[NM-210] Joubert Syndrome: A rare pediatric genetic disease, revisited.

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Introduction:
Joubert syndrome is a rare, autosomal recessive disease characterized by partial or complete agenesis of the cerebellar vermis with molar tooth sign on MRI of the brain[1,2]. Manifestations include ataxia, abnormal breathing patterns (hyperpnea alternating with apnea), nystagmus and abnormal tongue movements. There is sensitivity to inhaled anesthetics, and opioids worsening episodes of apnea and hyperpnea2, and treatment is supportive. We report the case of a child requiring renal transplant for End Stage Renal disease caused by cystic renal dysplasia.

Case Report:
A three-year-old male with Joubert's Syndrome and end-stage renal disease from Cystic renal dysplasia presented for work up for renal transplant. He was born full-term but required a three-week NICU stay for work- up of renal insufficiency. At four months of age, he was noted to have nystagmus and MRI of the brain showed molar tooth sign. Genetic testing revealed a mutation in CEP290 gene, diagnosing Joubert Syndrome. He had mild anemia and metabolic alkalosis on preop labs and echocardiogram showed no congenital abnormalities.
Intravenous induction and intubation were uneventful, and within nine hours of surgery, patient had left renal allograft placed and right native nephrectomy. He received a total of Fentanyl 250 mcg and decision was made to keep him intubated because of concerns of post-op apnea. In the PICU he was extubated the following day, and discharged home nine days later without major issues.

Discussion:
Joubert syndrome is now being described as Joubert Syndrome and Related Disorders (JSRD) and has been linked to ten causative genes, all of which encode for proteins of the primary cilium. Multiple cell types such as neurons, retinal photoreceptors, and kidney tubules are affected. Our patient carried the CEP290 mutation, which is associated with JS with oculorenal defects[3].
CNS manifestations can include hypotonia with ataxia, development delay, tremors and epilepsy. Ocular manifestations mostly involve the retina causing retinal dystrophy. Congenital heart defects have not been associated with JSRD. Respiratory abnormalities occur after birth, and improve with age. Stress, inhalational agents and opioids have been described to cause episodes of apnea-hyperpnea. Renal manifestations occur in 25% of JSRD and hepatic involvement is less common[3].
Total IV anesthesia may not exacerbate these episodes of hyperpnea or apnea . Opioids should be avoided when possible and use of regional anesthesia should be considered. If surgery can't be delayed until later in life, the use of caffeine for respiratory stimulation can be considered.

References:
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