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Case Report

Anesthesia in a patient with dyskeratosis congenita presenting for urgent subtotal gastrectomy

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Abstract Dyskeratosis congenita is a rare and complex congenital disease that may complicate surgical treatment and impact anesthetic care. We present the perioperative management of a patient with severe pancytopenia, respiratory dysfunction, and oral leukoplakia who presented for urgent surgery for removal of a gastric hemorrhagic malignant tumor. Important issues in the management of this patient include choice of anesthetic technique, correction of pancytopenia (thrombocytopenia in particular), judicious perioperative fluid management to avoid dilutional coagulopathy, antibiotic prophylaxis, and strict aseptic technique. Careful management of a potentially difficult airway and a higher likelihood of respiratory insufficiency further complicate patient care. Knowledge of this rare disease process and its potential impact on anesthetic management is paramount for safe perioperative patient care.
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1. Introduction

We report a case of gastric adenocarcinoma in a patient who presented for urgent surgery. The patient had been diagnosed with dyskeratosis congenita, which represents a complex challenge for both the anesthetic and surgical management. Dyskeratosis congenita, also known as *Zinsser-Engman Cole syndrome*, is a rare congenital disease with an overall incidence of about 1 in 1,000,000 people. Patterns of inheritance have shown genetic heterogeneity, mutations of the RNA component of telomerase producing an autosomal-dominant form, mutations in other uncharacterized genes producing an autosomal recessive form, and mutations in the gene dyskeratosis congenita 1 (*DKC1*), which produces an X-linked recessive form of disease [1–3]. The root of the disease process centers on bone marrow failure. Thus, clinically, this multisystem disease is seen in organ systems that depend on cell turnover and normal stem cell proliferation [4]. Manifestations of pathology vary in severity and importance: skin (reticular pigmentation) and

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mucosal (oral leukokeratosis) changes; nail and hair dystrophy; advanced dental disease, including alveolar bone loss and gingival inflammation; hematopoietic bone marrow dysfunction characterized by pancytopenia (anemia, leucopenia, and thrombocytopenia); stenoses of the esophagus, urethra, and lacrimal ducts; malignant neoplasms of the mouth and nasopharyx; and a particular susceptibility to infections. Furthermore, patients with dyskeratosis congenita may manifest differing degrees of pulmonary fibrosis with subsequent deterioration in respiratory function [3].

Because of the rarity of dyskeratosis congenita, anesthetic and surgical experience with these patients is limited to a small number of published cases (2 cases of dental surgical management) [5,6].

Written, informed consent was obtained from the patient for publication of this case report. A copy of the written consent is available for review by the Editor-in-Chief of this journal. Approval was obtained from our institution's review board.

2. Case report

A 43-year-old man with a known history of dyskeratosis congenita was referred to gastroenterology for an investigation of dysphagia and 1 episode of melena. Upper endoscopic evaluation revealed stenosis of the lower esophagus as well as an ulcerative, infiltrative lesion in the gastric antrum. Histopathological examination of biopsies confirmed the diagnosis of gastric adenocarcinoma. Given the ulcerative nature of the lesion with potential for hemorrhage, the patient was admitted for urgent surgical intervention.

A preoperative physical examination revealed a mildly malnourished patient weighing 55 kg with a body mass index of 17.5 and exhibiting signs of dyskeratosis at the level of the skin and nails. Although oral examination revealed no obvious oral tumors, leukoplakia of the piriform recess, severe dental caries, and gingival hyperplasia were present. Skin and mucosal inspection revealed no hematoma, petechiae, or other signs of blood dyscrasia. Preoperative testing included a complete blood count that revealed leucopenia: white blood cell count $1.57 \times 10^3/\mu L$ (reference range, 4-10 $\times$ $10^3/\mu L$), severe thrombocytopenia: platelet count $26 \times 10^3/\mu L$ (reference range, 140-440 $\times$ $10^3/\mu L$), and mild anemia: hemoglobin levels 10 g/dL (reference range, 14-18 g/dL). All coagulation parameters—prothrombin time (PT) of 16.3 seconds (reference range, 13-17 seconds), international normalized ratio (INR) of 1.05 (reference range, 0.84-1.1), and bleeding time of 2 minutes (reference range, 1-4 minutes)—were in reference range. A chest radiograph and computed tomographic scan revealed diffuse pulmonary fibrosis with signs of increased pulmonary artery size consistent with secondary pulmonary hypertension.

Pulmonary function tests showed a mild restrictive respiratory insufficiency (vital capacity = 78%, forced expiratory volume in the 1st second = 72%, forced expiratory flow between 25%-75% of FVC = 83%, peak expiratory flow rate = 78%).

Echocardiography revealed mild mitral regurgitation and prolapse with mild pulmonary hypertension (systolic pulmonary artery pressure = 40 mm Hg).

The day before the surgical intervention, 9 U of platelet concentrate and 2 U of packed red blood cells were administered to the patient. This transfusion increased the platelet count to $80 \times 10^3/\mu L$ at the time of surgery without a significant increase in the other cellular lines (leucocyte count, $2.45 \times 10^3/\mu L$; hemoglobin level, 9.0 g/dL). The patient was deemed adequate for surgery and moved to the operating theater. Before surgical incision, intravenous cephalexin and metronidazole were administered. Following placement of standard electrocardiogram and pulse-oximetry monitors, induction of general anesthesia was achieved with propofol and fentanyl. After paralysis was accomplished using rocuronium, endotracheal intubation was performed with mild difficulty under direct laryngoscopy using a Machintosh 4 blade (Mallampati class 1, Cormack-Lehane grade 2a). A double-lumen central venous catheter was then placed aseptically under sterile protocol into the patient's right internal jugular vein. Anesthesia was maintained throughout the surgical procedure using sevoflurane and fentanyl. The surgical procedure was uneventful; and in 90 minutes of operative time, a subtotal gastrectomy, Roux-en-Y reconstruction, and D2 lymphadenectomy were completed. Intraoperative blood loss was minimal and estimated to be 200 mL. Fluid resuscitation during the procedure included 500 mL isotonic sodium chloride solution, 1 U packed red blood cells, and 2 U of platelet concentrate. At the conclusion of surgery, the patient was stable both hematologically and hemodynamically.

The residual neuromuscular block was reversed with 2 mg/kg of sugammadex (Merck) administered before endotracheal extubation. The patient was awakened and trachea extubated without incident. He was discharged to the intensive care unit for postoperative management.

Six units of platelet concentrate were administered during the first postoperative day.

In the postoperative period, the patient's platelet count dropped over a 7-day period from $131 \times 10^3/\mu L$ to $14 \times 10^3/\mu L$. The patient's hemoglobin (9.3 g/dL) and leukocyte count ($1.50 \times 10^3/\mu L$) remained low but stable throughout his recovery. His coagulation profile had slightly improved between postoperative day 3 (PT = 20.6 seconds, INR = 1.37) and postoperative day 9 (PT = 17.4 seconds, INR = 1.13). Remarkably, the patient did not present clinical signs of hemorrhage throughout the postoperative period. Pain management was achieved via a multimodal intravenous analgesic regimen with morphine and acetaminophen. Throughout the admission, the patient was under antibiotic protection and strict aseptic precautions. He was placed in a private room under isolation, and the care staff wore dedicated protective clothing. He had no respiratory complications and was discharged from the hospital in satisfactory condition after 10 days from admission.