



# Fanconi Anaemia, What Anaesthesiologists Should Consider about

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## Abstract

Fanconi anaemia (FA) is a rare autosomal recessive inherited disease, which consists of bone marrow failure, skeletal deformities and multi organ involvement. Here, we report an emergency surgery on a FA patient with mucormycosis infection of paranasal sinuses. Since there are a few reports about FA, there is no general consensus on anaesthetic considerations so we deal with recommendations that are based on the pathology of FA. Possible difficult air way is the main concern, and low oxygen concentration and avoidance of nitrous oxide are among recommendations.

**Keywords:** Fanconi anaemia, anaesthetic consideration, emergent surgery, mucormycosis

## Introduction

Fanconi anaemia (FA) is an extremely rare (three in million) autosomal recessive disease, which involves bone marrow and all lineage of blood cells.<sup>1-3</sup> Patients may have some congenital abnormalities that might be of concern to anaesthesiologists such as airway management or patient positioning. Due to its scarcity, clinical trials about anaesthetic considerations are not available and most of the data have been obtained through case reports. Most of these reports are about optimal preparation and management for elective procedures. Here, we report a case of FA which was referred to our hospital for an emergent endoscopic sinus surgery.

## Case Presentation

A 5 year old boy from rural area of Iran was admitted to a primary care hospital due to progressive mucormycosis infection of paranasal sinuses and received antibiotic treatment, which was not effective then referred to our hospital for emergent functional endoscopic sinus surgery. He had received different medical treatments since 2 years old which were not effective. His parents were not relatives. He showed growth retardation (100 cm height and 12 kg weight) but had no obvious skeletal deformity in head, neck or limbs.

In the operating theatre, the boy was agitated with tachypnoea and tachycardia with a large necrotic oedematous lesion on nose and ulcerative lesions on tongue and oral cavity (Figure 1). Moreover, he had ecchymosis on limbs (Figure 2). In CT scan, mild cardiomegaly and mild bilateral pleural effusion at bases were reported.

Lab data showed pancytopenia (haemoglobin (Hb) 7.2 g dL<sup>-1</sup>, platelet less than 1,000  $\mu$ L<sup>-1</sup> at admission which reached 85,000  $\mu$ L<sup>-1</sup> after receiving 10 units of platelets, WBC 1,100  $\mu$ L<sup>-1</sup>), international normalisation rate 1.8'', prothrombin time 18'', partial thromboplastin time 40'', magnesium 1.5 mg dL<sup>-1</sup>, calcium 7.7 mg dL<sup>-1</sup> and potassium 5.2 mg dL<sup>-1</sup>.

Standard monitoring attached (electrocardiogram, noninvasive blood pressure, oxygen saturation on pulse oximetry (SpO<sub>2</sub>) and end tidal carbon dioxide) and difficult airway management devices prepared (videolaryngoscope, laryngeal mask airway, fiberoptic bronchoscope and paediatric bougie). His room air SpO<sub>2</sub> was 92% with RR (Respiratory Rate) 45 breath min<sup>-1</sup>, HR (Heart Rate) 161 beat min<sup>-1</sup>, BP (Blood Pressure) 110/70 mm Hg.



**Figure 1. Necrotic oedematous lesion on the nose and ulcerative lesions on the tongue and oral cavity.**



**Figure 2. Ecchymosis on the forearm.**

Although oral mucosa was ulcerative, primary evaluation of airway seemed to be satisfactory without any sign of difficult airway.

Fentanyl 25 µg injected intravenously as premedication while preoxygenating the patient with 8 L min<sup>-1</sup> of oxygen via sealed face mask. Anaesthesia was rapidly induced by using intravenous propofol (30 mg) and cisatracurium 4 mg, and direct laryngoscopy showed a good view of glottis (Cormack Lehane Score 2). A 4.5 armoured tracheal tube inserted uneventfully, secured and fixed at 12 cm after confirmation by capnography and auscultation.

Anaesthesia maintained by 1.4% of isofluran in air/O<sub>2</sub> mixture. Five minutes later, the surgeon was ready to insert the rigid sinuscope and start the surgery. Immediately after insertion, heart rate decreased to 50 beat min<sup>-1</sup> which was unresponsive to atropine. By reaching 30 beat min<sup>-1</sup>, chest

compression and intravenous epinephrine (100 µg) applied and in less than 2 minutes HR returned to 120 beat min<sup>-1</sup> and surgery continued and a specimen for biopsy obtained and patient transferred to ICU under mechanical ventilation. Unfortunately, patient died the day after due to massive infection, unresponsiveness to antibiotics and coagulopathy.

