INTRODUCTION: Creating an anesthetic plan is often referred to as an art incorporating a myriad of factors. As genetic testing becomes less costly, more popular, and increasingly covered by health insurance, it is creating new challenges for the anesthesia provider [1][2]. We report a challenging case which used genetic mutation data to guide the anesthetic plan.

Robert Kale MD, Rahila Bilal MD
SUNY UpState Medical University, Department of Anesthesiology

DISCUSSION: The ND1 gene affects mitochondrial complex I and is the origin of Leber’s Hereditary Optic Neuropathy (LHON). Her mutation at the ATP8 gene affects mitochondrial complex V. Together these mutations greatly inhibit the respiratory chain of the mitochondria which generate ~90% of the cell’s energy [3]. Adding drug induced suppression to the already suboptimal mitochondrial output can lead to deleterious consequences such as arrhythmias and lactic acidosis. Both propofol and volatile anesthetics have been shown to inhibit complex I of the mitochondria. Some benzodiazepines and acetaminophen cause uncoupling of oxidative phosphorylation and increase oxidative stress on the mitochondria. NSAIDs and certain antibiotics directly inhibit complex V. Furthermore, fasting guidelines result in depression of the body’s fuel stores which increases the workload on the mitochondria. Our approach created an anesthetic plan that did not dampen the already stunted mitochondrial energy output to below the bioenergetic threshold of the body’s cells. This was accomplished by limiting direct drug suppression of the mitochondrial complexes, avoidance of medicines known to uncouple oxidative phosphorylation and by decreasing oxidative stress. Dextrose 5% in 0.45% normal saline was used for fluid replacement. A single agent technique with sevoflurane was used for induction and maintenance of anesthesia to avoid multimodal suppression of the mitochondria. This resulted in the patient tolerating general anesthesia without complication.

CASE PRESENTATION: A syndromic 5yo female presented for MRI under general anesthesia. Her phenotypic characteristics of optic neuropathy, Wolff-Parkinson-White syndrome (WpW), clinodactyly, short stature, triangular face and microcephaly pointed toward a genetic origin. Genetic testing revealed mutations at two loci, the ND1 gene at m.3460G>A and ATP8 gene at m.8476dupC.

REFERENCES:

http://flipper.diff.org/app/items/6965