

Practical Pediatric Cardiology

Case-Based
Management of
Potential Pitfalls

Alan G. Magee
Jan Till · Anna N. Seale
Editors



Springer

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We would like to dedicate this book to the children and their parents who place their trust in us. Although we are only human, it is our duty to appraise and evaluate our experiences so that we can provide the best possible care.

Preface

In an era when most essential information about the morphology, diagnosis, management and prognosis of heart disease in children and adolescents is so easily available on the Internet, is there any longer a place for textbooks? So rapid has been the progress in almost all aspects of medicine that most books are out of date by the time they are published. The results of drug trials or the efficacy of new devices is instantly available to health professionals, patients and families as soon as they appear in one of the many journals available online. Classical teaching from traditional or reference textbooks can be found in an instant.

With this background, the publishers and editors have bravely taken on the challenge of delivering a new book devoted to ‘pitfalls’ in diagnosis and management of heart disease in the young. The editors Jan Till, Alan Magee and Anna Seale are not only personal friends and current or former colleagues for whom I have the greatest respect and admiration but also relatively young paediatric cardiologists; yet, they have a wealth of experience of success and failures in the treatment of children.

They have gathered together personal reminiscences about adverse events or diagnostic challenges from clinicians working in many paediatric cardiology centres. Each of their accounts provides lessons and insights of potential value to all of us. These types of personal descriptions devoted to a single medical specialty are not easily found online. And at a time when there is so much emphasis on informed consent, duty of candour and clinical risk this is a book which should have a place on the bookshelf of all health professionals involved in cardiology. Read it, reflect on your own clinical practice and take the opportunity to learn from the experiences of others.

London, July 2015

Michael Rigby

Acknowledgements

We wish to thank all our contributors, who have worked extremely hard to make this happen, and also to Springer for commissioning the project in the first place and to our partners and colleagues who put up with us during the final stages

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Chapter 1

It's Enough to Make You Anxious

Merlin Ranald McMillan, Jan Till, and Ferran Roses

Abstract The case of a 14-year-old boy with a 3-year history of palpitations and shortness of breath is described. His symptoms were thought to be due to anxiety, and he underwent inpatient psychiatric therapy. Eventually an ECG was performed as part of a screening programme and was recognised to be abnormal. Subsequent investigations confirmed permanent junctional reciprocating tachycardia. The child underwent radiofrequency ablation and was cured.

Keywords Palpitations • Anxiety • Tachycardia • Radiofrequency ablation • Persistent junctional reciprocating tachycardia

Case Description

A 14-year-old boy was referred to an electrophysiologist with a 3-year history of episodes of shortness of breath, palpitations and anxiety. Initially these were thought to be related to asthma; however, symptoms were not relieved by use of beta agonists. Eventually he was referred to psychiatric services and received treatment as

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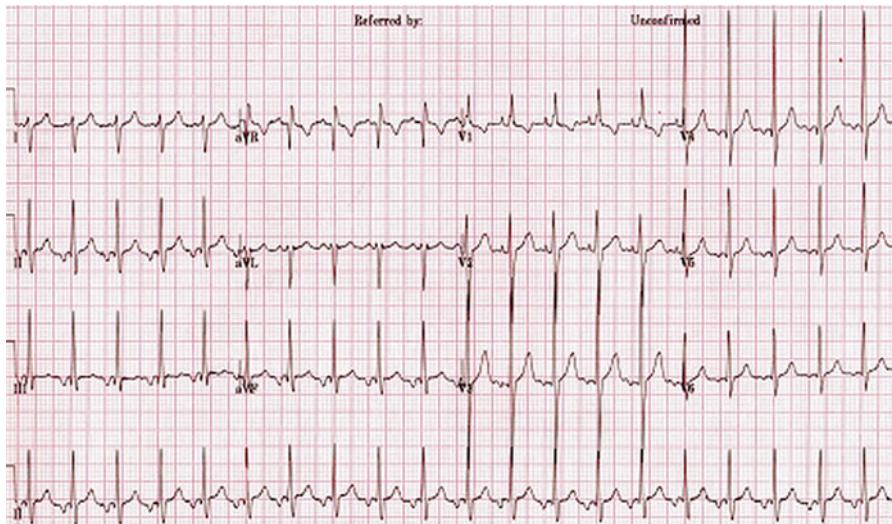


Fig. 1.1 An ECG at rest showing a narrow complex tachycardia of rate. Although the P wave appears immediately before the QRS, the rhythm is not sinus as the P wave axis is inverted (negative in lead II, III and aVF)

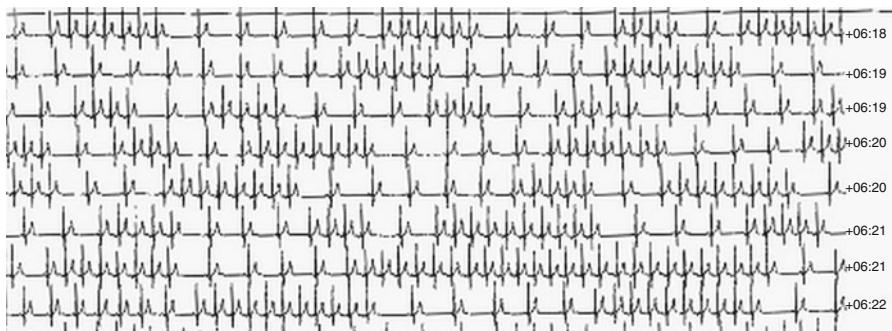


Fig. 1.2 Section of 24-h tape displaying the persistent nature of this tachycardia

an inpatient for anxiety and panic attacks, undergoing psychological therapies to provide coping strategies, which brought some symptomatic improvement.

These episodes did not prevent physical activity, and he continued playing football for a local team and training several hours a week. Only one episode necessitated stopping activity. There was no history of syncope or collapse. When taking deep breaths he used to feel a large “kick” in his chest. These symptoms continued for 3 years when at the age of 14 years he attended screening by the charity Cardiac Risk in the Young. His ECG was found to be abnormal and believing this may be an atrial tachycardia, he was referred to a paediatric electrophysiologist (Fig. 1.1).

His 24-h tape confirmed a persistent tachycardia, continually starting and stopping throughout the 24 h, and vagal manoeuvres performed during the tape reproduced the sensation of a “kick” in his chest and showed termination of tachycardia (Figs. 1.2 and 1.3). The differential diagnosis was atrial tachycardia or permanent

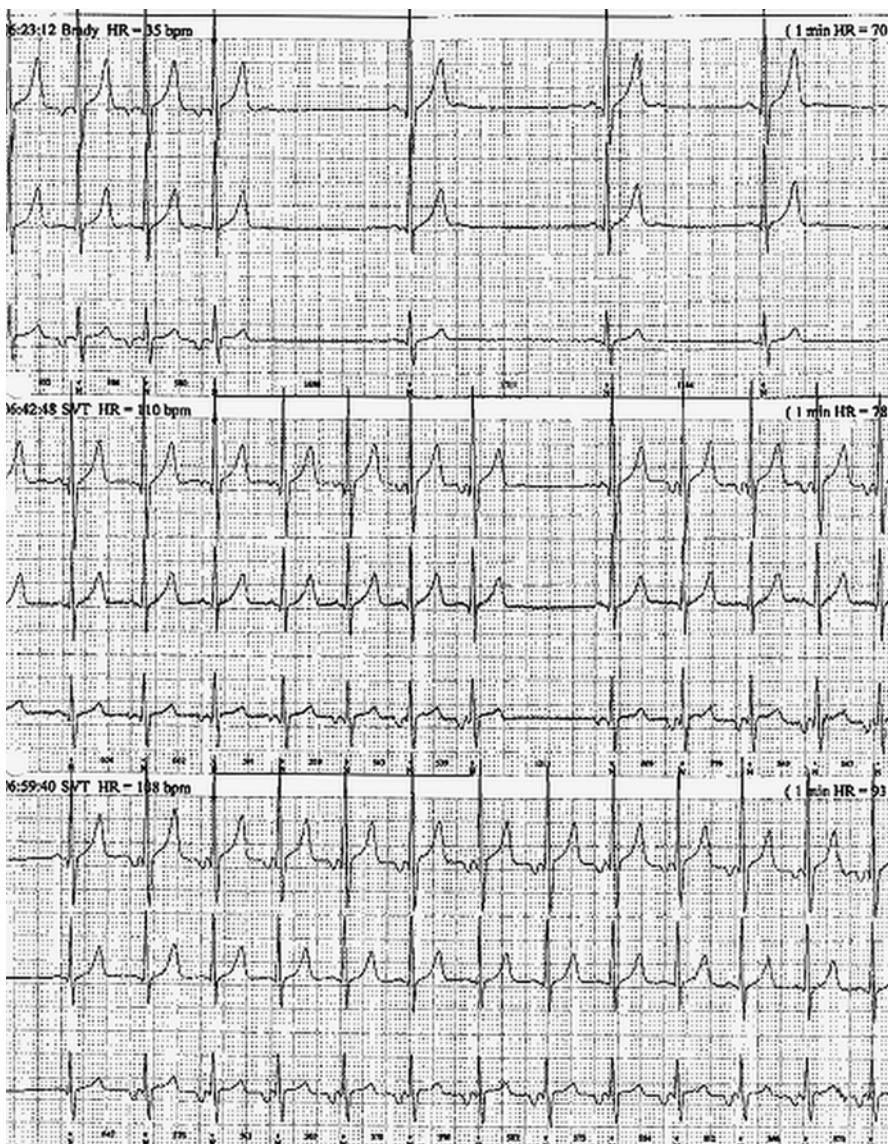


Fig. 1.3 Further sections from the 24-h tape showing in more detail the tachycardia stopping and restarting. The tachycardia appears to stop in the retrograde limb i.e. the accessory pathway limb of the re-entrant circuit. Atrial tachycardia could not be ruled out but was felt unlikely from the short PR interval

junctional reciprocating tachycardia (PJRT). This was discussed with the patient and his parents. A two-dimensional echo showed no structural abnormalities, and his function was within normal limits.

