CASE REPORT

Right ventricular cavity near obliteration in neonatal severe biventricular hypertrophic cardiomyopathy

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SUMMARY

A newborn presenting with cyanosis at day 9 of life was admitted to the local hospital. Initial local echocardiography confirmed a cardiac issue and the patient was transferred to a tertiary cardiac hospital. Further imaging confirmed a rare presentation of cardiomyopathy with severe right ventricular outflow tract obstruction. Surgery was performed but with postoperative haemodynamic instability complicated by incessant ventricular tachycardia. Following discussion with the family, care was withdrawn. Postmortem demonstrated a rare form of hypertrophic cardiomyopathy with right ventricular outflow tract obstruction not previously described in a neonate.

BACKGROUND

Cyanosis is a distressing predicament for any new family to deal with. An infant patient's cyanosis was well recognised and subsequently seen by the local team. The diagnosis was however unexpected. Unlike the 'common' causes of congenital heart disease causing cyanosis, the diagnosis in this case report was more sinister.

The presentation of this case presented challenges even for experienced cardiologists. This case report also highlights not only the importance of ensuring what is best for the child by involving the family in the difficult decision-making, but also demonstrates the presentation of a complex heart condition not seen previously with such severity.

CASE PRESENTATION

A full-term female baby born by elective caesarean section with normal antenatal ultrasound scans was discharged home at day 3 of life. The baby was noted to have a soft systolic murmur on day 1 of life with no other cardiovascular concerns and was due to be reviewed at 6 weeks of age. On day 10 of life the family brought her to hospital with concerns of dusky colour changes when the baby was lying on her side. A complete history, examination and review during her presentation revealed a healthy baby with a murmur as previously noted. The baby was noted to have saturations of 94% in air however on balance was thought to be well and the murmur considered to be that of a ventricular septal defect. The baby was therefore discharged following reassurance to the family and a plan for community midwives to review the baby in the next 2 days to monitor weight gain. On day 12 of life community midwives noted the baby to have a bluish discolouration while weighing the baby. The baby was subsequently referred back to the local

hospital. Review by the paediatrician confirmed cyanosis with low oxygen saturation of between 89% and 91% with no preductal or postductal difference and normal 4-limb blood pressures. The patient was thought at this point to have an ejection systolic murmur in the left upper sternal edge with a parasternal heave and no evidence of cardiac failure or respiratory distress. A brief echocardiogram in the local hospital suggested a diagnosis of hypertrophic cardiomyopathy. There was no family history of cardiomyopathy or any heart condition. The patient was discussed and transferred to the tertiary unit at the Royal Brompton Hospital in the early hours of the following morning.

INVESTIGATIONS

Admission ECG (figure 1) demonstrated right atrial dilation but no overt features of hypertrophy. Chest X-ray (figure 2) showed cardiomegaly and increased lung markings. Echocardiography (videos 1a, b) demonstrated severe right ventricular (RV) cavity muscular overgrowth with mid-cavity outflow tract obstruction causing an effective double chambered right ventricle (Doppler-derived peak gradient of 100 mm Hg) and severe RV systolic dysfunction. Both the ventricles were effectively slit-like with the mitral valve annulus Z score of -2.8 (Boston) and tricuspid valve annulus Z score -2.2 (Boston).¹ The RV anterior wall measured 10 mm. This resembled the appearances found in the luminal overgrowth of pulmonary atresia with intact ventricular septum but on balance it was felt to represent hypertrophic cardiomyopathy. On subsequent CT imaging she was noted to have small multiple cysts in the liver with no abnormalities elsewhere in the body. There were no genetic features pointing to a syndrome and chromosomal analysis revealed normal chromosomes, including negative testing for Noonan syndrome. Comprehensive cardiomyopathy screening was also negative.

TREATMENT

On admission (day 13 of life), the patient was started on a β -blocker (propranolol) following intermittent ECG rhythms suggestive of ventricular tachycardia with no haemodynamic compromise. Following discussion with the electrophysiologists it was felt the patient would benefit from this even with reduced RV function. She continued to have ventricular tachycardia and was started on amiodarone. She remained stable with an oxygen requirement to keep her saturations above 95%. As she was haemodynamically stable she underwent comprehensive investigation as to the cause of her cardiomyopathy and was

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Figure 1 A 12-lead ECG performed during admission demonstrating right axis deviation, right atrial enlargement and no evidence of right or left ventricular hypertrophy for age, but lateral repolarisation changes are present (inverted or flattened T waves).

subsequently discussed in the multidisciplinary team meeting (MDT) on day 18 of life. It was felt that relieving the RV outflow obstruction although high risk would improve the RV hypertrophy. On day 19 of life she had surgical relief of the RV outflow tract obstruction with a myectomy, infundibular patch across the outflow tract and patent ductus arteriosus ligation leaving the patent oval foramen open. At the end of surgery the infundibular lumen measured 15 mm having been 2 mm at the start of surgery.

