Clinical Report

Journal of INTERNATIONAL MEDICAL RESEARCH

Journal of International Medical Research
2018, Vol. 46(2) 895–900
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DOI: 10.1177/0300060517734680
journals.sagepub.com/home/imr
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Hypoxia due to positive pressure ventilation in Edwards' syndrome: A case report

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Abstract

Edwards' syndrome also known as trisomy 18 is a congenital disorder associated with cardiovascular issues including ventricular septal defect (VSD), atrial septal defect (ASD) and patent duct arteriosus (PDA). An emergency colostomy was performed on a neonate born with an imperforate anus. Pre-operative transthoracic echocardiography showed presence of VSD, a patent foramen ovale (PFO) or ASD. Even though the baby had a good general condition and optimal peripheral oxygen saturation (SpO_2), during positive pressure ventilation, she suffered severe hypoxia (50% SpO_2). The cause of the hypoxia was thought to be the right-left shunt and so during a second attempt at anaesthesia a vasopressor (noradrenaline 0.03 μg/kg/min) was infused to increase systemic vascular resistance. Thereafter, SpO_2 increased to 80–90% and the surgery was completed. The baby recovered without any neurological complications. Genetic testing post-partum showed she had Edwards' syndrome.

Keywords

Trisomy 18, Edwards' syndrome, heart septal defect, R-L shunt, positive pressure ventilation.

Date received: 16 June 2017; accepted: 11 September 2017

Introduction

Edwards' syndrome also known as trisomy 18, is a chromosomal abnormality associated with abnormalities in several parts of the body.[1] Affected individuals often have multiple organ defects, such as congenital heart disease (i.e., ventricular septal defects, valvular heart disease and patent duct

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arteriosus [PDA]), an imperforate anus, a prominent occiput and finger deformities.[1–3] Due to the presence of several life-threatening medical problems, a patient's life span is usually less than one year.[4]

We report here on a neonate who had an imperforate anus which required an emergency colostomy. During anaesthesia, positive pressure ventilation caused severe refractory hypoxia. Genetic testing post-partum showed she had Edwards' syndrome.

Case report

A 2-day-old baby girl was born by caesarean section at 38 weeks. Her mother was 33 years old and healthy. The baby weighed 2710g (10 ∽ 25 percentile) and had a 1-min and 5-min Apgar score of 8 and 10, respectively. Results from an integrated test performed at 12 weeks gestation had raised suspicion of Edwards' syndrome. Therefore, antenatal investigations were performed which included amniocentesis for karyotyping and three obstetric ultrasound examinations. However, apart from a single umbilical artery, no abnormalities were detected.

At birth, the baby was found to have a single umbilical artery and vein and no anus. Edwards' syndrome was suspected because of low birth weight, single umbilical artery, clenched hand and a gastrointestinal anomaly. Abdominal prone X-ray and invertogram (Figure 1) were taken but abdominal ultrasound was not performed. Liver function test results were normal and blood test results were as follows: white blood cell, 15,200/mm^3; haemoglobin, 19.7 g/dL; haematocrit, 58%; platelets, 275,000/mm^3; prothrombin time, 12.7 sec; activated partial thromboplastin time, 55.4 sec; blood urea nitrogen/creatinine, 10.5/0.7 mg/dL; Na/K/Cl, 141/5.0/106 mEq/L. Following a complete physical examination and review of the invertogram, an independent paediatric surgeon confirmed the presence of a high type imperforate anus with rectovaginal fistula. The neonate was transferred to the surgical department for an emergency colostomy.

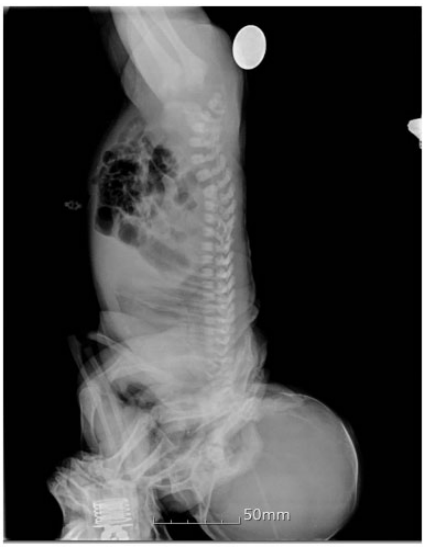

Figure 1 Invertogram of the neonate for demonstration of the extent of rectal atresia and delineation of rectal gas. The radiograph shows a large amount of bowel gas in the baby's abdomen.