After counselling he underwent diagnostic electrophysiological study and catheter ablation. At electrophysiological study performed with propofol, the tachycardia was easy to see. The re-entrant nature of the tachycardia was confirmed and the retrograde

limb of the circuit mapped characteristically to the mouth of the coronary sinus. The findings were consistent with PJRT. The VA interval was long in tachycardia as predicted, but when the earliest VA interval was identified, radiofrequency energy was applied and the tachycardia terminated. Thereafter tachycardia was not inducible despite pacing manoeuvres and isoprenaline. He made a prompt and uncomplicated recovery. Further follow-up has shown that the ablation has cured the condition and he is symptom-free with no evidence of tachycardia recurrence on 24-h monitoring.

Discussion

Permanent Junctional Reciprocating Tachycardia

PJRT is a rare type of atrio-ventricular (AV) re-entrant tachycardia. There is antegrade conduction through the AV node and slow retrograde conduction through a concealed accessory pathway. The retrograde pathway is often located at the mouth of the coronary sinus posteriorly. Microscopically the pathway has been described as having a serpiginous appearance, which may account for the delayed conduction through it. The pathway cannot conduct antegradely from atrium to ventricle and so there is no delta wave. PJRT is thought to account for around 5 % of childhood supraventricular tachycardia (SVT) and can present at any time from the foetal period until adulthood. It has been suggested that the majority of patients present during the first year of life, but cases can often present in later childhood.

The slow retrograde conduction facilitates the installation of this arrhythmia, which is often incessant from infancy, with few intermittent periods of sinus rhythm. The ventricular rate is variable, and whilst it can be over 250 bpm in neonates may also be seen at rates of 100–150 bpm in adolescents.

Clinical Presentation and Diagnosis

As the tachycardia may be persistent, and relatively slow, it can be relatively well tolerated. Syncope, for this reason, is unusual. Palpitations may not be recognised and if undiagnosed for prolonged periods, patients may present with fatigue or heart failure, which can result in a dilated cardiomyopathy. Whilst not so in our case, some patients have presented in the advanced stages of heart failure and have only been recognised when on the list for transplantation.

PJRT is diagnosed by a standard 12-lead electrocardiogram (Table 1.1).

The ECG in PJRT is frequently misinterpreted as sinus tachycardia or atrial tachycardia. The AV ratio is 1:1 and the P wave occurring just before the QRS may lead clinicians to believe the rhythm is sinus. The tachycardia, particularly in adolescents, may be mild (120–130 bpm) and so combined with symptoms of palpitations and anxiety the clinical picture, as in this case, may be confused with a sinus

Table 1.1 ECG alterations in PJRT^a

| | |
|-------------|--|
| Rhythm | Regular, can be confused with sinus tachycardia as the p wave is just before the QRS |
| Rate | Tachycardia, varies with age but may not be as rapid as “normal SVT” |
| P wave | Negative P wave in inferior leads, reflecting low atrial origin |
| PR interval | Short PR, with long R-P |
| QRS | Narrow complex |
| QT interval | Normal |

^aDuring periods of sinus rhythm a delta wave should not be present

tachycardia associated with a panic attack. The key to the diagnosis is the abnormal P wave axis with negative P waves in inferior limb leads II, III and aVF, reflecting conduction through the atria from the AV junction to the top.

Vagal manoeuvres and atropine may terminate the tachycardia, but the effects are usually short-lived (i.e., a few beats). However, this may aid in confirming the diagnosis.

Treatment

Emergency treatment is rarely needed as PJRT normally does not cause immediate haemodynamic instability. Radiofrequency ablation can offer a cure in the majority of cases. If a cardiomyopathic picture has evolved, then ablation can usually stop any further decline in cardiac function and in most cases cardiac function will eventually recover.

In younger children or children unsuitable for catheter ablation, medical management can be used to suppress or control the rate of the tachycardia. Beta-blockers, class IC anti-arrhythmics (Flecainide or Propafenone) and Digoxin can be used as therapeutic options. For those with poor heart function, Amiodarone (with or without digoxin) shows success rates of over 80 %. Successful medical control of the arrhythmia may be used in small children and neonates or those with poor heart function, allowing the delay of catheter ablation, to decrease complication rate of the procedure.

Palpitations

Palpitations refer to the *subjective* perception or awareness of the heartbeat. As such it is important to clarify exactly what the patient means when they use this word. Children may say that their heart is “jumping,” “skipping,” “racing,” or even “stopping,” and younger children may not be able to explain the sensation with some saying that it hurts.

Palpitations in children are a very common symptom, and one that can be very preoccupying for parents, particularly in families with a history of cardiac illness (i.e., recent death of a grandparent due to myocardial infarction, or atrial fibrillation in an elderly relative). However, evidence from adults with known tachyarrhythmias shows a poor correlation between reported symptoms and monitored tachycardias, suggesting poor proprioception of heart rhythm.

Palpitations in children are usually caused by physiological stimuli such as fever, exercise, anxiety or anaemia. Conversely, serious and potentially fatal arrhythmias may not be associated with palpitations. The lack of association between palpitations and arrhythmia combined with the parental/patient anxiety surrounding “a problem with the heart” make assessment a delicate process.

Children present to general practice, or accident and emergency, where the initial assessment is undertaken by a paediatrician or generalist. Examination is often normal, but a careful history may allow high-risk patients to be identified.

Children with a serious cause underlying their palpitations often have a history of syncope, heart surgery or congenital heart disease: palpitations in this population should be investigated. Other indications for referral to Paediatric Cardiology include a family history of sudden death.

Initial assessment should include a standard 12-lead ECG. This is particularly useful if the patient is symptomatic at the time of examination. The value of the ECG, however, depends on the experience of the interpreting physician. For example, in our case the ECG may have been erroneously thought to be in sinus rhythm because of the 1:1 AV ratio, a P wave before every QRS and normal, regular QRS complexes. If there is any doubt, the ECG should be discussed with local cardiology services.

Further investigations should be tailored to the clinical examination, i.e., haemoglobin and thyroid function. If the child is in tachycardia during the examination (and haemodynamically stable), then efforts should be made to record the rhythm and its response to vagal manoeuvres (allowing the diagnosis to be made retrospectively, if necessary).

Investigation of Paroxysmal Symptoms

In the case of persisting intermittent symptoms it may become important to try and capture the cardiac rhythm during one of these episodes. To do this there are various techniques available beyond the standard 12-lead electrocardiogram.

Holter monitoring or ambulatory ECG recording is one of the first-line investigations. It involves recording a continuous trace of the heart rhythm, for example, during 24 h. It is useful in children whose symptoms are relatively frequent (i.e., daily), and can also be used to assess response to treatment, i.e., number of episodes of tachycardia during the day, maximum and minimum heart rate.

Children can use event recorders, which can be used in two main ways. The child can wear the recorder constantly, and when symptomatic pushes a button to record

the cardiac rhythm. Most loop recorders will also save the 45 s preceding the button press. The other option is that the recorder is applied when symptomatic, and so only records the subsequent period.

The new generation of event recorders are designed to attach to the patient's (or parents') phone. By employing a previously downloaded APP the patient can record a rhythm strip of an ECG by placing his fingers on the metal bars on the device. In a child contact with the two metal feet can be made by placing the phone device on the child's back or leg.

If the child has infrequent episodes, and there is a concern that they may represent a serious arrhythmia, a loop recorder can be implanted to try and capture the rhythm during these episodes. The device has a battery life of up to 3 years, and is inserted subcutaneously in the left axilla or the left anterior chest wall.

Learning Points

- PJRT is a rare type of atrio-ventricular (AV) re-entrant tachycardia with antegrade AV node conduction and slow retrograde conduction through a concealed accessory pathway.
- The ECG in PJRT shows a narrow complex tachycardia, inverted P wave axis with a short PR and can be confused with sinus tachycardia.
- Untreated, the prolonged tachycardia can cause heart failure, and the child may develop a tachycardia-mediated cardiomyopathy. Ablation is usually curative with a good success rate.
- The sensitivity of ECG is dependent on the experience of the interpreting physician – a systematic approach should be used. However, if in doubt contact local cardiology services.
- Further investigation of palpitations should be considered in children with a history of congenital heart disease, cardiac surgery, syncope or a family history of sudden cardiac death.

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Chapter 2

Fetal AVSD or Maybe Not?

Victoria Jowett

Abstract Fetal echocardiography enables prenatal diagnosis of congenital heart disease. However, imaging and interpretation can be difficult for a variety of different reasons. We describe a case illustrating how a left superior vena cava to coronary sinus can be falsely interpreted as an atrioventricular septal defect in the fetus.

Keywords Prenatal diagnosis • Fetal echocardiography • Left superior vena cava • Atrioventricular septal defect

Case Description

A lady was referred to fetal cardiology at 19 weeks gestation in her second pregnancy. First trimester combined screening had given a low risk of trisomy 21. At the time of the anomaly scan a normal four-chamber view could not be obtained by the sonographer. In addition, a single umbilical artery was noted but no other extra cardiac abnormalities.

Imaging at the first consultation was limited by a suboptimal fetal lie with the fetal spine anterior. The conclusion was that the appearance was suggestive of situs solitus with concordant connections in the setting of a complete atrio-ventricular septal defect (AVSD). There appeared to be a moderate primum component and small ventricular component to the AVSD. The diagnosis and anticipated postnatal management was explained to the patient, including the strong association of AVSD with trisomy 21. The patient was offered amniocentesis, which they accepted, and a further appointment was made for 3 weeks later.

The patient returned for the second scan at 23 weeks gestation. Amniocentesis performed at the initial appointment had confirmed a normal karyotype. The fetal

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lie was more favorable for fetal echocardiography and it was clear that there was situs solitus with concordant atrio-ventricular and ventriculo-arterial connections with no AVSD. Normal offset of the AV valves could be demonstrated, as could a bi-leaflet normal appearance of the mitral valve in short axis. There were bilateral superior caval veins (SVC) with the left SVC draining to an enlarged coronary sinus (Figs. 2.1 and 2.2). Additionally, there was a small perimembranous VSD and mild disproportion in the size of the ventricles and great arteries, with left-sided structures slightly smaller than right.

