

Anaesthesia in Naxos disease: First case report

Hüseyin Yıldız¹, Emin Silay^{1*}, İsmail Coskuner¹, Kemal Ozyurt², Seref Olgar³, Nimet Senoglu¹, Hafize Oksuz¹

¹ Department of Anaesthesiology and Reanimation, Faculty of Medicine, Kahramanmaraş Sutcu Imam University, Yoruk Selim. Mah. Hastane Cad. No:32, 46050, Kahramanmaraş, Turkey. ² Department of Dermatology, Faculty of Medicine, Kahramanmaraş Sutcu Imam University, Yoruk Selim. Mah. Hastane Cad. No:32, 46050, Kahramanmaraş, Turkey. ³ Department of Paediatrics, Faculty of Medicine, Kahramanmaraş Sutcu Imam University, Yoruk Selim. Mah. Hastane Cad. No:32, 46050, Kahramanmaraş, Turkey.

ABSTRACT

Naxos disease is a recessively inherited arrhythmogenic right ventricular cardiomyopathy in which the cardiac phenotype is associated with palmoplantar keratoderma and woolly hair. The hair phenotype is unique, characterized by congenital woolly, curly, rough, and light-colored scalp hair and sparse eyebrows. However, arrhythmias and severe cardiomyopathies are causes of severe life threatened intracardiac thrombus. Thrombus therapy needs private care and sedative, operative processes need to give close attention to these patients. In this case report, sedation given a patient who having severe congestive heart failure with huge mural thrombus in left ventriculus has been presented.

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KEY WORDS: intracardiac mural thrombus, congestive heart failure, Naxos disease, anaesthesia

INTRODUCTION

Naxos disease is a rare autosomal recessive disease that consists of an associated triad of woolly hair, thickened palms and soles (keratoderma), and heart involvement. The hair phenotype is unique, characterized by congenital woolly, curly, rough, and light coloured scalp hair and sparse eyebrows [1]. However, arrhythmias and severe cardiomyopathies are causes of severe life threatened intracardiac thrombus. Thrombus therapy needs private care and sedative, operative processes need to give close attention to these patients [2-4]. In this case report, sedation given a patient who having severe congestive heart failure with huge mural thrombus in left ventriculus has been presented.

CASE REPORT

A nine years old girl patient who has treated with anti-congestive therapy for dilated CMP and arrhythmias due to Naxos disease since two years admitted to hospital due to palpitation, syncope and edema. On her electrocardiographic examination ventricular tachycardia had been detected and it was ceased with antiarrhythmic drugs but on echocardiographic examination huge

mural thrombus on left ventricle has been detected. The thrombus was adherent to left ventricle free wall and apex (Figure1). The apical part of thrombus was mobile. The clinician made a decision to insert jugular vein catheterization for intensive monitorization however. She is taking Acetylsalicylic acid, carvedilol, digoxine, amiodarone and furosemide drugs by peroral. On her physical examination she was nervous and pale. In addition, her pulse rate was 146 in minute; blood pressure was 70/50 mmHg. She had woolly hair (Figure 2), palmoplantar keratoma (PPK) and ikterus. On chest examination, there were crepitant ralles and gallop rhythm. She had hepatomegaly and pretibial edema. After examination and verbal and written consent taking from her family and patient she prepared for process-



FIGURE 1. Echocardiography in left ventricular thrombus

* Corresponding author: Emin Silay,
Department of Anaesthesiology and Reanimation, Faculty of
Medicine, Kahramanmaraş Sutcu Imam University, Yoruk Selim.
Mah. Hastane Cad. No:32, 46050, Kahramanmaraş, Turkey
Tel: +090 344 221 23 37; Fax: +090 344 221 23 71
e-mail: eminsilay@hotmail.com

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FIGURE 2. A. Palmar keratoderma, B. Plantar keratoderma, C. Woolly hair

ing in operation room. Due to hypotension ketamine was given and after then midazolam and fentanyl were given. Her blood pressures were in normal range and after the sedation a guide wire inserted to the internal jugular vein. During guide wire locating ventricular tachycardia has been developed and it has ceased after guide wire pulling. A 5 Fr three-lumen catheter was inserted on guide wire and the processing has been completed after the bleeding and localization control and then patient waken up without complication.

