Journal of Clinical Anesthesia (2015) **27**, 419–422

ELSEVIER

Journal of Clinical Anesthesia

Case Report

Proteus syndrome: what the anesthetist should know☆,☆☆,★,★★

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Received 22 August 2014; revised 20 March 2015; accepted 30 March 2015

Keywords:
Hamartomatous;
Hemihypertrophy;
Macrodactyl;
Proteus syndrome

Abstract Proteus syndrome (PS), a rare hamartomatous disorder, manifests itself in asymmetric and disproportionate overgrowth of multiple body tissues. Because of complexity of the disorder, the anesthetic problems encountered during patients' perioperative management are very varied. We discuss the case of a 14-year-old adolescent boy diagnosed with PS who underwent corrective osteotomy of right knee joint under subarachnoid block. The salient points the anesthetists need to be aware of while caring for patients with PS are highlighted.

1. Introduction

Proteus syndrome (PS) is a rare hamartomatous disorder manifesting itself in asymmetric overgrowth of multiple body tissues. Its global incidence is estimated to be less than 1 in a million, with very few cases reported from the Indian subcontinent. The syndrome, first described by Cohen and Hayden in 1979, was named by Wiedemann in 1983 after the Greek sea god "Proteus" who could transform into any shape. Earlier, it was hypothesized that the condition is caused by postzygotic mutation of somatic genes that is lethal in its nonmosaic state. Recently, researchers have identified point mutation in the AKT1 gene as the cause of the unregulated growth of cells involving all 3 germ layers in PS [1].

The syndrome is sporadic, is nonfamilial, and has a progressive course. The abnormal growth becomes apparent in the first few years after birth, accelerating rapidly during childhood but tending to slow down after adolescence. The diagnosis of PS is a clinical one, its most frequent and striking feature being disproportionate skeletal overgrowth leading to abnormalities such as asymmetric limb overgrowth, vertebral anomalies, macrodactyl, and limb length discrepancy. Other features of PS include asymmetric muscle development, lipomas or lipoatrophy, hyperpigmented skin lesions, epidermal nevi, cerebriform connective tissue nevi, vascular malformations, tumors of ovary or parotid glands, and visceral involvement such as cystic lung disease [2–4]. Resulting complications of deep venous thrombosis (DVT) and chest infections can prove fatal [5].

Proteus syndrome is extremely rare; however, affected patients often need orthopedic or reconstructive plastic surgeries for physical rehabilitation. Patients may also require genitourinary, gastrointestinal, otolaryngological, or tumor excision surgeries for treatment of complications of PS

☆ Funding source: none.
☆☆ Presentation at meetings: none.
★ Conflict of interest: none.
★★ Authorship: The author has contributed in the preparation of the manuscript to qualify for authorship rights.
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http://dx.doi.org/10.1016/j.jclinane.2015.03.038
0952-8180/

[6]. Because of the complexity of the disorder, the anesthetic problems encountered during patients' perioperative management are very varied. We thought it relevant to report this case so as to highlight the salient points the anesthetists need to be aware of while caring for patients with PS.

2. Case report

A 14-year-old boy diagnosed with PS was referred to our institute for corrective osteotomy of right knee joint. The boy had difficulty in walking due to the enlarged right knee joint. On physical examination, we noted hypertrophy of right half of the body, enlarged hands and feet, syndactyl of digits of right foot, facial asymmetry, and hyperpigmented skin lesions over the body (Figs. 1 and 2). The patient's skeletal scan showed bone and soft tissue hypertrophy of right side of the body, mild lumbar scoliosis, right hip joint deformity, and bilateral genu valgum (Fig. 3). Results of magnetic resonance imaging scans of the patient's chest and abdomen were normal. His pulse was irregular; the electrocardiograph showed bigeminy with occasional ectopics, although echocardiography was normal.

Fig. 1 Macrodactyl of hands and feet.

The airway evaluation was normal except for right-side maxillary overgrowth, protruding teeth, and slightly high-arched palate.

We conducted the surgery using subarachnoid block. After securing intravenous access and connecting all monitors (electrocardiograph, noninvasive blood pressure, pulse oximetry), 2 mL of 0.5% hyperbaric bupivacaine with 20 μg fentanyl was injected intrathecally through 25-gauge spinal needle in L3-L4 interspace with the patient in sitting position. Even though the patient had mild lumbar scoliosis, the block could be performed easily. The surgery lasted for 2 hours and was completed uneventfully. Postoperatively, the patient was given DVT prophylaxis with subcutaneous low–molecular weight heparin that was continued until he became ambulatory. He was counseled for regular medical follow-up with medicine and orthopedic departments after discharge.

3. Discussion

Overgrowth of soft tissues of the airway and asymmetric hyperplasia of tonsils and adenoids occur commonly in PS, causing airway obstruction and difficult intubation [5]. A previous case report has described enlarged epiglottis causing difficult laryngoscopy and necessitating use of McCoy laryngoscope [7]. In another case, a boy with torticollis was intubated using fiberoptic scope [8]. Hyperostosis and enlarged cervical vertebrae may cause external compression of the airway. Craniofacial disfigurement from abnormal skull and facial growth can make airway management challenging [4]. Hence, there is a need for thorough evaluation of the airway in such cases.