Fig. 2.1 Four-chamber view of the fetal heart. *LV* left ventricle, *RV* right ventricle, *RA* right atrium. ★ denotes marks enlarged coronary sinus giving false impression of a primum septal defect

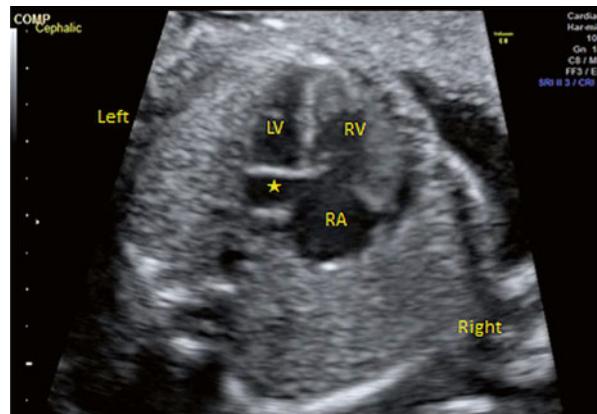
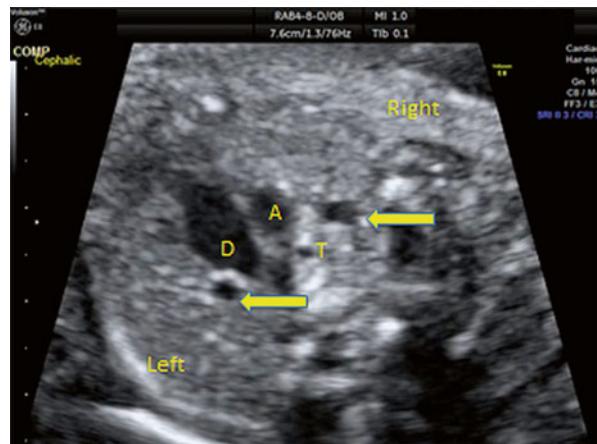


Fig. 2.2 Fetal three-vessel and tracheal view. *Left* and *right* SVC denoted by arrows. *A* aorta, *D* duct, *T* trachea



Discussion

Optimal fetal cardiac imaging depends on a combination of factors, including fetal lie and maternal habitus. Whilst in some patients it is possible to get pictures of comparable quality to a postnatal echo, in others this can be challenging. It is in these situations we have to be particularly wary of common pitfalls in diagnosis.

The coronary sinus lies behind the left atrium and becomes enlarged when it receives additional flow. The most common cause of additional flow is drainage of a left SVC to the coronary sinus that occurs in approximately 0.3 % of the population. More rarely the coronary sinus may appear enlarged due to anomalous pulmonary venous drainage.

Screening of the fetal heart involves a caudal-cranial sweep including five standard imaging planes from the abdominal situs, through the four-chamber view, the left and right ventricular outflow tract and finally the three-vessel and tracheal view. In tertiary fetal cardiology this is complemented by the use of different planes in particular sagittal views.

In imaging of the four-chamber view of the heart, which is a standard view obtained at screening, it is important to be in the correct plane as posterior four-chamber view can create the illusion of a primum atrial septal defect. In addition, in a posterior cut of the AV valves, the offset of the AV valves is not appreciated.

In order to differentiate between the two conditions, the four-chamber view of the heart needs to be visualized in a more anterior position. In addition to this in the three-vessel and tracheal view, the left SVC will be seen as a fourth vessel lying to the left of the arterial duct. Further confirmation of the AV connection can be gained by a short-axis view of the AV valve in which you would expect to see a normal bileaflet mitral valve.

The importance of diagnosing this correctly is that this had quite different implications; therefore, the counseling varies for the two conditions. Isolated left SVC connecting to the coronary sinus is a variation of normal; however, an AVSD is an important form of congenital heart disease. In the setting of an AVSD there is a strong association with trisomy 21. The patient will be offered amniocentesis whereby a small sample of fluid is taken from the amniotic sac. This procedure carries a risk, albeit small, of approximately 0.5 %, of procedure-related miscarriage. Clearly in the setting of a lesion with a very high risk of trisomy 21 the patient may well feel that the risk more than justifies the benefit.

It is, of course, possible for an AVSD and left SVC to coronary sinus to coexist!

Learning Points

A left SVC draining to an enlarged coronary sinus can give the false impression of an atrioventricular septal defect, particularly with suboptimal imaging. To minimise the risk of this pitfall:

- Sweep anteriorly from the four-chamber view giving the appearance of a primum AVSD.
- Look at the AV valve in short axis to examine the morphology of the left AV valve
- Look for a fourth vessel in the three-vessel and tracheal view to confirm the presence of a left SVC.

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Chapter 3

Mind the Gap

Srinidhi J.V. Rao

Abstract Lesions causing a left to right shunt are among the most common congenital heart defects. Whilst ventricular septal defect (VSD), atrial septal defect (ASD), atrioventricular septal defects (AVSD) and patent arterial duct (PDA) form the majority of these lesions, several less common but significant lesions may arise. The presenting symptoms of heart failure, growth faltering and frequent infections are similar to those of common lesions and having a high index of suspicion is important to facilitate early diagnosis and treatment. We describe a patient who had an uncommon cardiac shunt which can be easily missed.

Keywords Aortopulmonary window • Left to right shunt • Congestive heart failure • Ruptured sinus of valsalva aneurysm • Coronary artery fistulae

Case Description

A term baby was born weighing 3.5 kg (50th centile) following normal pregnancy with no perinatal complications. The baby developed respiratory distress a few hours following birth with significant subcostal recession, good bilateral air entry and clear lung fields. The oxygen saturations were >95 % with no difference between preductal and postductal measurements. Femoral pulses were well felt. Heart sounds were normal with no murmurs. The liver edge was palpated 1 cm below the right costal margin. The baby was commenced on non-invasive airway support and commenced on antibiotics following screening for sepsis. Chest radiograph showed cardiomegaly with increased pulmonary vascular markings. Initial transthoracic echocardiogram showed a normally connected heart with a moderate sized perimembranous outlet ventricular septal defect (VSD) with left ventricular

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volume overload. Diuretic therapy was commenced to manage symptoms of pulmonary over-circulation.

Over the next few days, the baby continued to require airway support and was failing to gain weight despite aggressive diuretic therapy along with caloric supplementation. As the symptoms were out of proportion to the size of the VSD, a repeat echocardiogram was performed which showed, in addition, a large proximal aortopulmonary window. The baby was referred to the local tertiary paediatric cardiac surgical centre where successful repair of both lesions was undertaken.

Discussion

A number of cardiac and vascular defects can cause left to right shunt, the most common being VSD, AVSD, ASD, and PDA. Less common cardiac lesions that cause a left to right shunt are listed below; these can occur in isolation but may co-exist with other cardiac lesions:

1. Aortopulmonary window
2. Large coronary fistulae
3. Ruptured sinus of Valsalva
4. Partial anomalous pulmonary venous return

Aortopulmonary Window

Aortopulmonary window (APW, also called aortopulmonary septal defect or aortopulmonary fenestration) refers to a communication between the ascending aorta and the main pulmonary artery in the presence of two separate semilunar valves. The defect is distal to the leaflets of the semilunar valves and can be found in any position where the two vessels are contiguous. Embryological origin is thought to be abnormal septation of the intrapericardial distal common arterial trunk. The defect varies in size and shape. It is a rare cardiac abnormality, comprising about 0.1 % of all congenital cardiac anomalies.

Classification

Richardson classified APW as simple defects between the ascending aorta and pulmonary trunk (type I), defects extending distally to include the origin of the right pulmonary artery (type II), and anomalous origin of the right pulmonary artery from the ascending aorta with no other aortopulmonary communication (type III). The classification scheme recommended by the Society of Thoracic Surgeons Congenital

Heart Surgery Database Committee for APW is as follows: Type I is a proximal APW located just above the sinus of Valsalva, close to the semilunar valve with little inferior rim separating the APW from the semilunar valves. Type II is a distal APW located in the uppermost portion of the ascending aorta. This would correspond to the Richardson type 2 lesion, where the defect overlies a portion of the right PA. Distal defects are noted to have a well-formed inferior rim but little superior rim. Type III is a defect involving the majority of the ascending aorta. Type IV is the intermediate defect. This has adequate superior and inferior rims and is the rare group most suitable for catheter device closure.

Simple APW is a defect without any significant associated anomalies, or anomalies requiring minor or simple repair (patent ductus arteriosus, atrial septal defect, patent foramen ovale). Complex APW is a defect occurring with more complex associated anomalies such as ventricular septal defect, interrupted aortic arch, transposition of great arteries, tetralogy of Fallot, tricuspid atresia, hypoplastic left heart syndrome or anomalous origin of the coronary arteries.

Presenting Features

Antenatal diagnosis has been reported but is exceedingly rare. The postnatal presenting features of an isolated AP window, similar to PDA or VSD, are dependent upon the size of the defect and the pulmonary vascular resistance. As pulmonary vascular resistance falls, the infant becomes symptomatic with tachypnoea, poor feeding, growth faltering, recurrent chest infections and diaphoresis. Pulmonary vascular resistance usually falls after the first 2 weeks of life and patients are often relatively well prior to this becoming symptomatic as the pulmonary resistance falls. Our patient was relatively unusual in that the resistance fell shortly after birth and the baby became quickly symptomatic. Untreated, there may be progression to irreversible pulmonary vascular disease with shunt reversal shunt (Eisenmenger syndrome).

Clinical examination may show a tachypnoeic infant with poor nutritional status. Cyanosis may be present in the setting of increased pulmonary vascular resistance, either in the early newborn period where the pulmonary vascular resistance has not fallen or in the setting of late presentation when pulmonary vascular disease is established. Pulses may be bounding, the precordium will be active, the pulmonary component of the second heart sound is often accentuated and there will be either an ejection systolic murmur at the left upper sternal border or a continuous murmur similar to a PDA. There may also be a rumbling mid-diastolic murmur at the apex due to increased flow across the mitral valve. In the setting of pulmonary vascular disease, the pulmonary component of the second heart sound will be loud and the systolic murmur unimpressive.

Chest radiograph typically shows an enlarged heart with pulmonary plethora related to the size of the defect and the net shunt. ECG may show biventricular hypertrophy and left atrial hypertrophy due to increased pulmonary venous return.

