

# ANESTHETIC CHARACTERISTICS AND AIRWAY EVALUATION OF PATIENTS WITH WEILL- MARCHESANI SYNDROME<sup>†</sup>

WALEED RIAD<sup>\*</sup>, MARWAN ABOUAMMOH<sup>\*\*</sup>  
AND MAMDOUH FATHY<sup>\*\*\*</sup>

## Summary

Weill-Marchesani syndrome (WMS) is a genetic connective tissue disorder associated with fibrous tissue hyperplasia. Weill-Marchesani syndrome is characterized by short stature, broad head and other facial abnormalities such as hypoplastic maxilla and distinctive ocular abnormalities. Joint stiffness is one of the features of this syndrome. We report 5 cases with classical features of WMS who were subjected to different ophthalmic procedures. To the best of our knowledge, this is the first series on the anesthetic management of this rare syndrome. We observe that patients with WMS can present for cataract, glaucoma as well as retinal surgery. Special consideration should be given to difficult intubation, cardiac abnormalities and patient positioning.

**Key Words:** Weill-Marchesani syndrome, difficult intubation, patient positioning.

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<sup>\*</sup> MD, AB, SO Consultant Anesthesiologist, Department of Anesthesia.

<sup>\*\*</sup> MD, Consultant Anesthesiologist, Department of Anesthesia.

<sup>\*\*\*</sup> Resident, Department of Ophthalmology, King Khaled Eye Specialist Hospital, Riyadh, Saudi Arabia.

Address for correspondence: Dr Waleed Riad, Department of Anesthesia, King Khaled Eye Specialist Hospital. P.O. Box: 7191, Riyadh 11462, Kingdom of Saudi Arabia. Tel: 9661-1-482-1234-3215. Fax: 966-1-482-1908. E-mail: waleed\_riad@yahoo.com.

## Introduction

Weill-Marchesani syndrome (WMS), also known as Spherophakia-Brachymorphia Syndrome, is a rare genetic connective tissue disorder associated with fibrous tissue hyperplasia. It was first described by Georges Weill in 1932<sup>1</sup> and further delineated by Oswald Marchesani in 1939<sup>2</sup>. It has been suggested that it may have autosomal recessive (AR) or autosomal dominant (AD) inheritance. Autosomal dominant families with WMS were linked to chromosome 15q21.1, the fibrillin-1 gene, while autosomal recessive WMS has recently been mapped to chromosome 19p13.3-p13.2<sup>3</sup>. This syndrome has not been adequately brought to the attention of the anesthesiologist because of its rarity.

The Weill-Marchesani syndrome is characterized by an unusually short stature, broad head (brachycephaly), and other facial abnormalities such as hypoplastic maxilla, thickened skin, hand defects, including unusually short fingers (brachydactyly), and distinctive ocular abnormalities<sup>4</sup>. The latter typically include unusually small, round lenses of the eyes (spherophakia) that may be prone to dislocating lens (ectopia lentis), as well as glaucoma and detached retina<sup>3</sup>. Due to such abnormalities, affected individuals may have varying degrees of visual impairment when presenting for anesthesia and surgery. Joint stiffness is one of the features of this syndrome. Rennert described an affected 9-year-old boy with joint stiffness who had difficulty in extending his arms over his head<sup>5</sup>. Another report pointed to the difficulty of tracheal intubation due to facial abnormalities and joint stiffness<sup>6</sup>.

While only 2 case reports have been described in the anesthesia literature<sup>6,7</sup>, we report 5 cases with classical features of Weill-Marchesani syndrome (Table 1) who were subjected to different ophthalmic procedures between 1992 to 2004. To the best of our knowledge this is the first case series on the anesthetic management of this rare syndrome.

