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Case Report / Olgu Sunumu

Anaesthesia Management in a Patient with Waardenburg Syndrome and Review of the Literature

Waardenburg Sendromlu Hastanın Anestezi Yönetimi ve Literatür İncelemesi

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Abstract / Öz

Waardenburg syndrome is a rare autosomal dominant disease that may cause hearing loss, pigmentary abnormalities, neurocristopathy and partial albinism. Incidence is estimated as 2%–3% among the cases of congenital deafness and 1/42,000 of the general population. Children with Waardenburg syndrome usually require anaesthesia for the cochlear implant operation in early age. The features of the syndrome that may bear importance for anaesthetic management are laryngomalacia, multiple muscle contractures, limited neck movements, cyanotic cardiopathy and electrolyte imbalance. Patients with Waardenburg syndrome stand for difficult airway. We aimed to report anaesthetic management of a child with Waardenburg syndrome who underwent surgery for cochlear implantation.

Keywords: Waardenburg syndrome, anaesthetic management, airway

Waardenburg sendromu, işitme kaybına, pigment anormalliklerine, nörokristopatiye ve parsiyel albinizme neden olabilen nadir bir otozomal dominant hastalıktır. İnsidansı, kalıtsal işitme kaybı olguları arasında %2-3 ve genel popülasyonda 1/42.000 olarak tahmin edilmektedir. Waardenburg sendromlu çocuklar sıklıkla erken dönemde koklear implantasyon için anesteziye ihtiyaç duyarlar. Sendromun anestezi yönetimi için önem arz eden belirtileri laringomalazi, multipl kas kontraksiyonları, kısıtlı boyun hareketleri, siyanotik kardiyopati ve elektrolit dengesizliğidir. Waardenburg sendromlu hastalar zor havayolu için adaydırlar. Koklear implantasyon için cerrahi geçiren Waardenburg sendromlu bir çocuğun anestezi yönetimini bildirmeyi amaçladık.

Anahtar kelimeler: Waardenburg sendromu, anestezi yönetimi, havayolu

Introduction

Waardenburg syndrome (WS) is a rare, inherited and genetic disorder of neural crest cell development that is most often characterized by achromia of the hair, skin (or both), congenital deafness, partial or total heterochromia iridis, hypertrichosis of the medial part of the eyebrows, broad and elevated nasal root and dystopia canthorum. Furthermore, WS has features that can be important for anaesthetic management, including laryngomalacia, multiple muscle contractures, limited neck movements, cyanotic cardiomyopathy and electrolyte imbalance. D. J. Waardenburg, an ophthalmologist, first defined the syndrome in 1951 (1).

Literature regarding the anaesthetic management of patients with WS is limited. We aimed to report the anaesthetic management of a 4-year-old child with Waardenburg syndrome who underwent surgery for cochlear implantation, and we reviewed the literature.

Case Presentation

A 4-years-old girl was admitted to the department of ear, nose and throat with bilateral decreased hearing, more on the right side. Abnormality of the inner ear was detected in the radiological examination. Because of hypoplasia and the absence of the posterior semicircular channel and hypoplasia of the cochleas, clinicians decided the implantation of cochlea.

Her hearing loss was diagnosed when she was 8 months old, and a hearing aid was fitted. She was using the hearing aid in her left ear since childhood. When she was 1.5 years, it was found that she had short bowel syndrome and no development of speech. The child had a definitive diagnosis of WS since the age of 1.5 years according to the Waardenburg Consortium at

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a university hospital (2). She had a white forelock on the anterior scalp, characteristic brilliant blue iris, broad nasal root, dystopia canthorum, hypoplastic nasal alae, bilateral sensory neural deafness and short bowel syndrome. The patient did not have any neurological defect or any skin abnormality. She has not received any medication and had no drug allergies and no family history of anaesthesia-related problems. Her laboratory tests, electrocardiography and chest radiography were assessed as normal. Mallampati classification was assessed as Class 3. The airway examination was normal except the view of mouth. However, we prepared difficult airway materials, such as laryngeal mask airway, endotracheal cuffed and uncuffed tubes at different sizes, and also fiberoptic bronchoscopy for possible difficult airway.

