Rev Bras Anestesiol. 2014;64(3):201–204

REVISTA BRASILEIRA DE ANESTESIOLOGIA

Official Publication of the Brazilian Society of Anesthesiology
www.sba.com.br

CLINICAL INFORMATION

Anesthesia for a patient with Fanconi anemia for developmental dislocation of the hip: a case report

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Received 12 November 2012; accepted 19 December 2012
Available online 23 October 2013

KEYWORDS
Fanconi anemia;
Caudal anesthesia;
Developmental dislocation of the hip

Abstract Fanconi anemia is a rare autosomal recessive inherited bone marrow failure syndrome with congenital and hematological abnormalities. Literature regarding the anesthetic management in these patients is limited. A management of a developmental dislocation of the hip was described in a patient with fanconi anemia. Because of the heterogeneous nature, a patient with fanconi anemia should be established thorough preoperative evaluation in order to diagnose on clinical features. In conclusion, we preferred caudal anesthesia in this patient with fanconi anemia without thrombocytopenia, because of avoiding from N_2O, reducing amount of anesthetic, existing microcephaly, hypothyroidism and elevated liver enzymes, providing postoperative analgesia, and reducing amount of analgesic used postoperatively.

Introduction

Fanconi anemia (FA) is a rare autosomal recessive inherited bone marrow failure syndrome, characterized by increased chromosomal fragility, and generally associated with multiple congenital anomalies.[1]

Literature regarding the anesthetic management in these patients is limited. A management of a developmental dislocation of the hip was described in a patient with FA.

Case report

A 2.5-year-old girl, born to first degree consanguineous parents presented with a failure to walking. She was delivered by spontaneous vaginal in time and small for gestational age, weighing 2.2 kg at birth. Physical examination revealed retarded growth (5.7 kg weight and 68 cm height) with absence of bilateral thumb (Fig. 1), café au lait spots at back, hypopigmentation at abdomen, generalized hyperpigmentation, and microcephaly.

Laboratory investigations did not reveal pancytopenia; hemoglobin, WBC and platelets were 11.4 g dl^{-1}, 7.4×10^3 dl^{-1} and 162×10^3 dl^{-1} respectively. ALP, GGT and

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0104-0014
http://dx.doi.org/10.1016/j.bjane.2012.12.006

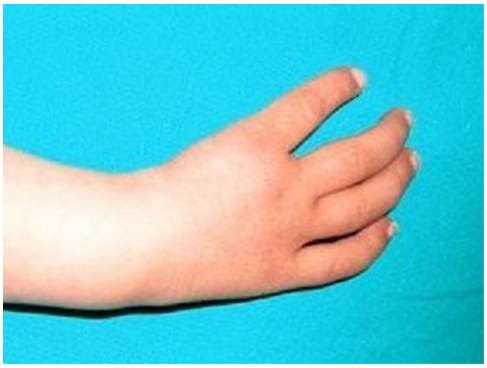

Figure 1 Absence of the thumb.

LDH levels were elevated and 564, 420 and 289 respectively. Ultrasound revealed hypoplastic horseshoe kidney.

The patient's medication included only levothyroxine because of hypothyroidism. No medication exist related FA.

Other investigations including electrolytes, BUN, creatinine, bilirubins, AST, ALT, prothrombin time, partial thromboplastin time, ECG, and echocardiogram were normal. The patient was euthyroidic.

Evaluation of the airway revealed no abnormalities, except microcephaly, and vital signs were normal. Adequate blood and blood products ordered depending on the procedure.

No medication was administered prior to the procedure apart from antibiotics. Anesthesia was induced with 8% sevoflurane in 100% oxygen. After peripheral i.v. access was secured, she was turned to the left lateral position. Caudal block was performed by using a 22-gauge short-bevel needle under aseptic conditions with bupivacaine 0.25% at 1 ml kg^{-1}. After caudal blockade inhalation anesthesia was lowered to 0.5–1% in 100% oxygen in order to obtain 4 or 5 point of Ramsay sedation scale. Peripheral oxygen saturation (SpO_2), heart rate, noninvasive blood pressure and end-tidal CO_2 ($ETCO_2$) were monitored throughout surgery. Ventilation was maintained via an oxygen mask with 2 L dk^{-1} fresh flow spontaneously (Fig. 2).

