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# General anesthesia for Marinesco-Sjögren syndrome

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## Introduction

The Marinesco-Sjögren syndrome is a rare autosomal recessive disorder characterized clinically by cataracts, cerebellar ataxia, mental retardation, and muscle weakness. Cataracts may be apparent at birth. Cerebellar ataxia with hypotonia is the most prominent neurological feature. Mental retardation is apparent from infancy. Alter et al. [1] reported clinical features of this syndrome for the first time in 1962 [1–3]. Although about 100 cases have been reported, there has been no report of anesthesia for patients with Marinesco-Sjögren syndrome. We report two cases of general anesthesia for Marinesco-Sjögren syndrome patients and discuss the problems of the anesthetic management for this syndrome.

## Case report

### *Case 1*

A 26-year-old woman, weighing 42 kg and 154 cm in height, was scheduled for a partial resection of the iris. She was a product of a full-term, uneventful pregnancy and normal delivery, but suffered from mental and physical retardation from infancy. Eparterial cataracts, cerebellar ataxia, mental retardation and muscle weakness had been apparent since she was 5 years old, and neurological symptoms had been stable until 22 years of age.

At 23 years of age, she was not able to walk and was admitted to our hospital. Physical examination on admission revealed apparent ataxia and mild proximal muscle weakness. In the lower extremities, the muscles were wasted and the tendon reflexes were barely detectable. Hand movement was grossly ataxic. Hematological studies and blood counts were normal. Biochemical studies showed her serum creatine phosphokinase (CPK) to be normal and electromyography (EMG) demonstrated a myopathic pattern. Muscle biopsy indicated myopathic changes including a marked variation in fiber size, fiber splitting, and scattered necrotic and regenerating fibers. Brain computed tomography (CT) revealed marked cerebellar atrophy. She was diagnosed as having Marinesco-Sjögren syndrome by clinical symptoms and these examinations.

A month after admission, she complained of headache and bilateral eye pain. On ophthalmological examination, an increase in the intraocular pressure was noticed. She was scheduled for a partial resection of the iris.

At first, slow induction with sevoflurane and nitrous oxide was planned, but she refused this treatment. Therefore, we rapidly induced anesthesia. A neuromuscular blockade monitor (Relaxsograph, Datex, Helsinki, Finland) was set during the anesthesia for monitoring the effect of vecuronium. Two milligrams of vecuronium was injected slowly. Following that, the train-of-four ratio decreased to zero and the trachea was intubated. Anesthesia was maintained using 60% nitrous oxide in oxygen and low concentrations of isoflurane (0.5%–1.0%). It took 90 min until the train-of-four ratio recovered to 90%. She was hemodynamically stable during the operation.

After the operation, 1.0 mg of neostigmine and 0.5 mg of atropine were injected. Four minutes after the injection, the train-of-four ratio reversed to 100% and

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spontaneous breathing occurred. Complete awareness was confirmed and she was extubated.

She was transferred to the recovering room. During 3 h of observation, she did not complain of dyspnea and was neurologically stable.

Six months later, she was scheduled for a partial resection of the iris because of recurrent increasing intraocular pressure.

Premedication consisted of famotidine 20 mg orally 2 h preoperatively. A neuromuscular blockade monitor was set during anesthesia. Anesthesia was induced with 250 mg of thiopental and the trachea was intubated without vecuronium. Anesthesia was maintained using 60% nitrous oxide in oxygen and low concentrations of isoflurane (0.5%–1.0%). During the operation, the train-of-four ratio was over 90%. Complete awareness was confirmed and her trachea was extubated. During the 3-h observation period in the recovering room, she did not complain of respiratory depression.

*Case 2*

A 34-year-old man, weighing 35 kg and 150 cm in height, was scheduled for a transplantation of the sclera. He was a product of a full-term and uneventful pregnancy and normal delivery. He suffered from mental and physical retardation from infancy. Cerebellar ataxia and mental retardation had been apparent since he was 5 years old. Eparterial cataracts were diagnosed when he was 12 years old. Muscle weakness in the extremities has been progressing since 30 years of age.

At 34 years of age, he entered our hospital suffering from poor visual acuity. Physical examination revealed apparent ataxia and proximal muscle weakness on admission. In his extremities, the muscles were wasted. Hand movement and walking were grossly ataxic. Hematological and biochemical studies were normal. EMG demonstrated a myopathic pattern and muscle biopsy showed myopathic changes. Brain CT revealed marked cerebellar atrophy. He was diagnosed as having Marinesco-Sjögren syndrome by clinical features. After an ophthalmological examination, he was scheduled for transplantation of the sclera.

Anesthesia was induced intravenously with thiopental and the trachea was intubated. Anesthesia was maintained using 60% nitrous oxide in oxygen and low concentrations of sevoflurane (0.5%–1.0%). The train-of-four ratio was about 90% during the operation and he was hemodynamically stable.

After the operation, the train-of-four ratio reversed to 100% and spontaneous breathing occurred. Complete awareness was confirmed and he was extubated. During 3 h of observation in the recovery room, he did not complain of dyspnea.

## Discussion

Problems of anesthetic management for Marinesco-Sjögren syndrome are: (1) risk of malignant hyperthermia, (2) respiratory depression with minimal muscular reserve, and (3) mental retardation.

General anesthesia was chosen for these two cases because immobilization is crucial during an ophthalmological operation. It may be difficult to obtain immobilization in mentally retarded patients without general anesthesia. In case 1, anesthesia was induced with thiopental and vecuronium. Vecuronium was used because several investigators previously reported that depolarizing neuromuscular blockades are likely to be related to malignant hyperthermia in muscular disorders [4]. A proper dose of vecuronium for this syndrome has not been reported. Therefore, we referred to reports of anesthesia for Friedreich's ataxia, which has similar clinical features [5,6]. In this case, the effect of the vecuronium was two times longer than that in a normal patient (Fig. 1). This data suggests that patients with Marinesco-Sjögren syndrome are more sensitive to non-depolarizing neuromuscular blockade. A neuromuscular blockade monitor was useful for monitoring the effect of neuromuscular blockade. As postoperative respiratory depression may be another issue for this syndrome because of muscle weakness, it is necessary to carefully observe the patient during postoperative recovery.

If operations are in the lower limbs or lower abdomen, local anesthesia may be used to prevent malignant hyperthermia and postoperative respiratory depression [7].

In summary, we have described two cases of general anesthesia for patients with Marinesco-Sjögren syndrome. Patients with this syndrome are hypersensitive

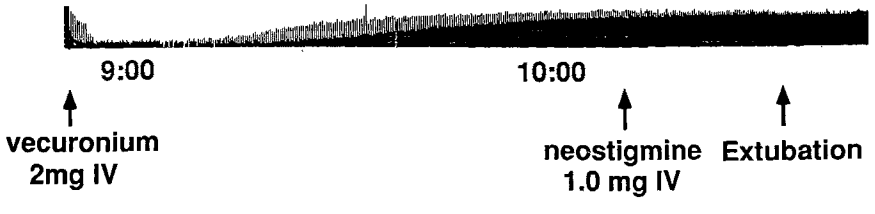


**Fig. 1.** The effect of vecuronium on a patient with Marinesco-Sjögren syndrome (case 1). The data was recorded by 50 Hz tetanus-stimulation at the left ulnar nerve with a neuromuscular blockade monitor. The effect of the vecuronium was two times longer than that in a normal patient

to neuromuscular blockade. Therefore, it is necessary to use a neuromuscular blockade carefully during an operation to avoid malignant hyperthermia and postoperative respiratory depression.

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