Prior to the general anaesthesia, a transthoracic echocardiography (TTE) was performed to evaluate any cardiac abnormality. The TTE showed a perimembranous ventricular septal defect (VSD) with a bidirectional shunt (diameter 1.8 mm) (Figure 2a) and a patent foramen ovale (PFO) or secundum atrial septal defect (ASD) with a left-to-right shunt (diameter 1.9 mm) (Figure 2b). A PDA was not observed and cyanosis or abnormal breathing patterns were not present. Oxygen saturation monitored by pulse oximetry showed 90–95% peripheral oxygen saturation (SpO_2) at room air without additional oxygen administration. General anaesthesia was induced by 2 mg/kg of thiopental sodium in 100% oxygen and rocuronium bromide (1 mg/kg) was administrated intravenously to facilitate later endotracheal intubation.

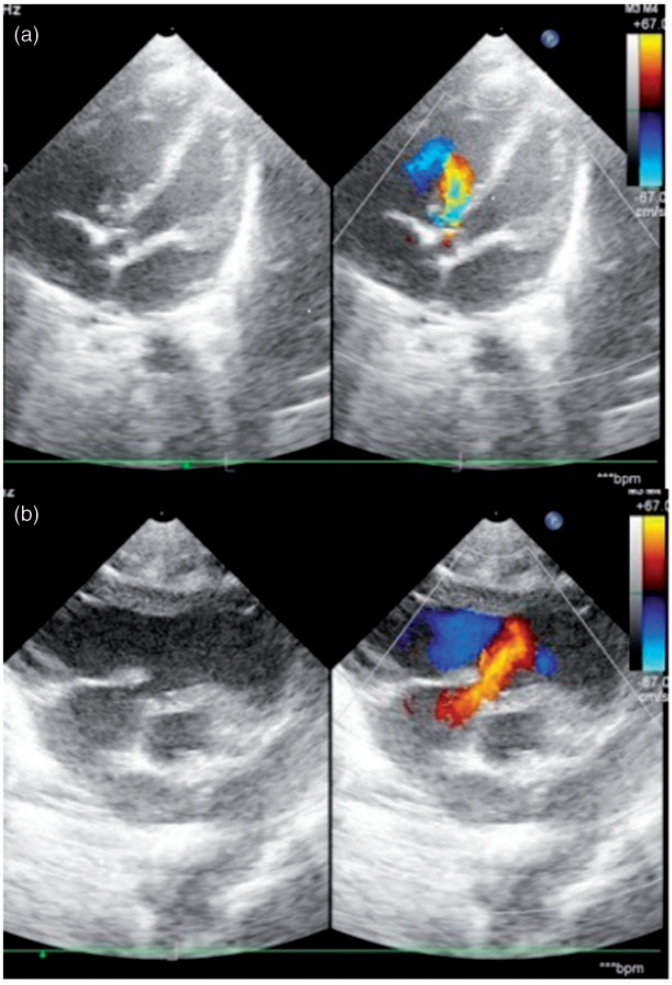

Figure 2 (a) Transthoracic echocardiography. Perimembranous ventricular septal defect (VSD) with bidirectional shunt (diameter 1.8 mm). (b) Transthoracic echocardiography. Patent foramen ovale (PFO) or secundum atrial septal defect (ASD) with left to right shunt (diameter 1.9 mm)
The colour at the right side of both figures indicates the amount of intracardiac shunt.
bpm, beats per minute

After positive pressure ventilation with mask bagging, oxygen saturation decreased slowly and so an endotracheal tube was inserted. Although correct positioning of the endotracheal tube was confirmed by clear breath sounds, following the intubation, SpO_2 increased but then gradually decreased to approximately 50%. Indeed, despite use of vigorous ventilation with 100% oxygen, SpO_2 did not increase above 80%, Therefore, surgery was postponed and the patient was awoken using pyridostigmine (0.2mg/kg) and glycopyrrolate (0.01mg/kg) as reversal agents. Spontaneous breathing returned and the SpO_2 increased immediately to 100%.

The following day, surgery was rescheduled. Despite the previous day's

desaturation events, prior to anaesthesia SpO_2 was 95% at room air without additional oxygen. Even with preoxygenation at 100% oxygen, SpO_2 remained at 95%. Radial artery cannulation failed and so arterial blood gas analysis was not possible. As on the previous day, after positive pressure ventilation with mask bagging, oxygen saturation once again, slowly decreased from 95% to 85–90%. Endotracheal intubation was undertaken and a portable x-ray was used to confirm the location of the tube. (Figure 3)

After the correct positioning of the endotracheal tube was confirmed, positive end-expiratory pressure (PEEP) of 5 cm H_2O was applied to correct the hypoxia. SpO_2 fell to 80–85% despite vigorous ventilation with 100% oxygen and the end tidal CO_2 was hypocapnic at 30–32 mmHg. The cause of the hypoxia was assumed to be the right-to-left shunt via VSD and ASD. Therefore, noradrenaline at 0.03 μg/kg/min was infused to increase systemic vascular resistance (SVR) and PEEP was stopped. SpO_2 was consistently maintained at approximately 85–90% during the operation.

