction was done using sedation with midazolam (0.6 mg) and fentanyl (30mcg) followed by IV propofol and rocuronium (0.8 mg/kg). A 6 mm ID endotracheal tube was passed under vision and ventilation was controlled using circle system. Anesthesia was maintained with oxygen (50%), air and sevoflurane (1-2%). Oro-gastric tube was passed to empty the gastric contents and to keep the stomach deflated during the laparoscopic procedure. A second IV line (20 G jelco) was placed to infuse fluids perioperatively. Patient was kept warm using a hot line intravenous fluids and a warm blower. Maintenance dose of levipil (10 mg/kg) was infused over 15 minutes. Pre-op ABG was within normal range, however no intra-arterial catheter was maintained. Peri-op monitoring also included end tidal carbon dioxide, inspired and expired oxygen and sevoflurane concentration, oro-pharyngeal temperature and urine output.

The laparoscopic procedure was abandoned after an hour in view of technical difficulty and converted to open ovarian cystectomy and hysterectomy. The estimated blood loss was approximately 300 ml which was replaced with crystalloids. Her temperature dropped to 34 degrees Celsius, which was managed by aggressive warming using hotline for fluids and an additional warm air blower. All other hemodynamic parameters were maintained throughout the surgery. For postoperative pain relief, bilateral transverse abdominis plane (TAP) block with 12 ml of 0.25% bupivacaine on either side was performed.

At the end of the surgery, anesthesia was reversed with neostigmine (0.05mg/kg) and glycopyrrolate (8mcg/kg). The patient was extubated on confirming wakefulness, good muscle tone, regular respiration without any apnoeic spells along with adequate respiratory efforts. She was shifted to the paediatric intensive care unit for post-operative monitoring and transferred to the ward the following day. She had an uneventful recovery and was discharged on the fifth post-operative day.

DISCUSSION

Rett syndrome, described in 1966, is a rare genetic neurological disorder affecting exclusively girls.[1] It is hypothesised that it is an X-linked dominant mutation on $MECO_2$ gene.[2]

Clinical features are remarkably consistent with affected infants having normal development until approximately 6 to 18 months of age following which there is rapid developmental regression leading to dementia and autism.[1] Acquired skills

such as purposeful hand movements and ability to communicate are lost and is replaced by a constant, repetitive hand washing movement a hallmark feature of Rett syndrome.[7] Wide range of disability are exhibited including choreoathetosis, dystonia, myoclonic jerk and stereotypic automatism.[1,8] Chronic disease leads to diffuse and progressive muscle wasting, growth stagnation of weight, stature and particularly microcephaly.[1] Seizures may be present in some cases (15%).

The estimated prevalence of Rett syndrome is 25% among the population of severely retarded females[9] and are very likely to present for some procedure before a practicing anesthesiologist. Hence, it is pertinent that we understand its pathophysiology and anesthetic implications.

Anesthetic challenges of significance in managing these children are lack of cooperation, muscle wasting, abnormal continuous limb movement, abnormal respiratory control, difficult positioning due to scoliosis and chest deformity, vasomotor instability, metabolic abnormalities increased lactic acid level, altered sensitivity to painful stimuli.

Non-depolarizing muscle relaxant is preferred over succinylcholine for its potassium increasing property in such patients with neuro-muscular dystrophy.[10] Risk of aspiration also exists. Hence, we did rapid sequence intubation using rocuronium in this case.

There is respiratory impairment in these patients due to muscle wasting and thoracic deformity. Polygraphy studies reveal normal breathing during sleep compared to irregular breathing with periods of apnoea during wakefulness which may lead to oxygen desaturation and loss of consciousness during a severe episode.[11] This has been explained by the impairment of behavioural control of breathing a forebrain function. Cirignotta et al. have suggested that the frequent desaturations may cause permanent hypoxic damage and contribute to the progressive cerebral deterioration.[12] Rapid desaturation has been explained by high expiratory position of the diaphragm during apnoea. However, the influence of anesthetic agents on respiratory patterns during perioperative period is not known. The increased sensitivity to sedation has been reported by Konarzewski in his patient who was administered trimeparazine and ketamine premedication followed by halothane-nitrous-oxygen induction and maintenance in a spontaneously breathing child.[4] The prolonged sedation is expected with this type of premedication into the postoperative period although a causal relation between the two has not been established. The parents of our patient did not give history of irregular breathing pattern or apnoea, although a negative history is usually unreliable and all RS patients should be considered as having disorder of respiratory control and resultant episodes of desaturation. We administered minimum dose of sedation and opioid as per weight of the patient and she did not exhibit any breathing variability during the post-operative period and continued to maintain normal oxygen saturation.