After written informed consent was obtained from her parent, she was orally premedicated with 0.5 mg kg^{-1} midazolam. Upon arrival in the operating room, standard monitoring, including ECG, pulse oximetry and end-tidal capnography, was applied. During bilateral auscultation of the lungs, neither rales nor rhonci were heard, and heart rhythm was normal. Preoperative vital signs, including heart, respiration rate and blood pressure, were within normal limits. Her preoperative vital signs were heart rate 132 bpm, arterial blood pressure 95/50 mmHg and SpO_2 was 97% in room air. Anaesthesia induction with 6% sevoflurane in O_2 was initiated and a 24-gauge intravenous (IV) cannula was inserted after induction. No problem occurred during a mask ventilation of the patient. Rocuronium bromide (0.6 mg kg^{-1}) was administered to facilitate orotracheal intubation. Twenty micrograms of fentanyl was administered. We calculated the endotracheal tube size by the known formulas as age/4+4 for uncuffed and age/4+3 for cuffed. However, the patient could not be intubated with number 5.0 or 4.5 endotracheal tube. Therefore, the patient was intubated with a number 3.0 uncuffed endotracheal tube (Laryngeal view of the patient was Cormack–Lehane Grade 2). Anaesthesia was maintained with nitrous oxide 50% in oxygen and end-tidal CO_2 was maintained at 30–35 mmHg. Intraoperative vital signs were stabile: blood pressure: 92–115/50–60 mmHg, heart rate: 110–127 bpm, oxygen saturation: 100% and temperature: 36.3°C. Operation continued for 90 min. Intraoperative electrolyte values were within normal range. At the end of the operation, the patient spontaneously opened her eyes, responded to verbal command and was able to lift her head. She was subsequently extubated. Both the extubation and postoperative course were uncomplicated.

Discussion

Waardenburg syndrome is a rare autosomal dominant syndrome that was first described by Petrus Johannes Waardenburg. It is classically characterised by lateral displacement of the medial canthi and lacrimal punctae, broad and high nasal root, hypertrichosis of medial part of the eyebrows, partial or total heterochromia iridis, white forelock and congenital deaf mutism (1, 3). It equally affects both male and females and all races with an incidence of one in 40,000 (4). It is clinically and genetically heterogeneous and is classified into four types (type 1–4) on the basis of the presence of variable clinical characteristics and additional symptoms (3). Types 1 and 2 are the most common types of the syndrome, whereas types 3 and 4 are rare. Type 4 is also known as Waardenburg–Shah syndrome (association of Waardenburg syndrome with Hirschsprung disease). Type 1 (WS 1) is characterised by congenital sensorineural hearing loss; heterochromia iridis; partial hypopigmentation of the hair, including premature greying and lateral displacement of the inner ocular canthi (dystopia canthorum). Type 2 (WS 2) is distinguished from WS 1 by the absence of dystopia canthorum. WS 3 or Klein–Waardenburg syndrome is similar to WS 1 but includes upper limb muscle abnormalities. WS 4 or Waardenburg–Shah syndrome has features of Hirschsprung disease in addition to WS 2.

A careful clinical evaluation is necessary to differentiate the various types of WS. According to the diagnostic criteria proposed by the Waardenburg Consortium, a person must have two major or one major plus two minor criteria to be diagnosed as WS type 1 (2). In our case, there was no history suggestive of familial childhood neurosensory hearing loss, maternal gestational infections, birth asphyxia, head trauma, postnatal pathological hyperbilirubinaemia, neonatal meningitis and exposure to drugs causing otologic dysfunctions such as aminoglycosides and diuretics.

This case met the above-mentioned criteria for the diagnosis of WS 1. The presence of white forelock on the anterior scalp, characteristic brilliant blue iris, broad nasal root, dystopia canthorum, hypoplastic nasal alae, bilateral sensory neural deafness and short bowel syndrome establishes the diagnosis of WS 1.

Waardenburg syndrome remains, at the fundamental level, a disorder of the abnormal neural crest cell differentiation and migration. Congenital anomaly of the larynx has been previously reported in association with the syndrome (5). In that case, direct laryngoscopic visualisation revealed an omega-shaped, floopy epiglottis with redundant aryepiglottic folds and prominent arytenoids bilaterally. Therefore, they reported that both pigment-producing cells and laryngeal cartilages have a common source of origin, which is the neural crest cells; aberrant differentiation and migration of the neural crest-derived cells may thus explain the occurrence of laryngomalacia in WS. In this case, formal direct laryngoscopic visualization revealed normal. However, we could not insert an endotracheal tube with normal size into trachea; therefore, we used a number 3.0 uncuffed tube. We could not describe tracheal narrowing or subglottic stenosis. Only the knowledge may be relevant on this matter that theracheal and laryngeal cartilages are the part of the mesectoderm which derivated by neural crest, and WS is caused by a mutation in the PAX-3

gene, which in mice was found to be expressed in the dorsal part of the neural tube, including the neural crest (6). For anaesthesia practice, the importance of this syndrome is the possible difficulty of airway management, e.g. the case report of WS reported by Michalek et al. (7). They have described the successful management of difficult airway in an adult patient with WS using fiberoptic bronchoscope with I-gel as conduit. Spastic dysplegia with multiple muscle contractures and microcephaly, spastic torticollis, short thyromental distance and significantly limited neck movements were the difficulties of the insertion of a laryngoscope blade with the fixed position of the head and muscle contractures (7).

