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Case Report Open Access

Respiratory and Cranial Complications during Anaesthesia in Pfeiffer Syndrome

Ela Erten[1], Nedim Çekmen[2*], Ferruh Bilgin[1] and Mehmet Emin Orhan[1]

[1]*Gülhane Medical School, Department of Anesthesiology and Reanimation, Ankara, Turkey*
[2]*Guven Hospital, Department of Anesthesiology and Reanimation, Ankara, Turkey*

Abstract

Pfeiffer syndrome (PS) is rarely encountered, even at major craniofacial centers. Published reports indicate high mortality rates (25-85%) for severely affected subtypes. PS is characterized by bilateral coronal craniosynostosis, midface hypoplasia, beaked nasal tip, broad and medially deviated thumbs and great toes. We present a case of a 12-months-old male infant with PS type 2 who underwent a craniosynostosis and advancing a supraorbital bar operations during general anesthesia. For simple procedures, a safe anesthetic plan can be formulated if the anatomic factors affecting the airway are carefully considered. More extensive and prolonged patient diagnosed to have PS require more monitoring and include all the problems associated with difficult intubation, hazardous airway management, massive blood loss and fluid shift, shunt-dependent hydrocephalus, and long anesthetic times. This case is presented since anaesthesiologists should be aware of the high incidence of respiratory and cranial complications in PS.

Keywords: Pfeiffer syndrome; Difficult intubation; Respiratory and cranial complications; Anaesthesia

Introduction

Pfeiffer syndrome (PS) is an autosomal dominant condition comprising bilateral coronal craniosynostosis, midface hypoplasia with a beaked nasal tip, and broad and medially deviated thumbs and great toes [1]. It is a clinically variable disorder and has been divided into three subtypes by Cohen in 1993 [2]. Originally, it was described in eight persons from three generations in a pedigree consistent with an autosomal dominant transmission. Since then, several reports have documented its high clinical and genetic heterogeneity [3]. The condition is usually detected in the newborn period or later, and very few prenatal ultrasound diagnoses have been reported. Type 1 is called classical PS, and its findings are moderate. Type 1 frequently shows normal neurological and mental development, and has better surveillance. Type 2 has an appearance of cloverleaf (trefoil) skull, and central nervous system is frequently involved. Type 3 differs with absence of cloverleaf skull from Type 2. Type 2 and 3 has a bad prognosis due to severe neurological complications and various visceral anomalies, and frequently death is seen at early ages [4-7]. These patients are candidates of difficult intubation because of existence of maxillary hypoplasia and associated narrow larynx and pharynx together with possibility of tracheal anomalies [8].

Clinical experience with anaesthesia for a series of patients with PS has not been reported previously. We present a case of a 12-months-old male infant with PS Type 2 who underwent a craniosynostosis and advancing a supraorbital bar operations during general anesthesia.

Case Report

Our case is a baby of 12 months old and 8 kg weight born from healthy parents who has had the diagnosis of PS Type 2 (Figure 1). The case had clover skull and craniosynostosis. A collective operation was planned by plastic and reconstructive surgery and brain surgery for the purpose of frontal craniectomy and advancing a supraorbital bar on the case having severe exophthalmia, choanal atresia, midfacial hypoplasia and low ear anomaly. It was learnt from the preoperative history of the patient that the patient has difficulty in breathing especially during sleep. On the physical examination for the patient whose malampathy score was evaluated to be III, narrow oral (mouth) structure, maxillary and choanal hypoplasia were observed, and a fiber optic bronchoscope and laryngeal mask (LMAs) with various sizes were prepared by considering a difficult intubation. Besides, surgery team was asked to be prepared as to open a tracheostomy in an emergency case. The patient was taken into operation hall and sevoflurane induction was started after monitorization with ECG, pulse oxymeter and noninvasive blood pressure, and peripheral vascular route was opened with 24 G angiographic catheter, fentanyl 10 µg was administered via i.v. route, and then it was observed that the patient is ventilated with mask,

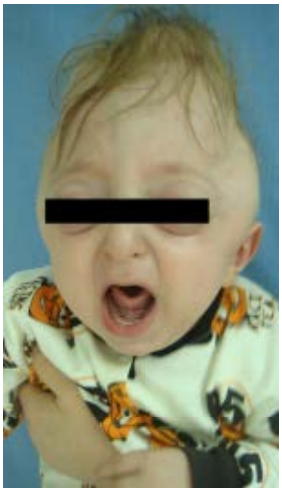

Figure 1: diagnosis of PS Type 2.

