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Turner-Kieser Syndrome or Iliac Horn Syndrome, a Challenge for the Anesthesiologist: A Case Report and a Literature Review

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Abstract

Turner-Kieser syndrome is a rare genetic disorder, autosomal dominant, which is related to variable gene expression and high penetrance, due to mutations in the LMX1B gene that affects connective tissue. The clinic has characteristics with alterations in nails, knees, elbows, and presence of iliac horns that makes the anesthetic difficult. However, data in the literature needs more research on this area. The present report is a cesarean section under general intravenous anesthesia performed on a 32-week pregnant woman due to the maternal risk of full-term evolution and aims to contribute to the anesthetic management of patients with this syndrome as there are few descriptions in the literature regarding the anesthetic management in Turner-Kieser syndrome. Even being a case report, the syndrome is rare and it is important to be reported to all anesthesiologists becoming aware of its management and to choose the best technique and anticipate possible complications. During general anesthesia, the technique chosen for the case reported here, we emphasize the risks of possible difficulty in intubating and positioning the patient, as well as renal impairment caused by changes in the cardiovascular autonomic response due to the choice of some drugs. Therefore, in the present report, our option was intravenous general anesthesia because of maternal complications, where fast acting drugs without renal metabolism or excretion were selected, resulting in an uneventful anesthetic procedure.

Keywords

Rare Disease, Pregnancy, Anesthesia, Neuraxial Block, Case Report

1. Introduction

Turner-Kieser syndrome, nail-patella, fong syndrome, hereditary onico-osteodysplasia, or even iliac horn syndrome is a rare genetic disorder, described in 1820, with an incidence of 1:50,000 live births, with an autosomal dominant character and gene variable expressiveness, but with high penetrance due to LMX1B gene mutations, which is being reported in several ethnic groups, with no preference for gender. It is a disease that affects the connective tissue, affecting organs of ectodermal and mesodermal origin [1], and patients may present distinct signs and symptoms. On the other hand, its characteristic is the clinical tetrade of nail, knees and elbows changes, and the presence of iliac horns [1]. The syndrome can be diagnosed from birth with nail changes, and can also present kidney disease, which ranges from mild proteinuria to renal failure, usually in the third decade of life, being the most serious complication of the disease [2]. In the present case, we highlight the anesthetic difficulty with regards to the mother-fetus binomial, making the technique more difficult. The patient agreed with the report and provided us with a free and informed consent form.

2. Case Report

The subject is a young person that is in 32 weeks gestational time, with Fong syndrome and previous history of glaucoma and proteinuria, already under follow-up with a nephrologist, with preserved function. Physical examination revealed the paleness, moderate dyspnea, palpitations, and presence of a systolic murmur in mitral focus. In addition, a decreased breath sounds in the right hemithorax also was observed and an important lower limb edema and bent elbows. BP= 110 × 80 mmHg, HR= 115 bpm, fetal heart beat present. The echocardiogram report described in the medical record showed a prolapse of the mitral valve, a mitral and tricuspid reflux with mild pulmonary hypertension and mild pericardial effusion. Laboratory tests such as normal blood count and biochemistry compatible with the gestational period, total serum proteins 6.9 g/dl, albumin of 1.3 g/dl and negative sorology for infectious diseases. The interruption of pregnancy by cesarean section was programmed, with the reservation of an intensive care unit for mother and fetus, being performed under general intravenous anesthesia, due to the maternal risk and anesthetic difficulty for neuraxial block. Intravenous induction was performed after pre-oxygenation under a mask, using propofol 150 mg, fentanyl 150 μgr, atracurium 25 mg, following the maintenance with continuous intravenous infusion of propofol and remifentanil. Next step, the procedure of patient's orotracheal intubation was realized with no complication and maintenance of oxygen saturation of 100%. The female child was born immediately after anesthetic induction, with signs of intrauterine fetal distress and apparently having the same maternal syndrome. After cesarean section, the patient was extubated after returning respiratory control and protective reflexes. Postoperative analgesia was performed with 6

mg methadone and was released from hospital one week later. The child remained in intensive care, but evolved well, being discharged one month later.

3. Discussion

During the gestational period, a series of physiological changes occur in the maternal organism, as a way of adapting to pregnancy and labor. Regarding the musculoskeletal system, there is an important relaxation and greater mobility of the sacral iliac joints, widening of the pubic symphysis as well as anteriorization of the pelvis, which begin between 10 and 12 weeks of gestation, becoming maximum close to term [3], aiming to expand the diameter of the birth canal in order to facilitate the passage of the fetus by increasing serum levels of relaxin, hormone secreted by the corpus luteum, deciduous endometrium and placental tissue [3] . These physiological changes are compromised in the present case.

Turner-Kieser syndrome is a rare genetic syndrome, affecting 1:50,000 individuals, with loss of function of the LMX1B gene located on chromosome 9, the same as the AB0 blood system. Its expression is variable with the tetrad of nail hypoplasia, changes in knees and elbows, as well as the presence of iliac horns, which is considered pathognomonic for the disease, being classic (**Table 1**). Other findings described are lordosis, kyphosis, elbow pterygium, absence of fibula, dysplasia of the first rib, clavicular and skull malformations. Changes in ligaments, tendons and muscles with hypotrophy and weakness have been reported. Glaucoma, caused by the presence of a shallow ocular anterior chamber and renal dysfunction, by malformation of type IV collagen in the basement membrane, are systemic findings that may be present. Renal histology studies in

Table 1. List of frequent anatomic changes in Turner-Kieser Syndrome (adapted from Hennessey TA *et al. Can J Anaesth.* 2007).