Table 1

*Patients with Weill-Marchesani Syndrome who underwent general anesthesia at the King Khaled Eye Specialist Hospital between 1992-2004*

| No | Year | Sex | Age (years) | Type of Surgery   | Difficult Intubation |
|----|------|-----|-------------|-------------------|----------------------|
| 1. | 1992 | M   | 44          | Glaucoma/Cataract | No                   |
| 2. | 1995 | M   | 17          | Glaucoma          | No                   |
| 3. | 2000 | M   | 13          | Retina            | No                   |
| 4. | 2001 | M   | 25          | Retina            | Yes                  |
| 5. | 2004 | M   | 66          | Glaucoma/Cataract | Yes                  |

### Case Reports

**Case 1:** 44-year-old male patient, ASA class II, was admitted in 1992 with total retinal detachment in the right eye with no light perception. The left eye suffered from open angle glaucoma, lens subluxation and pre-senile immature cataract with visual acuity of 20/200 after correction. The planned procedure was trabeculectomy combined with extracapsular cataract extraction in the left eye under general anesthesia.

Anesthesia was induced with thiopental 5mg/kg, fentanyl 2µg/kg and tracheal intubation was facilitated by atracurium 0.5mg/kg. Tracheal intubation was easy. Anesthesia was maintained with N<sub>2</sub>O, O<sub>2</sub> mixture with isoflurane. Surgery lasted 180 minutes. Residual muscle block was antagonized by neostigmine and glycopyrrolate. The patient was discharged on the second day with no complications.

**Case 2:** A 17-year-old boy, ASA class II, was admitted in 1995 with microspherophakia with partial lens subluxation in both eyes. The right eye suffered from uncontrolled intraocular pressure with severe optic nerve cupping. His right visual field was severely constricted while the left one was moderately constricted. A laser iridotomy was performed in both eyes under general anesthesia. Visual acuity was 20/50 right eye and 20/160 left eye, and family history was negative for Marchesani syndrome. Anesthesia was induced with propofol 2mg/kg, fentanyl

2 $\mu$ g/kg and tracheal intubation was facilitated by atracurium 0.5mg/kg. Tracheal intubation was easy. Anesthesia was maintained with N<sub>2</sub>O, O<sub>2</sub> mixture with isoflurane. Surgery lasted for 135 minutes. Residual muscle block was antagonized by neostigmine and glycopyrrolate.

**Case 3:** A 13-year-old boy, ASA class I, was admitted in 2000 for pars plana lensectomy and pars plana vitrectomy in both eyes, secondary to dislocated lenses and retinal detachment. Halothane was used for inhalational induction. Fentanyl 2  $\mu$ g/kg and atracurium 0.5mg/kg were administered after loss of consciousness followed by smooth intubation. Anesthesia was maintained with N<sub>2</sub>O, O<sub>2</sub> mixture with isoflurane. Surgery lasted for 55 minutes. Residual muscle block was antagonized by neostigmine and glycopyrrolate. The child was discharged successfully in the same day.

**Case 4:** A 25-year-old male patient, ASA class I, admitted in 2001 with a diagnosis of dislocated lens, to undergo para plana lensectomy and pars plana vitrectomy in the left eye. Prior to this admission, he was involved in a road traffic accident 9 months earlier with severe head and facial trauma. His body mass index (BMI) was 24 kg/m<sup>2</sup>. The patient had a history of surgical correction of LeFort I fracture, and a tracheostomy scar was observed. Anesthesia was induced by fentanyl 2 $\mu$ g/kg and propofol 2mg/kg. As it was easy to maintain manual ventilation, atracurium 0.5mg/kg was given. During intubation, only the tip of the epiglottis was visualized with great difficulty. With the help of McCoy laryngoscope and gum elastic bougie, the trachea was intubated at the fifth attempt. Anesthesia was maintained with N<sub>2</sub>O, O<sub>2</sub> mixture with sevoflurane. Surgery lasted for 150 minutes. Extubation was done when the patient was fully awake.