Our patient had no symptoms of respiratory impairment and pre-op chest X-ray revealed no evidence of recurrent aspiration. Preoperative ABG was within normal range, hence we did not maintain a continuous arterial line perioperatively, although it is indicated in patients who have respiratory symptoms. Presence of mild thoracic scoliosis in our patient, a classic feature of Rett syndrome, is usually neurogenic in origin, cautioning careful positioning under anesthesia.[13]

Cardiovascular considerations include vasomotor instability leading to cool extremities. Our patient had inadvertent hypothermia intraoperatively possibly due to heat loss contributed by large exposure of the operative field in a frail habitus. Hypothermia was managed using hotline and an additional warm air blower. All other hemodynamic parameters were normal. Aggressive attempts to prevent heat loss should be practiced.

Metabolic abnormalities include excessive ammonia level; however, it is not a consistent finding.[1]

Pain management was done using TAP block before reversal and institutional pain management protocol was followed postoperatively with intravenous and/or oral non-opioid analgesics.

CONCLUSION

Rett syndrome is a debilitating neurological disorder, exclusively occurring in young females. Anesthetist awareness of its peculiar anesthetic challenges involving control of seizures, abnormal continuous movement of limbs and vasomotor instability is important. Minimal use of sedatives and using regional analgesia for post-operative pain relief is the technique of choice. This report spotlights this rare syndrome and the successful management of such a patient with coordinated team work with specialities like paediatrics, pediatric intensivists, gynecologists along with the anesthesiologists.

Acknowledgement: Nil

Conflict of Interest: Nil

REFERENCES

1. Rett A. Ueber ein eigenartiges hirnatropisches syndrome bei Hyperammonamie im kindersalter. Wien Med Wochenschr 1966; 116: 723-38.
2. Campos J, Fernandez D, Munoz N, San Antonio V. Rett syndrome: 50 years' history of a still not well known condition. Medicina (Buenos Aires). 2007;67(6/7):531-42.
3. Ella wayC, Christodoulou J. Rett syndrome: clinical update and review of recent genetic advances. J Pediatr Child Health 1999;35: 419-426.
4. Konarzewski WH, Misso S. Rett syndrome and delayed recovery from anesthesia. Anesthesia. 1994;49:357.
5. Dearlove OR, walker RWM. Anesthesia and Rett syndrome: a case report. Pediatr Anesth. 1996;6:155-8.
6. Pierson J, Mayhew JF. Anesthesia in a child with Rett syndrome: a case report and literature review. AANA J. 2001;69:395-6.
7. Al-Mateen M, Phillipar M, Shields WD. Rett syndrome: a commonly overlooked progressive encephalopathy in girls. Am J Dis Child 1986; 140: 761-5.
8. Hagberg B, Aicardi J, Dias K, Ramos O. A progressive syndrome of autism, dementia, ataxia and loss of purposeful hand use in girls: Rett's syndrome: Report of 35 cases. Ann Neurol 1984; 14: 471-9.
9. Hagberg B. Rett's syndrome: prevalence and impact on progressive severe mental retardation in girls. Acta Paediatr Scand 1985; 74: 405-8.
10. Miller RD. Anesthesia, 2nd ed. New York: Churchill Livingstone, 1986: 920.
11. Lugaresi E, Cirignotta F, Montagna P. Breathing in the Rett syndrome. Brain Dev 1985; 7: 329-33.
12. Cirignotta F, Lugaresi E, Montagna P. Breathing impairment in Rett syndrome. Am J Med Genet 1986; 24:167-73.
13. Hagberg B. Rett syndrome: clinical peculiarities and biological mysteries. ActaPaediatr 1995; 84:971-6.