Anesthetic Management of an Infant with Cockayne's Syndrome

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Cockayne's syndrome (CS) is a rare autosomal recessive disorder characterized by clinical manifestations including dwarfism, progeria, photosensitivity, and mental retardation. This syndrome is also one of the congenital diseases suspected by the typical facial features with small skull, sunken eyes, prominent nose, and small mandible. Patients with CS frequently need general anesthesia for their co-existing diseases, and their physical characteristics in face may lead to difficulties in tracheal intubation.

Although the clinical features of this syndrome are well described in the pediatric literatures, only one report made reference to anesthetic management[1]. This report presents an anesthetic management of an infant with CS undergoing operation for congenital cataract at his ages of 5 and 8 months old. Furthermore we review and discuss clinical characteristics of CS that are likely to pose anesthetic problems.

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Case Report

A 5-month-old male infant (weight 5.5 kg and height 0.58 m) was admitted to our hospital for elective surgery to repair congenital cataract. He demonstrated signs, such as growth retardation, flexion deformity of the extremities, and small mandible, within the first year of life and the diagnosis was confirmed by the biological examination: hyperphotosensitivity of cultured fibroblasts to ultraviolet light. Physical examination on admission revealed a slightly retracted chin. Laboratory examination showed liver dysfunction; aspartate aminotransferase (AST) 84 $IU \cdot l^{-1}$, alanine aminotransferase (ALT) 107 $IU \cdot l^{-1}$, and lactic dehydrogenase (LDH) 664 $IU \cdot l^{-1}$. His previous illness included pneumonia and hepatitis due to cytomegalovirus infection at the age of 2 months, and pneumonia at the age of 5 months.

The patient was premedicated with 0.1 mg atropine i.m. 30 min before induction. Anesthesia was induced via a mask with halothane and nitrous oxide (N_2O) in oxygen. We could ventilate the patient's lungs via a mask though with slight difficulty because of his poorly developed mandible, and then administered 1.0 mg vecuronium intravenously. At laryngoscopy using

a straight-bladed pediatric laryngoscope, the posterior aspect of the vocal code was visualized without restricted mouth opening and without noticing any abnormality of the larynx or upper airway. His trachea was successfully intubated with a 3.5 mm size reinforced silicone rubber endotracheal tube with air leak at peak inspiratory pressure over 20 cmH_2O. During intubation, changes in the heart rate, blood pressure, and hemoglobin O_2 saturation as measured by a pulse oximeter were unremarkable. Anesthesia was maintained with halothane and N_2O in oxygen. No remarkable changes in hemodynamics were observed throughout the surgical procedures. After the completion of operation, muscular blockade was antagonized with intravenous 0.15 mg atropine and 0.3 mg neostigmine. His trachea was then extubated without any complications. His postoperative course was uneventful without any airway obstruciton or respiratory infection.

At his age of 8 months (weight 6.3 kg and height 0.63 m), he again underwent surgery for extraction of cataracta under general anesthesia. No remarkable changes were observed in physical findings. Biochemical examination revealed AST 107 $IU \cdot l^{-1}$, ALT 139 $IU \cdot l^{-1}$, and LDH 694 $IU \cdot l^{-1}$. Anesthesia was induced and maintained with halothane and N_2O in oxygen, and muscule relaxation was obtained with vecuronium. His trachea was intubated with an ID 3.5 mm reinforced silicone rubber endotracheal tube without any complications than those accompanying previous anesthetic management. The perioperative course was uneventful.

Discussion

The CS, first reported in two siblings in 1936,[2] is an autosomal recessive hereditary syndrome including progeria, ophthalmologic complication, and photosensitivity. The life expectancy of patients with CS is limited, and only a small number of patients can survive up to adolescene or early adulthood[3]. Joint contracture, exotropia, cataracta, undiscended testis, and carious teeth have been reported to be frequently found in CS[4]. Because of multiple surgical procedures required for these complications, recognition of risks and problems in anesthesia for CS is of paramount importance.

The accompanying table gives clinical manifestations of CS reported in Japanese literatures (table 1). Severe dwarfism, progeria, ataxia, mental retardation, and skin photosensitivity were present in all cases. More than half patients with CS have been reported to have carious teeth and joint deformity, for which surgical treatments under general anesthesia are needed.

The CS is one of the causative disorders of difficulty in intubation. A small mandible, large teeth for the oral cavity, high arch palate, and a restricted range of mandibular motion cause a difficulty in intubation. Cook reported a case of CS presenting problems in intubation[1]. They included difficult direct laryngoscopy and a severe postcricoid airway narrowing, requiring a tracheal tube of much smaller size than usual. Narrowing of the trachea, however, is not a common feature of CS, and we did not recognize it in our case.

Because disorders of metabolism associated with CS may give rise to renal and/or hepatic failure, and diabetes mellitus, preoperative evaluation of the extent of these conditions are highly recommendable. Renal function may decrease, even if proteinuria is absent or slight. From the fact that renal findings resembled those of an aged kidney, a prematurely aged metabolic state was supposed to cause the changes[5].

Disorders of calcium metabolism are

Table 1. Frequencies of clinical manifestations of Cockayne's Syndrome A review of the literatures in Japan

Symptoms	Incidence	(%)
General appearance		
Dwarfism	61/61	(100)
Small mandible	48/57	(84)
Small skull with sunken eyes	60/61	(98)
Skeletal		
Joint deformity and contraction	54/57	(95)
Kyphosis and/or ankylosis	40/51	(78)
Metabolic disorder		
Liver dysfunction	24/45	(53)
Renal dysfunction	22/25	(88)
Diabetes mellitus	11/22	(50)
Hypertension	5/11	(45)
Osteoporosis	14/16	(88)
Intracranial calcification	45/53	(85)
Neurological		
Mental retardation	61/61	(100)
Optic nerve atrophy	46/50	(92)
Auditory disturbance	47/56	(84)
Ataxia	59/60	(98)
Peripheral neuropathy	35/38	(92)
Miscellaneous		
Carious teeth	33/45	(73)
Cataract	22/38	(58)

likely to result in weakness of bones and teeth, consequently in carious teeth and pathological fracture without an apparent trauma. Caution should therefore be exercised in changing the patient's posture during surgical procedure. Furthermore, overextension of the cervical spine to cope with difficulties in tracheal intubation may cause spinal injury due to cervical fracture.

Visual disturbance due to optic nerve atrophy, cataracta, and corneal ulcers or opacities, hearing loss, and mental retardation are common among patients with CS. Anesthsiologists should reduce anxiety of children with CS prior to undergoing surgery and obtain the better co-operation.

In our case, we encountered only a minor difficulty in airway management and this is probably because the disease was in its early stage. From the progressive nature of the pathologic condition, one can readily infer that airway management will become more difficult on the progress of aging, even though there was no significant difference in difficulty in anesthetic management between 5 and 8 months of life, an interval too short to draw any inference. In recent years, a laryngeal mask has been effectively used in patients with difficulty in intubation[6]. Although ophthalmologic surgery did not encourage us to use this mask, it's use may be justifiable in the patients with CS undergoing other surgical procedures that do not involve the head, face, or neck.

In conclusion, the difficulty in airway management probably has priority over many other complications in the anesthetic management of patients with CS. The progression of this dis-

ease is likely to make tracheal intubation increasingly more difficult with elapse of time and a complete preoperative evaluation of the airway and selection of an appropriate approach to the anticipated problems become all the more important, accordingly. In addition, preoperative examination for metabolic disorders including renal and hepatic dysfunction and diabetes mellitus is also highly recommended.

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