

# Anesthesia in a Child with Pyruvate Dehydrogenase Deficiency: A Case Report

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*We report a case in a pediatric patient with pyruvate dehydrogenase deficiency who presented for a left foot tendon transfer with an Achilles tendon lengthening secondary to left ankle equinus. The pathophysiology of pyruvate dehydrogenase deficiency is dis-*

*cussed as well as anesthetic management in patients with pyruvate dehydrogenase deficiency.*

**Keywords:** Ketogenic diet, lactic acidosis, pyruvate dehydrogenase, thiamine.

**P**yruvate dehydrogenase deficiency is an x-linked mutation causing deficiencies of pyruvate dehydrogenase phosphatase.<sup>1</sup> This results in elevated lactic acid which is aggravated by the ingestion of carbohydrates in the diet.

There are 3 types of presentation of this disease. The first occurs in the neonatal period with significant lactic acidosis. The infantile form presents with psychomotor retardation and cystic lesions in the brainstem and in the basal ganglia.<sup>1</sup> Later childhood presentation occurs predominately in males and ataxia associated with a high carbohydrate diet is common.<sup>1</sup>

## Case Summary

An 8-year-old boy weighing 29 kg was scheduled for a left tendon transfer with an Achilles tendon lengthening secondary to a left ankle equinus. His physical status was designated as an American Society of Anesthesiologists class 3, because of a history of asthma and pyruvate dehydrogenase deficiency. He was medically managed on a ketogenic diet and thiamine. He had no surgical history, only magnetic resonance imaging completed under conscious sedation. Because the patient was not seen in the preoperative clinic, there were no preoperative laboratory results. The patient had fasted for 8 hours. Premedication included 10 mg of midazolam by mouth.

The patient was brought to the operating suite and an electrocardiograph, noninvasive blood pressure, pulse oximetry, and temperature monitors were placed. The patient was induced with 70% nitrous in 30% oxygen and 8% sevoflurane. When an adequate depth of anesthesia was achieved, 1 mL of 2% lidocaine was sprayed on his vocal cords, and his trachea was intubated with a 6.0-mm cuffed endotracheal tube without difficulty. Anesthesia was maintained with 1.5% minimum alveolar concentration of desflurane, 50% nitrous in 50% oxygen. A 22-gauge intravenous line was started in the dorsum of his left hand, and intraoperative fluid management consisted of 0.9% normal saline. A total of 250 mL of normal saline was infused. Analgesia consisted of a total of 37.5 µg of fentanyl.

Flat nasal bridge  
Long indistinct philtrum  
Thin upper lip  
High arched palate

**Table.** Characteristics of Facial Dysmorphism Specific to Pyruvate Dehydrogenase Complex Deficiency<sup>2</sup>

A blood gas analysis at the beginning of surgery revealed a base deficit of -5.7 and a lactate level of 2.6. The procedure lasted 2.5 hours. At the conclusion of surgery, another blood gas analysis was obtained and revealed a base deficit of -3.2 and a lactate level of 3.4.

When the patient was fully awake and responding to commands, his trachea was extubated and he was taken to the postanesthesia care unit. His postoperative course was uneventful, and he was discharged home in the care of his mother.

## Discussion

Pyruvate dehydrogenase complex (PDHC) deficiency is an x-linked mutation causing deficiencies of pyruvate dehydrogenase phosphatase that results in elevated lactic acid and is aggravated by carbohydrates. The enzyme pyruvate dehydrogenase has some activity in the later childhood onset category, thus a lesser degree of lactic acidosis is observed. These children have a normal intelligence but may exhibit facial dysmorphic features.<sup>1</sup> Medical treatment consists of thiamine and a ketogenic diet.

The enzyme pyruvate dehydrogenase is the catalyst in the conversion of pyruvate to acetyl coenzyme A. It is necessary for the production of adenosinetriphosphatase. The PDHC consists of 5 components. If there are any deficiencies in any of the 5 components, there are associated syndromes that range from benign ataxia to life-threatening lactic acidosis, failure to thrive, delays in development, hypotonia, and infant death (Table).<sup>2</sup>

## Conclusion

There are few reports of anesthesia in children with pyruvate dehydrogenase deficiency. Dierdorf and McNiece<sup>3</sup> report an infant with the severe form that is usually fatal in the neonatal period. Since their report, it appears that treatment of the disease has changed from that described in their patient. The use of thiamine and a ketogenic diet is now the treatment of choice. Our patient also had the milder form of the disease and was well controlled with diet and thiamine.

Although Dierdorf and McNiece<sup>3</sup> suggested the avoidance of halogenated agents that could interfere with glucose metabolism as could thiopental, this is based on theory alone.<sup>4</sup> However, since our experience with anesthesia in these children is limited; the only recommendations may be to (1) avoid lactate containing solutions and solutions that contain glucose, which may aggravate the lactic acidosis, to provide the least stress-producing anesthetic, and (2) monitor the acid-base status during the course of the surgery. (A brief correspondence of this case report was published in *Paediatric Anaesthesia*.<sup>5</sup>)

## REFERENCES

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