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Brief Report

Punch-out lesion following regression of a large left ventricular outflow rhabdomyoma

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Abstracts We report the case of a patient with rhabdomyoma of the left ventricular outflow tract, causing severe obstruction at birth. The tumour regressed completely by 6 years of age, leaving a punch-out lesion. The potential for spontaneous regression of these tumours and the formation of a myocardial lesion following rhabdomyoma regression are discussed.

Keywords: Rhabdomyoma; cardiac; regression

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CARDIAC RHABDOMYOMAS ARE ONE OF THE COMMON presentations of tuberous sclerosis in the neonatal period. These lesions are known to diminish in size or completely disappear after birth. The myocardium is thought to be restored following rhabdomyoma disappearance.

Report of a case

We present the case of an 8-year-old boy with tuberous sclerosis who was born with multiple cardiac rhabdomyoma. At birth, the echocardiogram revealed a large lobulated hyperechogenic mass within the intraventricular septum extending into the left ventricular outflow tract and causing severe flow obstruction (Fig 1a). The functional diameter of the outflow tract was 2 mm and the pressure gradient reached 70 mmHg at 1 week of age. Multiple other hyperechogenic masses were observed within the interventricular septum and the free wall of the left ventricle. Tuberous sclerosis was confirmed using wood-lamp skin scanning, brain ultrasound, and CT. Continuous ambulatory electrocardiograpy demonstrated premature atrial and ventricular beats that disappeared at later follow-up. The tumours gradually regressed.[1] At 4 months of age there was very mild left ventricular outflow obstruction with a 5-mm flow diameter causing a 11 mmHg pressure gradient. Regression of the septal mass was minimal. At 4 years of age, the protruding portion of the mass disappeared and the septal portion measured 4 mm. At 6 years of age the tumour in the left ventricular outflow tract had completely disappeared leaving a punch-out lesion (Fig 1b and c). The residual wall of the left ventricular outflow tract at the site of the punch-out lesion measured 2 mm in thickness. The other tumours also disappeared, and were replaced by myocardium, as observed in the echocardiogram, with no tissue defects.

Discussion

In the case presented, rhabdomyoma of the left ventricular outflow tract spontaneously disappeared, leaving a punch-out lesion. To the best of our knowledge, this phenomenon has not been described before.

Spontaneous rhabdomyoma regression has been described in many cases in the past. In some cases surgical resection is performed.[2] Recent reports suggest that drug therapy with a mechanistic target of rapamycin inhibitors, sirolimus,[3] or everolimus,[4] accelerated tumour regression. As these drugs have a potential side effect,[5] their use is restricted

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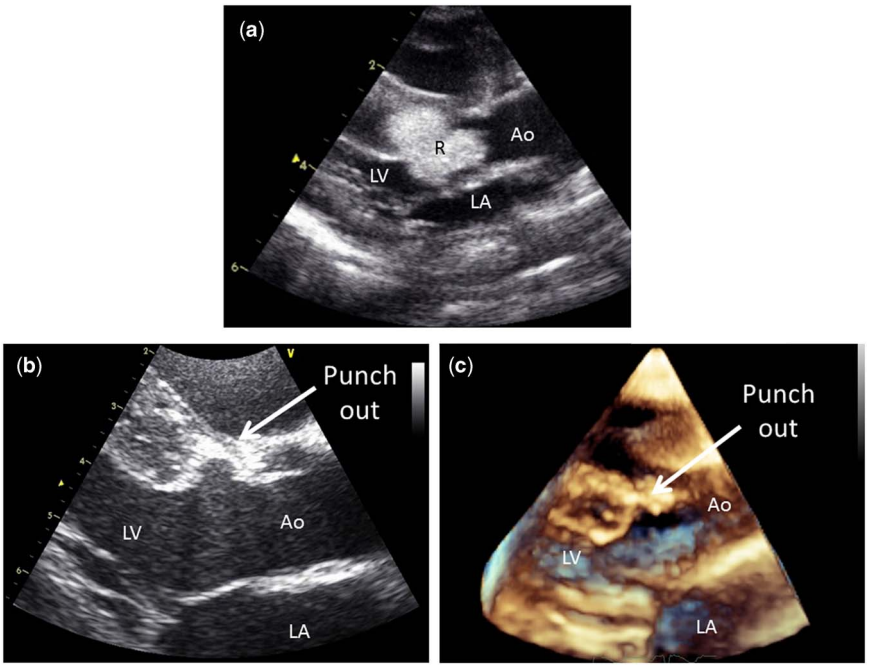

Figure 1.
*(**a**) Echocardiographic parasternal long-view aged 9 days showing a large, lobulated rhabdomyoma causing severe LVOT obstruction. (**b**) Two-dimensional and (**c**) three-dimensional echocardiographic parasternal long-view aged 6 years showing LVOT punch-out lesion at the site of the rhabdomyoma. Ao = Aorta, LA = left atrium, LV = left ventricle, R = rhabdomyoma.*

to patients with tumours causing life-threatening haemodynamic compromise. The potential for spontaneous regression, which may occur in months, should be kept in mind when faced with a new-born with significant obstruction.

Rhabdomyoma regression and disappearance is usually associated with myocardium replacing the rhabdomyoma space. Regression with no myocardial replacement, creating punch-out lesion, may potentially result in a tissue defect, such as a ventricular septal defect. In our case, the residual septum was 2 mm in size, with no septal defect. The reason the space left behind by the regressed tumour was not replaced by myocardium may be the late involution of the tumour – that is, after the age of 4 years – or the location of the left ventricular outflow tract. Does the myocardium return to a normal state with rhabdomyoma regression? A recent report found, on cardiac MRI, a region of fatty replacement in the interventricular septum in a 16-year-old boy with tuberous sclerosis.[6] The authors speculate that a regressed rhabdomyoma has been replaced by fat.

The International Tuberous Sclerosis Consensus Group[5] recommends that with rhabdomyoma disappearance there is no need for echocardiographic follow-up. Our report and the MRI finding suggest that the heart may not be completely normal after rhabdomyoma regression. Myocardial lesions may result in arrhythmia, septal defects, or impaired function. Thus, follow-up may be warranted.

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Conflicts of Interest

None.

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