Berry Syndrome in Association with Familial Limb Malformation

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Abstract
We describe a newborn boy diagnosed with Berry syndrome consisting of a distal aortopulmonary septal defect, aortic origin of the right pulmonary artery, and interruption of the aorta. The child was noted to have reduplication of the right thumb. The childâ s mother had a claw malformation of her left hand but a normal cardiovascular status. Genetic analysis for TBX5 and SALL4 mutations were negative in both the patient and his mother. This case describes the first ever report of Berry syndrome in an infant with reduplication of the right thumb and familial limb malformation.

Introduction
Berry syndrome refers to the novel constellation of cardiac findings including interrupted aortic arch, distal aortopulmonary septal defect, right pulmonary artery branch originating from the ascending aorta and intact ventricular septum, which was first described in 1982.[1] There have been no previous reports in the literature of any limb abnormalities or family history of this associated with Berry syndrome. We now describe the first report of Berry syndrome in an infant with reduplication of the right thumb and familial limb malformation.

Case Report
A boy weighing 3.3kg was delivered at 38 weeks gestation and noted to have thumb reduplication of the right hand and hypospadias (Figure 1). Of note, the mother of the child had a lobster claw malformation of the left hand but a normal cardiac examination (Figure 2). There were no other limb malformations or asymmetries. The infant was admitted to the intensive care unit at two days of age because of poor feeding and tachypnoea. On examination the heart rate was 130 beats per minute, respiratory rate was 65 breaths per minute and there was a 50 mm Hg systolic gradient between right upper and lower limbs. There was a normal first and split second heart sound.

Figure 1 Photograph of the infantâ s thumb reduplication of the right hand.

Figure 2 Photograph of the motherâ s lobster claw malformation of the left hand.

There was a 2/6 systolic murmur at the left sternal edge. The liver was palpable 2 cm below the costal margin. The femoral pulses were markedly diminished. The child was commenced on a prostaglandin E infusion at a rate of 0.05 micrograms/kilogram/minute. Transthoracic echocardiography demonstrated situs solitus with atrioventricular and ventriculoarterial concordance. There was a 5 mm perimembranous ventricular septal defect with left to right shunting. There was an 8 mm distal aorto-pulmonary window with origin of the right pulmonary artery from the ascending aorta (Figure 3). The left pulmonary artery arose from the main pulmonary trunk. There was a left aortic arch with an interruption of the aorta distal to the origin of the left subclavian artery (type A). The arterial duct was patent with right to left shunting. There was a left superior caval vein to coronary sinus connection. The origin of the coronary arteries was normal. There was normal biventricular systolic function.

Figure 3 High parasternal short-axis view demonstrating the distal aorto-pulmonary window and anomalous origin of the right pulmonary artery from the ascending aorta.

A complete one-stage surgical repair was performed under hypothermic circulatory arrest through a median sternotomy. The child was placed on cardiopulmonary bypass. Direct end-to-end anastamosis of the interrupted aortic arch was performed. The aorto-pulmonary window and anomalous right pulmonary artery were divided and the pulmonary artery was re-constructed using native pericardial patch. The ventricular septal defect was closed with a pericardial patch and the ductus was ligated and divided. Delayed sternal closure was performed four days after surgery. The child was discharged home two weeks after surgery. Five months after surgery the child underwent balloon angioplasty of the right pulmonary artery for moderate arterial stenosis with a peak instantaneous gradient of 45 mm Hg. The child is currently alive and well.

Discussion
Berry first described the constellation of findings including aortopulmonary defect, hypoplastic transverse arch, origin of the right pulmonary artery from the aorta and intact ventricular septum.[1] In contrast to the conventional proximal aorto-pulmonary septal defect, the distal defect in patients with Berry syndrome may represent a partial persistence of the common arterial trunk. The pulmonary arterial bifurcation may malattach to this undivided truncal segment and consequently the right pulmonary artery may be partially or completely shifted into the aorta. This abnormal right pulmonary arterial origin may lead to â stealâ from the aortic flow during embryogenesis resulting in hypoplasia of the aortic arch.[1] Although the constellation of anomalies characteristic of Berry syndrome can be elucidated by echocardiography alone as in this case,[2,3] MRI may assist in the diagnostic evaluation.[4]

The first one-stage surgical repair was reported by Boonstra et al.[5] This approach has now become the recommended operative repair with six such reports in the literature to date.[5-7] Although both mother and child had limb anomalies involving the contralateral upper limb, the motherâ s cardiac status was normal. Genetic screening of the mother and infant was negative for mutations in TBX5 and SALL4, the former genetic mutations previously described in association with limb anomalies including Holt-Oram syndrome. This report describes the first association of Berry syndrome with a familial limb malformation.

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