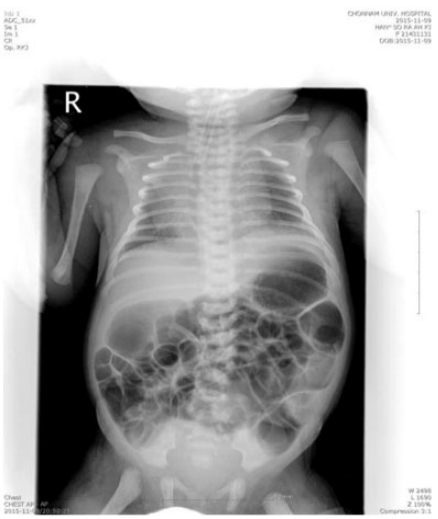

Figure 3 Chest radiograph after orotracheal intubation. Radiograph confirmed correct placement of endotracheal tube.
R, right side

At the end of surgery, an abrupt, severe desaturation occurred and despite increasing the dose of noradrenaline and giving a bolus injection of adrenaline (0.01 mg/kg) SpO_2 did not increase. The neuromuscular reversal drug, sugammadex (4 mg/kg), was administered to recover spontaneous breathing and when restored SpO_2 increased to 95%. The patient was transferred to the neonatal intensive care unit and SpO_2 was maintained at 95% during spontaneous breathing. The baby did not show any signs of focal motor weakness or altered mental state during her hospital stay. Approximately one month after surgery, genetic karyotyping test results confirmed a diagnosis of Edwards' syndrome.

Discussion

Edwards' syndrome or trisomy 18 is a rare but serious genetic disorder. [1] The live born prevalence is estimated as 1/6,000–1/8,000, but the overall prevalence is thought to be higher (1/2500–1/2600) due to the high frequency of fetal loss and pregnancy termination after prenatal diagnosis.[5] This chromosomal disorder is associated with multiple organ system deformities and only a small number of affected children survive the first year.[1] Cardiovascular issues include VSD, ASD and PDA, central nervous issues include cerebellar hypoplasia, brain oedema and enlarged cisterna magna and gastrointestinal system issues include, omphalocele, oesophageal atresia, trachea-oesophageal fistula, diaphragmatic hernia and rectal- or large-bowel atresia. Other physical deformities include clenched hands, rocker bottom feet and malformed ears.[1–3] Median survival time for live births with full trisomy 18 has been reported to be 14 days with a one-year survival rate of 8%[4].

Genetic screening is available for women with high risk of chromosomal abnormalities and a quadruple screening test is advised during the second trimester of pregnancy. Levels of serum alpha-fetoprotein [AFP], total human chorionic gonadotropin [hCG], unconjugated oestriol and inhibin A are significantly lower in pregnancies with trisomy 18 compared with normal pregnances.[5,6] Non-invasive pre-natal testing has been reported to have an overall sensitivity and specificity for detecting trisomies 21, 18 and 13 of 99.61% and 99.91%, respectivley.[7] While the quadruple screening test result in this present case suggested Edwards' syndrome (1:14), the karyotyping result obtained via amniocentesis at 17 weeks gestational age was negative for Edwards' syndrome. Chromosomal analysis of amniotic fluid cells is considered to be the 'gold standard' in prenatal testing because error rates are exceedingly low at less than 0.01–0.02%.[8] The neonate was eventually diagnosed with the condition following gene karyotyping after birth and the prenatal screening test result was assumed to be a false negative.

The imperforated anus required urgent surgery but positive pressure ventilation during anaesthesia caused the SpO_2 to fall. Positive pressure ventilation increases pulmonary vascular resistance which results from compression of the pulmonary vascular bed. The increased pulmonary vascular resistance leads to an increase in a right ventricular afterload.[9,10] In patients with intra-cardiac shunt, such as VSD, ASD, or PDA this causes shunting of the blood to the left atrium or ventricle of the heart without contacting the pulmonary vessels.[11] The surgery was abruptly terminated but because of the baby's worsening condition, it was re-scheduled for the following day. Refractory hypoxia occurred during general anaesthesia again but this time noradrenaline was infused to counteract the assumed right-to-left shunt via VSD or ASD. Noradrenaline increases SVR,[12,13] and in this patient, it appeared to reduce the right to left shunt which improved SpO_2 allowing surgery to be completed. The operation was successful and the baby recovered and was discharged without any neurologic complications.

A limitation of this case report was that arterial blood gas analysis was not obtained because of a failure to insert an arterial line. In addition, the assumed changes in direction of intra-cardiac shunting during general anaesthesia following positive pressure ventilation or noradrenaline could not be verified because intraoperative TTE was not performed.

In conclusion, the case report has highlighted that applying positive pressure ventilation to a patient with an intra-cardiac shunt, such as a VSD, ASD, or PDA, requires caution. Refractory hypoxia is likely to occur but using a vasopressor to increase SVR may alleviate the problem.

Declaration of conflicting interests

The authors declare that there are no conflicts of interest.

Funding

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

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