Introduction: Rett syndrome is characterized by repetitive hand wringing, autistic features, bruxism, episodic apnea, gait ataxia, tremors, microcephaly, encephalopathy and scoliosis. Rett syndrome is usually found in females. Atypical Rett syndrome is increasingly observed as MECP2 mutations have been identified in patients previously diagnosed with autism, mild learning disability, clinically suspected but molecularly unconfirmed Angelman syndrome, or mental retardation with spasticity or tremor (1). Males meeting the clinical criteria for Rett syndrome have been identified in association with 47,XXY karyotype and postzygotic MECP2 mutations result in somatic mosaicism (1). Males with a 46,XY karyotype and a MECP2 mutation may have a severe neonatal encephalopathy with a short life span (1). Seizures occur in 50% of females with Rett syndrome and more frequently when the disease stabilizes. A prolonged QT interval and sudden death in patients with Rett syndrome have also been reported (2). Khalil and colleagues reported a 14 year old girl with Rett syndrome and seizure disorder was unusually sensitive to sedating drugs and was slow to recover from anesthesia (3). Contrary to their findings, we report a case of a 16-year-old male with Rett syndrome who required a normal or even higher dose of opiates and volatile agent intraoperatively.

Results: A 16 year old male was diagnosed with Rett syndrome, seizure disorder, gastroesophageal reflex disease, scoliosis, developmental delay, encephalopathy, cerebral palsy, and incontinence of bowel and bladder. His seizure disorder was treated with valproic acid and tegretol. He has no history of opiate intolerance. Upon chromosomal analysis at the Baylor University School of Medicine, he was diagnosed to have a missense mutation on X-linked MECP2 encoding methyl-CpG-binding protein 2. During general anesthesia for an open Nissen Fundoplication procedure, he required 0.15 mg/kg of morphine in addition to 15 mcg/kg of fentanyl along with 1 to 1.6 MAC of isoflurane and full dose of muscle relaxant. Surgery lasted for 4 hours and neuromuscular block was readily reversible. The electrocardiogram rhythm strips showed normal QT interval throughout the entire case. He was awake and the endotracheal tube was removed in the operating room. Patient was awake and comfortable in the recovery unit.

Discussion: Rett syndrome is seen almost exclusively in females although a few males with clinical phenotype resembling this disorder have been reported (4). Anticonvulsant therapy increases the fentanyl requirement during anesthesia for craniotomy (5). Valproate can inhibit gamma-aminobutyric acid (GABA) transaminase and activate glutamic acid decarboxylase, which can decrease the degradation and increase the synthesis of GABA (6). GABAergic agents presumably potentiate the activity of narcotic analgesics and decrease its sensitivity. The normal-to-high anesthetic requirements in this case may be related to anticonvulsant use, but may also be a characteristic of male-variant Rett syndrome. This differs from Khalil’s observation of a female Rett syndrome patient who experienced a lower analgesic and anesthetic requirement (3). Our case also extends Coleman’s observation of normal analgesic requirements in female with Rett syndrome to a patient with male-variant Rett syndrome (7).

References: