A Patient with Cornelia De Lange Syndrome

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A six year old female with a history of Cornelia De Lange Syndrome (CdLS) presented for developmentally-delayed and presented with the craniofacial bilateral hamstring lengthening. She was non-verbal, anomalies that are associated with CdLS. She was born pre-term with a moderate ventricular septal defect which resolved without surgical intervention. Her medical history was also significant for pulmonary stenosis, and significant ventilation was maintained. She had an uneventful intra-gastroesophageal reflux disease (GERD). She also had a known history of difficult intubations. Following an inhalational induction, a difficult intravenous line was placed. Her airway was secured with a laryngeal mask airway and spontaneous operative course without any postoperative complications.

Craniofacial Features Associated with Cornelia De Lange Syndrome 1-6
- Flattened Mid-Face
- Short Stature
- Micrognathia
- Low hairline with confluent eyebrow
- Wide Spaced Teeth
- Low Set Ears
- Cleft palate
- Square Chin
- Micrognathia
- Macroglossia
- Short Stature
- Flattened Mid-Face

Anesthetic Concerns 1-3,6
- Pre-operative Anxiety : Hearing Loss, Developmental Delay, Non-Verbal, Tendency to self-harm 5
- Difficult Airway: Craniofacial abnormalities
- Even though a difficult intubation is a concern and anticipated, in a retrospective analysis of 42 patients, only one patient had a difficult intubation.1
- Congenital Cardiac Abnormalities : VSD, ASD, Pulmonary Stenosis, Tetralogy of Fallot, Hypoplastic Left Heart Syndrome
- Difficult Intravascular Access
- Aspiration risk: Gastrointestinal anomalies (GERD, intestinal malrotation, pyloric stenosis)3
- Increased Susceptibility to infections 4

Discussion
Cornelia De Lange Syndrome is a rare genetically heterozygous and sporadic syndrome with a prevalence of 1 in 10,000 - 30,000. 6 It is characterized by distinctive facial features and a wide range of cardiac, gastrointestinal, developmental, neuropsychiatric and musculoskeletal anomalies. This syndrome can either be autosomal dominant of inheritance or X-linked dominant pattern of inheritance depending on the specific gene mutation. Approximately 60% of cases are due to a mutation of the NIPBL gene, autosomal dominant of inheritance.1 Other gene mutations (10%) associated with this syndrome are SMCL (X-linked), SMCM (autosomal dominant), HDACR (X-linked), and RAD21 (autosomal dominant).1 The proteins produced by these genes are associated with the function of the cohesin complex which plays a direct role in early development. This complex helps regulate the structure and organization of chromosomes, stabilize the cell’s genetic information and repair damaged DNA.4 However, the specific genes involved in the genetic mutation causes a wide spectrum of characteristics. SMCL, RAD21, SMCM are all associated with mild symptoms when compared to a mutation to the NIPBL gene.4 In a study of 120 subjects, 47% were found to have a mutation involving NIPBL.6 Multiple types of mutations were identified including missense, splice site, nonsense and frameshift; these different genetic mutations correlated with a spectrum of phenotypes.6

References
6. Department of Anesthesiology at Baystate Medical Center/University of Massachusetts Medical School.