Fig. 3.1 High parasternal short-axis image at the level of ascending aorta demonstrating a large proximal AP window



Echocardiography is usually diagnostic. The parasternal short-axis view at the level of ascending aorta and main pulmonary artery can demonstrate a defect using 2D imaging (Fig. 3.1), which can be confirmed on colour Doppler. Sometimes colour Doppler can mislead as it can ‘bleed’ between adjacent structures. Parasternal long-axis and subcostal views are also useful to delineate the defect. A comprehensive echocardiogram looking for associated lesions is essential.

Cardiac catheterisation is reserved for those patients with suspected pulmonary vascular disease in order to assess reversibility of the pulmonary vascular resistance and whether surgery is appropriate. Cardiac MRI scan can be a useful adjunctive investigation.

Imaging Pitfalls

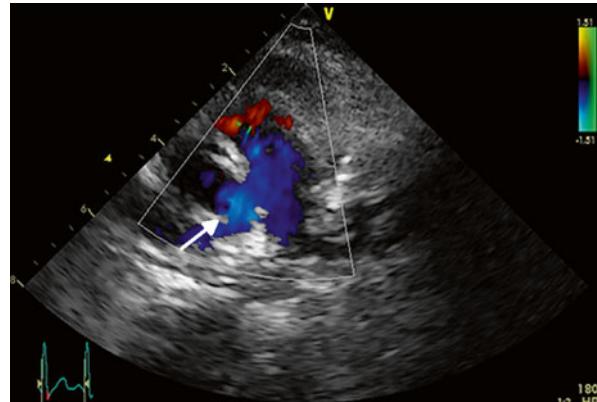
It is common to see echo “dropout” in the blood vessel walls where AP windows can exist; this can make assessment by 2D imaging difficult. In addition, as the pulmonary vascular resistance is high in the newborn period, little flow across the defect may be observed on colour-flow Doppler. This might explain why the diagnosis is often missed. As in our case, it is easy to attribute the clinical findings to an associated patent arterial duct or ventricular septal defect (Fig. 3.2). In the absence of a shunt, if left heart volume overload is suspected, an unusual shunt such as an AP window must be actively ruled out.

It is also easy to mis-diagnose a patient as having an AP window when the lesion is not present because of the problem of “dropout.” Figure 3.3 shows the parasternal short-axis view of a 9-month-old child who presented with mild tachypnoea and failure to thrive. He had a pansystolic murmur and enlarged liver. Echocardiogram demonstrated a moderate-sized doubly committed subarterial ventricular septal defect with left heart enlargement. In addition, the echo appearances were suspicious of a distal AP window (see Fig. 3.3). The child underwent VSD closure under

Fig. 3.2 Apical four-chamber view showing dilated left atrium and left ventricle secondary to the large left to right shunt caused by the AP window. Note the prominent pulmonary veins which are secondary to the significantly increased pulmonary blood flow



Fig. 3.3 The arrow points towards colour continuation between the aorta and right pulmonary artery. Flow was not seen continuously throughout cardiac cycle. There was no AP window when inspected at the time of surgery to close the associated doubly committed subarterial VSD



cardiopulmonary bypass, but direct inspection by the surgeons ruled out a distal AP window. The volume overload was secondary to the VSD alone.

Management

Management of an AP window is surgical in most cases, although a few reports of use of transcatheter techniques have been reported for small AP windows or residual AP windows following surgical repair. Operative results are good in isolated AP windows, while reported mortality increases to 20–25 % with associated lesions depending on complexity. Reported surgical complications include distortion of the coronary arteries, distortion of the pulmonary artery and damage to aortic valve causing aortic regurgitation. The post-operative period can be complicated by pulmonary hypertensive crises. It is important to bear in mind that this lesion is a very rare condition and even in high surgical volume centres, there are no more than a

few cases annually. Diagnosis depends on a high index of suspicion. Prognosis depends on the timing of diagnosis and associated lesions.

Learning Points

- In cases that present with pulmonary over-circulation with apparently normal cardiac structure, one should look for rarer congenital heart conditions.
- Rare lesions such as AP window can co-exist with other more common intracardiac lesions.

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Chapter 4

Dilated Cardiomyopathy: If You Don't Suspect, You Can't Diagnose!

M. Kanagaratnam and Pavanasaam Ramesh

Abstract Dilated cardiomyopathy (DCM) is a rare but a serious disease in children. The majority of children present in heart failure due to impaired left ventricular function, a smaller proportion present with arrhythmia and rarely some are found to have asymptomatic cardiomegaly. Treatment of dilated cardiomyopathy is rarely curative but is directed at improving symptoms and long-term outcome. We present the case of a 17 month old boy who presented with cardiac failure secondary to dilated cardiomyopathy; a number of his symptoms and signs were similar to common childhood conditions resulting in a delay in diagnosis and initiation of appropriate anti-failure therapy.

Keywords Heart failure • Congestive cardiac failure • Dilated cardiomyopathy • Viral illness • Liver failure • Diuretics • Lactic acidosis • Peripheral oedema

Case Description

A 17-month-old boy, weighing 13.4 kg, presented to his local children's assessment unit, with nonspecific symptoms. He was born at full term after an uneventful pregnancy with a birth weight of 4.1 kg and had been growing and developing appropriately. He was not taking regular medications and his immunisations were up to date. He had a 3 ½-year-old brother who was fit and well. Both parents were white Caucasians and non-consanguineous.

Further history revealed that he has been unwell with lethargy, decreased appetite and reduced fluid intake for 2–3 weeks. His mother had noticed mild puffiness around his eyes and hands for the past week and a rash had developed the day he

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presented to the assessment unit. History was negative for cough, coryza, diarrhoea, vomiting or fever. He had travelled abroad 6 weeks previously.

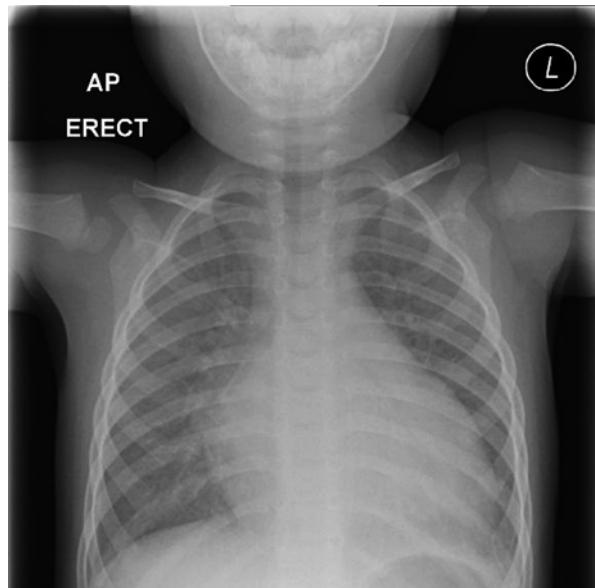
Observations on admission showed respiratory rate 52 breaths per minute, percutaneous oxygen saturations of 95 % in air; heart rate, 135 beats/min.; blood pressure, 117/86 mmHg; capillary refill time, 3 s; and axillary temperature, 36.5 °C. On examination he looked unwell, lethargic and pale, preferring to keep his neck in an extended position. His eyes and hands were mildly puffy. There were scattered blanching and non-blanching maculopapular lesions over his back, abdomen and face. There was no generalised lymphadenopathy. Kernig's sign was negative and there was no sign of neck stiffness. He had good peripheral pulses, normal first and second heart sounds with no murmurs. There was no subcostal or intercostal recession and chest was clear with no crackles or wheeze. Abdomen was soft, not distended, and non-tender with a palpable soft liver edge 3 cm below the right costal margin. There was no splenomegaly. Neurological, ENT and musculoskeletal examination were normal.

The initial impression was infection most probably due to a virus; the differential diagnosis was acute leukaemia. His urine was negative for protein and infection. Blood results were: haemoglobin 117 g/L; white cell count 15.6×10^9 /L; neutrophils 7.2×10^9 /L; platelets 318×10^9 /L; INR, 1.9; APTT, 1.08; blood glucose 5.2 mmol/L; serum sodium 136 mmol/L; potassium 4.8 mmol/L; urea 7.2 mmol/L; creatinine 31 µmol/L; C-reactive protein <4 mg/L; albumin 38 g/L; alkaline phosphatase 263 IU/L; alanine transaminase 134 IU/L; gamma glutamyltransferase 64 IU/L; bilirubin 19 µmol/L. Peripheral blood smear was normal with no blast cells.

He was treated with intravenous antibiotics (cefotaxime) for possible bacteraemia, especially in view of presence of the non-blanching rash and a dose of vitamin K was given for his deranged INR. He was allowed to have free fluids and a normal diet. On the following day, there was no obvious change in his clinical status however he remained very lethargic. He was clingy, not playful and reluctant to engage in any activity. All his vital signs (heart rate, respiratory rate, blood pressure and temperature) remained stable. His Paediatric Early Warning Score (PEWS) was one meaning a clinically stable child (a high score over of four requires clinical review).

As his clinical condition remained static with no improvement after 24 h, a second opinion was sought from another general paediatric consultant. His history and examination findings were the same as those on admission with the liver still palpable 3 cm below the right costal margin and no extension of the rash. Repeat blood tests showed an increase in alanine transaminase to 167 IU/L from 134 IU/L, gamma glutamyl transferase 109 IU/L from 64 IU/L, INR 1.5, and APTT 0.98, with renal function and full blood count remaining unchanged. Capillary blood gas showed a pH 7.36, pCO₂ 4.4 kPa, pO₂ 5.38 kPa, base deficit 6.1 mmol/L, bicarbonate 19.1 mmol/L, lactate 4.0 mmol/L, glucose 6.2 mmol/L, sodium 138 mmol/L, potassium 5.1 mmol/L, calcium 1.16 mmol/L and chloride 110 mmol/L. The raised lactate and high normal potassium values were attributed to a squeezed capillary

Fig. 4.1 Chest radiograph showing cardiomegaly and pulmonary venous congestion



blood sample obtained from his foot. A laboratory sample sent later showed normal potassium of 4.5 mmol/L, repeat lactate was not performed.

