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CLINICAL REPORT

Anesthetic management of a child with both Marfan syndrome and Turner syndrome

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Abstract Marfan syndrome is an autosomal dominant heritable disorder of the connective tissue that involves primarily the skeletal, ocular, and cardiovascular systems. Turner syndrome is a genetic disorder resulting from partial or complete X chromosome monosomy. We report the anesthetic management of a case of Marfan–Turner syndrome, which is the first such case to appear in the literature to our knowledge. A 3 year old ASA III girl was scheduled to undergo minor plastic surgery. She had a short webbed neck, prognathism, micrognathia, low-set ears, and a high palate. Her anterior and posterior facial heights were long. She had growth retardation, pectus excavatum, and joint laxity. She also had high-degree mitral insufficiency, mitral valve prolapse, and an atrial septal defect. After sevoflurane induction, the airway was secured using a size 2 LMA without any difficulty in the spontaneously breathing patient. Her blood pressure was within normal limits, no arrthymia occurred, and anesthesia was uneventful. Special care should be given to syndromic patients. Prior medical evaluations and any prior anesthetic history can help to focus preoperative evaluations and planning. Preoperatively targeting relevant organ systems, any anatomic or laboratory abnormalities that can be optimized, and perioperative airway management are all key to a successful outcome.

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Introduction

Marfan syndrome (MFS) is an autosomal dominant heritable disorder of the connective tissue that involves primarily the skeletal, ocular, and cardiovascular systems [1]. Cardiovascular involvement is present in 70–80% of cases, including aortic dilatation with the risk of aortic dissection, and mitral valve prolapse with or without mitral regurgitation [1]. The leading cause of premature death in MFS patients is progressive dilatation of the aortic root and ascending aorta, causing aortic incompetence and dissection [2, 3]. MFS has an incidence of at least 1:10,000 [4]. The most common cardiovascular manifestations of MFS affect the atrioventricular valves and the aorta. Mitral valve disease may be the earliest of the cardiovascular manifestations of MFS. Progressive dilatation of the aortic root is responsible for most cases of aortic incompetence.

Turner syndrome (TS) is a genetic disorder resulting from partial or complete X chromosome monosomy [5, 6]. It is the single most common chromosomal abnormality in human beings, but it has an incidence of approximately only 1:3,000 live births because of the high degree of spontaneous abortion. Common physical abnormalities associated with the disorder include short neck (caused by cervical vertebral hypoplasia) and micrognathia [6–8]. Clinical features include short stature, gonadal dysgenesis, renal malformation, facial anomalies, and pterygium colli [9]. A cardiac defect is present in 20–40% of patients with this syndrome, including partial anomalous pulmonary vein connection, bicuspid aortic valve, aortic coarctation and other types of left-sided obstruction, up to full hypoplastic

left heart syndrome [9]. In children with Turner syndrome and aortic coarctation, friability of the aortic wall with a higher risk of hemorrhage has been reported at surgery [10]. The sequelae of Turner syndrome thus pose a challenge to the anesthesiologist, particularly with respect to airway management.

We report the anesthetic management of a case of with Marfan syndrome associated with Turner syndrome, which is the first such case to be reported in the literature, followed by a case discussion.

Case presentation

A 3 year old ASA III girl with both Marfan syndrome and Turner syndrome, weighing 10 kg, and 85 cm in height, was scheduled to undergo minor plastic surgery. A diagnosis of Marfan–Turner syndrome had been made on the basis of the association of Marfan syndome and Turner syndrome in pediatric clinics of a university hospital. Family history was negative for Marfan syndrome.

She had an contracture in her right hand at the fourth and fifth fingers in the first and second interphalangeal spaces. She had a short webbed neck, prognathism, micrognathia, low-set ears, and a high palate. Her anterior and posterior facial heights were long. She had growth retardation, pectus excavatum, and joint laxity. She had dyspnea, palpitation, and anginal pain at rest, but she did not have any arrhythmia in her ECG. Airway examination revealed a large tongue. In her chest auscultation, breath sounds were normal. She had a rhythmic heart sound with a systolic murmur on the left border of the sternum. In her echocardiography, there was high-degree mitral insufficiency, mitral valve prolapse, minimal mitral stenosis, and an atrial septal defect. She had minor mental retardation. After written informed consent had been obtained from the patient's parents for anesthetic management and for publication of this report, she was premedicated with 0.5 mg/kg oral midazolam on the day of surgery. She had bacterial endocarditis prophylaxis 30 min before the operation. After standard monitoring, she had sevoflurane for induction. No muscle relaxation was induced, and the airway was secured using a size 2 PVC LMA without any difficulty. The laringeal mask airway was securely taped and the case proceeded smoothly thereafter. The laringeal mask location was confirmed intraoperatively with periodic visual inspection of the relationships of tape and teeth, as well as auscultation of the chest. Oxygen, nitrous oxide, and sevoflurane (1 MAC) were administered to maintain anesthesia in the spontaneously breathing patient. Alfentanil 200 μg was added to the anesthetic regimen. No muscle relaxant was given. Appropriate positioning was provided to avoid joint dislocations and injuries, and her extremities were supported. Her blood pressure was within normal limits, and no arrhythmia occurred during anesthesia. The antiemetic regimen consisted of metoclopramide 5 mg, and paracetamol 170 mg was given to suppress pain after the procedure. The total operative and anesthetic times were 50 and 60 min, respectively. Anesthesia was uneventful. Postoperatively cardiopulmonary parameters were stable in the postanesthesia care unit. The patient was transferred to the service after 60 min.

