Cardiol Young 2009; 19: 111–112

ISSN 1047-9511
doi:10.1017/S1047951108003363
First published online 2 December 2008

Brief Report

An infant with transposition and cystic fibrosis

Amanda L. Cook, Kari L. Crawford, Derek A. Williams

Department of Pediatrics, Division of Pediatric Cardiology, Wake Forest, University School of Medicine, Winston-Salem, North Carolina, United States of America

Abstract We report an infant with transposition, with the circumflex artery arising from the right coronary artery, who was diagnosed with cystic fibrosis following the occurrence of meconium ileus in the postoperative period.

Keywords: Congenital heart disease; genetic syndromes; meconium ileus

Transposition, produced by the combination of concordant atrioventricular and discordant ventriculo-arterial connections, is the most common cyanotic congenital cardiac defect presenting in the neonatal period. In treating this entity, the arterial switch procedure has now been shown to have excellent results when performed in infants at low risk. Associated genetic disorders. and extracardiac anomalies, can significantly exacerbate the clinical management and outcome of these infants. Clinicians who care for children with this lesion must remain aware of the potential for other anomalies which can complicate the care. In this report, we describe an infant with both transposition and cystic fibrosis.

Case Report

The infant, born at full term, was a male weighing 2.8 kilograms. Pregnancy was complicated by insulin dependent diabetes mellitus, and a positive family history of cystic fibrosis in a sibling. Prenatal screening for the cystic fibrosis mutation was refused. Initially vigorous, he was noted to be cyanotic by the end of the first day of life. An arterial blood gas was obtained, and showed a pH of 7.34, with a partial pressure of oxygen of 12 torr. The infant was started on supplemental oxygen and prostaglandins, with saturations increasing to mid 80%. The infant was transferred to a tertiary medical facility. Upon arrival, the repeat arterial blood gas showed a pH of 7.36, and a partial pressure of oxygen of 61 torr.

Examination revealed a right ventricular lift, a normal first heart sound, a single second heart sound, and no murmurs. The lungs were clear, with shallow respirations, and no retractions. Pulses were equal and full in all extremities. There was no hepatomegaly.

A chest radiograph revealed a normal cardiac silhouette with a narrow mediastinum. The pulmonary vasculature was normal in appearance, and there were no infiltrates seen. Echocardiography demonstrated transposition, with concordant atrioventricular and discordant ventriculo-arterial connections. There was low velocity left-to-right shunting at the level of the patent oval foramen. Inspection of the coronary arteries revealed the circumflex artery arising from the right coronary artery. There was a large patent arterial duct, with left-to-right shunting. The infant was continued on 100% inspired oxygen via a hood. He passed some meconium stool on the first day of life, and received no enteral feeds preoperatively.

At 9 days of age, the patient underwent the arterial switch procedure. He initially tolerated the

Correspondence to: Amanda L. Cook, MD, Wake Forest University School of Medicine, Medical Center Boulevard, Winston-Salem, NC 27157, United States of America. Tel: (336) 716 3232; Fax: (336) 716 0533; E-mail: acook@wfubmc.edu

Accepted for publication 19 October 2008

procedure well, and returned to the paediatric intensive care unit intubated. At 16 days of age, he was noted to have abdominal distension, having passed no stool over the previous 10 days. A barium enema was performed, which demonstrated a large meconium plug and a small left colon. His treatment for the meconium plug included broad spectrum antibiotics and acetylcysteine. After several days of treatment, he passed the meconium plug, and began tolerating enteral feeds. His blood tested positive for cystic fibrosis with ΔF508 homozygosity being found on a separate test prior to receiving the results of neonatal screening. On the 29th day of life, he was discharged home.