All tests for infection were negative, including meningococcal and pneumococcal PCR, throat swab, blood cultures and viral screening for CMV and EBV.

In view of the hepatomegaly and deranged liver function tests, abdominal ultrasound was arranged. This showed dilated hepatic veins, echogenic enlarged liver and peri-cholecystic fluid suggesting congestion. The radiologist suggested the team explore the possibility of any underlying cardiac cause which prompted the chest radiograph shown in Fig. 4.1. The heart was significantly enlarged, with an increased cardiothoracic ratio and evidence of pulmonary venous congestion.

Echocardiogram showed normal situs and cardiac connections, with poor biventricular function and marked tricuspid and mitral valve regurgitation. There was no outflow tract obstruction or aortic coarctation and the coronary arteries had normal origins. Ejection fraction was estimated at 24 % and fractional shortening at 10 %. A diagnosis of severe dilated cardiomyopathy was made. Clinically he remained stable; however, there was some increase in capillary lactate to 4.9 mmol/l, and he was transferred to the paediatric intensive care for initiation of dobutamine and diuretics prior to transfer to the regional tertiary cardiac centre.

He is currently aged 4 years and remains on medical therapy. Since diagnosis he has had two cardiac arrests both precipitated by viral illnesses, he is known to the cardiac transplant team but has not been listed for cardiac transplant.

Discussion

Dilated cardiomyopathy (DCM) is the most common form of heart muscle disease in children accounting for approximately 55–60 % of all childhood cardiomyopathies. Outcomes are difficult to predict and depend on cause, severity and age at presentation.

It is a relatively rare condition in general paediatric population with a reported incidence of 0.57 cases per 100,000 in the United States and 2.6 cases per 100,000 in Finland. Infants can present with a history of irritability, poor feeding leading to failure to thrive, increased breathing effort, pallor, decreased urine output and sweating on activity. Older children tend to present with reduced exercise tolerance, shortness of breath on minimal exertion, recurrent chest infections and chronic cough. Presentation of DCM can also sometimes be in the form of arrhythmia with symptoms of syncope, palpitations, and seizures and in some cases the first presentation can be a cardiac arrest.

Signs of heart failure include tachycardia, prolonged capillary refill time, gallop rhythm, heart murmur (functional mitral regurgitation), elevated jugular venous pressure, hepatomegaly, peripheral oedema and tachynoepea. Diagnosis of cardiomyopathy is made easier once a cardiac diagnosis is suspected. Chest radiograph and ECG are required; however, the principal investigation is echocardiography, which confirms the diagnosis, assesses degree of dysfunction and rules out structural underlying causes. It is vitally important to diagnose treatable causes of cardiomyopathy (see other chapters); however, in most cases the cardiomyopathy screen is negative. Rarely, DCM can be secondary to metabolic, storage, neuromuscular, endocrine, and mitochondrial disorders. Blood tests may include: lactate, glucose, ammonia, amino acids, carnitine, acylcarnitine, cholesterol, triglycerides, thyroid function, full blood count, creatinine kinase, thiamine, selenium, calcium, vitamin D and parathyroid hormone. Urine should be checked for amino-acids, organic acids and glycosaminoglycans to further exclude metabolic disease. DCM may be due to previous myocarditis and it may be useful to check viral titres against enterovirus especially coxsackie B, parvovirus B19, human herpes virus 6, adenovirus, rubella and HIV. Dilated cardiomyopathy can be familial, and it is estimated that 20–30 % of children with DCM have a relative with the disease. However, unlike hypertrophic cardiomyopathy, DNA mutation analysis is not currently useful clinically and is of low yield.

Angiography, CT, or cardiac magnetic resonance imaging is preformed when there is diagnostic uncertainty in ruling out anomalous coronary artery from the pulmonary artery. Cardiac catheterisation and haemodynamic studies are also used to assess pulmonary vascular resistance to help assess suitability for heart transplantation. Myocardial biopsy is controversial and not without risk; however, it can help to investigate cause of myocardial dysfunction (e.g., myocarditis).

Whatever the cause, management of dilated cardiomyopathy is to give supportive relief which may include positive pressure ventilation and inotrope support. Phosphodiesterase III inhibitors (e.g., milrinone) are particularly helpful as they

reduce afterload and improve ventricular contractility without increasing myocardial oxygen consumption. Diuretics should also be started; furosemide is often required intravenously as well as oral spironolactone. Any reversible causes of cardiomyopathy must be aggressively treated, e.g., vitamin D deficiency. When stability has been reached, the child is converted to oral diuretics, ACE-inhibitors and beta-blockers such as carvedilol. The child should also be anticoagulated with aspirin or warfarin to reduce the risk of thrombosis forming in the ventricle with risk of embolus. Detailed discussion of management is beyond the scope of this chapter. In some cases, children fail to thrive and deteriorate despite medical support and cardiac transplantation should be considered.

In our patient the diagnosis of dilated cardiomyopathy was delayed. This could be attributed to the nature of his presentation, which was similar to a variety of common paediatric diseases. He did not have abnormal cardiovascular examination findings such as a murmur or absent femoral pulses, and his vital signs were relatively normal. This put a cardiac cause low in the differential diagnosis. However, there were subtle signs that should have prompted consideration of a cardiac problem, including pallor, lethargy, puffy eyes and mildly enlarged liver with raised enzymes. Instead each symptom was linked to a specific condition (e.g., puffy eyes and nephrotic syndrome, abnormal liver function tests to primary liver pathology) rather than considering the single aetiology of heart failure.

Another key lesson from this case description is the assessment of blood lactate level. An isolated rise in lactate with clinical presentation such as lethargy should always prompt the possibility of an underlying cardiac cause. Although plasma lactate also can be raised in non-cardiac conditions such as global or regional ischaemia, high metabolic states (seizure, exercise, increased work of breathing), drugs, toxins, malignancy, liver disease, diabetic ketoacidosis, thiamine deficiency and inborn errors of metabolism. Since we did not suspect an underlying cardiac condition, we ignored the high lactate level and attributed it to be secondary to a squeezed capillary blood sample causing release of lactate from lysed red blood cells. Ideally, if there is any suspicion, then a repeat free flowing capillary or venous sample should be checked to confirm the lactate levels. The gold standard in assessing lactate is an arterial blood sample, but this can be difficult to obtain and can cause significant discomfort to the child.

Learning Points

- Underlying cardiac disease must be considered in any unwell child.
- Heart failure in children may present with non-specific symptoms and signs.
- Any child presenting with high blood lactate should have an underlying cardiac condition ruled out.
- Cardiac failure should be included in the differential diagnosis for hepatomegaly and liver dysfunction

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Chapter 5

Syncope: It's All in the History

Sarah Boynton and Vinay K. Bhole

Abstract Syncope is a transient, but complete, loss of consciousness due to global cerebral hypo-perfusion. It has rapid onset, short duration and recovery is generally quick, spontaneous and complete. Most syncopal episodes in children are benign in nature and are secondary to reflex syncope and orthostatic hypotension. Clues to the cause of syncope are usually found from taking a careful history. “Red flag” symptoms for cardiac syncope that need direct referral to a paediatric cardiologist include syncope during exertion or emotional stress. We describe two children who had syncope due to a serious arrhythmia. In both children history taking was vital.

Keywords Sudden death • Syncope • Catecholaminergic polymorphic ventricular tachycardia

Case Description 1

A previously fit and healthy 10-year-old girl presented on two occasions to her local hospital following episodes of syncope. The first episode occurred while running in a race at school and was witnessed by her teacher. During the race she collapsed to the ground, was incontinent and then recovered quickly. She described feeling light headed at the time. She was taken to her local hospital, and a 12-lead ECG was performed that showed a sinus bradycardia.

The second episode occurred 4 months later. Again she had been running in a race, and she had just finished running when she collapsed. She was unconscious for a few seconds, and witnesses described that she made a few shaky movements

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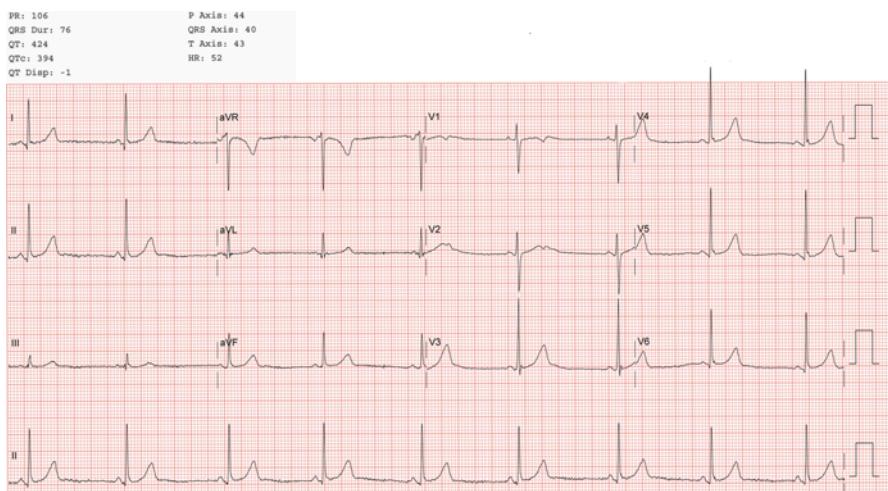


Fig. 5.1 Baseline 12-lead ECG showing sinus bradycardia

whilst she was unconscious. Upon waking she was pale and disorientated. She was taken to her local hospital and on arrival had completely recovered. Another 12-lead ECG was performed that showed a sinus bradycardia. Following this second episode she was referred to paediatric cardiology outpatients.

She was an active and healthy girl who enjoyed multiple activities including swimming, tennis and skating. She did not have any known medical conditions, and there was no cardiac family history. There was no history of palpitations or chest pain. She had a normal cardiovascular examination.

Twelve-lead ECG showed sinus bradycardia (Fig. 5.1), heart rate 52 beats per minute. Echocardiogram showed normal cardiac structure and function.

