DOI: 10.7860/JCDR/2023/63928.18456

Case Report

Anaesthesia Section

Anaesthetic Considerations for General Anaesthesia in an Adult Patient with Wolf-Hirschhorn Syndrome and Kyphoscoliosis: A Case Report

MANJU BALA[1], RASHMI[2], RISHMEET KAUR[3], AJITHKUMAR[4], VINAY JANGRA[5]

ABSTRACT

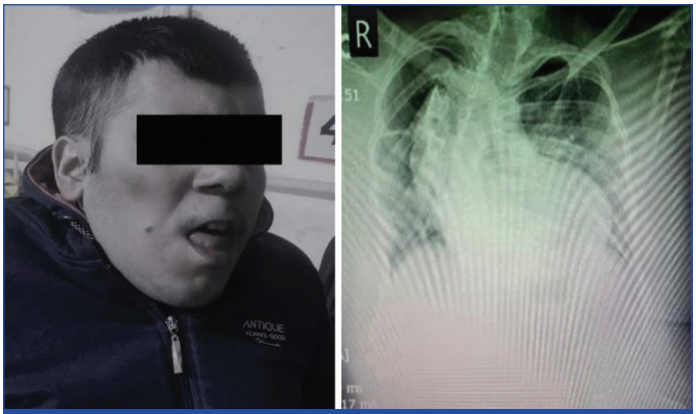
Wolf-Hirschhorn Syndrome (WHS), also known as Chromosome 4 deletion syndrome, is a rare hereditary disease with a prevalence of 1 in 50,000. It occurs due to the microdeletion of the short arm of chromosome 4, specifically the 4p16.3 domain. Patients with WHS exhibit diverse phenotypes, including growth retardation, developmental delay, congenital heart disease, and convulsions, depending on the amount of deleted genetic material. This case report focuses on a 25-year-old male who was admitted for cataract extraction and posterior chamber intraocular lens implantation. Through gene analysis, the patient was diagnosed with WHS. He exhibited micrognathia, a short neck, kyphoscoliosis, seizure disorder, and mental retardation. Additionally, he had severe kyphoscoliosis, which resulted in restrictive lung disease. These abnormalities posed significant challenges for anaesthetic management. To ensure a successful general anaesthesia, adequate preparedness for difficult airway management was crucial. A thorough cardiovascular and neuromuscular examination was conducted preoperatively to rule out associated anomalies and minimise complications. The patient's perioperative antiepileptic cover was continued. Extubation proved challenging due to the patient's mental retardation and restrictive lung disease. This case underscores the importance of effective anaesthetic management for patients with this rare condition undergoing cataract surgery under general anaesthesia.

Keywords: Cataract surgery, Difficult airway, General anaesthesia, Seizure disorder

CASE REPORT

A 25-year-old male, with a height of 168 cm and weight of 65 kg, was admitted to our hospital for cataract extraction and posterior chamber intraocular lens implantation under general anaesthesia. Consent for publication was obtained from the patient's father, as the patient was mentally retarded. Through gene analysis, he was diagnosed with Wolf-Hirschhorn Syndrome (WHS) at the age of four. He presented with micrognathia, a short neck, restricted neck extension, kyphoscoliosis, and a seizure disorder. The patient has been experiencing seizures since he was three years old, and he has been taking phenytoin 100 mg tablets three times a day to manage his seizures. The most recent seizure occurred two and a half years ago. There is no history of any other chronic medical illness. Despite his mental retardation, he is able to ambulate without support and does not have muscular dystrophy. Due to breathing difficulties, the patient is unable to lie supine and sleeps in a propped-up position, experiencing snoring for the past five years. He underwent cataract extraction and posterior chamber intraocular lens implantation in the opposite eye under general anaesthesia one year ago, with a history of delayed recovery from anaesthesia.

During the physical examination, the patient exhibited facial features typical of WHS, including micrognathia, macroglossia, and a short neck with restricted neck extension. The patient was unable to follow commands, making it impossible to evaluate the Mallampatti grading. Severe kyphoscoliosis was present, and decreased air entry was noted in all lung fields. Heart sounds were normal upon auscultation, and muscle power was normal in all four limbs. Blood investigations yielded results within normal limits. Echocardiography revealed a normal study with an ejection fraction of 55-60%. A chest X-ray in the posteroanterior view showed kyphoscoliosis with restricted lung volumes bilaterally [Table/Fig-1]. Pulmonary function testing could not be performed due to the patient's lack of cooperation. His oxygen saturation level was 91%. Chromosomal analysis conducted during childhood revealed the following: 46XY, del 4 (q22), t(4:10), q22.



[Table/Fig-1]: Preoperative image of patient with Wolf-Hirschhorn Syndrome (WHS) (left); preoperative chest X-ray of patient showing scoliosis (right).

