

# THE ANESTHETIC MANAGEMENT OF A PATIENT WITH DORFMAN-CHANARIN SYNDROME

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## Abstract

Dorfman-Chanarin syndrome (DCS), which is also known as neutral lipid storage disease, is a rare autosomal recessive inherited lipid storage disease with congenital ichthyotic erythroderma. Since the Dorfman-Chanarin syndrome is a multisystemic disease the choice of drugs and the conduct of anesthesia in these patients are important. Preoperative evaluation should be performed in detail and anesthetic method and drugs to be used should be chosen carefully in accordance with affected system, overall state of patient and characteristics of the operation, in order to decrease perioperative morbidity rates in these patients. We report the anesthetic management of a child with DCS operated for correction of strabismus under general anesthesia.

**Keywords:** Dorfman-Chanarin syndrome, anesthesia.

## Introduction

Dorfman-Chanarin syndrome (DCS), which is also known as neutral lipid storage disease, is a rare autosomal recessive inherited lipid storage disease with congenital ichthyotic erythroderma<sup>1</sup>. Peripheral blood smears showing lipid vacuoles in neutrophils (Jordans' anomaly) in patients with ichthyosis leads to the diagnosis<sup>2</sup>. Other organ systems, such as liver, muscle, central nervous system, ears and eyes are frequently involved<sup>1,3</sup>. Therefore, patients with DCS may present anesthetic challenges to the anesthesiologist. To our best knowledge, there is no information available on the anesthetic management of a patient with DCS. Here, we report the anesthetic management of a child with DCS operated for correction of strabismus under general anesthesia.

## Case Report

A 5-year-old, 18 Kg, 108 cm (at 25th-50th percentiles) girl with DCS was scheduled for elective strabismus surgery. Review of the previous medical records revealed that the patient had presented with ichthyosis since birth. At age 16 months, she had demonstrated hepatomegaly, hepatosteatorrhea, elevated liver enzymes, renal parenchymal hypertrophy and motor-mental retardation. After observing the peripheral blood smears showing lipid vacuoles in neutrophils (Jordans' anomaly) she had been diagnosed with DCS. The patient's preoperative physical examination revealed

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generalized squamous skin lesions and horizontal strabismus in the right eye. The liver was palpable to 2 cm below the costal margin, smooth and firm, but the spleen was not palpable. She also had slight motor-mental retardation. Other systemic examination was normal. Results of laboratory studies showed a normal blood count, lipid profile, serum electrolytes, urea, creatinine, total protein, albumin, globulin, total bilirubin, direct bilirubin, indirect bilirubin, glucose, prothrombin time, activated partial thromboplastin time, INR and urinalysis. The liver functions were deranged with elevated enzymes [serum aspartate aminotransferase (AST): 122 U/L, alanine aminotransferase (ALT): 145 U/L, alkaline phosphatase (ALP): 312 U/L, gammaglutamyl transferase (GGT): 36 U/L]. The muscle enzyme, serum creatine phosphokinase (CPK) has also been increased (CPK: 722 U/L). However, there were no myopathy evidence clinically and she was electromyographically normal. Ultrasound of the abdomen showed hepatomegaly with grade II hepatosteatosis, splenomegaly and bilateral grade I renal parenchymal hypertrophy. A chest x-ray showed a normal heart size, and clear lung fields. Liver biopsy showed 80% fatty changes, minimal portal fibrosis with mixed type inflammatory infiltrates. Skin biopsy revealed marked hyperkeratosis and sporadic acanthosis. The peripheral blood smear showed lipid vacuoles in the cytoplasm of leucocytes consistent with Jordans' anomaly, characteristic of DCS. Lipid vacuoles were also found in cells of the myeloid series in bone marrow smear. The patient received no preoperative medication.