## Discussion

FA could involve anaesthesiologists since they might need anaesthesia for elective procedures such as Hickman catheter insertion,<sup>2</sup> dental treatment,<sup>3</sup> splenectomy or correction of skeletal deformities.<sup>1</sup> Due to its rarity (1 in 340,000), trials are not available in literature and most of the data come from case reports about elective surgeries. Anaesthetic considerations and case reports about emergent cases are even scarcer.

FA first described in 1927 by Swiss paediatrician Guido Fanconi, which consist of bone marrow failure and high propensity to develop acute myelogenous leukaemia (700 times higher than normal population) and different types of solid tumours (malignancies 50 times higher than general population) with a median survival of 20 years.<sup>1,2,4</sup> Researchers have found 22 different gene mutations among these patients.<sup>4</sup> Its rarity made it a challenge for us to manage an

### Main Points

- Anaesthesiologists should be concerned about difficult airway in FA.
- High inspired oxygen concentration should be avoided in FA.
- Nitrous oxide (and any bone marrow suppressing medication) is better to be avoided in FA.

emergent endoscopic sinus surgery for a patient with FA since we could not find sufficient data concerning its management for emergent situations.

Clinical findings could be divided in two main categories: first, bone marrow failure leading to pancytopenia and second, congenital deformities including short stature, poly or oligodactyly, mental retardation, skin hyperpigmentation and hearing loss.<sup>1,3</sup> However, one-third of patients may not have congenital deformities.<sup>2</sup> Chemotherapy and stem cell transplant are the corner stone of treatment.<sup>3</sup> The main causes of death are bone marrow failure, leukaemia and solid tumours.<sup>5,6</sup>

Since skeletal deformities are common among FA patients, difficult airway is anticipated in at least 25% of patients.<sup>3</sup>

Pancytopenia as the main finding is another threat to the patient candidate for surgery as the platelet count more than  $50,000 \mu\text{L}^{-1}$  is the least acceptable level<sup>3</sup> whilst FA patients usually have platelet counts much less, even as low as  $1,000 \mu\text{L}^{-1}$ . Low Hb level should be corrected. Leukocytopenia may increase the risk of infection, ending to increased mortality. Cardiovascular, renal and hepatobiliary system involvement should be sought since they may aggravate the situation and be a challenge for anaesthesiologist.<sup>2</sup> Fortunately, our patients had no skeletal deformities which may indicate difficult airway and only suffered pancytopenia.

Anaesthesia technique and choice of medications for FA patients may have repercussions, for example, due to bone marrow failure some authorities believe that nitrous oxide should not be administered because of its potential suppression on bone marrow<sup>3</sup>; however, others believe that it is safe to use it for surgeries less than 6 hours. Although a high oxygen concentration could provide luxury oxygenation to end organs, it could be dangerous as well since surges reactive oxygen species and aggravates apoptosis.<sup>2</sup> Knowing these points, we avoided nitrous oxide and used air-O<sub>2</sub> mixture.

It is recommended to avoid anaesthetics that have adverse hepatorenal effects such as Halothane, long acting muscle relaxants, nonsteroidal anti-inflammatory drugs (NSAIDs) and long acting opioids such as morphine and meperidine.<sup>2</sup> Antithyroid drugs, antiepileptic drugs and NSAIDs are recommended to be avoided secondary to the possibility of acquired aplastic anaemia.<sup>2</sup> In the light of these points, we obtained muscle relaxation by using cisatracurium and did not prescribe any long acting opioid.

Other comorbidities such as hypothyroidism (36%) and hyperinsulinemia (72%) may accompany FA, which should be considered by anaesthesiologist in charge of patient man-

agement and blood sugar should be monitored especially in patients under steroid treatment.<sup>2</sup>

Our patient suffered a transient bradycardia which could be due to autonomic system insufficiency or inappropriate depth of anaesthesia. We recommend that diluted epinephrine ( $10 \mu\text{g mL}^{-1}$ ) should be at hand.

## Conclusion

Anaesthesiologists should be familiar with FA and possible challenges should be well thought in advance. All things together, avoidance of high inspired oxygen concentration, nitrous oxide, long acting muscle relaxants and opioids is the general recommendation; moreover, NSAIDs, antithyroid medications and antiepileptics are better to be avoided due to the likelihood of inducing acquired aplastic anaemia. Platelet transfusion may be needed to increase the platelet count to at least  $50,000 \mu\text{L}^{-1}$  before surgery.

**Informed Consent:** Written informed consent was obtained from the guardian (parents) of patient who participated in this case.

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