Discussion

Cystic fibrosis is a disorder of a membrane glycoprotein, which is present in some types of epithelia cells and contributes to the regulation of the flux of ions across cell surfaces. There is often severe injury to the organs involved, including the lungs, the exocrine pancreas, the liver, and the intestines. The pulmonary disease is often chronic and progressive, resulting in most of the morbidity and mortality. Other complications can also be severe, including infection, malnutrition, electrolytic disturbances, diabetes, nasal polyps, and vasculitis.

The life expectancy for those born with cystic fibrosis has improved from about 6 months when initially described in the 1930s, to more than 30 years.[1] Advances in survival include the recognition of milder cases, supplementation of pancreatic enzymes, use of antibiotics, and centralized care in centres promoting aggressive treatment. It is the most common lethal autosomal recessive disease found in Caucasians in the United States of America, with an incidence of 1 per 3200 live births in Caucasians, and 1 in 15,000 in African-Americans.[2] The gene resides on chromosome 7, with the most common disease-causing allele in the United States of America being ΔF508.[3] This gene codes for the cystic fibrosis transmembrane conductance regulator, which is a membrane glycoprotein, and results in abnormal cAMP-regulated chloride channels.

Transposition accounts for 5 to 7% of all congenitally malformed hearts, having an incidence of 0.2 per 1000 live births, making it the most common cyanotic cardiac lesion encountered in the neonatal period.[4] The lesion has known associations with certain genetic conditions including 22q11 deletions, acrocephalopolysyndactyly type II, CFC1 mutations, and PROSIT240 mutations.[5–8] Furthermore, there is risk of familial recurrence in siblings of 1.8%, suggesting monogenic or oligogenic inheritance.[9] There has been one previous case report from Saudi Arabia of an infant with transposition with a positive sweat chloride test, albeit with a normal analysis for deoxyribonucleic acid.[10]

Although it is well recognized that transposition is frequently associated with extracardiac anomalies, including 22q11 syndrome, in our review of the literature we were unable to find other reported patients with both transposition cystic fibrosis in the United States of America. Clinicians must remain aware of genetic disorders, which may alter the clinical management of patients with congenitally malformed hearts.

References

1. Davis PB. Cystic fibrosis since 1938. Am J Respir Crit Care Med 2006; 173: 475–482.
2. Orenstein D, Rosenstein B, Stern R. Diagnosis of cystic fibrosis. In: Orenstein D, Rosenstein B, Stern R (eds). Cystic fibrosis medical care. Lippincott Williams & Wilkins, Philadelphia, 2000, pp 21–54.
3. Accurso FJ. Update in cystic fibrosis 2006. Am J Respir Crit Care Med 2007; 175: 754–757.
4. Hoffman JI. Incidence of congenital heart disease: I. Postnatal incidence. Pediatr Cardiol 1995; 16: 103–113.
5. Goldmuntz E, Clark BJ, Mitchell LE, et al. Frequency of 22q11 deletions in patients with conotruncal defects. J Am Coll Cardiol 1998; 32: 492–498.
6. Balci S, Haytoglu T, Ozer S. A case of a four-day-old male with Carpenter's syndrome with transposition of great arteries. Turk J Pediatr 1998; 40: 461–466.
7. Goldmuntz E, Bamford R, Karkera JD, de la Cruz J, Roessler E, Muenke M. CFC1 mutations in patients with transposition of the great arteries and double-outlet right ventricle. Am J Hum Genet 2002; 70: 776–780.
8. Muncke N, Jung C, Rudiger H, et al. Missense mutations and gene interruption in PROSIT240, a novel TRAP240-like gene, in patients with congenital heart defect (transposition of the great arteries). Circulation 2003; 108: 2843–2850.
9. Digilio MC, Casey B, Toscano A, et al. Complete transposition of the great arteries: patterns of congenital heart disease in familial precurrence. Circulation 2001; 104: 2809–2814.
10. Baslaim GM, Jamjoom AA. Extracorporeal membrane oxygenation support post-arterial switch procedure for a child with cystic fibrosis: case report. J Card Surg 2006; 21: 419–420.