In the operating room, electrocardiography, heart rate, non-invasive arterial blood pressure, temperature, pulse oximetry and end-tidal (ET) CO<sub>2</sub> were monitored. In addition, neuromuscular function was monitored by using a peripheral nerve stimulator (TOF Watch SX Monitor, Organon, Oss, Holland). Heating blanket was used and room temperature was kept at normal limits, in case of a possibility of hypothermia development. Anesthesia was induced with sevoflurane in 50% nitrous oxide (N<sub>2</sub>O) and oxygen (O<sub>2</sub>). After induction of anesthesia, intravenous access was established and TOF Watch SX Monitor was calibrated. Then, endotracheal intubation was performed easily without using muscle relaxants, at the first attempt. Following tracheal intubation, anesthesia was maintained with

isoflurane 1.0-1.5% in a combination of 50% N<sub>2</sub>O and O<sub>2</sub>. Ventilation was controlled mechanically and was adjusted to maintain an ET CO<sub>2</sub> at 30-40 mmHg. Muscle relaxants were not used, since the patient had not moved during surgical operation, which was completed in 50 minutes, and the operation had been performed easily. After stopping the administration of anesthetic agents the patient waked up rapidly, began to breathe spontaneously; she could raise her head and grasp forcefully. Her trachea was extubated uneventfully.

In recovery room, patient was evaluated with "Modified Aldrete Scoring System". When her Modified Aldrete Score reached to 10 she was transported from recovery room to the ward. Patient's hemodynamic variables, SpO<sub>2</sub> and body temperature were within normal limits both during the operation and in recovery room. There were no differences in laboratory values on the second day postoperatively compared with preoperatively. The patient was discharged successfully on the third day postoperatively.

## Discussion

Since the Dorfman-Chanarin syndrome is a multisystemic disease the choice of drugs and the conduct of anesthesia in these patients are important. Extracutaneous manifestations variably include fatty liver, myopathy, cataracts, hearing impairment, mental retardation and a variety of neurologic symptoms<sup>1,3,4</sup>. In this presented case, the perioperative anesthetic care has been performed by considering the ichthyosis, hepatosteatosis, hepatic dysfunction and raised serum CPK, present in the patient.

There is a tendency to increased skin fragility in the ichthyotic patients, therefore adhesive pads or electrodes can cause trauma during application or removal. However, the fixation of endotracheal tube and ECG monitor electrodes to the skin was well tolerated by the patient. It is very important to maintain normothermia during surgery, because patients with ichthyosis have risk of developing inadvertent hypothermia during anesthesia<sup>5</sup>. Using of heating blanket and at normal room temperature, the intraoperative body temperature of the patient could be maintained at a normal level during operation.

Patients with liver disease are at risk for perioperative morbidity and mortality. General anesthesia can reduce total hepatic blood flow and hence can increase the present parenchymal damage<sup>6</sup>. Therefore drugs decreasing the hepatic blood flow, hypotension and hypercapnia should be avoided. Isoflurane and desflurane may be the most appropriate for use in patients with liver disease, as they seem to decrease liver blood flow less than the other volatiles, and have a low degree of metabolism<sup>7</sup>. In the present case, we preferred isoflurane in maintenance of anesthesia, stabilized the hemodynamic variably and adjusted the ventilation to maintain normocapnia. Besides, since hepatic dysfunction affects the distribution, metabolism and excretion of anesthetic drugs<sup>8</sup>, drug doses and dosing schedules should be set accordingly. The coagulopathy, associated with liver disease, may cause bleeding of friable nasopharyngeal structures, such as application of an endotracheal tube.

Abnormally high levels of serum CPK can be reflect an occult myopathy<sup>9</sup>. Patients with myopathy are very sensitive to paralyzing agents. If a muscle relaxant had been required in these patients, a short-acting non-depolarizing muscle relaxant should be administered carefully, using small incremental doses and under close monitoring of neuromuscular function, since some reports have described prolonged effect

of neuromuscular blocking agents in patients with myopathy<sup>10,11</sup>. Furthermore, use of succinylcholine should also be avoided because of risk of severe hyperkalemia development or malign hyperthermia<sup>12</sup>. Although she is clinically and electromyographically normal, we monitored the neuromuscular function, since serum CPK level was high in our case. We did not use muscle relaxants for endotracheal intubation, since sevoflurane could provide sufficient muscle relaxation. Moreover, we did not use neuromuscular blocking agents during surgery, because the surgical operation did not require further muscle relaxation and the patient was sufficiently anesthetized with volatil anesthetic agent. Although strabismus correction in children is associated with a high incidence of postoperative nausea and vomiting (PONV) without prophylactic antiemetic medication<sup>13</sup> PONV was not occurred in our patient.

Anesthesia for these patients should be carried out carefully, because DCS is a multisystemic lipid storage disease. Preoperative evaluation should be performed in detail and anesthetic method and drugs to be used should be chosen carefully in accordance with affected system, overall state of patient and characteristics of the operation, in order to decrease perioperative morbidity rates in these patients.

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