Case Report

# Maffucci syndrome and anaesthesia: Case report

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**Access this article online**

Website: www.ijaweb.org

DOI: 10.4103/ija.IJA_779_18

Quick response code

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## ABSTRACT

Maffucci syndrome is an extremely rare, sporadic, and nonhereditary disease characterized by enchondromatosis, bony abnormalities, and vascular malformations. We report a successful anaesthetic management of an adult patient with Maffucci syndrome scheduled for surgical excision of parathyroid adenoma, who had numerous enchondromas, haemangiomas, and skeletal deformities involving different parts of the body and posing significant challenge during positioning, securing intravenous access, and insertion of endotracheal tube. Awareness about this syndrome is of paramount importance because similar patients may be encountered with increasing frequency for incidental or corrective surgeries.

**Key words:** Deformities, enchondromas, haemangiomas, maffucci

## INTRODUCTION

First described by Angelo Maffucci in 1881, Maffucci syndrome, a subtype of enchondromatosis is an exceedingly rare, sporadic, and nonhereditary disease characterized by multiple enchondromas, haemangiomas, and lymphangiomas.[1] Although the etiology is unclear, heterozygous somatic mutations in isocitrate dehydrogenase 1 and 2 (*IDH1*/*IDH2*) genes are thought to be associated with Maffucci syndrome.[2] Long bones and phalanges are commonly affected, and pathological fractures are more common in the affected bones. We report a successful anaesthetic management of a patient with Maffucci syndrome posted for surgical excision of parathyroid adenoma.

## CASE REPORT

A 22-year-old female patient (height, 1.31 m; weight, 39 kg), a known case of Maffucci syndrome, presented with complaints of bone pain, fatigue, and weakness, diagnosed with right inferior parathyroid adenoma was scheduled for surgical excision of tumor. Written informed consent was taken from the patient before surgery. On physical examination, the patient had multiple enchondromas and haemangiomas involving right upper limb, right lower limb, and right anterior chest wall [Figure 1]. Also, deformities of the right upper limb, anterior chest, and bowing of lower limbs with limb length discrepancy were observed. Airway examination revealed a Mallampati class II airway, full range of motion of her neck, adequate mouth opening, and a thyromental distance of three finger breadths. Radiographs showed multiple well-defined radiolucent lesions corresponding to enchondromas on the phalanges of right hand, right foot, right anterior chest wall, and skeletal deformities of lower limbs, right upper limb, pelvis and spine [Figure 2]. X-ray spine revealed thoraco-lumbar scoliosis with cobb's angle of 45°. Relevant laboratory investigations along with pulmonary function tests, echocardiography, and computed tomography (CT) head were within normal limits. Fiber-optic bronchoscopy and intubation was planned because of possibility of accidental trauma to the undetected intraoral haemangiomas. Difficult airway cart was kept ready. Patient was kept nil per oral from midnight and premedicated with tab alprazolam



**How to cite this article:** Naik BN, Sujith J, Kajal K. Maffucci syndrome and anaesthesia: Case report. Indian J Anaesth 2019;63:400-2.

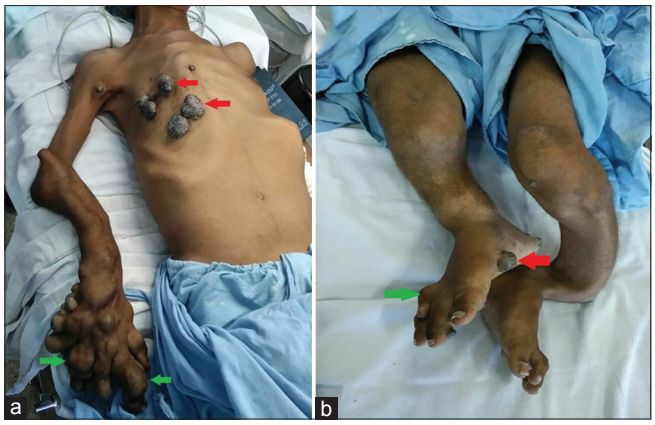


**Figure 1:** Image of the patient showing enchondromas (*green arrows*), haemangiomas (*red arrows*), and skeletal deformity of (a) right upper limb and anterior chest wall and (b) lower limbs

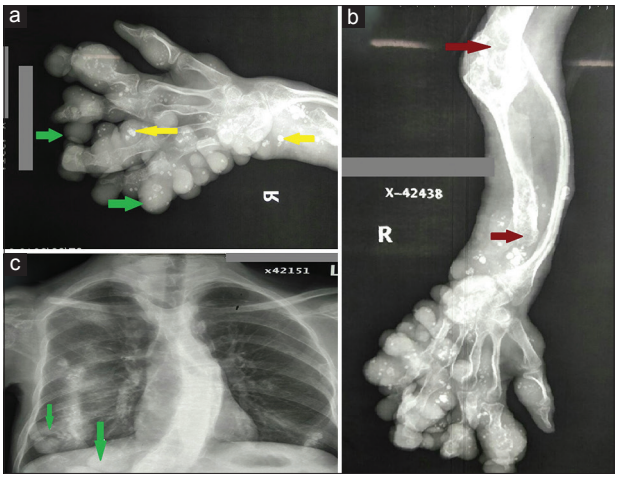


**Figure 2:** Radiograph showing (a) multiple enchondromas (*green arrows*), phleboliths (*yellow arrows*) and deformities of phalanges and metacarpals of right hand (b) deformity and radiolucency (*brown arrows*) of right radius and ulna (c) enchondromas (*green arrows*) over lower zone of right side of chest

