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Anesthesia

Waardenburg Syndrome Anesthetic Management: Two Cases Experiences

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Abstract **Case Report**

This article presents the anesthetic management of two cases of Waardenburg syndrome (WS) in patients undergoing cochlear implantation. Mutations in genes that control the neural crest cell migration and division during the embryonic period are the hallmark of the hereditary disorder known as WS. White forelock, bright blue eyes, heterochromia from iris coloring deficiency, unusual facial deformities, and sensorineural deafness are the key phenotypic characteristics of WS. The anesthetic management of patients with WS poses challenges due to their multisystem abnormalities.

Keywords: Waardenburg syndrome, anesthetic management, cochlear implantation.

INTRODUCTION

Described first in 1951 by Waardenburg, The Waardenburg syndrome (WS) is caused by mutations in one of the multiple genes that control the movement and division of neural crest cells during the embryonic period. The primary phenotypic features of this syndrome are a white forelock, shining blue eyes, or heterochromia because of iris pigmentation deficiency, different facial anomalies (laterally moved inner canthi and a broad nasal bridge), loss of sensorineural hearing, and anomalies of the upper extremities [1].

The anaesthetic management of patients with WS constitutes a challenge for anesthesiologists due to multisystem abnormalities. As a result, we describe the anesthetic care given to two WS patients who had cochlear implantation.

CASE REPORT

Case 1

An ear, nose, and throat department admission involved a 4-year-old kid. for congenital deafness. Given to characteristic phenotypic findings (wide nasal root, bright blue iris [Figure 1], bilateral sensory deafness), At the age of nine months, type II WS was identified. Airway exam during the pre-anesthesia assessment showed a Mallampati class II with a 3 fingerbreadth thyromental distance. Neck flexibility and mouth openness were both normal. The respiratory and cardiac were normal. Abdominal exam finds umbilical hernia [Figure 2]. His serum electrolytes and hemogram both fell within acceptable ranges.

After a preoperative consent was obtained, the patient was kept nil per os for 6 hours. Standard monitoring was put into effect as soon as we entered the arrival operation room. After starting anaesthesia induction with 8% sevoflurane in O2, a 24 gauge intravenous (IV) cannula was placed.Ventilation was simple. Following the administration of Fentanyl 3 mg/kg, Propofol 2 mg/kg, and 0.6 mg/kg of Rocuronium bromide, his trachea was intubated using a 3 mm cuffed trachea tube. The patient was extubated at the end of the operation after responding to orders, opening her eyes, and raising her head. The post-anesthesia care unit received him after his relocation. The postoperative course went without incident.

Fig. 1: Bilateral blue iris

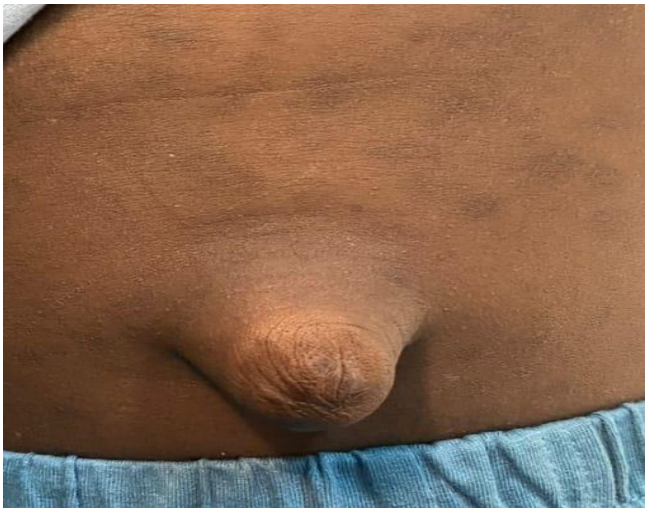

Fig. 2: Umbilical hernia

Case 2

Due to congenital deafness, a 6-year-old girl was admitted to the ear, nose, and throat department. At the age of 13 months, type II WS was identified due to the morphological characters (heterochromia irides [Figure 3], broad nasal root, and bilateral sensory deafness). Airway examination during the pre-anesthesia evaluation revealed a Mallampati class I with a 3 fingerbreadth thyromental distance. The range of motion of the neck and the mouth opening were acceptable. The cardiovascular and respiratory systems were also normal. His serum electrolytes and hemogram both came back normal. The anesthetic administration was identical to the previous case. His trachea was intubated with a 3.5mm cuffed endotracheal tube. Body motions and spontaneous breathing began 15 minutes after the operation. Following that, the patient was extubated and transported to the post-anesthesia care unit.