Spine deformities including asymmetric vertebral bodies, kyphoscoliosis, and scoliosis are frequently seen in PS; these add to difficulties in giving central neuraxial blocks [3,4]. Neurological compromise of spinal cord due to infiltration of the spinal canal by angiolipomatous mass or canal stenosis from vertebral hypertrophy is also reported; in these patients, central neuraxial block is best avoided [9]. Skeletal overgrowth and restricted joint mobility add to difficulties in positioning patients on operating table. As vascular malformations in PS predispose patients to DVT and pulmonary embolism, they are particularly at risk during the period of convalescence after surgery; therefore, DVT prophylaxis should be considered at this time [10].

Although visceral involvement is less common in PS, it is recommended that all the patients be screened for it with magnetic resonance imaging scan of the abdomen and chest. Hamartomas and lipomas of the gut can lead to intussusceptions, acute intestinal obstruction, gastric outlet obstruction, and rectal prolapse [6]. There is also a risk of increased bleeding during gastrointestinal surgeries due to intestinal vascular malformations. Primary involvement of lung causes cystic emphysematous lung disease, whereas rapidly progressive scoliosis can cause secondary restrictive lung

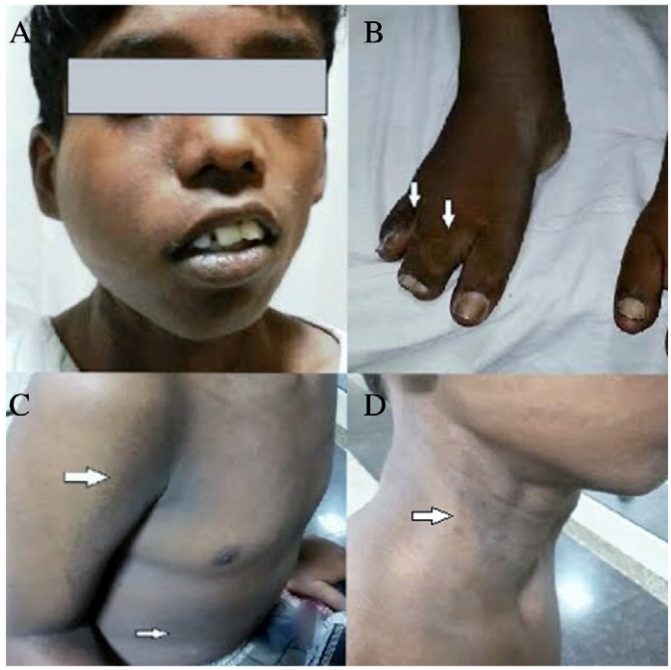

Fig. 2 A. Facial asymmetry; B. Syndactyl of digits of right feet; C and D. Hyperpigmented skin lesions over arm, abdomen and neck.

disease, predisposing patients to repeated pneumonias and lung atelectasis [11]. As poor respiratory reserves may lead to perioperative respiratory failure, aggressive chest physiotherapy is needed for clearing the respiratory tract secretions and preventing the complication [12]. Hemimegalocephaly and white matter abnormalities of brain causing developmental delay or seizure disorder are also seen in PS [13]. Although cardiac involvement is rare, hypertrophic cardiac rhabdomyomas and conduction abnormalities of heart have been reported [14]. Our patient too had a conduction block, but no structural abnormality of the myocardium was detected on echocardiography.

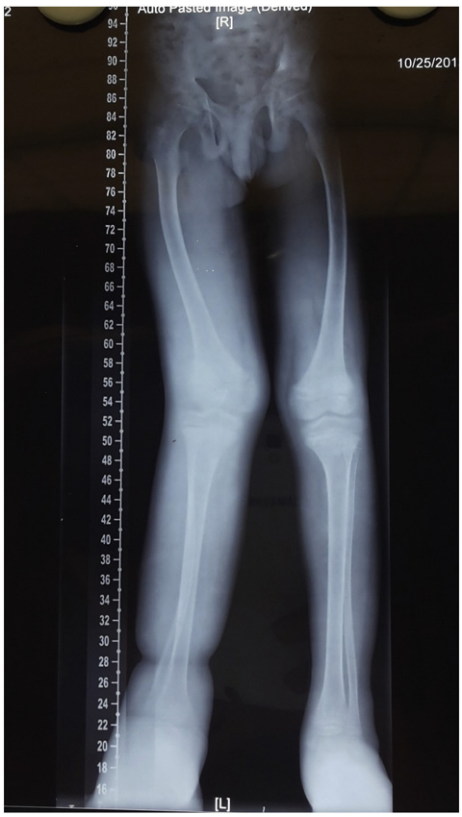

Fig. 3 Skeletal scan showing hemihypertrophy of right side of body.

For orthopedic and reconstructive surgery in patients with PS, use of regional anesthesia with central or peripheral neuraxial block as opposed to general anesthesia has the advantage of avoiding possible problems of difficult airway and respiratory complications. Besides providing excellent analgesia and facilitating patients' early movement, regional anesthesia can help reduce the incidence of DVT. However, we need to be cautious about technical difficulties that could arise in performing the block due to anatomical abnormalities. Besides, timing of perioperative anticoagulants should be such as to prevent hemorrhagic complications of regional blocks.

In conclusion, while planning anesthesia for a patient with PS, a careful evaluation comprising history taking, physical examination, and relevant imaging studies is important. The musculoskeletal and soft tissue overgrowth can create problems for the anesthetist in airway management and regional anesthesia procedures, besides hampering proper positioning of the patient on the operating table. Moreover, complications may arise from visceral involvement of other organ systems such as cystic degeneration of lung compromising respiratory functions. Also, perioperative DVT prophylaxis with subcutaneous low–molecular weight heparin should be considered in view of the high reported incidence of mortality from DVT and pulmonary embolism in these patients.

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