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CASE REPORT

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Anesthesia management in Bart's syndrome: A case report

Bart's sendromunda anestezi yönetimi: Bir olgu sunumu

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ABSTRACT

Bart's syndrome is a genetic disorder that is also associated with epidermolysis bullosa (EB), which is characterized by congenital focal absence of the skin, mechanical bullous, and nail dystrophy. We present the anesthesia management of a male neonate with congenital localized skin absence, nail dystrophy, and ear atrophy who underwent surgery due to congenital pyloric atrophy. Palliative measures were perforemed paying attention to skin lesions in areas of rich-red peeling on the hands and feet of the patient, and standard general inhalation anesthesia was administered. As far as I could research in the English literature, this case report may be the first case report to present the management of anesthesia in a patient with Bart syndrome

Keywords: Bart's syndrome; case report; congenital absence of the skin; congenital pyloric atresia; epidermolysis bullosa.

ÖZ

Bart's sendromu, derinin konjenital fokal yokluğu, mekanik büllöz ve tırnak distrofisi ile karakterize epidermolizis bülloza (EB) ile de ilişkili genetik bir hastalıktır. Konjenital pilorik atrofi nedeniyle ameliyat edilen doğuştan lokalize cilt yokluğu, tırnak distrofisi ve kulak atrofisi olan erkek yenidoğanın anestezi yönetimini sunuyoruz. Hastanın el ve ayaklarında zengin kırmızı soyma bölgelerindeki cilt lezyonlarına dikkat edilerek palyatif önlemler alındı ve standart genel inhalasyon anestezisi uygulandı. İngilizce literatürde araştırabildiğim kadarıyla bu olgu sunumu Bart sendromlu bir hastada anestezi yönetimini sunan ilk olgu sunumu olabilir.

Anahtar Kelimeler: Bart's sendromu; olgu sunumu; cildin doğuştan yokluğu; konjenital pilorik atrezi; epidermolizis bülloza.

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Introduction

Bart's syndrome is a genetic disorder that is also associated with epidermolysis bullosa (EB), which is characterized by congenital focal absence of the skin, mechanical bullous, and nail dystrophy. It was described by Bart et al. in 1966 [1]. Raw and red plaques, which are a sign of localized skin absence, appear on different parts of the patient's body. There is a sharp boundary between affected and normal skin. Any part of the skin may be affected, but the disease tends to be more common in parts of the body that are subject to friction and trauma, such as the skin around the mouth and extremities [2].

With this case report, anesthesia management in a neonate with Bart syndrome who underwent surgery with a diagnosis of congenital pyloric atresia is described.

Case Presentation

A 35-week, 1800 grams, 1-day-old male neonate was taken to surgery for pyloric atresia. A physical examination revealed that there was no skin extending from both knees on the anteromedial aspect of the lower leg to the dorsal and medial plantar directions of the feet (Figure 1). There were also similar lesions involving both elbows. A sudden transition to normal skin where the lesions ended was striking (Figure 2). In addition to the absence of skin, hypoplastic nails (Figure 3) and ear atrophy were present. No brain anomaly was detected in transfontanelle ultrasonography (USG). There was no erosion in the mouth or nasal cavity. Complete blood count, liver and kidney function tests, and electrolyte results were within normal limits. Immunohistologic and genetic linkage studies of the sample taken from the skin after birth could not be performed in the period until the surgery (within 24 hours). Hydrocortisone + mupirocin mixed pomade was applied to the patient's body before being taken to the operating table, and he was carried on a plastic pad. Thus, pathologies that could occur with the friction of his body were avoided. Electrocardiography (ECG) pallets were placed on the back of the patient because the palettes were not self-sticking, and in this way heart rate and ECG monitoring were achieved. A pulse oximeter for SpO2 was connected to the lower extremity. The cautery plate was placed on the back. Vascular access was provided through the umbilical vein. After induction, anesthesia was provided with 3 mg.kg-1 propofol, 1 mcg.kg-1 fentanyl, 0.6 mg.kg-1 rocuronium, and endotracheal intubation was provided at the appropriate time with a No.3 endotracheal tube. No intraoral lesions were observed. The endotracheal tube was fixed to the rim with a hypoallergic silk patch. Anesthesia was maintained with 40% O2 and 3.2% sevoflurane. The patient, who underwent gastrojejunostomy, was transferred to the neonatal intensive care unit (NICU) as intubated at the end of the procedure. The neonate died on the 7th postoperative day.