0.25 mg and tab ranitidine 150 mg night before surgery and on the day of surgery in the morning. Upon arrival in the operating room, patient positioning was done carefully by padding placed under the head, arms, and heels with a pillow placed under the knees, standard ASA monitors were placed for monitoring, and an 18G intravenous line was secured in left upper limb using portable infrared vein finder device. The patient was preloaded with 10 ml/kg of ringer lactate before induction of anaesthesia, and maintenance fluid of 80 ml/h was given intraoperatively. Anaesthesia was induced with intravenous (IV) morphine 0.1 mg/kg, propofol 2 mg/kg, and after ensuring smooth mask ventilation, IV vecuronium 0.1 mg/kg was administered and fiber-optic endotracheal intubation was performed. During fiber-optic bronchoscopy, though we found multiple small nodular projections in the posterior wall of trachea, exposure of the larynx and trachea was not difficult. However, these small projections did not impede intubation, and a 7.0 mm internal diameter cuffed endotracheal tube was railroaded over the fiber-optic bronchoscope without resistance. Anaesthesia was maintained with isoflurane [1.2 minimum alveolar concentration (MAC)] in a mixture of oxygen and nitrous oxide, along with intermittent boluses of IV vecuronium. Surgery lasted for 2 h and intraoperative course was uneventful. At the end of the surgery the patient was extubated and shifted to recovery room by transferring patient gently from the operation table to the shifting trolley having soft foam mattress by using sliding board. Postoperative course was uneventful.

## DISCUSSION

Maffucci syndrome is characterized by enchondromatosis, bony abnormalities, haemangiomas, and phlebolithiasis. It is a very rare disease with less than 225 cases reported worldwide. Clinical problems caused by the cartilaginous lesions include skeletal deformities, such as short stature, scoliosis, bowing of limbs, and limb-length discrepancy, as were noted in our patient. Complications include pathological fractures and potential risk for malignant change.[3] Radiographic images of the patient, as shown in Figure 2, display well-demarcated radiolucent skeletal lesions and remodeling of the affected bone with predominant thinning of the cortex, which demands a close attention to careful positioning for surgery and during shifting out to recovery room, such as gently shifting onto operation table, using cotton pads underneath pressure areas and bony joints, using patient sliding boards, and soft mattress trolleys for shifting off to postoperative recovery room. Our patient had difficult intravenous access due to numerous enchondromas and haemangiomas involving hand and feet along with multiple skeletal deformities [Figure 1]. Obtaining intravenous access can become further more difficult particularly when they present for repeated surgeries. Intravenous cannula should be secured by using infrared vein finder devices or under ultrasound guidance.

It has also been reported that patients with Maffucci syndrome have vascular malformations in tongue, buccal mucosa, trachea, lips, gingiva, and palate.[4] One of the important anaesthetic concerns implicated in this syndrome is to check for the presence of these haemangiomas in the upper airway during preanaesthesia checkup. Of primary concern to

anaesthesia providers is the potential for rupture of unseen haemangiomas present in the trachea during endotracheal intubation. Fiber-optic bronchoscopy and intubation must be preferred in these patients and contact of the bronchoscope or the endotracheal tube with haemangiomas must be avoided to prevent rupturing of any haemangiomas. Awake intubation should preferably be avoided. Moreover, we were able to successfully accomplish endotracheal intubation using fiber-optic bronchoscope, given the above-mentioned problems. Preoperative CT scan or MRI is a good modality to visualize and provide accurate information about the location and extension of the lesions.[5] In addition, the appropriate equipment for dealing with the difficult airway should be kept ready.

Tracheal chondromas and chondrosarcomas arising from trachea-cartilaginous rings may lead to obstructive symptoms depending on whether the tumor grows intraluminal or extraluminal. Frequent debulking of cartilaginous tumor discovered in larynx or trachea might be required to maintain patency of the airway. However, complications such as tracheomalacia and tracheal stenosis might occur.[5]

## CONCLUSION

The main anaesthetic concerns in patient with Maffucci syndrome are difficult intravenous access, risk of pathological fractures, and accidental trauma to the undetected intraoral haemangiomas. Therefore, it necessitates a proper preoperative assessment and careful anaesthetic techniques regarding airway management, securing intravenous access, and gentle positioning to avoid potential complications.

### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

### Financial support and sponsorship

Nil.

### Conflicts of interest

There are no conflicts of interest.

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