An exercise test was performed, and she exercised for 11 min and 59 s on the Bruce protocol. Her resting heart rate before starting the test was 55 beats per minute rising to a maximum of 173 beats per minute during peak exercise. There was an appropriate blood pressure response to exercise reaching 165/67 and returning to baseline upon rest. At the start of the test her ECG showed normal sinus rhythm but on exertion once her heart rate reached 140 bpm frequent ventricular ectopics began to appear. At 150 bpm ventricular couplets were seen and as her heart rate rose further these became more frequent and bi-directional in nature. At that point the test was terminated (Figs. 5.2 and 5.3).

A diagnosis of catecholaminergic polymorphic ventricular tachycardia (CPVT) was made and she was prescribed atenolol. Since the introduction of medical therapy repeat exercise testing has shown a reduced amount of ventricular ectopy on exercise and she has had no further episodes of collapse.

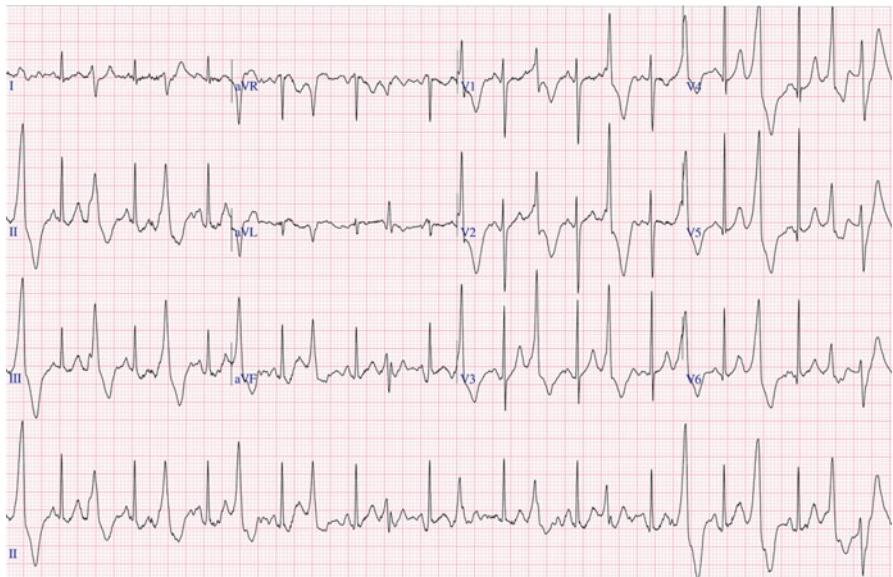


Fig. 5.2 Frequent ventricular ectopic beats during exercise ECG



Fig. 5.3 Frequent ventricular couplets of bidirectional nature during exercise ECG

Case Description 2

An 8-year-old boy who had been living with his grandparents abroad moved to the UK to live with his parents. Three months later, the boy had gone to bed as usual when his mother heard a scream coming from his room. When she went to investigate she found him collapsed on the floor. She called for an ambulance and started CPR. The first responder arrived after 5 min and found the boy to be in pulseless electrical activity; he continued basic life support. When the paramedic crew arrived the boy was intubated, resuscitation was continued, and by 17 min cardiac output had returned. The boy was air-lifted to the children's hospital for further management.

On arrival in the emergency department the boy had a heart rate of 140–190 bpm, showing a variable rhythm with runs of monomorphic and polymorphic ventricular tachycardia (Fig. 5.4), supraventricular tachycardia, bigeminy and trigeminy. The rhythm changes were noticed to be associated with the painful stimuli such as suction and blood sampling. The boy tolerated the rhythm changes well with no further loss of cardiac output. He was transferred to the intensive care unit where he was cooled for 24 h. During this time his electrolytes were optimized and he was started on lidocaine and esmolol infusions. After 24 h he was re-warmed, sedation was reduced, and he was successfully extubated. The esmolol infusion was changed to oral propranolol and other infusions discontinued. He went on to make a good recovery, and was discharged with no evidence of neurological injury. He has continued to do well and is currently managed on oral flecainide and propranolol.

More information of the child's background began to emerge while he was in hospital. A letter from his grandparents indicated that the boy had fainted several times before. He had been referred to a cardiology centre in his home country where he had undergone an exercise test and subsequently diagnosed with CPVT. He had been prescribed 10 mg propranolol three times a day. His grandparents understood that the propranolol would not cure the problem and had decided to stop propranolol and treat him with traditional alternative local medications. When he arrived in the UK to live with his parents he registered with a General Practitioner. His parents mentioned he had a heart condition and a non-urgent outpatient referral was made to paediatric cardiology. He was awaiting the appointment and had remained symptom free in the UK until the night he collapsed.

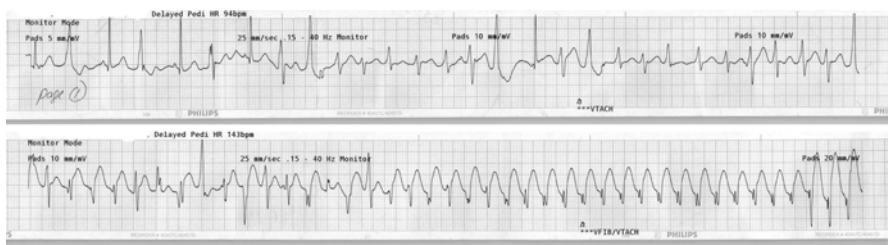


Fig. 5.4 Rhythm strip in A&E showing ventricular ectopy and runs of ventricular tachycardia

Discussion

Background

Catecholaminergic polymorphic ventricular tachycardia is an inherited disorder characterised by adrenergic-mediated induction of polymorphic ventricular tachycardia in a structurally normal heart.

CPVT results from inherited defects of intracellular calcium handling in cardiac myocytes. Four genetic variants of CPVT have been identified (RyR2, CASQ2, TRDN, CALM1). A mutation in RyR2 or CASQ2 is identifiable in approximately 60 % of cases of CPVT. TRDN and CALM1 are much rarer and account for less than 1 %. Patients that are gene-positive for RyR2 are known as having CPVT1, and those gene-positive for CASQ2 are sometimes referred to as having CPVT2.

Presentation

CPVT usually presents with a syncopal episode in response to exercise or emotional stress. Approximately 60 % of cases will have had their first episode of syncope or cardiac arrest before they are 20 years old. The mean age of onset is between 7 and 12 years of age, but cases presenting in the fourth decade of life have been reported.

Diagnosis

The key presenting factor is syncope on exercise or during emotional distress, so history is vitally important to make the diagnosis. However, episodes can happen at other times, as demonstrated in second case. Clinical examination, resting ECG and echocardiogram are usually normal. The arrhythmia is reproducible on exercise, so exercise stress ECG is the diagnostic test of choice, with the characteristic polymorphic VT (bidirectional at times) becoming apparent upon stress testing as in our first case. The onset of arrhythmia tends to become apparent when heart rates reach 100–120 bpm. With increased workload the complexity of the arrhythmia progresses from isolated ectopic beats to bigemeny to runs of non-sustained ventricular tachycardia, and if exercise continues there may be progression to sustained VT and cardiac arrest.

Molecular genetic testing is available and due to the patterns of inheritance family testing is also advised.

Management

Life Style Changes

- Limit /avoid competitive sports.
- Limit/avoid strenuous exercise.
- Limit exposure to highly emotional or stressful situations.

Medical Therapy

Recommended for all patients with a symptomatic diagnosis of CPVT. The first-line therapeutic option for patients with CPVT is beta-blockers without intrinsic sympathomimetic activity combined with exercise restriction. Beta blockade is thought to be effective in 60 % of cases. Nadolol is the preferred option for prophylactic therapy and has been found to be clinically effective. The dosage used is usually quite high (1–2 mg/kg) and control of the arrhythmia is often dependent upon compliance.

If beta blockade alone is not sufficient to control arrhythmia, recent evidence suggests that Flecainide should be considered as a second-line agent.

ICD Implantation

Although pharmacological treatment is documented as being highly effective, religious compliance is necessary. Research has shown that a minority of patients continue to have symptoms despite medication and an implantable defibrillator may be considered. ICD may also be considered for secondary prevention in patients who have suffered a cardiac arrest. However, recent retrospective analyses of patients with CPVT with an ICD have shown a high burden of both appropriate and inappropriate shocks in addition to the usual problems of ICD technology in young patients, so such decisions to implant must be carefully considered. If an ICD is implanted it is important that medical therapy needs to be continued to reduce the frequency of appropriate shocks. ICD shock itself can trigger ventricular tachycardia storm and hence decision to implant ICD should be weighed carefully in this condition.

Left Cardiac Sympathetic Denervation (LCSD)

This may be considered in CPVT patients who experience recurrent syncope, polymorphic/bidirectional VT, or multiple appropriate ICD shocks while on medications or who are intolerant of beta-blocker therapy. Recurrence of cardiac events has

been reported in those who have undergone LCSD; therefore, medical therapy must continue.

Learning Points

- Careful history taking is vital when assessing a patient with syncope.
- Syncope on exertion and/or emotional stimuli are red flags that there may be an important underlying arrhythmia.
- Baseline ECG and echocardiogram are usually normal in CPVT.
- Increasing polymorphic ventricular ectopy on exertion is a sign of CPVT.
- CPVT is an important diagnosis and cause of cardiac arrest, patients need to be on medications.

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Chapter 6

Chest Pain in Children: Not Always Benign

Paraskevi Theocharis and Alan G. Magee

Abstract A 15-year-old African male presented with rheumatic heart disease after moving to the UK. He developed severe mitral and aortic valve disease and underwent replacement of both valves using mechanical prostheses. Compliance and clinic attendances were poor, but he remained well for 20 months when he began to experience chest pains with dizziness. Echocardiogram was unremarkable and while awaiting outpatient exercise testing he collapsed with severe left ventricular failure. ECG showed ischaemia and troponin was significantly elevated. Angiography showed mechanical left main stem compression caused by the mechanical aortic valve ring.