DISCUSSION

Naxos disease is a rare autosomal recessive disease that consists of an associated triad of woolly hair, thickened palms and soles (keratoderma), and heart involvement. The hair phenotype is unique, characterized by congenital woolly, curly, rough, and light coloured scalp hair and sparse eyebrows [1]. The nonepidermolytic keratoderma appears during the first years of life and involves mainly pressure areas in the palms and soles. The heart manifestations appear during the teenage years and are severe and progressive and may end with arrhythmia and premature sudden death. The disease was originally described in individuals from the Greek Island Naxos by Protonotarios et al in 1986. The heart condition is known as arrhythmogenic right ventricular dysplasia (ARVD) [1-2]. It is characterized by progressive replacement myocytes by adipose tissue and fibrosis. These results in a frequent and moderate ventricular dilatation associated with ventricular, or sometimes supraventricular, arrhythmia [4, 5]. In this case report, sedation for central vein catheterization a girl patient who having severe congestive heart failure with huge mural thrombus in left ventriculus has been presented. In patients with hemodynamic instability, heart failure and arrhythmia, or thrombosis, performing Anaesthesia and sedation is not preferred and requires close attention due to serious complications. The drugs that will be used in such cases,

should not lead to myocardial depression and hypotension. Our study represents the first case report in the literature focusing on anaesthesia in patients with Naxos disease. Naxos disease usually presents itself by syncopes during the adolescent ages. However, the first symptom in our case was swelling in the hands and feet. Cardiac symptoms arise at late puberty in the form of dysrhythmia (mostly ventricular tachycardia) in 91%, sudden death in 28%, and heart failure in 30% of cases [6]. Since the disease had a tendency to show progression, the treatment is of symptomatic character. Implanting automatic defibrillator may be life-saving during ventricular tachycardia and fibrillations. Final treatment option is heart transplantation. Because ventricular fibrillation may develop during anaesthesia, defibrillator should be readily available and inotropic support should be continued before and after the procedure due to risk of cardiac failure [7]. Premedication with standard doses of any of the commonly used agents is desirable and well tolerated in patients with normal or near normal ventricular function. Patients with poor ventricular function, on the other hand as in our case, tend to be very sensitive to most agents, and premedication doses should be reduced in proportion to the severity of ventricular impairment. These patients should receive premedication with oxygen support under monitorization [8]. Anaesthetic management should be tailored to the severity of regurgitation as well as the underlying left ventricular function. Factors that exacerbate the regurgitation, such as slow heart rates (long systole) and acute increases in after load, should be avoided. Bradycardia can increase the regurgitant volume by increasing left ventricular end-diastolic volume and acutely dilating the mitral annulus. The heart rate should ideally be kept between 80 and 100 beats/min [7, 8]. On the other hand, excessive volume expansion can also worsen the regurgitation by dilating the left ventricle and lead to pulmonary oedema. Therefore, fluid replacement should be carried out carefully. Any hemodynamic instability can

cause sudden ischemia and cardiac arrest in these patients [8]. While choosing the drugs and techniques for anaesthesia, optimization of cardiac output should be considered. Ventricular tachycardia independent from hypotension can be corrected by intravenous delivery of procainamide, lidocaine, or amiodarone, whereas symptomatic ventricular tachycardia is best treated by external electrical cardioversion. Our cases did not require any of those [8]. Thus, while delivering our anaesthetics, we combined low doses of midazolam, fentanyl, and ketamine in order to prevent an excessive response that could be triggered by them individually at normal doses. Etomidate can be used, since generally it does not change the myocardial contractility and cardiac output. Volatile anaesthetics can also be used at low doses, however, one should be careful in preventing excessive cardiac depression. Propofol can reduce arterial blood pressure associated with the decreased cardiac contractility and preload and inhibition of the sympathetic vasoconstrictor activity [8]. In conclusion, anaesthetic procedures in patients with Naxos disease require routine monitorization and close attention, because heart failure and dysrhythmia can lead to death. Also it should be kept in mind that anaesthetics may lead to myocardial depression and sudden hypotension.

DECLARATION OF INTEREST

There is no conflict of interest.

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