[NM-170] Rare COCKAYNE Syndrome Revisited: a Case Report

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Cockayne syndrome [1] is a rare autosomal recessive disorder characterized primarily by growth failure, premature aging, leukodystrophy, and abnormal sensitivity to sunlight. Underlying genetic disorder is related to a defect in a DNA repair mechanisms [2-5]. It is unlikely for these children to survive past the 2nd decade of life [6, 7].

We present a case of 18 year-old female with Cockayne syndrome Type I. She had classical presentation of the disease: cachectic short stature, premature aging appearance, blindness, profound bilateral hearing loss, and a global developmental delay. She also had hypertension, chronic renal dysfunction, dental caries, as well as chronic failure to thrive. Patient's height was at 50th % for a 5-year old child. Instead of growing and gaining weight, patient actually lost weight during the span of several years (14.4kg in 2011 to 12.4kg in 2013). Her cachectic appearance was becoming more apparent. To improve the feeding, she underwent complex dental rehab and laparoscopic gastrostomy tube placement.

Anesthesia Considerations. Patient underwent inhalational induction with easy mask ventilation. Skin. Despite loss of subcutaneous fat and poor skin turgor, placement of a PIV was noted to be difficult. Airway. For the previous procedures, technical difficulties of intubation were overcome with the use of glydescope size 2. This time, airway attempts with direct laryngoscopy with a generous crycoid pressure achieved vocal cord grade 1 view. However, patient required placement of a smaller tube. Specifically, she was intubated with cuffed ETT 4.5 (low-pro) at age 15, but required one size smaller cuffed ETT (4.0 low pro) at age 18. Although size 4.0-cuffed ETT was adequate for the smaller laryngeal inlet, we provided higher cuff pressures (30 mm H20) to achieve adequate ventilation. Multiple organ involvement. Her hypertension secondary to renal fibrosis was adequately controlled with oral amlodipine and enalapril. We maintained her intraoperative blood pressures in the range that was appropriate for the 5-year old child. Positioning. She had progressive spasticity and numerous contractures for which we provided generous padding.

Summary. We present a case of a girl with advanced age for Cockayne syndrome. Recognition of patients' advanced physiologic age associated with adult diseases [8, 9], vis-à-vis undeveloped somatic appearance is critical for successful and adequate anesthesia management, including a difficult airway management [10, 11].

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