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Case report

Cardiac rhabdomyoma with LVOT obstruction and anorectal malformation in a neonate: A rare association

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ABSTRACT

Cardiac rhabdomyomas are described as sporadic as well as associated with Tuberous sclerosis. Association with LVOT Obstruction with anorectal malformation or imperforate anus has not been reported so far in published literature. In this case report we describe one such case.

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1. Introduction

Cardiac rhabdomyomas are described as sporadic as well as associated with Tuberous sclerosis. Association with LVOT Obstruction with anorectal malformation or imperforate anus has not been reported so far in published literature. In this case report we describe one such case.

2. Case report

The patient was 2.57 kg full term male baby born to a 27 yrs old multigravida mother at district hospital at periphery. Antenatal history was insignificant. The patient was born by

vaginal delivery and did not require any resuscitation. The patient was referred to our centre for complaint of not passing stool. The patient was investigated (invertogram done) and diagnosed to have anorectal malformation (low ARM-membranous type), for which urgent anoplasty was done in emergency operation theatre (EOT No.872/436/9). Post-operative phase was uneventful. On 16th postoperative day patient was referred to cardiology department for evaluation of asymptomatic ejection systolic murmur grade 3/6 over the precordium with maximum intensity at left upper parasternal area. Rest all systemic examination as well as routine blood investigations (CBC, RFT, LFT, Urine, ASLO Titre, CRP) were within normal limits. Two Dimensional Echocardiography (Movies 1–7) revealed multiple homogenous, well

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Fig. 1

Fig. 2

circumscribed, echogenic, intramural as well as intracavitary masses in LV & RV with speckled appearance. Pedunculated mass over Aortic valve was producing significant LVOT Obstruction, with Doppler derived gradient Peak 47.7 mm of Hg (Fig. 1). There was no echolucent area noted in the mass. On basis of above said Echo findings a provisional diagnosis of cardiac rhabdomyoma was made. Both mother and patient were examined for any feature of Tuberous sclerosis. But there were no finding suggestive of Tuberous sclerosis, found in both of them. CT head of patient was normal. Family history was insignificant. In view of asymptomatic presentation and natural history given in literature, patient was advised conservative management and observation in form of advice of follow up echocardiography.

Supplementary video related to this article can be found at <http://dx.doi.org/10.1016/j.ihj.2012.08.006>.

Review Echocardiography (Movies 8–13) was done after approximately 10 months, showing regression in number and size of previous cardiac masses. Doppler derived Peak LVOT gradient was also reduced to Peak 8.5 mm of Hg (Fig. 2). This confirmed the diagnosis of cardiac rhabdomyoma.

Supplementary video related to this article can be found at <http://dx.doi.org/10.1016/j.ihj.2012.08.006>.

3. Discussion

Primary cardiac tumours are rarely encountered in neonatal period. Of these, cardiac rhabdomyomas are the most common (Fig. 3). Although cardiac rhabdomyomas are associated with Tuberous sclerosis in more than 50% cases but sporadic cases are equally prevalent.^{1–4} Cardiac rhabdomyomas are intramural or pedunculated well-circumscribed tumours, generally multiple, found mostly in the ventricles, but may be found anywhere in heart. During prenatal period cardiac rhabdomyomas may be found incidentally during routine foetal ultrasound scan or may present as hydrops foetalis. In postnatal period presentation may be varied from completely asymptomatic detection to may be as

asymptomatic cardiac murmur or may present variously as congestive cardiac failure, low cardiac output due to intra-cardiac flow obstruction,^{1,2,5} arrhythmias of various types^{1,2,5} or as sudden infant death. Left ventricular outflow tract obstruction seems to be one of the rarer presentations of cardiac rhabdomyomas in infants.⁵ But unlike our patient, most of these children are ill at diagnosis. As per natural history of cardiac rhabdomyomas in infants and children, these tumours have a propensity for spontaneous regression. Younger the age at diagnosis, higher the chance for spontaneous regression, complete regression being more common in the first 4 years of life.⁵ This has important implications for therapy. Surgical intervention is indicated only in the event of haemodynamic compromise or intractable arrhythmias. Anorectal malformation is neither described as a part of presenting feature of Tuberous sclerosis and cardiac rhabdomyoma nor with sporadic cardiac rhabdomyoma. As far as the authors are aware, this is one of the very rare cases detected to have a cardiac rhabdomyoma in a clinically stable neonate

Fig. 3 – Invertogram.

with LVOT Obstruction and anorectal malformation without any antenatal suspicion for cardiac rhabdomyoma. Whether this is a rare association or a chance association, cannot be commented as cardiac rhabdomyoma is itself a rare entity.

Conflicts of interest

All authors have none to declare.

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