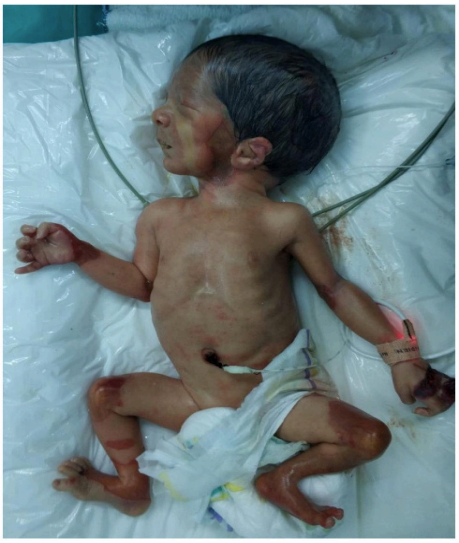

Figure 1. Absent skin, on face and neck, bilaterally on the upper and lower extremities

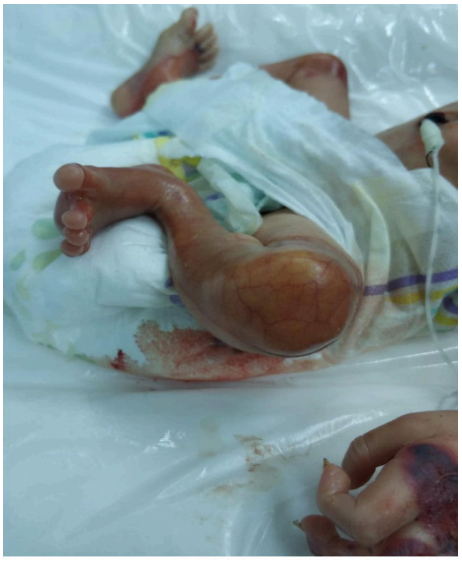

Figure 2. Sharply demarcated lesion margin.

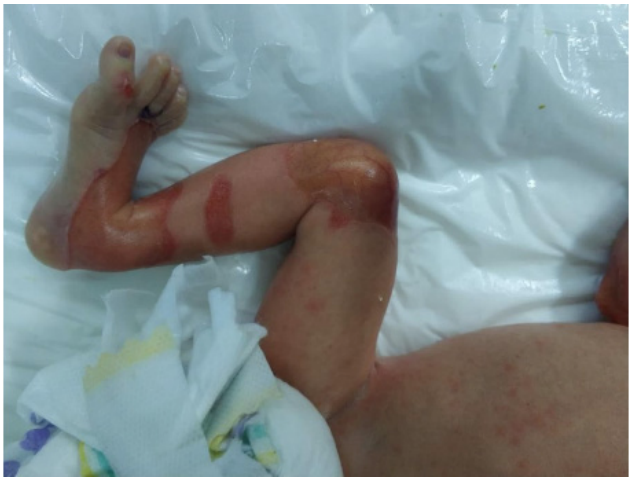

Figure 3. View of hypoplastic nails

Discussion

Bart's syndrome is known to be one of the subtypes of EB [3], which initially presents with congenital absence of skin in the lower leg and common skin, mucous membrane, and nail dystrophy [4]. Duran-McKinster et al. suggested that the congenital absence of localized skin in Bart's syndrome might follow Blaschko's lines due to physical trauma to the uterus [5]. Zelickson et al. demonstrated various abnormalities of anchorage fibrils, which are predominantly composed of VII type collagen, at the dermal-epidermal junction in Bart's syndrome [6]. The autosomal dominant inheritance of this syndrome has been demonstrated. However, some patients with unaffected parents are believed to be due to sporadic mutations [7]. Chiaverini et al.[7] reported that collagen VII gene was associated with mutations in the triple helix area in sporadic cases. This mutation can lead to Bart's syndrome, associated with the synthesis of a thermolabilin Col 7. In the present case, there was no family history of congenital localized skin absence and raised lesions. Bart's syndrome is sometimes accompanied by nail abnormalities. Our patient had a congenital localized absence of skin, involvement of raised lesions, and partial nail involvement. Bart's syndrome may also be associated with other anomalies such as pyloric atresia, primitive ear development, flattened nose, broad nasal root, and wide-set eyes [8]. There were associated findings in the present case.

Bart's syndrome has been associated with pyloric atresia [9,10]. In our case, the reason for the surgery was congenital pyloric atresia and gastrojejunostomy was performed. During the procedure, the patient was on a plastic pad and because the hydrocortisone + mupirocin mixed pomade was applied to his body, it was not possible to stick the ECG pallets, so they were placed on the patient's back. We think that the use of a cuff for pulse pressure may pose a risk because patients with Bart's syndrome are quite susceptible to skin trauma. In addition, the patches used to fix the vascular access can cause skin trauma. In the present case, an umbilical venous catheter was used for volatile anesthetics, opioid and neuromuscular blockers for anesthesia induction, and maintenance. Treatment management in Bart's syndrome is generally conservative. Treatment aims to prevent infection of the affected area, accelerate healing, and reduce the risk of scarring [11]. Close monitoring is essential for serious complications such as bleeding, infection, hypothermia, and hypoglycemia. The prognosis is good and depends on the effectiveness of the treatment. However, death occurred on the 7th postoperative day in the present case. The possible cause of sudden death in our case may have been of metabolic origins, such as hypothermia and hypoglycemia due to congenital absence of skin or concomitant infection.

Conclusion

This case report has shown that preoperative use of moisturizing + antibiotic ointments avoided possible advanced skin trauma. The key components of this case were anesthesia induction including optimal preoperative preparation and controlled induction of anesthesia. Standard general anesthesia techniques and anesthetic drugs (including sevoflurane, fentanyl, remifentanil and rocuronium) were used with success. This case report is the first on anesthetic management in Bart's syndrome in the literature.

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