

Anesthetic Management of a Patient with Cri Du Chat Syndrome. Case Report

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Summary: Santos KM, Rezende DC, Borges ZDO – Anesthetic Management of a Patient with Cri Du Chat Syndrome. Case Report.

Background and objectives: Cri Du Chat syndrome is a chromosomal disorder with peculiar clinical characteristics including airways abnormalities that require special care by anesthesiologists when handling those patients.

Objective: To present a case of outpatient anesthesia in a patient with Cri Du Chat syndrome and discuss the anesthetic aspects related to this disorder.

Case report: Male patient, 14 years old, 25 kg, with Cri Du Chat syndrome, physical status ASA P2, was admitted for upper gastrointestinal endoscopy and esophageal dilation. The patient had mental retardation, a few episodes of seizures, and marked hypertonia of the limbs. Airways exam showed limited cervical mobility and thyromental distance less than six centimeters. The patient was unable to comprehend verbal commands, making it difficult to undertake a complete assessment of the airways. Other findings on physical exam included microcephaly, micrognathism, subtle strabismus, limb hypertonia with flexion, and protrusion of the tip of the tongue. Intravenous fentanyl 50 µg, midazolam 1 mg, and propofol 60 mg were administered. The patient was maintained on spontaneous ventilation. The procedure lasted 5 minutes, without interurrences.

Conclusions: Patients with Cri Du Chat syndrome have clinical characteristics that are very important for their anesthetic management, being the responsibility of the anesthesiologist to consider carefully the structural particularities of each patient.

Keywords: ANESTHESIA: ambulatorial; DISEASES, Genetic: Cri Du Chat syndrome; SEDATION.

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INTRODUCTION

Cri Du Chat syndrome was first described in 1967 by the French geneticist Lejeune et al ¹. The cry of newborns with this disorder is similar to that of a meowing kitten, to which this syndrome owes its name.

This is a chromosomal disorder that affects approximately 1:50,000 live born; however, in the literature this incidence varies considerably. For patients with mental retardation the incidence is 1.5:1,000 ². It originates from the terminal deletion of the short arm of chromosome 5p; however, chromosomal translocations and inversions can contribute for the etiology of this syndrome.

Mental retardation is a common clinical finding, being noticeable in the first year of life. Other signs vary according to the phase of development of the individual and include: microcephaly, hypertelorism, low-set ears, hypertonicity, scoliosis,

flatfoot, facial asymmetry and laxity (maintaining an open mouth with protrusion of the tongue), prominent orbital arch, poor dental occlusion, elongated facies (in newborns it is usually rounded), palpebral fissures, divergent strabismus, flat nasal bridge, and repetitive respiratory infections ³. Seizures are rare. Cryptorchidism may be seen in some patients. Malformations are less common and they may include cardiac (patent ductus arteriosus and septal defects) ⁴, neurologic, and renal manifestations.

Anesthesia for patients with Cri Du Chat syndrome involves some peculiarities, especially when managing the airways. This article reports the case of an adolescent with Cri Du Chat syndrome undergoing intravenous sedation for upper gastrointestinal endoscopy and esophageal dilation, besides reviewing some questions about the anesthetic management of those patients.

CASE REPORT

This is a 14-year old male patient with Cri Du Chat syndrome weighing 25 kg, physical status ASA P2, admitted as an outpatient for upper gastrointestinal endoscopy and esophageal dilation. Preoperative exams were within normal limits. He had a history of prior admissions for pneumonia, orchiopexy, and esophageal dilations. The patient had mental retardation, a few episodes of seizures, and important hypertonia of the limbs. His medications included topiramate, nitrazepam, and pantoprazole. Exam of the airways showed limited cervical mobility, thyromental distance lower than six centime-

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ters, non-invasive blood pressure 90/60 mmHg, heart rate 112 bpm, and normal breath sounds. The patient was unable to follow verbal commands, which hindered full evaluation of the airways; however, family members reported good mouth opening and no history of difficult intubation in prior surgeries. Other findings included microcephaly, micrognathism, subtle strabismus, limb hypertonia in flexion, and protrusion of the tongue.

Monitoring included electrocardiograph (DII and V5 derivations), non-invasive blood pressure, and pulse oximetry. Peripheral venous access was established with a 22G Teflon catheter. Fentanyl 50 µg, midazolam 1 mg, and propofol 60 mg were administered IV. The patient was maintained on spontaneous ventilation with peripheral oxygen saturation ranging from 93% – 100%. The procedure lasted 5 minutes and was performed without interurrences. During the first 45 minutes in the postanesthetic recovery room the patient was very sleepy and awakened only when he was called. After 90 minutes, the patient was discharged to be accompanied by his legal guardian with a maximal score in the modified Aldrete-Kroulik scale.

DISCUSSION

When patients with Cri Du Chat syndrome are scheduled to undergo surgical intervention or ancillary exams under anesthesia and present to the Anesthesia clinic for preanesthetic evaluation, they usually are being followed by a multidisciplinary team, which includes neurologists, occupational therapists, and physical therapists. Judicious evaluation of the airways and all organ systems is important to detect other specific areas that can influence the anesthetic plan for that patient.