Figure 2 Showing chest X-ray of patient following insertion of nasogastric tube to support feeding. Note the cardiomegaly and slightly increased lung markings (AP, anteroposterior).

Video 1 (A) Apical 4-chamber view showing severe biventricular hypertrophy with evidence of a double chamber in the right ventricle. (B) Short axis view of the two ventricles showing bilateral severe hypertrophy. The right ventricle shows such severity of hypertrophy that the moderator band splits the chamber in two.

Figure 3 A 12-lead ECG showing incessant ventricular tachycardia (VT). There is a prolonged QTc with wide QRS complexes and a relatively slow rate VT.

OUTCOME AND FOLLOW-UP

Postoperatively the patient was haemodynamically unstable with severe low cardiac output and multiorgan failure. Optimal intensive care management (inotropes, antiarrhythmics and medication to reduce afterload) failed to improve her condition, which was complicated by incessant ventricular tachycardia (figure 3). Following MDT discussion and being declined for heart transplant, her parents agreed to withdraw care at the age of 6 weeks.

Postmortem examination showed a normal appearing infant with marked biventricular hypertrophy. There was near obliteration of the RV lumen (figure 4). The myocardium was pale with hypertrabeculation throughout the right and left ventricle and subendocardial fibrosis (figure 4). This is an interesting

finding as there was no evidence of hypertrabeculation on multiple echocardiographic studies. Surgical resection created a small channel from the RV inlet to outlet (figure 5) but severe hypertrophy remained. There were no changes consistent with inflammatory or metabolic conditions. There was also circumferential left ventricular hypertrophy and microscopic evidence of myocyte disarray with fibrosis in both ventricles and evidence of hypertrabeculation (figure 6A, B). The irregular cysts noted

Figure 4 Postmortem section of heart at the mid-ventricular level demonstrating severe biventricular hypertrophy. Note the cavity of both the ventricles is slit-like and almost completely occluded by the hypertrophic myocardium. Both ventricles show pale myocardium with evidence of subendocardial fibroelastosis and large trabeculations (arrow; RV, right ventricle; LV, left ventricle).

Figure 5 Postmortem section of the opened RV demonstrating a severe hypertrophied wall of the RV (arrow) and the hypertrophied apex with hypertrabeculation. The PV measured 10 mm in diameter. The figure also demonstrates the channel from the RV inlet to outlet created by surgical resection of the myocardium (RV, right ventricle; PV, pulmonary valve).

Figure 6 (A) Microscopic plate of myocardium showing loss of normal parallel alignment of muscles. The image shows myocytes encircling around central foci of connective tissue (H&E stain $\times 600$). (B) Microscopic plate of myocardium demonstrating large trabeculations. There is subendocardial fibrosis (red). Van Gieson stain. (Magnification $\times 200$).

in the liver were mesenchymal hamartomas of the liver. No other abnormalities were noted at autopsy.

DISCUSSION

The diagnostic characteristic of hypertrophic cardiomyopathy is ventricular hypertrophy in the absence of conditions that produce the magnitude of hypertrophy present.² It is the commonest genetic cardiac condition with a prevalence of 1 in 500.³ The annual incidence in infants is even lower (1.89–3.2 in 100 000) perhaps due to late gene expression.⁴ RV involvement

occurs in just under a quarter of all cases but sufficient to cause RV outflow tract obstruction in only 12.5%.⁵ This degree of RV hypertrophy is extremely unusual and was sufficient to mimic the luminal overgrowth found in pulmonary atresia with intact ventricular septum. It is often due to a metabolic or syndromic aetiology but none was demonstrated in this case. Biventricular hypertrophy in an infant is known to have a poor prognosis.⁵

This rare combination of hypertrophic cardiomyopathy with RV outflow tract obstruction in infants has been previously described,⁵ but this is the first time one of such severity is reported in a neonate. Surgical rescue was attempted but to no avail in such a high-risk case.

Learning points

- ▶ Severe cardiomyopathy can be a cause of cyanosis in neonates.
- ▶ Surgical treatment in alleviating outflow tract obstruction in cardiomyopathy is a high-risk procedure.
- ▶ Arrhythmias in hypertrophic cardiomyopathy can be extremely challenging to manage.
- ▶ Hypertrophic cardiomyopathy with right ventricular outflow tract obstruction with such severity is extremely rare.

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Competing interests None.

Patient consent Parental/guardian consent obtained.

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