***Corresponding author:** Nedim Çekmen, İlko evleri 2786.sok No:12 Çayyolu Mah, Yenimahalle, Ankara, Turkey, Tel: +9005337133150; E-mail: nedimcekmen@yahoo.com

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and muscular relaxation was obtained with 0.5 $mg.kg^{-1}$ atracurium. A stenosis was determined at the subglottic region, while the patient, whose *Cormack-Lehane* score was considered as 2 by direct laryngoscopy, was being intubated with a tube having cuff number 3. Localization of tube was confirmed by auscultation and capnography and fixed. Maintenance of anaesthesia was continued with 50% nitrous oxide, 50% oxygen, 2% sevoflurane and 0.1 $mg.kg^{-1}$ atracurium I.V. as required. End-tidal CO_2 and temperature monitorizations were made on the patient. Antibiotic pomade was applied onto eye of the patient who experienced severe exophthalmia during all these procedures, and eye was closed and protected with cotton as to prevent compression onto it. Right subclavian central catheter was inserted to the patient, CVP monitorization and right radial artery cannulation were performed and arterial blood pressure monitorization was made, and hourly urine output was monitored with a urinary catheter placed. Patient's preoperative Hb value has been 10.1 $g.dl^{-1}$, and Hb value during postoperative period was measured to be 9.5 on the patient, who was administered total 130 cc erythrocyte suspension and 110 cc FFP (Fresh Frozen Plasma) during the operation lasting for about 11 hours. 1/3 mixed fluid and 0.9% NaCl and ringer lactate infusions were given to the patient during intraoperative and postoperative period in a balanced way by monitoring blood electrolytes, CVP value and urine output. Ca^{2+} replacement was made for the patient when Ca^{2+} value measured became 0.7 mmol/L at postoperative 5th hour, whose intraoperative and postoperative blood gas analyses and electrolytes monitored closely. The patient was hemodynamically stable maintaining a systolic blood pressure of 90-100 mmHg. After the completion of surgery, the patient was assessed and on ensuring that the patient was fully awake trachea was extubated. After the operation lasting approximately 11 hours, the patient was intubated and taken to postanaesthesia care ünit (PACU) following anaesthesia, who had adequate respiration following spontaneous respiration resumed and effect of residual myorelaxant was antagonized with neostigmine 50 $\mu g.kg^{-1}$ together with atropine 20 $\mu g.kg^{-1}$. The case, who had respiratory distress on follow ups and whose P_{CO2} value of 66 mmHg was determined on blood gas analysis, was re-intubated 1 hour later, and the patient was followed up by mechanical ventilator firstly in BIPAP mode and afterwards in CPAP mode after respiration became comfortable and P_{CO2} returned to normal limits in blood gas analysis. The patient was taken into T piece 6 hours after intubation, and was extubated without problem 1 hour later. 70 cc erythrocyte suspension and 50 cc FFP were given to the patient with Hb follow ups, who had oozing hemorrhage during postoperative period. Control cranial tomography was taken with a suspicion of raised intracranial pressure when bradycardia attacks occurred with crying and respiration became distressed during follow up in PACU. The case was taken into ventriculoperitoneal shunt operation, on whom a severe hydrocephalus developed and herniation risk appeared on cranial tomography (Figure 2). The case, who became more distressed due to airway edema in the operation room, was intubated with a tube no. 2,5 without cuff. On the chest x-ray taken because respiratory sounds could not be taken at the base of right lung and a pleural effusion was seen on the chest x-ray, the patient who was extubated without problem after operation was inserted a thorax tube by pediatric surgery, and it was observed on control x-rays that effusion was removed. It was thought that pleural effusion developed may have occurred during collection of bone and cartilage rib graft. The patient was handed over to the clinic without problem after followed in intensive care unit in an extubated state for 2 days during postoperative period. The patient was discharged to home without problem, who was followed up in the clinic of plastic and reconstructive surgery.

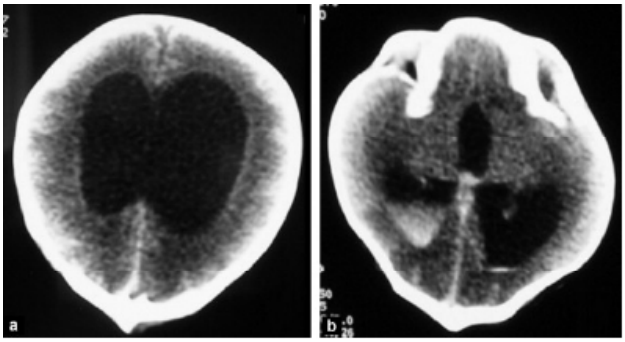

Figure 2: hydrocephalus developed and herniation risk appeared on cranial tomography