Anatomic changes in Turner-Kieser Syndrome
Nail, knee and elbow hypoplasia
Iliac horns
Dislocation or absence of patella
Lordosis or kyphosis
Elbow pterygium
Absence of fibula
First rib dysplasia
Clavicle and cranial malformations
Ligament changes
Muscular hypotrophy
Fragile teeth
Glaucoma
Kidney dysfunction

cases of renal failure demonstrate focal glomerulosclerosis [1]. In addition, we can find fragile dentition, as well as involvement of the autonomic and peripheral nervous system, with 30% to 40% of patients developing polyneuropathy in childhood or adulthood with potential for autonomic dysfunction because the genetic mutation in LMX1B is related to several changes in the central nervous system [2] including development of mesencephalic and serotonergic dopaminergic neurons in the hindbrain [4], how sympathetic pre-ganglionic neurons in the spinal cord's interiolateral nucleus are stimulated catecholaminergic and serotonergic brainstem, changes in the central nervous system associated with the syndrome can affect the cardiovascular autonomic response [2]. In fact, patients can demonstrate symptoms suggestive of vasomotor dysfunction such as cold extremities and Reynaud's phenomenon [2]. The mutation of the LMX1B gene is associated with abnormalities in the dorsal horn of the spinal cord [4] that would be responsible for the described reports of paresthesia, lethargy and decreased perception of pain and temperature in extremities of the upper limbs [4]. In a previous study in patients with Turner-Kieser Syndrome, 55% reported chronic low back pain due to severe lumbar lordosis or scoliosis, in addition to rare changes in the axial skeleton that included dural ectasia and hidden spina bifida [2], which can complicate the general condition if the patient is parturient.

In parturient, there is a case report of spontaneous aortic dissection that can be attributed to changes in the formation of collagen since collagen types I and III are related to the walls of the vessels and skin, and to hormonal changes in pregnancy that affect proliferation of smooth muscle cells, abnormalities in the proteoglycan matrix and reduced collagen synthesis, thus, parturient women would be at high risk [5]. Such changes are associated with pre- and perioperative complications, especially in relation to anesthesia. Among the main precautions we should take is the assessment of the airways for possible difficulties in direct laryngoscopy and tracheal intubation, checking for the presence of cleft lip or cleft palate. There may be no cooperation and positioning may be difficult in the presence of deformation of joints and should be performed with caution, paying attention to their joint mobility limitation and tendency to dislocations. In addition, in case of neuraxial block, we must perform prior radiological research to investigate the presence of hidden spina bifida and to evaluate scoliosis (**Table 2**). Conventional anesthetic techniques can be used in patients without renal impairment, but in the presence of this, we should check the extent of the kidney injury and the medications involved to treat it with anti-hypertensives, diuretics and steroids, if renal failure is present, optimization of the control of volume and blood pressure are indicated. Patients with renal failure need hydration control and cautious use of renal excretion drugs. In these patients, proteinuria can lead to hypoalbuminemia, resulting in higher doses of free drugs. Avoid succinylcholine if serum potassium is above 5 mmol/l or in the presence of glaucoma. Atracurium and cisatracurium are preferable since they do not depend on renal excretion and metabolism.

Table 2. Anatomical changes in Turner-Kieser syndrome that hinder laryngoscopy and makes anesthesia procedure difficulties (adapted from Hennessey TA *et al.* Can J Anaesth. 2007).

Anesthesia procedure	Anatomic changes in Turner-Kieser syndrome
Neuro-axis block	Scoliosis/lordosis/kyphosis
	Hidden bifid spine/dural ectasia
	Anomalies in the dorsal horn of the spinal cord (paraesthesia, lethargy, decreased perception of pain and temperature in extremities)
	ANS: 30% - 40% develop polyneuropathy and vasomotor dysfunction
	muscle weakness and hypotrophy
Orotracheal intubation	Fragile teeth
	Cleft lip
	Cleft palate
	Non-cooperation
	Displacement, limitation or ear deformation

4. Conclusion

There are few descriptions in the literature regarding the anesthetic management in Turner-Kieser syndrome. Even being a case report, the syndrome is rare and it is important to be reported to all anesthesiologists becoming aware of its management. Patients with this syndrome have several deformities, making the anesthetic-surgical management difficult; therefore, each case must be carefully evaluated to choose the best technique and anticipate possible complications. Neuroaxis blockade can be considered, since anatomical changes involving the nervous system or autonomic ones have been ruled out; however, a worsening of the condition such as polyneuropathy with autonomic dysfunction can be misinterpreted as anesthetic complication. During general anesthesia, the technique chosen for the case reported here, we must emphasize the risks of possible difficulty in intubating and positioning the patient, as well as renal impairment caused by the syndrome and changes in the cardiovascular autonomic response due to the choice of some drugs. Therefore, in the present report, our option was intravenous general anesthesia because of maternal complications, where fast acting drugs without renal metabolism or excretion were selected, resulting in an uneventful anesthetic procedure.

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The present work was developed with the authors' own resources.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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