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Chronic Pain Management in the Patients with Torg-Winchester Syndrome and Anesthesia Experience at Home

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Abstract

Torg-Winchester Syndrome is characterized by diffuse osteolysis, severe osteoporosis, progressive arthropathy of the joints, contracture and hard, multiple, painless, subcutaneous nodules. Patients also have hypertrichosis, skin hyperpigmentation, corneal opacities, gingiva hypertrophy, rough facial appearance and thickened skin. In this autosomal recessive syndrome, Matrix Metalloproteinase-2 (MMP2) mutation is present. Patients with Torg-Winchester Syndrome may have severe pain. The analgesia of these patients is difficult, but should not be neglected. It should be noted that there may be a difficult airway when there is a need for analgesia and anesthesia. In our country, only 2 male patients (siblings) are diagnosed in Torg-Winchester Syndrome, which is in autosomal recessive and rare diseases group. Only a few cases have been reported in the medical literature till now. This syndrome is very rare and unusual; less literature related to anesthesia management, the problems of these patients and their approach were presented.

Keywords: Torg-Winchester syndrome; Anesthesia at home; Pain management

Introduction

Torg-Winchester Syndrome (TWS) is characterized by diffuse osteolysis, severe osteoporosis and progressive arthropathic joints, and painless, subcutaneous nodules [1]. Classically, the patients have hypertrichosis, skin hyperpigmentation, corneal opacities, gingival hypertrophy, coarse facial feature, and thickened skin [2,3]. It is a very rare disease, only 2 patients (siblings) were diagnosed TWS in Turkey.

Considering the rarity of this condition and the lack of literature on anesthesia and pain management, we would like to present the problems of these patients and our approaches to them. Because both patients lived bedridden, they admitted to the home care unit due to severe pain.

Case Series

Case 1

A 24-year-old male patient was diagnosed with Torg-Winchester Syndrome at the age of 13 years [4]. He has suffered from severe joints pain since he was 5 years old. His pain is increased with movement and has a burning sensation. Physical examination revealed very discrete and painful subcutaneous nodules, polyarthropathy, massive osteolysis and osteoporosis of his hands and feet. In musculoskeletal examination, large feet with step deformities (higher and severe) were observed. There was a limitation of movement in the wrist, knee, and ankle joints. He had fusiform fingers, interphalangeal joint contractures and finger nail deformities. His height was 145 cm, but weight could not be measured because of immobilization. There was no hyperpigmentation, hypertrichosis, coarse facial feature or mental retardation. He was diagnosed with chronic papilledema. In the ophthalmologic examination, myopia (right cylindrical: -1.5 and left cylindrical: -1.5) was detected in both eyes and glasses prescription was given to him. While in motionless, the Visual Analog Scale (VAS) score in the bilateral limbs and arms was 8 points (VAS become 10 as he moved). VAS was 7 points for headache. Non-steroidal anti-inflammatory drugs (flurbirofen and dexketoprofen) were used orally for 2 months, but the pain did not decrease and NSAIDs were not used repeatedly due to the risk of nephropathy. In laboratory tests, Blood Glucose: 66cg/l, BUN: 19cg/l, Creatine: 0.4cg/l, AST: 15U/l, ALT: 9U/l, Hemoglobin: 16 g/dl, Hematocrit: 50%, Vitamin D: 5.6 ng/ml, Calcium: 10 mg/dl, Magnesium: 2 mg/dl. Oral vitamin D (100,000/8 weeks), Pregabalin 75 mg/24 h and tramadol 50 mg/48 h were ordered. One month later, when he was motionless, VAS was 2 points in bilateral limbs, while VAS for headache was 1 point. Because of the pain decreased,

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the drug doses were decreased gradually. Sertraline 25 mg/day was started with the suggestion of the psychiatric doctor. According to his pain level, he continued taking oral 50 mg tramadol daily for a week. After 1 year, his pain became more severe. Thus, the patient was thought to have spontaneous fractures of the left tibia. The VAS score was 10 on the left leg. Because of fear of the cleansing of stools due to the pain, he avoided defecation for 15 days. Because his pain increased with movement, he did not accept the transport to the hospital without taking anesthesia, although we mentioned all the risks and our insistence. Oral and written informed consent was obtained after the patient and his relatives were informed. Nasal oxygen was given to the patient with fasting for 8 h (3 lt/min). The aspiration device was kept ready. Portable mechanical ventilator, ambu mask, and other intubation materials were prepared for the usage. After emergency ambulance team came for the transportation and assistance, anesthesia was started to procedure with intravenous (iv) cannulation. Non-invasive blood pressure measurements were performed at 3-minute intervals, continuous finger-saturation follow-up with pulse oximetry, and cardiac monitoring with ECG (Electrocardiogram) was performed. Midazolam 2 mg, propofol 100 mg, and fentanyl 100 mcg iv were slowly given to the patient to keep spontaneous respiration (approximately 5 min). As soon as analgesia and anesthesia were provided, the patient was carefully transferred to the stretcher and taken to hospital. During anesthesia, fecal impaction was removed thorough rectal touch. In an orthopedic examination, tibia fracture was observed, but the patient was discharged after the fixation only, because operation for the fracture was not thought to be necessary. Anesthesia infusion was administered intermittently because of pain and agitation during transportation (for approximately 2 h). In total, midazolam 5 mg, fentanyl 250 mcg, propofol 350 mg, and paracetamol 1000 mg iv were administered. During this period, the patient's spontaneous breathing was never impaired. Any complication wasn't observed. Under anesthesia, bed sheets which could not be changed for a year due to fear of pain, fragility, and immobilization were changed and personal cleaning was done. Pregabalin 75 mg/day, tramadol 50 mg/72 h (when needed approximately for every 3 days) and sertraline 25 mg/day were administered to the patient whose VAS was 6 (at rest) and 10 (movement).

