

Anaesthetic Challenges and Difficult Airway Management in Noonan Syndrome

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Abstract

Noonan's syndrome was first described by Noonan and Ehmke in 1963. These patients have facial, cardiovascular, musculoskeletal and developmental abnormalities which may pose problems to the anaesthetist during surgery. It is a rare clinical entity representing the phenotype of Turner's syndrome with normal chromosome studies. The most common congenital cardiac lesion is pulmonary stenosis, either alone or in combination with a septal defect, which is usually atrial. The potential anaesthetic problems presented by a patient with Noonan's syndrome relate to impairment of cardiopulmonary function, the possibility of a difficult airway and the problem of technical difficulty with regional anaesthesia. We reported a case of 21 years old male, weighing 30 kg, who was diagnosed clinically as a case of Noonan syndrome and had undergone balloon pulmonary valvotomy in past. Patient had severe kyphoscoliosis. This rare case report represents to the successful difficult airway and anaesthetic management due to existence of kyphoscoliosis. Problem with airway management may be fatal for patients.

Keywords: Noonan syndrome; Kyphoscoliosis; Difficult airway management

Introduction

Noonan syndrome is a genetic disorder that is typically evident at birth (congenital). The disorder is characterized by a wide spectrum of symptoms and physical features that vary greatly in range and severity [1,2]. In many affected individuals, associated abnormalities include a distinctive facial appearance; a broad or webbed neck; a low posterior hairline; a typical chest deformity and short stature. Characteristic features of the head and facial (craniofacial) area may include widely set eyes (ocular hypertelorism); skin folds that may cover the eyes' inner corners (epicanthal folds); drooping of the upper eyelids (ptosis); a small jaw (micrognathia); a depressed nasal root; a short nose with broad base; and low-set, posteriorly rotated ears (pinnae). Distinctive skeletal malformations are also typically present, such as abnormalities of the breastbone (sternum), curvature of the spine (kyphosis and/or scoliosis), and outward deviation of the elbows (cubitus valgus).

Many infants with Noonan syndrome also have heart (cardiac) defects, such as obstruction of proper blood flow from the lower right chamber of the heart to the lungs (pulmonary valvular stenosis) and thickening of the ventricular heart muscle (hypertrophic cardiomyopathy). Additional abnormalities may include malformations of certain blood and lymph vessels, blood clotting and platelet deficiencies, learning difficulties or mild intellectual disability, failure of the testes to descend into the scrotum (cryptorchidism) by the first year of life in affected males, and/or other symptoms and findings. Noonan syndrome is an autosomal dominant genetic disorder caused by abnormalities (mutations) in more than eight genes. The five most commonly involved genes are: PTPN11 (50%), SOS1 (10-13%), RAF1 (5%), RIT1 (5%), and KRAS (less than 5%). Fewer individuals have a mutation in NRAS, BRAF, MEK2, RRAS, RASA2, A2ML1, SOS2, and LZTR1. The potential anaesthetic

problems with Noonan's syndrome may be due the possibility of a difficult airway and musculoskeletal abnormalities. In this rare case report the preoperative evaluation and anaesthetic management of a male patient with Noonan syndrome complicated by kyphoscoliosis, pulmonary stenosis posted for ASD closure pulmonary valvotomy is described [3,4].

Case Report

21 year old male (weight 30 kg, height 124 cm) who was diagnosed clinically as Noonan presented to us with complaints of Tachypnea, dyspnea on exertion and easy fatigability. He had short stature, sloping and broad forehead and hypertelorism ptosis, short webbed Neck, broad chest with widely spaced nipples, kyphoscoliosis and undescended testis on the left side (Figure 1). Past history revealed that the child had undergone Balloon pulmonary valvotomy under general anesthesia at age of 15 year. He had no history of drug allergy and blood transfusions in past. Hemoglobin level was 12.8 g/dl and renal function test, liver function test, blood glucose, serum electrolytes and rest of investigations were within normal limits. No coagulation or platelet defects.

Preoperative evaluation: Short stature, short webbed neck, buck teeth, micrognathia, dental malocclusion, widely spaced nipples, pectus excavatum, kyphoscoliosis (Figure 2). On clinically examining patient his pulse rate 98/min and regular, blood pressure 108/64 mmHg. Airway assessment revealed his mouth opening was 2 finger, mallampati airway grade 3, buck teeth present, thyromental distance was 6 cm, neck and spine examination showed webbed neck, severe kyphoscoliosis. On auscultating patient a systolic murmur was heard at aortic area and pulmonary area.



Figure 1: Front view of patient, showing pectus excavatum, wide set nipples.



Figure 2: Back view of patient, showing kyphoscoliosis.

Electrocardiogram (ECG) showed left axis deviation with dominant S wave in leads V1 to V6, Chest X-ray showed increased cardiac silhouette and deviation of trachea to right side. X-ray dorsal spine (AP) and (lateral) s/o severe kyphoscoliosis seen with convexity to left centered at D9 vertebra and cob angle 85°. MRI lumbo-sacral spine s/o kyphoscoliotic deformity noted in the thoracic spine with maximum convexity at D10 vertebral level. Pulmonary function test revealed severe restrictive pattern. 2D ECHO s/o 12 mm Ostium secundum ASD with left to right shunt (Figure 3). Dysplastic pulmonary valve with moderate to severe pulmonary stenosis. LVEF (Left ventricular ejection fraction) 60%.