**Case 5:** A 66-year-old male patient, ASA class II, was admitted in 2004 with the diagnosis of glaucoma in the right eye secondary to prior cataract surgery. He was to undergo pars plana lensectomy and Ahmed implant under general anesthesia. His weight, height and BMI were 60kg, 148cm and 27.4kg/m<sup>2</sup> respectively. Visual acuity was counting fingers at 1-2 feet right eye, and 20/40 left eye. Anesthesia was induced with

fentanyl 2µg/kg and propofol 2mg/kg. Because of the ease of manual ventilation with normal preoperative airway assessment, atracurium 0.5mg/kg was given. Nothing could be visualized during the first attempt of laryngoscopy. Even with the help of the McCoy laryngoscope, and with repositioning and maximum laryngeal compression, only the tip of epiglottis could be observed with great difficulty (Grade III of Cormak and Lehane classification). Tracheal intubation was successful with the use of a gum elastic bougie. Anesthesia was carried out in the same standard way as with other patients.

## Discussion

From these series, it is observed that patients with WMS can present for cataract and glaucoma surgery as well as repair of retinal detachment. These complications are an integral part of this syndrome. Some of these patients may be unexpectedly difficult to intubate but easy to ventilate. This type of patients may undergo elective surgery because their primary pathology is associated with this syndrome or they may present for any emergency procedure.

In this series, 2 of the patients were difficult to intubate their tracheas. Karabiyik<sup>6</sup> reported a case of WMS who was difficult to intubate and ventilate. His patient had joint stiffness and limited mouth opening. Tracheal intubation was performed with the help of an intubating laryngeal mask. Dal et al<sup>7</sup> report use of the laryngeal mask airway (LMA) to secure the patient's airway during surgery while he was breathing spontaneously without neuromuscular blockage. McKusick<sup>8</sup> reported that patients with WMS can have hypoplastic maxilla, joint stiffness and arthritis. Giordano et al<sup>9</sup> reported a case of WM syndrome showing progressive joint stiffness; an unusual feature was the presence of "primary" osteoporosis in the 28-year-old affected man who was 130cm tall. One feature of this syndrome is malformed and malaligned teeth<sup>7</sup>. Karabiyik<sup>6</sup> performed magnetic resonance imaging postoperatively which revealed laryngeal stenosis. All of these factors could attribute to intubation difficulty.

Contrary to the above no difficulty was met in ventilating any of our patients. Although joint stiffness is one of the fibrous tissue manifestation of WMS, preoperative airway assessment was normal in all our patients. Anesthesiologists who evaluated patients in the perioperative period failed to suspect and/or detect any airway abnormality.

Despite the well documented difficulty of intubation, in case no. 4 it was hard to differentiate whether the difficulty in intubation could be related to the syndrome itself or to the previous incident of facial trauma, surgical correction and tracheostomy, or to a combination of all these factors. Joint stiffness and joint prominence are due to dystrophia mesodermalis hyperplasia, and this may also lead to difficulty of airway control and intubation<sup>6</sup>.

Occasionally, cardiac abnormalities can complicate the clinical course of WMS. Ferrier et al<sup>10</sup> reported an affected 11-year-old girl who also had subvalvular fibromuscular aortic stenosis. Dal et al<sup>7</sup> reported a systolic murmur during patient's examination but pediatric cardiology consultation did not reveal any pathological finding. Although none of our patients had any cardiac abnormalities, comprehensive cardiac evaluation is highly recommended in these types of patients.

Weill-Marchesani syndrome is also associated with multiple skeletal abnormalities such as joint stiffness<sup>5</sup>, joint arthritis and limited movement of fingers and modeling defect<sup>8</sup>. Therefore, special attention should be directed to patient positioning during surgery and in the recovery room until the patient is fully awake.

## **Conclusion**

WMS is a rare condition which could present for surgery as a routine ophthalmic procedure or an emergency procedure. Special consideration should be paid for difficult intubation, cardiac abnormalities and patient positioning.

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