Discussion

Although definite incidence of PS isn't known, it is estimated to be 1 out of 100.000 live birth in western populations and rare in Asian population [1,3]. Type 2 PS is a condition associated with premature fusion of multiple cranial sutures, cloverleaf skull (kleeblatschädel deformity), prominent ptosis, thumb and first toe abnormalities, variable syndactyly, and mutated genes for type 1 or 2 fibroblast growth factor receptor [3,5]. Midfacial hypoplasia and hand-foot abnormalities, hydrocephalus, low ear, external auditory canal stenosis or atresia and sometimes anomalies such as pelvic kidney, hydronephrosis and hypoplastic urinary bladder as well as cloverleaf skull structure due to early closure of coronal sutures in these patients [5,6]. These children generally do poorly because of significant often severe neurologic and cognitive defects, and many die very young [7]. Although these children have bad prognosis, good results have been reported in some cases with surgical and medical interventions performed at early stage [7,8]. Our case had clinical findings consistent with PS Type 2. In general, purpose of surgeries can be summarized as: correcting shape of skull and providing decompression of brain, elongating and expanding ocular bones as to eyeballs located inside and eyelids close, also opening nasopharyngeal airway by advancing naso-maxillary-zygomatic complex [7,8]. Having patients with PS midfacial hypoplasias, choanal atresias, narrow larynx and pharynx anatomies may cause these patients to experience respiratory distress. Besides, tracheal anomalies have been reported in some cases.[9] When all of them taken into consideration, airway of the patient should be evaluated thoroughly before anesthesia, and it should be inquired if a patient has any respiratory distress by obtaining medical history, especially during sleep. Early rigid bronchoscopy is important in these patients when there are problems with the airway, as they have a relatively high incidence of airway anomalies. After assessment of all these specific needs, the anesthesiologist will be presented with a difficult but not insurmountable risk [9,10]. After we have taken required precautions for the aspect of difficult intubation with our physical examination and detailed medical history preoperatively, we performed induction of anesthesia.

An awake extubation is generally indicated in the difficult airway. Consider the possibility of airway oedema, especially after repeated attempts at intubation. A plan for a failed extubation should also be formulated. It should be remembered that patients with PS may enter into respiratory distress on smallest amount of edema in respiratory tract due to narrow airway, and they should be closely monitored during intraoperative and postoperative period. A thorough assessment of the individual is crucial for developing a safe anesthetic plan for

such patients [10]. Our case also required to be re-intubated during postoperative period for the patient entered into respiratory distress. We administered anti-edema treatment to the patient in postoperative follow up. Several disadvantages of early or delay extubation must be kept in mind absolutely in this cases. Ocular propitosis is frequently seen due to shallow orbital cavities in cases with PS. Care should be given for protecting eyes and not to cause compression on them during procedures performed [7]. We also spread antibiotic pomade to our patient's eyes and closed them with cotton as to prevent compression. Shunt-dependent hydrocephalus is predominantly associated with PS. The pathogenesis of progressive hydrocephalus remains somewhat obscure, a hypoplastic posterior fossa and a venous outlet occlusion at the skull base being the main causative factors discussed in literature. Clinical evaluation is mainly aimed at identifying progressive hydrocephalus, but diagnosis is hampered by the fact that classical clinical signs may be absent, and that ventricular dilatation will often become evident only after decompressive cranial surgery. Moreover, mild ventriculomegaly may in some cases coexist with intracranial hypertension from craniostenosis [8,11]. We report a infant with type 2 PS, who was not noted to have any hydrocephalus on initial brain imaging done at 12 months. Control cranial tomography (CT) was taken with a suspicion of raised intracranial pressure when bradycardia attacks occurred with crying and respiration became distressed during follow up in PACU. The case was taken into ventriculoperitoneal shunt operation, on whom a severe hydrocephalus developed and herniation risk appeared on CT. Because hydrocephalus may be present in these children [11], care should be given for not increasing intracranial pressure, especially at the time of anesthesia induction. Therefore, careful monitoring of intracranial pressure and ventricular size in the pre-operative and postoperative period is a diagnostic mainstay. Craniosynostosis is a disorder with genetic origin occurring as a result of early closure of skull bones, which is seen in 1 of 2000 live births [6]. The earlier craniosinostosis correction operations are made, the better the results are. Although such management should be the rule for PS types 2 and 3, it needs to be remembered that normal outcome is not the rule.[7] Favorable outcomes in children with types 2 and 3 PS were documented by Robin et al. [7]. These cases illustrate that while children with PS types 2 and 3 have an increased risk for neurodevelopmental difficulties, a favorable outcome can be achieved in some cases with aggressive medical and surgical management [7,8]. However, because large incision on the scalp, which is rich in vascular aspect in infants, and bone resection would lead to greater amount of blood loss, these are dangerous operations [8,12]. Volume and blood replacement should be made carefully in these operations, and patients should be closely monitored for bleeding diathesis, heat and electrolyte balance. Hb values of patients and volume balance should be closely followed up, and blood and fluid replacement shouldn't be neglected [12]. We monitored Hb values, urine outputs and CVP values closely in our cases during intraoperative and postoperative period, and performed proper fluid replacement.

Conclusions

Consequently, patient diagnosed to have PS require more monitoring and include all the problems associated with difficult intubation, hazardous airway management, massive blood loss and fluid shift, shunt-dependent hydrocephalus, and long anesthetic times. Syndromes with craniofacial anomalies, such as Pfeiffer's, are very rare and can be a challenge to anesthesiologists. These can be managed successfully with meticulous planning and execution. Especially, the pre-operative X-ray emage of the airway may help for anaesthesiologists to understand the hazardous airway management. Anaesthetists should be aware of this high incidence of respiratory and cranial complications in PS. A multidisciplinary care is required for perioperative management of these patients.

Conflict of Interest and Funding

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