

Anaesthesia Management in Waardenburg Syndrome with Hirschsprung's Disease

Dr. G. D. Sree Mounika¹, Dr. Y. Murali Prabakar², Dr. M. Sharada³, Dr. R. Kondareddy⁴,
Dr. C. G. Raghuram⁵

¹Post Graduate, Department of Anesthesiology and Critical Care, Kurnool Medical College and GGH

^{2,3}Assistant Professor, Department of Anesthesiology and Critical Care, Kurnool Medical College and GGH

⁴Associate Professor, Department of Anesthesiology and Critical Care, Kurnool Medical College and GGH

⁵Professor, Department of Anesthesiology and Critical Care, Kurnool Medical College and GGH

Abstract: *Waardenburg syndrome type 4 is a rarely occurring pediatric emergency in association with mega colon and Hirschsprung's disease. We hereby report a case of type 4 Waardenburg syndrome with Hirschsprung's disease coming up for an emergency decompressive colostomy. The anaesthetic implications and the multisystem involvement relevant to the syndrome are discussed and presented in this paper.*

Keywords: Shah-Waardenburg syndrome, stomach decompression, aspiration pneumonitis, Isolate-P

1. Introduction

- 1) Waardenburg syndrome is an uncommon autosomal inherited and genetically heterogenous disorders of neural crest cell development. Waardenburg syndrome belongs to neurocristopathies, disorders caused by an alteration in the migration of the neural crest cells during embryonic phase.
- 2) The neural crest cells are important for the formation of several parts of body i.e. melanocytes, inner ear and enteric nervous system. Other features very rarely associated with Waardenburg syndrome include vestibular symptoms, urinary system abnormalities, neural tube defects, Sprengel shoulder anomaly, cleft lip and palate, facial nerve palsy, twisted and plicated tongue, laryngomalacia, severe cyanotic cardiomyopathy, multiple muscle contractures, limited neck movements and electrolyte imbalance, which can lead to difficulties in anaesthetic management.
- 3) It equally affects both male and female, all races with an incidence of 1 in 42,000.
- 4) Four types of Waardenburg syndrome are described. Type 1 2 3 and 4. Type 4 is the association of Waardenburg syndrome with Hirschsprung's disease. This type is called Shah-Waardenburg syndrome. The classical presentation of Shah-Waardenburg syndrome includes Hirschsprung's disease, sensory neural hearing loss, depigmentation of hair, skin and iris. In Shah-Waardenburg syndrome, the aganglionic segment may be long and may have total colonic or total intestinal aganglionosis.

2. History and Examination

2.1 Preoperative Evaluation

A 3 days old term child weighing 3kg was admitted to the department of paediatric surgery with complaints of bilious vomiting and not passing meconium since birth. Baby had

depigmentation of hair and iris. Impression of ultrasound abdomen was intestinal obstruction.



2.2 Preoperative Preparation

After written consent from the parent; Upon arrival in operation theatre, standard monitoring viz ECG, pulse oximetry and EtCO₂ were established. Heart and lungs were clinically normal as per auscultation.

Preoperative vital signs including heart rate, respiratory rate and saturation were within normal limits. An infant feeding tube was passed to decompress the stomach and then removed before induction and meanwhile IV line was secured.

2.3 Intubation and Maintenance

After preoxygenation, anaesthesia was induced with thiopentone 15mg iv and continued with O₂ and sevoflurane by inhalation. Ability to expand the chest with mask ventilation was checked and found to be okay. Then, atracurium 1.5mg iv was administered to facilitate

Volume 9 Issue 7, July 2020

www.ijsr.net

Licensed Under Creative Commons Attribution CC BY

orotracheal intubation. After securing the endotracheal tube (3.0 uncuffed) fentanyl 4mcg iv was given and patient ventilated in Jackson ree's circuit, anaesthesia was maintained with oxygen, air, volatile and relaxant technique (O₂+ Sevoflurane 0.8%+ Inj atracurium 0.1mg/kg).