Keywords Exertional chest pain • Children • Aortic valve replacement • Coronary compression • Left ventricular failure

Case Description

A 15-year-old male emigrated from Gambia to the UK at the age of 15. His sister was known to have rheumatic carditis. He presented with a 2–3-week history of intermittent chest pain, dyspnoea on exertion and haemoptysis. ECG showed lateral ST depression, and echocardiogram showed mitral stenosis with a mean inflow gradient of 14 mmHg as well as mixed aortic valve disease with moderate regurgitation and a Doppler-derived pressure drop of 64 mmHg. The left ventricle was hypertrophied and the valve disease was thought to be rheumatic in origin.

Three months later elective double valve replacement was performed using a 27-mm On-X valve in the mitral position and a 21-mm On-X valve in the aortic position. A transoesophageal echocardiogram at the conclusion of the procedure

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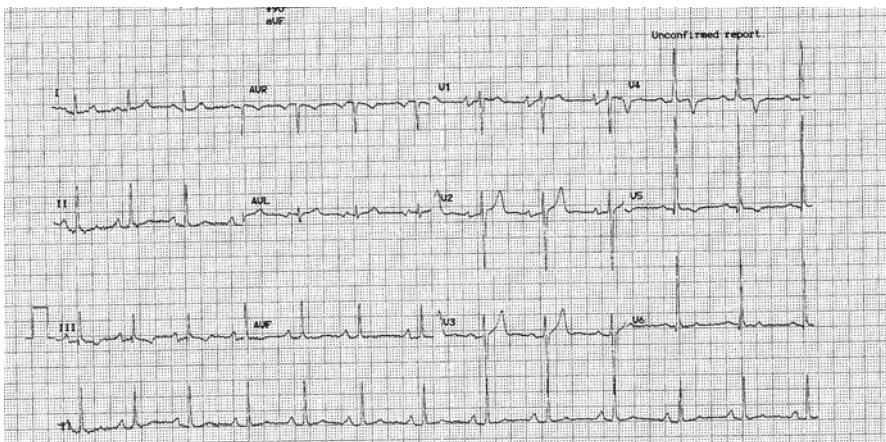


Fig. 6.1 ECG on presentation at the local hospital two and a half years after surgery showing T-wave inversion in mid-precordial leads

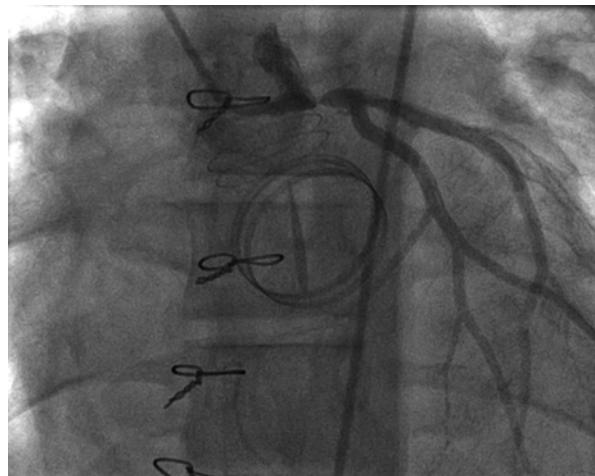
showed good biventricular function, well-seated prosthetic valves with no paravalvar leaks and normal proximal coronary flow. The post-operative period was uneventful and anticoagulation established. Compliance with medication and clinic attendances were poor, with no documented INR results within the reference range.

He re-presented at his local hospital two and a half years after surgery with chest pain and dizziness on exertion. ECG showed new T wave inversion in the mid precordial leads (Fig. 6.1) and a raised troponin level of 1.69 ug/L (reference range <0.04). He continued to experience chest pain and dizziness; however, troponin levels gradually decreased and he was allowed home. On review he appeared clinically well with no evidence of heart failure. ECG T-wave changes persisted but echocardiogram was unremarkable with no regional wall motion abnormalities. His symptoms were not thought to be cardiac in origin; however, an outpatient exercise tolerance test was arranged that he failed to attend.

Three months later he developed sudden shortness of breath while walking home; on arrival he collapsed and was coughing blood-stained sputum. He was brought to Accident and Emergency by ambulance, receiving 15 L O₂/min by face mask but remained hypoxic, although conscious. Arterial lactate was 4 mmol/l, troponin was grossly elevated and echocardiogram showed globally poor left ventricular function. Chest CT showed no evidence of pulmonary embolus but revealed bilateral lower lobe consolidation. Dobutamine was commenced and urgent cardiac catheterisation arranged. The working diagnosis was myocarditis or coronary embolism, possibly related to poor INR control.

Catheterisation was performed under local anaesthesia and he experienced chest pain during the procedure. The right coronary artery was normal; however, the left main stem was severely narrowed where it crossed over the ring of the On-X valve in the aortic position (Fig. 6.2), and the valves were functioning normally. The following day he was taken to the operating theatre, where he underwent patch

Fig. 6.2 Snapshot of the cardiac catheterization showing mechanical compression of the left main stem at the point of crossing over the ring of the On-X valve in the aortic position



repair of the left main stem and grafting of the left internal mammary to LAD. He made an uneventful recovery and left ventricular function gradually improved.

Discussion

Chest pain is common in children and accounts for 10–15 % of ambulatory cardiology visits but in contrast to adult practice very rarely represents cardiac pathology and is usually musculoskeletal, pulmonary, gastrointestinal or psychogenic in origin (Table 6.1). Cardiac causes of chest pain in children and adolescents are found in less than 5 % of presentations but one must remain alert to the possibility.

The lack of evidence-based standards for the evaluation of chest pain in paediatric patients has led to a widespread variation in practice among cardiologists, with exercise stress testing, echocardiography and ambulatory ECG commonly requested. In the absence of any cardiac history all appear to have a very low yield. Previous studies have shown that exercise stress testing was not able to detect any cardiac disorders in otherwise healthy children and adolescents. Ambulatory ECG is also low-yield, except in the presence of palpitations or syncope, and echocardiography is more likely to reveal incidental findings. However, in the presence of a concerning history such as previous cardiac surgery, exertional pain and/or collapse, together with clinical and ECG findings, echocardiography can lead to the diagnosis of significant cardiac lesions such as anomalous coronary artery from the pulmonary artery, cardiomyopathy, pulmonary hypertension, myocarditis, pericarditis, and left severe ventricular outflow tract obstruction. Cardiac biomarkers, including cardiac troponin, are not routinely recommended in the evaluation of chest pain in children. However, they are indicated for selected patients with suspected myocarditis, pericarditis and coronary ischaemia. Table 6.2 summarises the cardiac testing for paediatric chest pain.

Table 6.1 Causes of non-cardiac chest pain in paediatric patients

| Aetiology | |
|---------------------------|--|
| Musculoskeletal (50–68 %) | Costochondritis Muscle strain Trauma Slipping rib syndrome |
| Respiratory (3–12 %) | Asthma Pneumonia Bronchitis Pleuritic pain Pulmonary embolus Pneumothorax |
| Psychogenic (10–20 %) | Conversion disorder Panic/anxiety attack |
| Gastrointestinal (2–8 %) | Oesophagitis Gastritis Gastroesophageal reflux Pancreatitis Gastric ulcer Biliary colic/disease |
| Other (<10 %) | Acute chest syndrome of sickle cell disease Skin infection Breast disease |

Table 6.2 Cardiac investigations indicated for chest pain

| Test | Indication | Utilization | Suspected diagnosis |
|-------------------------|--|--|---|
| ECG | Abnormal physical examination Exertional chest pain | Selected patients | Cardiomyopathy, myocarditis, pericarditis, previous cardiac surgery, pulmonary HTN |
| Echocardiography | Abnormal physical examination Abnormal ECG Family cardiac history Exertional chest pain | Selected patients | Anomalous coronary artery origins, cardiomyopathy, myocarditis, pericarditis, previous cardiac surgery, pulmonary HTN, left ventricular outflow obstruction |
| Troponin | Suspected myocarditis, pericarditis | Selected patients | Myocarditis, pericarditis, coronary ischaemia |
| Ambulatory ECG | Chest pain and palpitations | Rarely useful for chest pain | Atrial or ventricular arrhythmias, conduction abnormalities/heart block |
| Exercise stress testing | Exertional chest pain and exertional syncope and/or palpitations | Rarely useful in patients with no previous cardiac history | Coronary ischaemia, exercise-induced arrhythmias |

In this case there were numerous red flag findings suggesting that the patient's chest pain was of cardiac aetiology. The previous history of cardiac surgery, the abnormal ECG findings and the positive cardiac troponin in a paediatric patient presenting with chest pain, although atypical, should have triggered further evaluation, particularly to assess the coronary arteries. Although the prevalence of coronary ischaemia as a cause of chest pain in paediatric patients is extremely low, it should never be completely discounted.

Previous aortic valve replacement is known to be associated with occlusion of both the left and right coronary arteries, resulting in myocardial ischaemia. Furthermore, the risk of coronary embolisation is high when mechanical prostheses are implanted and therapeutic targets for anticoagulation are not met. The presence of a mechanical prosthetic aortic valve, positive cardiac biomarkers and the sub-therapeutic levels of anticoagulation should have raised the suspicion of myocardial ischaemia. Investigations for chest pain in a paediatric patient with a previous cardiac surgery, especially when compliance with anticoagulation is poor, should be performed in a timely fashion.

Learning Points

- Chest pain in paediatric patients is common but is rarely associated with significant underlying cardiac pathology and cardiac investigations are usually of low yield. A detailed history is vital when evaluating patients with chest pain, especially concentrating on pain on exertion, associated collapse or palpitations, previous cardiac intervention and family history.
- There is no clear consensus, and there are significant variations regarding the approach to paediatric chest pain; however, diagnostic algorithms can help.
- A history of previous cardiac surgery, new ECG findings and a raised troponin level merit further investigation.

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Chapter 7

Coronary Artery Imaging Is Crucial

Georgia Spentzou and Benjamin G. Smith

Abstract A 6-month-old girl presented in cardiac failure and was found to have a large, poorly contractile left ventricle. A convincing appearance of a normal left coronary artery was subsequently found to be misleading, with the artery in fact arising from the main pulmonary artery. Left ventricular damage was severe at presentation and despite immediate surgery the child ultimately succumbed. Anomalous left coronary artery from the pulmonary artery, previously referred to as Bland-White-Garland syndrome, is a rare condition, easily missed if fastidious attention is not given to identifying the origin of the left coronary artery and direction of blood flow within it. Prompt recognition and surgery offer the best chance of a successful outcome.

