Arterial Tortuosity Syndrome

INTRODUCTION
Arterial Tortuosity Syndrome (ATS) is a connective tissue disorder characterized by elongation and tortuosity of major vascular structures, leading to kinks, aneurysms, dissections, and stenosis. It is a rare syndrome with only 100 individuals identified worldwide and no known documentation of anesthesia experiences. We present a patient with ATS that has undergone multiple procedures with general anesthesia.

CASE
An 8-year-old male with ATS who presented for interventional cardiac catheterization for severe bilateral branch pulmonary artery stenosis, pulmonary hypertension, & right ventricular hypertrophy. His other comorbidities included obstructive sleep apnea, joint hypermobility, scoliosis, and tracheomalacia. On physical exam he had a right upper sternal III/VI systolic ejection murmur. A transthoracic echocardiogram showed right ventricular hypertrophy. Previously, the patient's tortuous pulmonary vasculature and left pulmonary artery stenosis had been treated with a surgical patch angioplasty. Unfortunately, he had developed further stenosis of both branch pulmonary arteries, requiring multiple percutaneous balloon angioplasties and stent placements.

The patient received general anesthesia for cardiac catheterization with standard ASA monitors. He had an inhalational induction of anesthesia prior to peripheral venous access. Intubation and maintenance of anesthesia was achieved with a mix of sevoflurane, rocuronium, fentanyl and dexmedetomidine. The cardiac catheterization was uneventful, and the patient was extubated immediately and recovered in the post anesthesia care unit. He was discharged home the next day.

DISCUSSION
• ATS is a genetic disorder of the connective tissue. It is an autosomal recessive disease with the defect located on chromosome 20q13.1. A mutation of the SLC2A10 gene leads to production of defective GLUT10 protein, altering production of collagen and elastin in the body. The lack of functional GLUT10 causes upregulation of transforming growth factor beta (TGF-β) signaling in the developing body. TGF-β is involved with cell growth and proliferation, as well as blood vessel development and formation of the extracellular matrix. When TGF-β is upregulated, excessive elongation & tortuosity of the vasculature, especially arterial, can occur. Patients with this condition vary in severity of the affected vasculature. The disease is progressive, and older individuals can be more severely affected.

• A thorough preoperative workup is essential in patients with ATS. ATS can affect the respiratory, cardiovascular, skeletal, and integumentary systems as well cause craniofacial abnormalities. The most common signs are vascular anomalies of the pulmonary, great and cerebral vessels which require routine surveillance with echocardiography and cardiac and neural angiography. Treatment is typically surgical correction for focal issues.

• There are numerous perioperative considerations for patients with ATS:
  • Skin can be hyperelastic; when combined with the tortuous vasculature, arterial or venous cannulation can be difficult.
  • Craniofacial abnormalities and joint hypermobility or contractures can complicate mask ventilation and intubation.
  • Ventricular hypertrophy and pulmonary hypertension can result from increased vascular resistance from arterial tortuosity.
  • Tracheomalacia, recurrent pulmonary infections, and restrictive lung pathology can be present.

• In conclusion, patients with ATS are complicated and require a circumspect approach to their anesthetics.

REFERENCES
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