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CLINICAL REPORT

Spinal anesthesia in a child with Brachmann-de Lange (Cornelia de Lange) syndrome

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Abstract Providing anesthesia to patients with Brachmann-de Lange syndrome (BdLS) may be challenging, mainly because of intubation difficulty, gastroesophageal reflux disease, and aspiration complications. The use of spinal anesthesia (SA) in this population has not been reported. We report the uneventful administration of awake SA to a 7-month-old girl with BdLS who was scheduled for rectal biopsy. The current literature is reviewed to discuss the indications for SA in those patients.

Keywords Spinal anesthesia · Brachmann-de Lange syndrome · Children

Introduction

Brachmann-de Lange syndrome (BdLS), also known as Cornelia de Lange syndrome (CdLS), is a rare multiple malformation syndrome (1 in 10,000 live births), typically involving proportionate small stature, specific facial features with cleft palate and micrognathia, major malformations (particularly the cardiac, gastrointestinal, and musculoskeletal systems), developmental delay, and behavioral abnormalities [1]. Additionally, many of these children suffer from lung disease as a consequence of gastroesophageal reflux disease (GERD) with recurrent aspiration pneumonia [2]. Changes in three different genes have been identified as causing BdLS: NIPBL on chromosome 5, SMC1A on the X chromosome, and SMC3 on chromosome 10. Changes in the latter two genes seem to correlate with a milder form of the syndrome [3].

Providing anesthesia to patients with BdLS may be challenging. Previous reports mainly discuss the issues that arise when using general anesthesia (GA) in patients with BdLS, such as intubation difficulty, GERD and aspiration complications, pericarditis, and hyperthermia [4–14]. However, the use of spinal anesthesia (SA) in this population has not been reported. We report the uneventful administration of awake SA to a 7-month-old girl with BdLS, who was scheduled for rectal biopsy to exclude Hirschsprung's disease diagnosis.

Case description

The girl was born full term, at birth weight 3,090 g. The diagnosis of BdLS was made soon after birth from her characteristic face, hirsutism, cleft palate, and the presence of congenital heart defects, confirmed by cytogenetic analysis. Before the current operation she uneventfully underwent GA at the age of 2 days for atrial septal defect (ASD) closure and coarctation of aorta repair, and twice for central line insertion (at the age of 4 and 6 months, both because of aspiration pneumonia). At the time of the current operation she weighed 3,330 g, suffered from chronic lung disease (CLD) because of GERD, and was on treatment with budesonide and albuterol. Her CLD consisted of cough, wheezing, and recurrent aspiration pneumonia.

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Oxygen saturation was 95% on room air. The patient had mild mental retardation.

Physical examination showed a child with characteristic appearance of BdLS (micrognathia, synophrys, hirsutism, cleft palate). Wheezing was noted during auscultation of the lungs. Ultrasonographic examination of the abdomen revealed partial sacral hypoplasia with normal position of conus medullaris. As no neurological deficit was found during the preoperative assessment, magnetic resonance imaging (MRI) or computed tomography (CT) scan of spinal cord and vertebral column was not performed. Normal blood count, electrolyte and coagulation profiles, and electrocardiogram (ECG) were obtained. No premedication was prescribed. After placement of the routine monitors, the patient was placed in the lateral decubitus position and the skin aseptically prepared with chlorhexidine in tinted 70% alcohol. A neonatal lumbar puncture needle, 25 gauge (0.50 × 51 mm; Becton-Dickinson, Fraga Hueska, Spain), was used for lumbar puncture at the L3–L4 level. Spinal puncture was performed under the awake condition. After free flow of cerebrospinal fluid was obtained, 2.5 mg 0.5% normobaric bupivacaine hydrochloride without epinephrine (0.75 mg/kg) (Marcaine Spinal 0.5%; Astra Zeneca) was slowly injected using a 1-ml syringe. The patient was then repositioned on her back, and the adequacy of SA was determined by the presence of profound motor block (unable to move feet, knees, and legs) in the lower extremities and the absence of a skin prick response at the level of the perineum. The surgical time was 15 min. During the surgery cardiovascular parameters remained stable, with systolic blood pressure (BP), 76–93 mmHg, diastolic BP, 37–49 mmHg, and heart rate, 118–128 beats/min. There was no need for supplemental sedation during the operation. There were also no perioperative problems, and the child was transferred to the post-anesthesia care unit. Two hours later she was discharged to the ward. The surgeon rated the quality of anesthesia using an in-house grading system as 10 (scale from 0 to 10: 0 being the worst, 10 the best). The biopsy was not suggestive for Hirschsprung's disease, but showed signs of inflammation, and the child did not need further surgery at the current time.

Discussion

It has been suggested that the possibility of difficult intubation and recurrent respiratory problems may be indications for regional anesthesia in patients with BdLS [7, 15]. However, we were able to find only two case reports describing the use of combined (general and caudal) anesthesia in those patients. Lumb and Carli [16] reported a case of a 3.5-year-old boy who developed respiratory arrest after caudal injection of bupivacaine, but this complication most likely resulted from inadvertent subarachnoid injection [17, 18]. On the other hand, successful use of caudal block with bupivacaine for bilateral herniorrhaphy in an infant with BdLS was reported by del Rio et al. [19]. The main indication for caudal analgesia was optimal intra- and postoperative pain control. Both patients had no problems of GERD and CLD. Our patient was known to have recurrent aspiration pneumonia, and presented for surgery shortly after exacerbation of her lung disease. Based on the high incidence of GERD in BdLS patients, we postulated that diminishing pulmonary complications in this population is an important goal for the use of SA as well.

SA is a safe and effective alternative to GA in newborns and infants in experienced hands [20–22]. This technique has a low incidence of complications, a high degree of cardiorespiratory stability, and offers excellent relaxation. Additionally, SA provides the means for avoiding airway instrumentation, which is especially important in patients with possible difficult intubation or CLD, and allows avoidance of rapid sequence induction (RSI) and gastric aspiration in patients with GERD. Moreover, SA alone or in combination with GA was successfully applied in patients with cardiovascular anomalies that are common in patients with BdLS [22–24]. Even though in the past two decades the popularity of SA in pediatric patients has increased considerably, often it remains limited to situations in which GA poses a major risk, as in premature or ex-premature infants. The case described was performed in a center where SA is widely and routinely used in neonates and infants at elective program and in after-hours on-call operations as well. Previously we have reported 505 cases performed during an 8-year period with a high success rate and minimal complications [21].

Although skeletal anomalies are quite common in patients with BdLS, the usual place of prevalence are the upper extremities. Spinal cord and/or vertebral column abnormalities are rare. Among 34 patients in the series of Roposch et al. [25], only 4 had scoliosis, and Mutlu et al. [26] described one case of CdLS associated with thoracic meningocele. In view of these data and keeping in mind the absence of neurological abnormalities and presence of severe CLD with recurrent aspiration pneumonia from GERD in our patient, we decided not to perform further radiologic evaluation of the spinal column or cord. Furthermore, spinal column defects are not a contraindication for SA administration, if they are not located in the place of spinal puncture. Previously Viscomi et al. [27] reported the successful use of SA for repair of lumbar or sacral meningomyelocele in 14 neonates. In our case, lumbar puncture was performed at the L3–L4 level without difficulties, and no complications were observed intra- or postoperatively.

In conclusion, SA may be advisable for patients with BdLS scheduled for surgery, appropriate for this technique.

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