As the patient was mentally challenged and unable to stay still during cataract surgery, a plan was made to proceed with general anaesthesia, along with all the necessary preparations to manage a difficult airway. The patient continued taking their antiepileptic medications during the perioperative period. Preoperatively, they were given a 40 mg tablet of pantoprazole in the morning and a 0.25 mg tablet of alprazolam on the night before surgery and in the morning of the procedure. After obtaining informed written consent due to the high-risk associated with a difficult airway and severe restrictive lung disease, the patient was

transferred to the operation table. Routine monitors such as electrocardiography, non invasive blood pressure, and pulse oximeter were attached. The patient's baseline heart rate was 88 beats per minute, blood pressure was 126/72 mmHg, and oxygen saturation was 91% on room air. Intravenous access was established using a 20-gauge cannula in the left hand. Lignocaine 10% was sprayed in the oropharynx. Anaesthesia induction was performed using intravenous administration of glycopyrrolate injection (0.2 mg), fentanyl injection (130 µg), and propofol injection (130 mg). Bag-mask ventilation was challenging and could only be achieved after inserting an oral airway. An attempt was made to secure the airway using a size 4 I-gel supraglottic airway device, but it was unsuccessful. A second attempt was made using the reverse technique with the size 4 I-gel, but it also failed. A check laryngoscopy was performed to visualise the glottic view, followed by the administration of 5 mg of intravenous vecuronium. The airway was successfully secured using direct laryngoscopy and endotracheal intubation with a 7.5 mm internal diameter endotracheal tube. On direct laryngoscopy, a Modified Cormack Lehane grade 2B view was observed, and endotracheal intubation was confirmed using capnography and auscultation [1]. After the completion of the surgery, the patient displayed spontaneous respiratory efforts and was extubated with a nasal airway in place due to difficult bag-mask ventilation and a large tongue. The patient's oxygen saturation was not maintained in the supine position, so they were positioned at a 60-degree angle, provided with non invasive ventilation, and nebulised with levosalbutamol, ipratropium bromide, and budesonide. After approximately two hours in a sitting position, the patient started maintaining their preoperative oxygen saturation on room air. The patient was then transferred to the high dependency unit for further monitoring. In the evening, the patient was moved to a regular ward where their recovery was uneventful, and they were discharged on the third postoperative day.

DISCUSSION

WHS syndrome is a rare hereditary disease with a prevalence of 1 in 50,000, caused by a microdeletion of the short arm of chromosome 4, specifically the 4p16.3 domain [2]. Patients with WHS syndrome exhibit typical craniofacial features, including microcephaly, a high forehead with a prominent glabella, a nose with a "Greek warrior helmet appearance," ocular hypertelorism, highly arched eyebrows, epicanthus, a downturned mouth, a short philtrum, micrognathia, and underdeveloped ears [2,3]. These patients present with diverse phenotypes such as growth retardation, developmental delay, congenital heart disease, and convulsions, depending on the amount of deleted genetic material [4].

Patients with WHS require general anaesthesia for multiple corrective surgeries. However, general anaesthesia poses challenges due to various types of cardiac and nervous system anomalies, difficulties in airway management, muscle relaxation regulation, and maintenance of vital signs [5]. Therefore, a proper preoperative evaluation, including a comprehensive history, thorough physical examination with a focus on airway assessment, and relevant investigations, should be conducted.

The most challenging aspect of general anaesthesia in these patients is airway management [5,6]. Therefore, all preparations to manage a difficult airway were made, and a difficult airway cart was readily available. Smaller-sized endotracheal tubes were prepared considering growth retardation. These patients also have a risk of chronic aspiration, so proton pump inhibitors were administered preoperatively [5].

Ginsburg R and Purcell-Jones G reported a case of malignant hyperthermia in a 21-year-old female patient with WHS syndrome who underwent cleft palate repair. In this case, anaesthesia was maintained with halothane [7]. The anaesthetic drugs that can trigger malignant hyperthermia include halothane, isoflurane, sevoflurane, enflurane, ether, and succinylcholine [8]. Recent reports have shown that general anaesthesia can be safely performed with inhaled anaesthetics in patients with WHS [9,10].

WHS is often accompanied by convulsions [5,6,11]. Anticonvulsants should be administered on the day of surgery. In a case reported by Sari M and Cicekci F, phenobarbital was continued perioperatively in a 22-month-old patient with WHS who underwent undescended testis and colostomy surgery. This patient also had a history of convulsions, so anticonvulsants were continued on the day of surgery [6].

Since patients with WHS can also have associated congenital cardiac anomalies, preoperative echocardiography was performed. If the patient is mentally challenged or has visual or auditory impairment, measures should be taken to prevent postoperative agitation.

In cases of known difficult airway, fiberoptic intubation, video laryngoscopy, or an intubating laryngeal mask airway could have been alternative modalities to secure the airway.

