Cockayne syndrome: A case report

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A 4-year-old female with Cockayne syndrome presented for cataract extraction under general anesthesia. She was thin and frail; her neck, epiglottis and larynx were stiff; she was deaf and blind; and she could not speak, sit unaided, or perspire. At the time of her admission, she weighed 5.5 kg.

Cockayne syndrome is a disease of childhood characterized by mental retardation and premature aging. Although the underlying abnormality appears to be autosomal recessive inheritance or metabolic (possibly thymic) dysfunction, there is no consensus on etiology.

The multiple organ involvement carries significant implications for the anesthetist. Intubation can be technically difficult, and care of the skin can be problematic. Essential hypertension, hepatic deficiencies, osteoporosis, deafness, blindness, and other effects of premature aging may be encountered making perioperative management a challenge.

Key words: Cockayne syndrome, pediatric syndromes, progeria.

Introduction

Cockayne syndrome is a rare form of mental retardation that includes symptoms of premature aging. Administering anesthesia to children with this condition presents several challenges for the anesthetist. These challenges may include a compromised airway, allergic asthma, emphysema, liver and kidney dysfunction, deafness, blindness, and senile changes in the skin and bones. We present a case and discuss the history, incidence, major features, and anesthetic implications of the syndrome.

Case summary

A 4-year-old Caucasian female came to our operating room with a left lamellar cataract which had first appeared at age 16 months (Figure 1). Physical examination revealed a quiet, pale child with fine, thin hair and a wizened appearance. A prominent nose accented her sunken eyes. Lack of subcutaneous fat gave a wasted appearance to her extremities; she had flexion contractures and intention tremors. Her thorax was rigid. She could not sit or stand unaided. Neither could she hear nor speak. It was noticed that although dentition was normal, the teeth were large in relation to her mouth. Neck extension was limited and mouth opening was difficult to elicit. These signs are characteristic of Cockayne syndrome.

An interview with the mother revealed that the child choked easily on thin liquids and had limited ability to perspire. Communication with the child was primarily through touch. Although the weight of a 4-year-old female at the 50th percentile is 15.96 kg, this child was 5.5 kg.

After placement of appropriate monitors in the operating room, an inhalation induction was accomplished using a halothane/oxygen mixture. Satisfactory depth of anesthesia was achieved, adequacy of manual ventilation confirmed, degree of mouth opening ascertained, and the epiglottis visualized. Meanwhile, intravenous access was obtained using a 24-gauge catheter and an infusion of dextrose 5% in .2% saline was begun. To prevent bradycardia during laryngoscopy and during manipulation of the eyeball by the surgeon, atropine...
Cockayne syndrome is a childhood disease characterized by premature aging, microcephaly, and dwarfism. Onset follows normal birth and infancy; the first symptoms, usually cutaneous or ocular, appear around the second year. \(^1\) \(^5\) The child displays developmental delay, progressive spasticity, and ataxia secondary to basal ganglia calcification, \(^6\) \(^7\) mental retardation, deafness, \(^3\) and blindness from retinal involvement or cataracts. \(^9\) There may be hypertension from renal arteriosclerosis and other renal pathology typical of an aged kidney. \(^10\) A study by Higginbottom and associates described three children with essential hypertension (900/200 blood pressure in one case) in whom histopathologic studies showed immune complex deposits in the kidneys, although specific antigens had not been found. \(^11\) Examination by x-ray showed changes specific to the syndrome including patchy osteoporosis. \(^4\) \(^12\) \(^13\) Blood glucose regulation may be abnormal. \(^14\) Emphysema and allergic asthma have also been described. \(^4\) \(^13\) \(^15\) It is unusual for these children to survive past the second decade of life. \(^5\) \(^9\)

There is still no consensus on etiology. Transmission by autosomal recessive inheritance seems likely. \(^5\) \(^7\) \(^9\) "There may also be a metabolic component, possibly a thymic hormone deficit." \(^12\) \(^13\) However, a consistent genetic or metabolic abnormality has not been found. \(^9\) In contrast, the syndrome known as progeria has a clinical onset at 1 year. Osteoporosis is variable, the eyes are normal, and mental retardation is absent. The nature of inheritance is unknown. \(^13\)

Children with Cockayne syndrome have required anesthesia for surgical interventions including eye procedures, \(^9\) dental restorations, \(^4\) \(^16\) and renal biopsy. \(^10\) Cotton and colleagues mentioned briefly that a 12-year-old patient had died following the administration of anesthesia for dental extractions. \(^14\) However, anesthesia for these patients appears to have been described only once in the anesthesia literature when difficult intubation was encountered. \(^4\) It is clear that Cockayne syndrome patients may present with a constellation of abnormalities with important implications for the anesthetist.

**Airway management.** These patients may present with limited neck and jaw movement, small mouths, large teeth, narrow and high palate,
and immobile epiglottis and larynx. Asthma, emphysema, and the presence of a stiff "pigeon-like" thorax may further complicate management. Cook describes postcricoid narrowing requiring a 3.5 mm endotracheal tube on a girl 9 years old.4 Awake intubation is usually not considered because of the inability to secure the patient's cooperation. There is debate regarding the wisdom of using muscle relaxants on a patient with a compromised airway. Judgement must be used after careful examination of the patient and the circumstances under which induction will be accomplished. In any case, pediatric fiberoptic intubating equipment and tracheostomy tray should be readily available. Attention must also be paid to tidal volume and peak airway pressures.

Temperature control and care of the skin. The child with Cockayne syndrome typically has minimal subcutaneous fat, thin skin of poor turgor, and may develop severe skin rash in response to ultraviolet light exposure.17 Therefore, several approaches to temperature maintenance and skin preservation must be considered. Addition of heat by warming pads or overhead warming lamps must be approached cautiously, as must the use of ultraviolet lights. Care of thin, frail skin might include gentle handling, padding the blood pressure cuff, and judicious use and careful removal of tape and electrocardiographic electrodes.

Management of concurrent conditions. Children with Cockayne syndrome may present with multiple organ involvement consistent with progeria. Specifically important to anesthetic management would be essential hypertension, renal and hepatic deficiencies, emphysema, osteoporosis, visual impairments including blindness, and neurological impairments including deafness.

During the preoperative interview it is important to determine the patient's medication regimen and ensure that it will be properly managed throughout the perioperative period. Especially important are medications for the relief of asthma and emphysema and medication to control hypertension. Intraoperatively, propranolol has been proven effective for blood pressure control.11

Many of these patients have lost the ability to speak. If the child is also deaf and blind, all attempts to comfort the child must be accomplished through touch. If allowed by hospital policy, it is most helpful if a parent accompanies the patient to the operating room and remains through the initial phase of induction. The efficacy of this strategy was reported as early as 1967 when the presence of a well-prepared and calm mother was shown to be a valuable asset during the induction of a frightened child.18 Under prescribed circumstances, this practice may reassure both the child and her parent.19,20 If this is not possible, then gentle handling and a reassuring touch must be provided by the anesthetist or an assistant.

Cockayne syndrome renders the patient mentally retarded, with symptoms of progressive old age and involvement of many systems important to anesthesia. A knowledge of the condition will allow the anesthetist to plan strategies of care best suited to the needs of these unfortunate children.

REFERENCES


AUTHORS

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