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Brief Report

A rare case report of corrected transposition of the great arteries in association with tuberous sclerosis and cardiac rhabdomyomas

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Abstract The neuro-cutaneous syndrome tuberous sclerosis is commonly associated with rhabdomyomas in various organs including the heart. We are reporting a rare case of a 7-month old male child with congenitally corrected transposition of the great arteries associated with tuberous sclerosis and cardiac rhabdomyomas. To our knowledge, this rare association has not been reported so far.

Keywords: Tuberous sclerosis; rhabdomyomas; congenitally corrected transposition of the great arteries

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Congenital heart defects are often associated with various extra-cardiac malformations. In the Baltimore-Washington Infant Study, the frequency of associated extra-cardiac anomalies in live births is reported to be 20%.[1] Congenitally corrected transposition of the great arteries associated with tuberous sclerosis and multiple cardiac rhabdomyomas is a very rare finding and has not been reported so far.

Case report

A 7-month old male child presented to our institute with a 5-month history of failure to thrive. The patient was born at full term by spontaneous vaginal delivery and did not require any postnatal resuscitation. The patient demonstrated tachypnoea and diaphoresis with feeding and, though he had achieved normal developmental milestones, he had severe failure to thrive. The patient also had history of recurrent lower respiratory tract infections and recurrent generalised tonic–clonic seizures since 2 months of age. He was referred to our institute for cardiac work-up for his symptoms.

On clinical examination, he was underweight for his age at 4 kg (below 3rd percentile) and his transcutaneous oxygen saturation was 89%. He was tachypnoeic and irritable on presentation. There were multiple ash leaf macules on his trunk. His heart rate was 80 beats/min with irregularly irregular pulse.

The chest x-ray showed cardiomegaly with increased pulmonary blood flow. The electrocardiogram was suggestive of complete atrioventricular dissociation (3rd degree heart block). Ultrasonography of the abdomen showed multiple cysts in both kidneys.

Transthoracic two-dimensional and colour Doppler echocardiography showed atrial situs solitus, levocardia, atrioventricular and ventriculoarterial discordance, suggestive of congenitally corrected transposition of the great arteries. There was large non-restrictive inlet ventricular septal defect and multiple polypoidal masses in the right ventricle suggestive of rhabdomyomas (Fig 1).

Cardiac magnetic resonance imaging showed multinodular masses and pedunculated masses in the right ventricle suggestive of rhabdomyomas. Magnetic resonance imaging of the brain showed subependymal nodules and multiple tubers in various sites including both grey and white matter (Fig 2).

Discussion

Congenitally corrected transposition of the great arteries is a rare congenital heart defect with an estimated prevalence of less than 1% of all congenital

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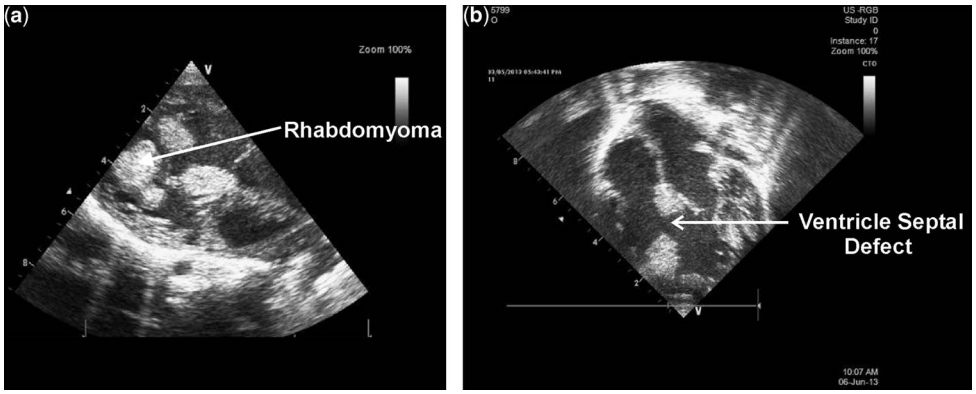