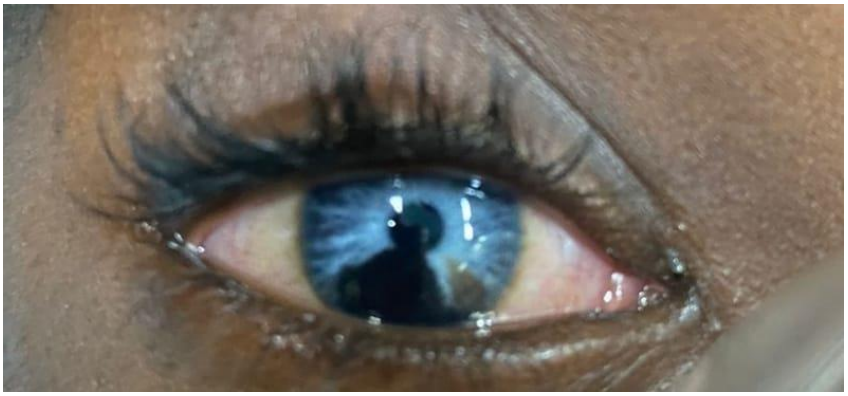

Fig. 3: Heterochromia irides

DISCUSSION

WS is an uncommon genetic condition that interferes with neural crest cells' ability to migrate and differentiate, especially the lineage-forming melanocytes. Melanocyte involvement causes a lack of pigmentation, which includes vitiligo and hypopigmented skin areas as well as the recognizable white hair tuft, heterochromia of the iris, or very blue eyes. Lack of melanocytes, organ of Corti malformations, or organ of Corti absence in the inner ear cause sensorineural deafness [2]. WS classifications, which are divided into four subclasses, Differentiate phenotypic presentations according to the location and intensity of gene mutation. Type 1 primarily affects facial and hair, skin, and eyes, with a lower proportion reporting sensorineural hearing loss. Type 2 WS has a higher rate of hearing loss but does not have the symptoms of dystopia canithorum seen in type 1 patients. A small subset, known as type 3 WS or Klein-Waardenburg syndrome, has extra musculoskeletal anomalies, most notably flexion contractures, carpal bone fusion, and/or syndactyly. Type 4 WS is a rare WS variant linked to long-segment Hirschsprung's disease [3, 4]. Given the prevalence of deafness, heterochromia iridis, and the lack of dystopia canthorum and Hirschsprung disease, both of our patients fit the diagnostic criteria for WS 2.

Anecdotally, it has been suggested by the limited literature on anesthesia care for these individuals that endotracheal intubation and airway control may present challenges [5].

Airway management difficulties are common symptoms of patients with WS. According to reports, Neural crest cells are the common ancestors of pigment-producing cells and laryngeal cartilage. Melanocytes and other Neural crest- derived cells exhibit abnormal proliferation, survival, migration, or differentiation due to improper development of Neural crest [6]. In WS, aberrant differentiation and migration of NC-derived cells may produce laryngomalacia and epiglottis redundancy. The preoperative airway evaluation in our patients was normal. We prepared different types of endotracheal tubes, For a possible problematic airway, video laryngoscopy and a laryngeal mask airway were used, and anesthesia induction was overseen by two anesthesiologists. Video laryngoscopy visualization was normal. Resistance was encountered during tracheal intubation after anesthesia induction, after rotating and adjusting the endotracheal tube's size, the tracheal tube was successfully intubated. In another report, a 4-year-old child had normal direct laryngoscopy but had problems passing a 4.5mm endotracheal tube because of sub-glottic resistance.. The trachea was eventually intubated with a 3.0 mm uncuffed endotracheal tube and the case proceeded uneventfully [7]. The same for our patients, We utilized a small size tube because the passage of the tracheal tube for intubation was difficult. According to the patient's preoperative history and physical examination, a preoperative consultation with otolaryngology may be recommended.

In addition to the possibility of difficulties during endotracheal intubation, congenital abnormalities of the glottis, larynx, and trachea, such as laryngomalacia and subglottic stenosis, have been observed [7, 8]. Congenital heart disease is another typical clinical symptom of WS [9, 10]. The WS described atrial septal abnormalities and embryologic structural persistence (persistent left superior vena cava, dilated cardiomyopathy) [11-13]. Both our patients were asymptomatic and the echocardiograms were normal.

CONCLUSION

Careful consideration must be given to preoperative evaluation, due to the presence of concomitant system defects, and difficulties of airway management. Pediatricians and surgeons should work together with anesthesiologists to manage these patients because they frequently need multiple surgeries.

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