The surgery was transverse colostomy. Duration of surgery was 60min, and appropriate iv fluids were administered as dextrose and ringer lactate.

2.4 Postoperative Phase

The infant reversed at the end of surgery, and continued appropriate fluid in postoperative period. One-fourth of postoperative fluid requirement was given as Isolate-P in the postoperative period. After reversal with glycopyrrolate + neostigmine patient shifted to the neonatal intensive care unit for nursing and postoperative care.

3. Discussion

Waardenburg syndrome is a genetic disorder (autosomal dominant) that may be evident at birth (congenital). Primary features include facial abnormalities, unusual depigmentation of the hair/skin and their iris and congenital deafness.

Hirschsprung's disease is the most often observed gastrointestinal abnormality and is seen typically with type 4 Waardenburg syndrome. The facial abnormalities are of relevance to the anaesthetist and can sometimes contribute to a difficult intubation, due to rounded upturned nasal tip, philtrum of the upper lip, fully pouted lips, mandibular prognathism, cleft palate and cleft lip and incomplete closure of the roof of the mouth. All the above abnormalities can lead to a difficulty in mask ventilation with O₂ before intubation, difficulty with laryngoscopy and subsequent technique of endotracheal intubation.

Hirschsprung's disease (also known as aganglionic megacolon) can lead to abnormality in peristalsis, a bloated abdomen, risk of vomiting, failure to thrive and grow and electrolyte abnormalities that can derail the process of recovery, at the end of surgery.

The condition of Waardenburg syndrome requires a well-coordinated treatment plan involving the dermatologist, orthopaedician, ophthalmologist, gastroenterologist, paediatrician and paediatric surgeon and not the least the anaesthesiologist, who has to bailout the infant from the crisis of anaesthesia and postoperative care. At a later stage, speech therapist and language therapist along with paediatric faciomaxillary surgeon to correct multiple abnormalities, may be needed.

Hirschsprung's disease should be corrected as early as possible, and if neglected can lead to toxic megacolon, peritonitis and bowel perforation, and is associated with the risk of aspiration pneumonitis at induction and intubation if not attended properly. The definitive diagnosis is made by the colon or rectal biopsy which depicts absence of ganglion cells. The treatment of such infants includes suitable iv fluids, antibiotics and colon decompression procedures.

Nowadays laparoscopy is also sometimes used and gaining in popularity to tackle the above conditions.

References

- [1] Peker K, Ergil J, Öztür i. Anaesthesia Management in a Patient Waardenburg Syndrome and Review of the Literature. *Turk J Anaesthesia Reanim*. 2015;4(5):360-362. doi:10.515/TJAR.2015.52714
- [2] Mahmoudi A, Rami M, Khattala K, Elmadi A, Afifi MA, Youssef B. Shah-Waardenburg syndrome. *Pan Afr Med J*. 2013;14:60. doi:10.11604/pamj.201.14.60.1543
- [3] Thapa R, Mallick D, Ghosh A, Ghosh A. Waardenburg syndrome associated with laryngomalacia. *Singapore Med J*. 2009;50(12):e401-e403.

Author Profile

Dr. C. G. Raghuram, M. D., Professor, Department of Anesthesiology and Critical Care, Kurnool Medical College and Government General Hospital, Kurnool, Andhra Pradesh.

Dr. R. Kondareddy, M.D., Associate Professor, Department of Anesthesiology and Critical Care, Kurnool Medical College and Government General Hospital, Kurnool, Andhra Pradesh.

Dr. M. Sharada MD., Assistant Professor, Department of Anesthesiology and Critical Care, Kurnool Medical College and Government General Hospital, Kurnool, Andhra Pradesh.

Dr. Y. Murali Prabakar, M.D. Assistant Professor, Department of Anesthesiology and Critical Care, Kurnool Medical College and Government General Hospital, Kurnool, Andhra Pradesh.

Dr. G. D. SREE MOUNIKA, Post Graduate, Department of Anesthesiology and Critical Care, Kurnool Medical College and Government General Hospital, Kurnool, Andhra Pradesh.