One of the other clinical features associated with WS is severe cyanotic cardiopathy (8). The importance of perioperative vital sign screening and preoperative evaluation is that the unusual combination of Waardenburg syndrome with severe congenital heart disease has been observed in a child with WS type 1 (8) and dilated cardiomyopathy in a child with type 2 (9). This child's preoperative cardiovascular physical examination was normal and vital signs were normal during anaesthesia.

Waardenburg syndrome type 4 is an unusual variant of WS associated with long-segment Hirschsprung's disease and has difficulties due to congenital deafness and blindness, demonstrated perioperative problems, malnutrition and electrolyte imbalance (10). A 5-year-old child with WS was reported by Kfoury et al. (11) with facial dysmorphism, operated cleft palate and spina bifida using anaesthetic technique of pudental nerve block. The experience of two cases with WS syndrome recommended no particular anaesthetic technique and, different volatile anaesthetics and muscle relaxants could be used (12).

Conclusion

Anaesthesia-related information regarding Waardenburg syndrome is very limited. However, specific facial features and muscle contractures may cause difficulties in both direct laryngoscopy and tracheal intubation. When anaesthesiologists encounter a child with white forelock, they should keep in mind the differential diagnosis and variants of WS. A careful preoperative clinical evaluation, determining other system abnormalities association with WS and keeping difficult airway equipment for anaesthetic management is required.

Informed Consent: Written informed consent was obtained from patient who participated in this case.

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References

1. Milunsky JM. Waardenburg Syndrome Type I. (Ed) Pagon RA, Adam MP, Ardinger HH. (1993-2014) GeneReviews® [Internet]. The web site: http://www.ncbi.nlm.nih.gov/books/NBK1531/
2. Farrer LA, Grundfast KM, Amos J, Arnos KS, Asher JH Jr, Beighton P, et al. Waardenburg syndrome (WS) Type I is caused by defects at multiple loci, one of which is near ALPP on Chromosome 2: First report of the WS Consortium. Am J Hum Genet 1992; 50: 902-13.
3. Krishtul A, Galadari I. Waardenburg syndrome: Case report. Int J Dermatol 2003; 42: 651-2. [CrossRef]
4. Read AP, Newton VE. Waardenburg syndrome. J Med Genet 1997; 34: 656-65. [CrossRef]
5. Thapa R, Mallick D, Ghosh A, Ghosh A. Waardenburg syndrome associated with laryngomalacia. Singapore Med J 2009; 50: 401-3.
6. Baldwin CT, Hoth CF, Amos JA, da-Silva EO, Milunsky A. An exonic mutation in the HuP2 paired domain gene causes Waardenburg's syndrome. Nature 1992; 355: 637-8. [CrossRef]
7. Michalek P, Hodgkinson P, Donaldson W. Fiberoptic intubation through an I-gel supraglottic airway in two patients with predicted difficult airway and intellectual disability. Anesth Analg 2008; 106: 1501-4. [CrossRef]
8. Mathieu M, Bourges E, Caron F, Piussan C. Waardenburg's syndrome and severe cyanotic cardiopathy. Arch Fr Pediatr 1990; 47: 657-9.
9. Baspinar O, Gungor M, Parlak M, Coşkun Y. Waardenburg syndrome type II and dilated cardiomyopathy in a child. Pediatr Int 2006; 48: 100-2. [CrossRef]
10. Mizushima A, Nitami K, Kikuchi T, Kugimiya T, Ohya T, Miyano T. Anesthetic problems in a child with Waardenburg's syndrome and Hirschsprung's disease. J Anesth 1996; 10: 144-6. [CrossRef]
11. Kfoury T, Staiti G, Baujard C, Benhamou D. Pudendal nerve block by nerve stimulation in a child with Waardenburg disease. Paediatr Anaesth 2008; 18: 1267-8. [CrossRef]
12. Ambi US, Adarsh ES, Hatti R, Samalad V. Anesthetic manegement of Shah-Waardenburg syndrome: Experience of two cases and review of literature. Saudi J Anaesth 2012; 6: 172-4. [CrossRef]