CONCLUSION(S)

The syndrome is a rare type of genetic disorder that occurs due to a microdeletion of the short arm of chromosome 4, specifically in the 4p16.3 domain. Patients present with varying phenotypes depending on the amount of deleted genetic material. This patient had micrognathia, a short neck, kyphoscoliosis, a seizure disorder, mental retardation, and severe kyphoscoliosis with restrictive lung disease. To ensure the safe conduct of general anaesthesia, a thorough cardiovascular, neuromuscular, respiratory, and airway examination should be performed. Antiepileptics should be continued during the perioperative period. Adequate measures should be taken to manage a difficult airway and to prevent malignant hyperthermia.

REFERENCES

[1] Koh LKD, Kong CF, Ip-Yam PC. The Modified Cormack-Lehane score for the grading of direct laryngoscopy: Evaluation in the asian population. Anaesth Intensive Care. 2002;30(1):48-51.
[2] Battaglia A, Filippi T, Carey JC. Update on the clinical features and natural history of Wolf-Hirschhorn (4p-) syndrome: Experience with 87 patients and recommendations for routine health supervision. Am J Med Genet C Semin Med Genet. 2008;148C(4):246-51.
[3] Battaglia A, Carey JC, South ST, Wright TJ. Wolf-Hirschhorn Syndrome. In: GeneReviews [PMID: 20301362]. Edited by Pagon RA, Bird TD, Dolan CR, Stephens K: Seattle (WA), University of Washington, Seattle. 2002 Apr [updated 2010 Jun]. Available from: http://www.ncbi.nlm.nih.gov/bookshelf/br.fcgi? book=gene & part =whs.
[4] Bosenberg AT. Anaesthesia and Wolf-Hirschhorn syndrome. South Afr J Anaesth Analg. 2007;13(3):31-34.
[5] Kim HJ, You JA, Park S, Kim EJ, Park SJ, Kim HY. Anaesthetic considerations for an adult with Wolf-Hirschhorn syndrome- A case report. Anaesth Pain Med (Seoul). 2020;15(1):120-23.
[6] Sari M, Cicekci F. Anaesthetic concerns in patient with Wolf-Hirschhorn syndrome: A case report. Austin J Clin Case Rep. 2021;8(3):1201.
[7] Ginsburg R, Purcell-Jones G. Malignant hyperthermia in the Wolf-Hirschhorn syndrome. Anaesthesia. 1988;43(5):386-88.
[8] Gerald AG, Isaac NP, Heila MM, Timothy JT. Malignant Hyperthermia. In: Miller's Anaesthesia. 6th ed. Edited by Miller RD: Philadelphia, Elsevier Churchill Livingstone. 2005; pp 1169-90.

[9] Mohiuddin S, Mayhew JF. Anaesthesia for children with Wolf- Hirshhorn syndrome: A report and literature review. Paediatr Anaesth. 2005;15(3):254-55.
[10] Iacobucci T, Nanni L, Picoco F, de Francisci G. Anaesthesia for a child with Wolf-Hirshhorn syndrome. Paediatr Anaesth. 2004;14(11):969.
[11] Tsukamoto M, Yamanaka H, Yokoyama T. Anaesthetic considerations for a pediatric patient with Wolf-Hirschhorn syndrome: A case report. J Dent Anaesth Pain Med. 2017;17(3):231-33.

PARTICULARS OF CONTRIBUTORS:
1. Associate Professor, Department of Anaesthesia, Pt. B.D. Sharma, PGIMS, Rohtak, Haryana, India.
2. Senior Resident, Department of Anaesthesia, Pt. B.D. Sharma, PGIMS, Rohtak, Haryana, India.
3. Junior Resident, Department of Anaesthesia, Pt. B.D. Sharma, PGIMS, Rohtak, Haryana, India.
4. Junior Resident, Department of Anaesthesia, Pt. B.D. Sharma, PGIMS, Rohtak, Haryana, India.
5. Junior Resident, Department of Anaesthesia, Pt. B.D. Sharma, PGIMS, Rohtak, Haryana, India.

NAME, ADDRESS, E-MAIL ID OF THE CORRESPONDING AUTHOR:
Dr. Rashmi,
H. No. 614/29, Gali No 17, Tilak Nagar, Rohtak-124001, Haryana, India.
E-mail: rashi.singh65@gmail.com

PLAGIARISM CHECKING METHODS: [Jain H et al.]
- Plagiarism X-checker: Mar 20, 2023
- Manual Googling: May 10, 2023
- iThenticate Software: Jul 15, 2023 (5%)

ETYMOLOGY: Author Origin

EMENDATIONS: 6

AUTHOR DECLARATION:
- Financial or Other Competing Interests: None
- Was informed consent obtained from the subjects involved in the study? Yes
- For any images presented appropriate consent has been obtained from the subjects. Yes

Date of Submission: **Mar 09, 2023**
Date of Peer Review: **May 01, 2023**
Date of Acceptance: **Jul 20, 2023**
Date of Publishing: **Sep 01, 2023**