Discussion

Anesthesia in Marfan syndrome requires careful consideration because of the possible presence of cardiac valve disease, thoracic aortic aneurysm, and joint laxity [11]. Cardiac insufficiency may cause dyspnea, anginal pain, and palpitation because of aortic insufficency caused by the enlargement of the aortic ring. As big blood vessels lack elastic fiber integrity, the whole cardiovascular system is influenced by the hyper- and hypotension caused by the anesthesia [10]. Patients should be evaluated preoperatively for aortic aneurysm, aortic regurgitation, cardiac decompensation, and arrhythmias, and if they have valve disease, bacterial endocarditis prophylaxis should be given [10]. Echocardiogram assumes great importance as a method of performing a preanesthetic evaluation to detect the aortic root diameter. Marfan syndrome requires special considerations regarding the anesthetic technique, in order to prevent hypotension and hypertension, conserve coronary perfusion, and prevent the development of a dissecting aneurysm. Intubation should be performed gently in these patients, and traction of the temporomandibular joint should be avoided. Appropriate positioning should be provided to avoid joint dislocations and injuries, and the extremities should be supported. Prognathism and a high palata may pose difficulties during tracheal intubation. Positive pressure ventilation may cause pneumothorax [12, 13]. The anesthetic approach employed in the present case took into account the association of Marfan sydrome with Turner syndrome.

The anesthesiologist should be aware of the most frequent anatomo-physiological changes in patients with Turner syndrome and their repercussions for the anesthetic procedure. The predominant physical abnormalities of Turner syndrome include webbed neck, low-set ears, multiple pigmented nevi, micrognathia, an increased carrying angle of the arms, and widely spaced nipples [6–8]. Systemic manifestations include cardiac defects, cardiovascular diseases such as hypertension and hypercholesterolemia, renal anomalies, liver disease, and inflammatory bowel disease [6–8]. The patient in this case presented with a classic short webbed neck, prognatism, a high palate, and

micrognathia. A small percentage of Turner syndrome patients can present with mental retardation, which can limit the relationship between the doctor and the patient. In those cases, it is important that the parents are present during the pre-anesthetic evaluation, and informed consent should be obtained from the patient, parents, or legal guardians [14]. Maxillary and mandibular hypoplasia, associated with a short and wide neck, may impose difficulties for tracheal intubation. It is important to recall that the contracture of the temporomandibular joint in patients with multiple congenital arthrogryposis might be responsible for difficult intubation in Turner syndrome patients [15]. In this particular case, airway management was our real concern. Because of difficult airway management, alternatives to general endotracheal anesthesia should be considered when appropriate. A laryngeal mask, fiberoptic endoscope, and material for a surgical approach to the airways should be readily available during tracheal intubation maneuvers [5, 16]. Tracheal intubation should be done preferentially on the awake patient, under topical anesthesia of the oropharynx and laryngeal or transtracheal nerve block, with the patient mildly sedated with low doses of benzodiazepines and opioids. A laryngeal mask airway may be beneficial in patients without reflux disorder and in cases in which neuromuscular blockade is not required. We chose to secure the airway using a laryngeal mask airway after sevoflurane induction. The trachea in these patients is shorter and the tracheal bifurcation is higher than usual, favoring endobronchial intubation and accidental extubation whenever the tube is under traction [5, 16]. One should suspect endobronchial intubation in the presence of a sudden increase in airway pressure, an absence of breath sounds on auscultation, and decreased peripheral oxygen saturation [17]. The increased incidence of cardiopathies requires a preoperative echocardiogram and in the presence of possible ischemic cardiopathy. In the rare reports of anesthetic procedures in patients with Turner syndrome, anesthetics and adjuvant drugs like atropine, thiopental, succinylcholine, nitrous oxide, meperidine, promethazine, midazolam, fentanyl, propofol, cisatracurium, and isoflurane have been safely used [5, 16]. Sevoflurane induction and maintenance therapy were given for anesthesia after sedation with midazolam. A laryngeal mask was used for airway control.

The operative and postoperative periods were uneventful with the anesthetic method that we employed in this patient. Special care should be given to syndromic patients. Prior medical evaluations and any prior anesthetic history can help focus preoperative evaluations and planning. Preoperatively targeting relevant organ systems, any anatomic or laboratory abnormalities that can be optimized, and perioperative airway management are all key to a successful outcome.

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