Introduction:
We present a patient with Schwartz-Jampel Syndrome and discuss its implications for anesthetic management.

Case Report:
C. W. presented at 13 months for failure to thrive, dysmorphism, and possible neuromuscular disease and was diagnosed with Schwartz-Jampel syndrome (SJS). He had prior surgeries for a paralyzed right hemidiaphragm and placement of myringotomy tubes. He first came to our OR at 2 years of age for adenotonsillectomy and Nissen fundoplication with gastrostomy tube. At that time he was noted to have a Mallampati class 3 airway with micrognathia. He underwent an uneventful inhalation induction and intubation and was given 1 mg of pancuronium. The adenotonsillectomy proceeded uneventfully but the surgeons aborted the Nissen due to abdominal wall rigidity, felt due to the patient’s myotonia. The patient was given reversal and extubated without incident. Approximately one year later the patient presented for EUA for evaluation of amblyopia. This was done with an inhalation induction and mask airway for the brief procedure. The patient had progressive sleep apnea and at 4 years of age he presented for an airway exam and tracheostomy. He tolerated an inhalation induction and had an adequate mask airway. He was intubated by the surgeon and the case once again proceeded without incident. The most recent anesthetic we performed on this patient was at 5 years of age for eye exam and dental restoration. The patient tolerated sevoflurane well. Since that time he has had progressive motor impairment due to contractures and stiffness but has not required any further surgical intervention.

Discussion:
SJS is a rare autosomal recessive myotonia caused by a mutation in the gene encoding perlecan, a component of basement membranes. This results in myotonia, blepharophimosis, and multiple joint contractures. The symptoms typically progress slowly or remain stable and the prognosis is good. Anesthetic concerns in these patients include airway difficulties and the potential risk of malignant hyperthermia.1 These patients commonly have micrognathia, decreased range of motion of the neck and jaw, and obstructive sleep apnea. The mouth is typically turned down, with puckered lips due to increased perioral muscle tone; this in combination with the other features may make mask ventilation difficult. That was not the case in our patient and mask ventilation was used successfully in 3 of his 4 anesthetics at our hospital.

Descriptions of SJS include increased risk for malignant hyperthermia. There is a single case report describing elevated temperature in a patient with SJS from 1978. The authors assert that this was MH, but their patient had only modestly elevated levels of CK and the temperature and muscle stiffness were consistent with myotonia and associated increased metabolism. Significantly, the anesthetic they described did not include any triggering agents.2 Parness et al discuss myotonias and MH and conclude that myotonias do not confer an increased risk of MH.3 Our patient has been exposed to sevoflurane multiple times with no evidence of MH. Regardless of the risk for MH, succinylcholine should be avoided because it can induce hyperkalemia and increase the degree of myotonia. Nondepolarizing muscle relaxants are safe, but their effectiveness may be unpredictable.

References:
1 Neuromusc Dis. 2003, 13; 347-351
2 J Pediatr. 1978, 93; 83-84
3 Anesth Analg. 2009, 109; 1054-1064