Abstract for the SSIEM 2018

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Interference of urinary bile acid profiling by propofol

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Background
Urinary bile acid analysis is routinely used to screen for cerebrotendinous xanthomatosis (CTX), an autosomal recessive disorder caused by CYP27A1 mutations. Recently, we identified a urinary bile acid profile suggestive of CTX in a boy with surgery for an epidural hematoma. Plasma was not available. Although not symptomatic, therapy with chenodeoxycholic acid was started. However, bile acid profile was normal in a second pre-medication sample and no pathogenic CYP27A1 mutations were found. Similarly, a CTX bile acid profile was identified in a boy undergoing endoscopy for failure to thrive and diarrhea. Bile acid excretion and plasma cholestanol were normal in previous samples from this patient. Both boys underwent propofol (2,6-diisopropylphenol) sedation prior to urinary sampling and because propofol is metabolized via cytochrome P450 enzymes, we hypothesized that the bile acid profiles were caused by pharmacological inhibition of CYP27A1 activity by propofol.

Methods
We analyzed bile acid profiles in urinary samples from 5 patients after sedation with propofol 8-12mg/kg/hour for at least 4 hours during surgery for idiopathic scoliosis.

Results
Similar to our cases, patients with propofol sedation showed bile acid profiles suggestive of CTX.

Discussion
Although many pharmacological agents are known to affect cytochrome P450 enzyme activities, we are not aware of any prior study showing that propofol inhibits CYP27A1, resulting in a diagnostic footprint for CTX. The clinical impact is clearly illustrated by our cases. Propofol is one of the safest and most commonly used anesthetic agents and urethral catheterization during anesthesia offers an opportunity for urinary sampling for metabolic screening. Misdiagnosis with CTX has significant impact. CTX is a severe progressive disorder that requires lifelong expensive treatment. To prevent further misdiagnoses, we suggest to add propofol to the list of known interferences of bile acid profiling.