Case 2

A 22-year-old male patient was diagnosed with Torg-Winchester Syndrome at the age of 11 years old. He has pain in all joints like burning sensation since he was 6 years old, which has become worse. In physical examination, discrete and painful subcutaneous nodules, polyarthropathy, massive osteolysis and osteoporosis of the hands and feet were observed. In musculoskeletal examination, large feet with step deformities in varying degrees (higher and severe) were observed. There was a limitation of movement in his wrist, knee, and ankle joints. He had fusiform fingers, interphalangeal joint contractures, and nail deformities. His height 152 cm but weight measurement could not be performed due to immobilization. There was no hyperpigmentation, hypertrichosis, coarse facial feature or mental retardation. He was diagnosed with chronic papilledema, nephrolithiasis, and secundum Atrial Septal Defect (ASD). Eye examination revealed myopia and astigmatism in both eyes (right cylindrical: -1.5 and left cylindrical: -1.75, right spherical: -1.0 and left spherical: -0.25). Glasses prescription was given to him. While at rest, the Visual Analog Scale (VAS) score in the bilateral limbs and arms was 6 points (VAS become 10 as he moved). VAS was 3 points for headache. Non-steroidal anti-inflammatory drugs (flurbirofen and dexketoprofen) were taken orally for 2 months, but the pain did not decrease and NSAIDs were not used repeatedly due to the risk of nephropathy. In laboratory test, Vitamin D: 6 ng/ml and then replacement treatment of Vitamin D was given by orally. Tramadol 50 mg/48 h were started to give to the patient, and 1 month later, when he was at rest, VAS was 1 point in bilateral limbs, while VAS for headache was 1 point. When the pain decreased, the drug doses were decreased gradually. Sertraline 25 mg/day was started with the suggestion of the psychiatric doctor. According to his pain level, he continued taking oral 50 mg/day tramadol in about 2 weeks and sertraline 25 mg/day.

Discussion

It was first described in two sisters (consanguineous marriage) aged 4 and 12 years by Winchester et al. [5], in 1969 as a new type of mucopolysaccharidosis with skeletal deformities mimicking rheumatoid arthritis. In the same year, Torg et al. [6], described a similar skeletal disorder characterized by diffuse osteolysis, arthropathy, and generalized osteoporosis in the hands and feet. Therefore, this syndrome has been named as Torg-Winchester. It is associated with Matrix Metalloproteinase 2 (MMP2) mutation and Nodule Arthropathy Osteolysis Syndrome (MIM605156) in skeletal dysplasia group [7].

Torg and Winchester Syndromes may have heterogeneous phenotypic findings and clinical features, and often mimic juvenile rheumatoid arthritis. For example, our patients did not have skin lesions as previous case reports published by Winchester et al. [5]. In cases presented by Hollister et al. [8], skin thickening, hyperpigmentation, and hirsutism were observed. There have been skeletal disorders in all cases presented in the literature. Although severe pain has not been asked in many cases, I think there has been severe pain in most of them. Bone fractures may be occurred easily due to osteoporosis and osteolysis. Severe pain may be occurred due to nodule compression, arthropathy, and contractures. In our cases, they had severe pain that would prevent them from doing personal hygiene and even defecation. Therefore, the presence and degree of pain should be asked in order to increase the quality of the daily life of such patients. Therefore, the presence and degree of pain should be asked in order to increase the quality of the daily life of such patients. When the electrolyte levels were normal, the vitamin D levels were low in control. Therefore, vitamin D levels can be checked regularly with 1-year intervals so it can be replaced or treated with a daily maintenance dose. Because osteolysis and osteoporosis are so common in patients with Torg-Winchester Syndrome, considering that mandibula and maxilla fractures may occur while applying the mask and cervical vertebrae fractures may occur while positioning the neck when intubation is needed, this syndrome should be considered as a difficult airway, and preventive measurements should be taken. Because of severe pain, these patients make voluntary spasm of sphincter about 10 days and try to eat less. Our patients stated that they felt severe pain in their extremities when moved during the cleansing of the stool (VAS 10 points). Therefore, they demand amputation or neurolytic block treatment. However, none of the orthopedists or algologists accepted the treatment, because they did not have enough experience about the syndrome, did not face complications such as phantom pain. Local neurolytic block therapy can be more effective and comfortable in terms of the side effects of systemic analgesic drugs in such patients. It is needed detailed studies

for ideal analgesic and anesthetic approaches.

Results

The management of pain may be difficult in Torg-Winchester Syndrome, but it should not be neglected. It should be kept in mind that it may be a difficult airway if patients need analgesia or anesthesia.

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