Anesthesia for surgery in a pediatric patient with familial recurrent rhabdomyolysis secondary to LPIN1 gene mutation

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CASE REPORT

We report the anesthetic management of a 5 year old, 17.5 kg male presenting for adenoidectomy and bilateral myringotomy and tube placement with a past medical history significant for LPIN1 gene mutation causing recurrent rhabdomyolysis/myoglobinuria.

Genetic diagnosis was established 6 months prior to surgery surrounding an upper respiratory illness associated with severe muscle pain and CK levels > 200,000 units/L.

There are no reports in the anesthesia literature regarding management of these patients.

WHAT IS LPIN1?

- 890 AA protein primarily expressed in muscle and adipose tissue
- Dual function 1:
  - Phosphotidyl phosphatase for triglyceride and phospholipid synthesis
  - Transcriptional co-activator of enzymes including those involved in fatty acid oxidation and mitochondrial respiratory chain
- Skeletal muscle expresses some of the highest levels of lipin 2
- Mutations also associated with:
  - Statin induced myopathy, hypertension, diabetes, and obesity

FAMILIAL RECURRENT RHABDOMYOLYSIS

Rare, autosomal recessive genetic deficiency in lipin-1 that can lead to life-threatening episodes of rhabdomyolysis. 1-4

Triggered by:
- Acute illness, Low caloric intake or fasting
- Anesthesia

Symptoms:
- Age < 5 yo
- Muscle weakness, tenderness, pain, swelling, refusal to walk or move
- Dark urine

Mechanism: Phospholipid membrane imbalance
- Defect in mitochondrial fatty acid oxidation
- Abnormal energy supply to muscle cells

- 1/3 of patients reported die during an acute crisis of rhabdomyolysis. 5

ANESTHETIC

Pt admitted evening prior to surgery
- IV placed D51/2 NS with 20 KCl at 54 ml/hr
- All vital signs and labs stable

0745 case start
- Premed midzolam 1 mg IV
- TIVA: fentanyl bolus 20 mcg, propofol 40 mg propofol infusion 200 mcg/kg/min remifentanil 0.2 mcg/kg/min
- CK drawn intraop. pacu, and overnight (263, 213, 178)
- Case duration 24 min
- 1 emesis in PACU

Uneventful recovery overnight
- Normal electrolytes
- Pain control with Lortab PO

DISCUSSION

Michot et al. observed that anesthesia and/or the fasting state were implicated as precipitating factors in 7 out of 17 cases of rhabdomyolysis in early infancy associated with LPIN1 gene mutations.

Current genetic guidelines include:

- NPO period requires IV fluid with glucose
- Prompt treatment of acute illness, even minor
- Run CK immediately if symptoms develop
- Better estimates course and treatment

At risk for:
- Hypovolemic shock
- Electrolyte disturbances: Hypo K, Hypo Ca
- Acute renal failure (especially CK > 15,000)
- Metabolic acidosis
- Less common: DIC, compartment syndrome, pulmonary edema, CHF, altered mental status

Treatment:
- Aggressive IV hydration
- Correct electrolyte imbalances
- Urine alkalinization to prevent precipitation of myoglobin in renal tubules (nephrology)
- Continued treatment until CK < 1000

We present an anesthetic approach to the management of a patient with this rare disease that went without complication.

- Limitations: short duration case, minimal intraoperative/postoperative pain requirements

REFERENCES

1. Online Mendelian Inheritance in Man (OMIM): http://omim.org/entry/608200