Figure 3: Dorsal X-ray of spine (AP /lateral) s/o kyphoscoliosis with convexity to left centered at D9 vertebra.

After written informed high risk consent, patient was scheduled for cardiac surgery for relief of pulmonary stenosis and ASD closure. In the operating room, ECG, pulse oximetry and blood pressure monitoring was instituted. Under all aseptic precaution femoral central venous catheterization was done with blind external landmark guided technique. Right internal jugular cannulation was tried but we were unsuccessful because of distorted anatomy due to severe kyphoscoliosis. Under aseptic condition femoral arterial lines was inserted to monitor blood pressure trends, titrate drug therapies and obtain blood samples for arterial blood gases and laboratory studies.

Difficult airway management in this rare Noonan syndrome exists due to dysmorphic craniofacial appearance, skeletal anomalies such as pectus deformities, cubitus valgus and vertebral defects. In case of intubation and airway management difficulties, we were prepared with difficult trolley equipments, McCoy laryngoscope, video laryngoscope, fiber optic laryngoscopy. The European Resuscitation Council guidelines for the management of the airway and ventilation during resuscitation suggest that both curved and straight laryngoscope blades, gum elastic bougie and stylet be available to undertake intubation as well as a laryngeal mask airway and combitube as alternatives where intubation is not possible [5]. In addition, these guidelines suggest that equipment be kept for both needle cricothyrotomy and surgical cricothyrotomy for use in the 'cannot intubate, cannot ventilate' situation [6-10].

General anaesthesia was initiated with glycopyrrolate 0.1 mg, fentanyl 150 mg, midazolam 1 mg, propofol 50 mg and scoline 80 mg. After establishing successful bag mask ventilation Cormack-Lehane score 3 were assessed with direct laryngoscopy (McCoy laryngoscope blade, size 3). Successful intubation was done with endotracheal tube number 7 mm internal diameter into the trachea without trouble in first attempt after 60 seconds. Maintenance of anaesthesia and adequate analgesia achieved with oxygen, nitrous oxide, sevoflurane 2.5% and vecuronium bromide [11,12].

Intraoperative according to kirklins score of pulmonary valve patients score was 18 mm. Pulmonary valve was dilated with Hegar's dilator number 18, adequate opening of pulmonary valve insured. ASD closure was done with pericardial patch. Cardiopulmonary bypass was

uneventful. Patient came of CP bypass with minimal inotropes adrenaline 0.05 microgram/per min. The entire procedure lasted for 2 h. Post-operative the patient was taken to CICU, mechanical ventilation was done in SIMV mode. The patient was put on fentanyl infusion to provide optimal post-operative analgesia [13-17].

The postoperative course of the patient was normal and the inotrope and vasodilatory medications were stopped and subsequently patient was extubated 6 h after shifting CICU. Patient was hemodynamically stable. Drains removed on 2nd POD, CVP & arterial line removed on 3rd POD. Patient was shifted to ward on 4th POD.

Discussion

Dr. Jacqueline Noonan in 1963, a paediatric cardiologist studied a series of 9 cases with congenital heart disease associated with a phenotype similar to Turner syndrome. Noonan syndrome is autosomal dominant rare genetic disorder with incidence estimated at 1:1000 to 1:2500 live births. The exact incidence is unknown. The principle characteristic features of Noonan syndrome are unusual facial features (broad forehead, ptosis, short webbed neck, micrognathia, and wide set nipples), short stature (restricted growth), musculoskeletal abnormalities (kyphoscoliosis) and heart defect present at birth (congenital heart disease). Anaesthetic challenges and management in such cases is due to difficult airway management and maintaining cardiovascular stability (Table 1).

Noonan syndrome should be considered in anyone who presents with two or more of the following	
Characteristic facial features	Short stature
Developmental delay and learning disability	Typical chest deformity
Undescended testes	Kyphoscoliosis
Heart defect	First-degree relative who has Noonan
Pubertal delay and /or infertility	syndrome or any of the above features

Table 1: Noonan syndrome is considered in anyone who presents with two or more of the following.

Successful difficult airway management needs early recognition, adequate preparation, planned and vigilant approach. Familiarity with special techniques which may be used for tracheal intubation of patients with a difficult airway is must.

Conclusion

Through evaluation of children and adolescents with unusual facial and skeletal abnormalities, short stature and congenital heart defects should be done. Preoperative evaluation should include a thorough evaluation of the airway and cardiovascular system. Intraoperatively

we must be prepared for a difficult airway and avoid the sympathetic nervous system activation. The entire anaesthesia management should be designed and planned considering all above anomalies to prevent complications. Vigilant and prompt anaesthesia management is necessary intraoperatively and postoperatively to avoid anticipated anaesthetic and surgical adverse effects and ensure successful outcome.

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