J Anesth (2013) 27:141–142
DOI 10.1007/s00540-012-1459-6

LETTER TO THE EDITOR

Successful perioperative management of a patient with primary systemic carnitine deficiency: a case report

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Received: 22 March 2012 / Accepted: 16 July 2012 / Published online: 21 September 2012

Keywords Carnitine transporter deficiency · Propofol infusion syndrome · Mitochondrial dysfunction

To the Editor:

Primary systemic carnitine deficiency (SCD), also called carnitine transporter deficiency, is a rare autosomal recessive disorder. Carnitine is an essential cofactor for the transportation of long-chain fatty acids into the mitochondrial matrix (Fig. 1). Because very little carnitine is synthesized de novo in mammals, SCD may cause metabolic decompensation and sudden death when the patient is exposed to prolonged fasting and drugs that affect mitochondrial function [1].

An asymptomatic 3-month-old boy with SCD (62 cm, 6104 g) with cleft lip and palate was referred to our hospital for plastic surgery. He was diagnosed with SCD on the basis of tandem mass spectrometry performed to screen for congenital metabolic disorders. A genetic test also revealed that he was compound heterozygous for mutations in the *SLC22A5* gene, which resulted in the production of defective OCTN2 protein. Consequently, he was prescribed L-carnitine supplementation. He did not have any symptoms or signs suggestive of congestive heart failure or skeletal muscle weakness.

He was administered oral carnitine and breast milk 4 h before admission to the operating room. General anesthesia was introduced and maintained with sevoflurane and nitrous oxide in oxygen, and his trachea was intubated using rocuronium. Acetate-Ringer solution supplemented with 10 % glucose was administered at a rate of 5 ml/kg/h. A labioplasty was performed for the labia and cleft palate. The anesthetic course, lasting for 3 h and 13 min, was unremarkable. For postoperative pain control, wound infiltration with lidocaine, 3 μg/kg intravenous fentanyl, and 40 mg/kg acetaminophen suppositories were used. After successful extubation, he was in a good mood and did not seem to be perturbed by the surgical wound. His postoperative blood glucose level was 125 mg/dl. Infusion of 10 % glucose was continued until 4 h after the surgery, when carnitine supplementation and oral intake were restarted. His subsequent clinical course was uneventful, and he was discharged on postoperative day 9.

A mainstay of the perioperative management of SCD patients is to avoid prolonged fasting because they easily develop metabolic decompensation due to an energy shortage [1]. Carnitine is mainly provided in the diet, but is synthesized at extremely low rates from trimethyllysine residues generated during protein catabolism and is excreted in the urine. Therefore, in SCD patients carnitine shortage develops very rapidly once supplementation is stopped. Adequate supplementation of energy is not always apparent, and energy demands vary for each patient. Here we infused acetate-Ringer solution with 10 % glucose both during and after surgery to prevent metabolic decompensation [2]. Rapid recovery from anesthesia and immediate restarting of the oral intake of L-carnitine are also critical.

Moreover, avoidance of propofol use is also important. Propofol infusion syndrome may easily occur in SCD patients [3]. An acute fat burden in the setting of inadequate delivery of carbohydrate and of carnitine deficiency

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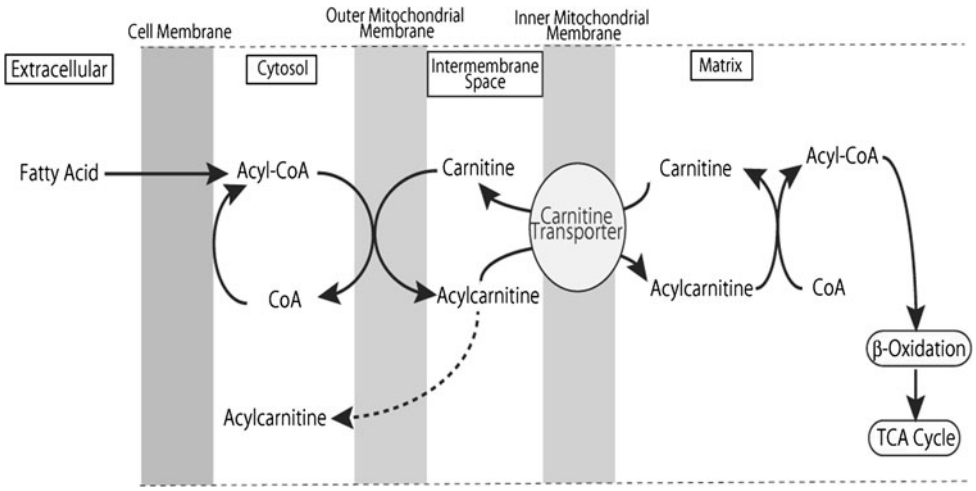

Fig. 1 Carnitine shuttle. *CoA* Coenzyme A, *TCA* tricarboxylic acid

may impair fatty acid oxidation and cause mitochondrial dysfunction in SCD patients.

In conclusion, we described the successful perioperative management of an SCD patient. Our experience emphasizes that, during the perioperative period, particular attention should be given to energy supply, the choice of anesthetic agent, and adequate pain control for SCD patients.

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