Figure 1.
*(**a**) Two-dimentional echocardiography image in parasternal long axis view showing multiple rhabdomyomas (**b**) Two-dimentional echocardiography image in apical 4-chamber view showing Ventricle Spetal Defect.*

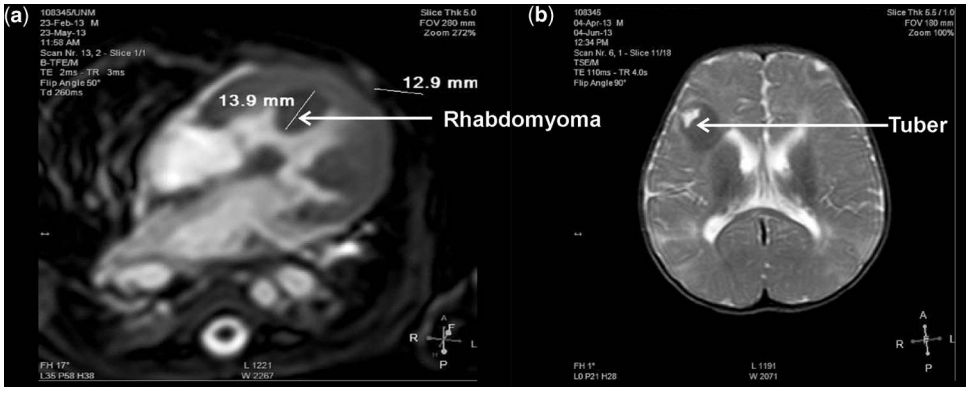

Figure 2.
*(**a**) T-2 weighted axial image showing multiple rhabdomyomas in right ventricle (**b**) T-2 weighted axial image showing tuber in brain.*

heart diseases and is often associated with various congenital anomalies such as ventricular septal defects, pulmonary stenosis, valvular malformation, conduction system dysfunction, and coronary anomalies.[2] The outcome and prognosis in congenitally corrected transposition of the great arteries depends upon the pressure within the systemic ventricle, progression of disease, and associated anomalies.[3]

Cardiac rhabdomyoma is the most common primary cardiac tumour in children and is considered to be a hamartoma of developing cardiac myocytes.[4,5,6] In all, 80% of cardiac rhabdomyomas are associated with tuberous sclerosis. Tuberous sclerosis is a rare (1:1000 to 1:6000) multi-system genetic disease associated with benign tumours of the brain, kidney, heart, eyes, and skin.[7] Cardiac rhabdomyomas are usually well tolerated in utero, and the natural history is to regress or completely resolve in the third trimester or postnatally in most of the cases.[8]

Rhabdomyomas in a close proximity to the conduction system can lead to various arrhythmias and various degrees of heart block. Sudden death has been reported with cardiac rhabdomyomas probably due to ventricular tachycardia. Our patient had the rare association of a large inlet ventricular septal defect with congenitally corrected transposition of the great arteries and multiple rhabdomyomas in the right ventricle. The associated finding of complete atrioventricular dissociation is probably due to congenitally corrected transposition of the great arteries and associated rhabdomyomas involving the conduction system. The symptoms and clinical course in congenitally corrected transposition of the great arteries depends chiefly on associated anomalies.[9] Young patients with isolated congenitally corrected transposition are often overlooked because symptoms are absent and clinical signs are subtle. Infant mortality is related to congestive heart failure. Survival is then relatively constant, with an attrition rate of ~1% to 2% a year.[10] However, the associated anomalies have a great impact on prognosis and outcome. Multiple rhabdomyomas may have an impact on the ultimate outcome in this infant and hence needs long-term follow-up. Meanwhile, we had advised our patient to undergo pulmonary artery banding to reduce his increased pulmonary blood flow and to improve his

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failure to thrive. Placement of an epicardial pacemaker was recommended to treat the complete heart block.

To the best of our knowledge, this is the first reported case of congenitally corrected transposition of the great arteries associated with cardiac rhabdomyomas and tuberous sclerosis. Owing to its wide clinical spectrum, it is prudent to anticipate unexpected abnormalities in congenitally corrected transposition of the great arteries, which may affect the treatment and outcomes in such cases.

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Conflicts of Interest

None.

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