Whole procedure including open reduction and putting the limb in a plaster cast from induction to recovery lasted 140 min and was uneventful. Recovery was fast and uneventful. Postoperative analgesia was not required for ten hours.

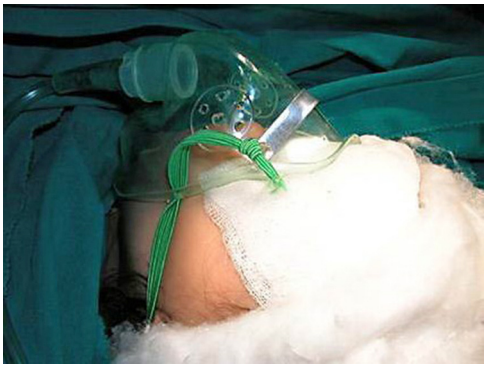

Figure 2 The patient with facemask during surgery.

Discussion

Fanconi anemia is an autosomal recessive disorder in over 99% of cases with 25% risk to siblings.[1] The incidence of FA is approximately three per million and the heterozygote frequency is estimated at one in 300 in Europe and the USA,[2] 1/100 in Ashkenazi Jews and South African Afrikaners due to ''founder effect''.[1,3] Consanguinity has been implicated in its incidence.[3]

Clinical features of FA can be broadly divided into two categories: congenital abnormalities and hematological abnormalities, which may include altered skin pigmentation and/or café au lait spots, short stature (impaired growth hormone secretion), thumb or thumb and radial anomalies, hip anomalies, vertebral scoliois, rib anomalies, male hypogenitalism, mental retardation, anorectal atresia, duodenal atresia, microcephaly, eye anomalies (microphthalmia, strabismus, ptosis, nystagmus), structural renal defects, low-birth weight, developmental delay, abnormal ears or hearing, and cardiopulmonary defects.[1–3] The most important clinical features of FA are hematological. Pancytopenia is the usual finding and typically presents between the ages of 5 and 10 years, the median age of onset being 7 years.[1] Clinically, the affected FA patient may present with bleeding, pallor and/or recurring infections.[2] The major cause of death in FA is bone marrow failure, followed in frequency by leukemia and solid tumors. The projected median survival from all causes for more than 1000 cases reported in the literature is age 20 years.[3] Of patients thirty or forty percent lack obvious physical abnormalities.[1–3] There is great clinical heterogeneity even within a genotype (sibling may be phenotypically different).[1] The gold-standard screening test for FA is based on the characteristic hypersensitivity of FA cells to the crosslinking agents, such as mitomycin C (MMC) or diepoxybutane (DEB).[5] FANCA is the most common complementation group, representing about 70% of cases.[1] Androgen therapy (oxymetholone), cytokines (G-CSF and GM-CSF), and supportive therapies such as blood and platelet transfusions form the mainstay of treatment prior to allogeneic hematopoietic stem cell transplantation. Gene therapy is experimental. E-amino caproic acid may be used for symptomatic bleeding. Steroids are occasionally used.[1–3] FA frequently terminates in myelodysplastic syndrome and/or leukemia.[1] Hematopoietic stem cell transplantation (bone marrow, cord blood, or peripheral blood stem cells) may cure aplastic anemia and prevent myelodysplastic syndrome or leukemia.[2,3]

Patients with FA may present for procedures such as Hickman catheter insertion for bone marrow transplantation, limb reconstructive surgeries, splenectomy, gastrointestinal anomalies, congenital heart defects, trauma or other related and/or non-related indications. The insufficiency of literature regarding anesthetic management of this condition makes it difficult to estimate the real impact of individual anomalies on the perioperative management.[6] It is important to emphasize that of patients thirty or forty percent have no abnormalities.[1–3]

Because of the heterogeneous nature of the condition, FA is difficult to diagnose on clinical features.[2] A thorough preoperative evaluation includes airway examination, and hematological, cardiopulmonary, hepatorenal