Preferably, patients with Cri Du Chat syndrome should undergo careful cardiologic evaluation by a specialist to detect the possible presence of cardiovascular abnormalities, including septal defects and stenosis of the pulmonary artery. Jugular vein engorgement, symptoms of congestive heart failure, and murmurs on auscultation should alert the anesthesiologist for the presence of concomitant cardiac disease and, therefore, the patient should be referred to a cardiologist for evaluation.

siologist for the presence of concomitant cardiac disease and, therefore, the patient should be referred to a cardiologist for evaluation.

The airways of a patient with Cri Du Chat syndrome require special attention by the anesthesiologist. Laryngeal (hypoplasia, narrowing, vocal cord asymmetry) and epiglottis (small, atonic, flaccid) abnormalities, besides neurologic involvement, represent peculiar characteristic present in those patients and seem to contribute for the characteristic cry². Those abnormalities, combined with micrognathism and alterations of the hard and soft palate, can contribute for a difficulty to intubate airways. In the case presented here, since it was a relatively fast exam in the hands of well-trained endoscopists, tracheal intubation was not necessary, but it is suggested that supraglottic mechanisms of airways management such as laryngeal mask be readily available in case of failure to intubate. In theory, the bougie could be another helpful device in those cases. According to Brislin et al.⁵, hypotonic pharyngeal muscles could also contribute for obstruction of the airways and, therefore, preanesthetic medication should be avoided. Fast acting neuromuscular blocking agents are preferable to long-acting ones due to the hypotonia component; however, reports on the association of this syndrome with malignant hyperthermia or succinylcholine-induced hypercalcemia were not found in the literature⁵.

Besides, also according to Brislin et al.⁵, it is recommended that patients with this syndrome handled on an outpatient basis should be observed for a longer period during postanesthetic recovery, until they are well awakened and free of residual anesthetic effects. In the case presented here, the patient was observed for 90 minutes and he was discharged with a maximal score in the modified Aldrete-Kroulik scale.

Therefore, one can see that patients with Cri Du Chat syndrome have clinically important characteristics for the anesthetic management, may it be on an outpatient basis or hospitalized. With the diagnosis made by a multidisciplinary team, the anesthesiologist should be careful when managing those patients, observing the structural particularities of each patient.

REFERÊNCIAS / REFERENCES

01. Lejeune J, Lafourcade J, Berger R et al. – Trois cas de délétion partielle du bras court d'un chromosome 5. *CR Hebd Seances Acad Sci*, 1963;257:3098-3102.
02. Niebuhr E – The Cri du Chat syndrome: epidemiology, cytogenetics and clinical features. *Hum Genet*, 1978;44:227-275.
03. Mainardi PC – Cri Du Chat Syndrome. *Orphanet J Rare Dis*, 2006;1:33.
04. Hills C, Moller JH, Finkelstein M et al. – Cri du Chat syndrome and congenital heart disease: a review of previously reported cases and presentation of an additional 21 cases from the Pediatric Cardiac Care Consortium. *Pediatrics*, 2006; 117:e924-927.
05. Brislin RP, Stayer SA, Schwartz RE – Anaesthetic considerations for the patient with Cri du Chat syndrome. *Paediatr Anaesth*, 1995;5:139-141.

Resumen: Santos KM, Rezende DC, Borges ZDO – Manejo Anestésico de Paciente con Síndrome de Cri Du Chat (Maullido del Gato). Relato de Caso.

Justificativa y objetivos: El síndrome de Cri Du Chat es un desorden cromosómico con características clínicas peculiares, que incluye anomalías en las vías aéreas, exigiendo de los anestesiólogos cuidados especiales en el manejo de esos pacientes.

Objetivo: Presentar un caso de anestesia ambulatorial en paciente con síndrome de Cri Du Chat y abordar los aspectos anestésicos relacionados con esa enfermedad.

Relato del caso: Paciente del sexo masculino, 14 años, 25 kg, portador de síndrome de Cri Du Chat, estado físico ASA P2, admitido para la realización de endoscopia digestiva alta y dilatación esofágica. Cuadro neurológico con retraso mental, algunos episodios de convulsiones e hipertonia acentuada de los miembros. El examen de las vías aéreas reveló movilidad cervical limitada y distancia tireoentoniana inferior a 6 cm. El paciente no respondía al comando verbal, siendo difícil la evaluación completa de las vías aéreas. Otros hallazgos en el examen físico incluyeron microcefalia, micrognatia, discreto estrabismo, hipertonia de los miembros en flexión y protrusión de la lengua. Fueron administrados por vía venosa, 50 µg de citrato de fentanila, 1 mg de midazolam y 60 mg de propofol. Se le mantuvo en ventilación espontánea. El procedimiento demoró 5 minutos y fue realizado sin interurrencias.

Conclusiones: Los pacientes con el síndrome de Cri Du Chat, presentan características clínicas de gran relevancia para el manejo anestésico, y el anestesiólogo tiene que considerar con cautela las particularidades estructurales de cada paciente.