Rare COCKAYNE Syndrome Revisited: a Case Report

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BACKGROUND

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

GENETICS

Cockayne syndrome has an autosomal recessive type of inheritance. It is a genetically diverse disorder. Mutations in the ERCC6 (~70% of cases) and ERCC8 genes are the cause of Cockayne syndrome. Certain types show some overlap with xeroderma pigmentosum, another disorder caused by defective DNA repair. The proteins made by these genes are involved in repairing damaged DNA. If either of the genes are altered, the damaged DNA is not repaired, ultimately leading to malfunctioning cells and cell death [8].

TYPES OF COCKAYNE SYNDROME

◆ Type I, the classic form, is characterized by normal fetal growth with the onset of abnormalities in the first two years of life. Impairment of vision, hearing, and the central and peripheral nervous systems progressively degenerate until death in the first or second decade of life.
◆ Type II (cerebro-oculo-facio-skeletal (COFS) syndrome) is characterized by minimal neurological development after birth. Death usually occurs by age seven.
◆ Type III, has late onset, and is rare and milder than Types I and II.
◆ Xeroderma pigmentosum - Cockayne syndrome, symptoms of both diseases are present.

CASE PRESENTATION

We present a case of an 18 year-old female with Cockayne syndrome Type I. She had classic 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, a likely swallowing dysfunction, as well as chronic failure to thrive. The patient’s height was at 50th % for a 5-year old child. Instead of growing and gaining weight, the patient actually lost weight during the span of several years (14.4kg in 2/2011 to 12.4kg in 8/2013). Her cachectic appearance was becoming more apparent. To improve her feeding, she underwent complex dental rehab and laparoscopic gastrostomy tube placement.

ANESTHESIA CONSIDERATIONS

◆ Skin. Despite loss of subcutaneous fat and poor skin turgor, placement of a PIV was noted to be difficult.
◆ Airway. Patient underwent inhalational induction with easy mask ventilation. For the previous procedures, technical difficulties of intubation were overcome with the use of gylcoseape size 2. This time, airway attempts with direct laryngoscopy with a generous crycoid pressure achieved a 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 H2O) to achieve adequate ventilation.
◆ Multiple organ involvement. Her hypertension secondary to renal fibrosis was adequately controlled with oral amloclilene and enalapril. We maintained her inotropers pressure 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 AND DISCUSSION

◆ We present a case of an 18-year of girl, considered advanced age for Cockayne syndrome.
◆ Possible difficult airway is a significant implication to consider when taking care of a child with Cockayne syndrome. Although case reports in literature describe airway difficulties due to limited neck extension, kyphoscoliosis, anterior larynx and need for cricoid pressure [9,10], in this report we highlight the importance of considering smaller size ETT despite child’s advancing age.
◆ Recognition of patients’ advanced physiologic age associated with adult diseases [11, 12], vis-à-vis undeveloped somatic appearance is critical for successful and adequate anesthesia management, including a difficult airway management [9, 13].

CASE FOLLOW UP

Patient was admitted in Sept 2013 with complaints of abdominal pain, as well as pulmonary edema, pleural effusions, and presumed aspiration pneumonia for which she received appropriate medical treatment.

IMPORTANT DRUG INFORMATION

◆ Metronidazole (Flagyl®; Metrolpyl®) may cause acute liver failure in children with Cockayne syndrome and should be avoided in this patient population.

REFERENCES