Keywords ALCAPA • Bland-white-garland syndrome • Left coronary artery • Dilated cardiomyopathy

Case Description

A 6-month-old girl presented with a 5-day history of poor feeding on a background of failure to gain weight over the preceding month. She was pale, desaturated, tachycardic, tachypnoeic and grunting, with a laterally displaced apex beat, parasternal heave and hepatomegaly. A chest x-ray demonstrated gross cardiomegaly. On ECG there was sinus rhythm with widespread T-wave inversion, no elevation of ST segments, a widened QRS complex of 120 ms, a left ventricular strain pattern and deep lateral q waves. Frequent ventricular ectopics were noted (Fig. 7.1). The echocardiogram demonstrated a very enlarged left ventricle measuring 60 mm at end-diastole, associated with severely reduced contraction. The left atrium was also

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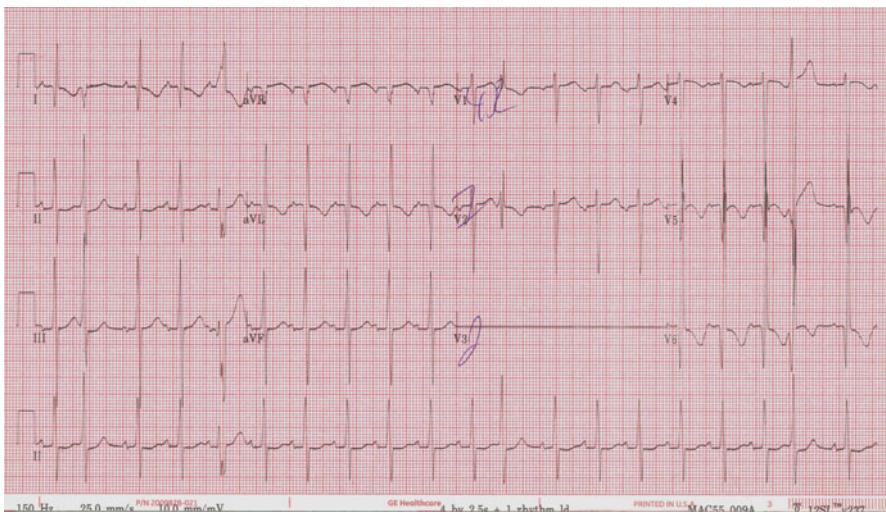


Fig. 7.1 ECG demonstrating sinus rhythm with lateral T wave inversion and ventricular ectopy

grossly dilated. There was moderate mitral regurgitation due to failure of the valve leaflets to co-apt. Both the right and left coronary arteries were demonstrated, with a convincing appearance of a normal left coronary artery (LCA) origin showing antegrade flow (Fig. 7.2a, b).

A diagnosis of dilated cardiomyopathy was made. The child was intubated and ventilated, and managed with inodilation, adrenaline and isoprenaline for bradycardia. A cardiomyopathy screen returned normal. The child's condition did not improve and the ECG developed progressive ST changes consistent with ischaemia as well as frequent ventricular ectopies and runs of tachycardia. The echocardiogram was repeated specifically to re-assess the LCA. The original appearance of a normal connection and direction of flow could be reproduced but was in fact misleading. The vessel originally thought to be the LCA was instead a small branch of the right coronary artery (RCA) coursing in the direction the LCA would ordinarily take, and therefore showed antegrade flow. A much larger vessel, the LCA, could be seen arising from the pulmonary artery. The direction of flow was retrograde (see Fig. 7.2c, d). A diagnosis of anomalous left coronary artery from the pulmonary artery (ALCAPA) was made. Reduction of the adrenaline and isoprenaline infusions resulted in less ventricular ectopy, and surgery was undertaken the same day.

At operation there was extensive collateralisation around the aorta and pulmonary artery, possibly accounting for the initial echocardiographic impression of a normal LCA. The LCA was transferred to the aorta, but despite good flow being seen on epicardial echocardiogram myocardial performance remained poor. The child was weaned off cardiopulmonary bypass, but due to the advanced state of left ventricular damage suffered persistent ventricular fibrillation and was placed on extra-corporeal membrane oxygenation (ECMO) prior to transfer to the intensive

Fig. 7.2 Initial echocardiogram demonstrating apparently normally arising LCA (arrow) from the aorta (a) with antegrade flow (b). Subsequent echocardiogram demonstrating true left coronary artery (c) and retrograde flow (d)

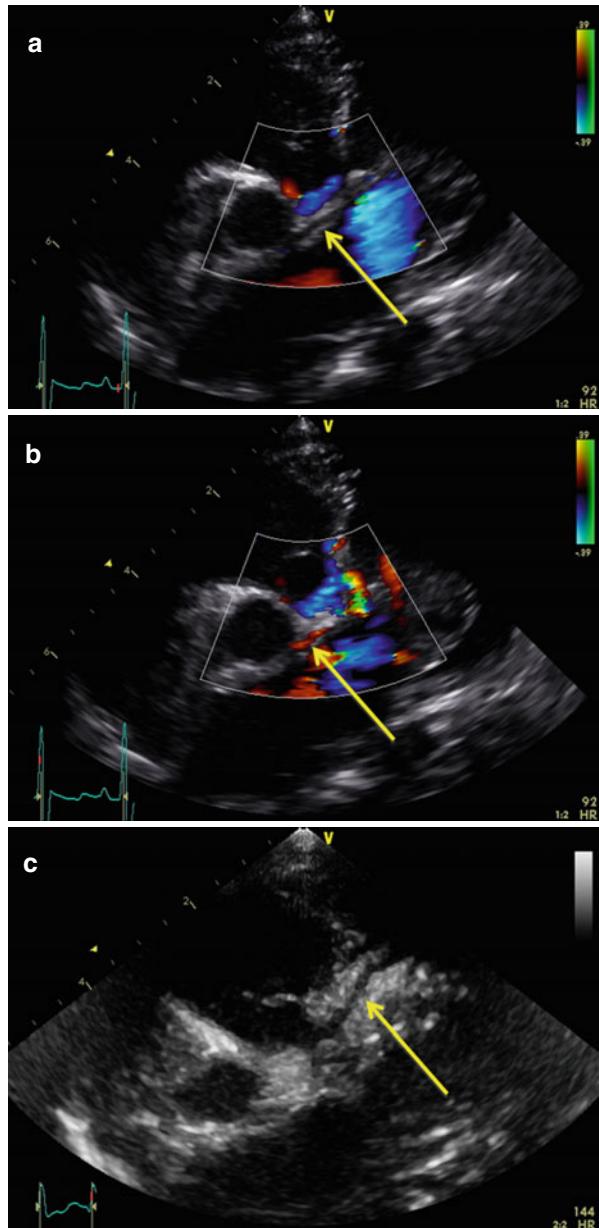
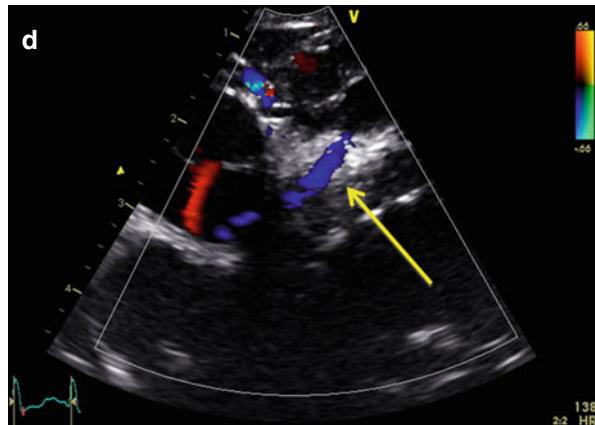


Fig. 7.2 (continued)

care unit. A week later the patient was separated from ECMO; however, she remained fully dependent on inotropic support and positive pressure ventilation, and experienced frequent ventricular tachyarrhythmias requiring resuscitation. Five weeks post-operatively one such arrest necessitated re-institution of ECMO support. Ventricular dysrhythmias continued despite a continuous amiodarone infusion and fastidious management of electrolyte balance.

There were grave concerns over the extent of irreversible myocardial damage by the time of presentation, and that transplantation represented the only possibility of survival. The patient was transferred to a transplant centre and support converted to a biventricular Berlin Heart. She was listed for urgent cardiac transplantation; however, she developed complications of mechanical circulatory support, including bacterial infection and evidence of neurologic injury manifested as choreoathetoid movements upon weaning of sedation. She also had persistent atelectasis of the left lower lobe due to gross cardiomegaly-induced compression of the left main bronchus. An initial CT demonstrated only a small sub-acute subdural haemorrhage; however, she subsequently became less responsive and developed fixed dilation of the left pupil. A repeat CT demonstrated a devastating intracranial haemorrhage. Following discussion with the patient's parents, active support was withdrawn and care re-oriented.

Discussion

Rapid diagnosis of ALCAPA is important for two reasons. Firstly, severe, irreversible damage to left ventricular myocardium may occur, resulting in death in up to 80 % of patients by 1 year of life. Secondly, this outcome can be avoided by prompt re-implantation of the left coronary artery into the aorta.

Normal Anatomy and Physiology

The LCA ordinarily divides into the left anterior descending (LAD) and the circumflex (Cx) arteries. The LAD artery lies anteriorly in the interventricular groove, while the circumflex artery courses posteriorly between the left atrium and ventricle. They account for the coronary arterial blood supply to the left atrium and ventricle. These arteries lie within the epicardium and give rise to smaller branches that penetrate the myocardium. The contraction of the myocardium in systole results in compression of these smaller branches, the highest degree of compression being in the subendocardial regions. Consequently, the filling of the coronary arteries is restricted during cardiac systole. In fact, filling of the LCA occurs exclusively in diastole. Subsequently, the diastolic aortic pressure and duration of diastole are important determinants of coronary artery perfusion. Any significant fall in the diastolic pressure may lead to ischaemia, the subendocardium being the area most susceptible to injury.

Pathophysiology

The exact location of the LCA ostium in ALCAPA varies. The most common site is the posterior-facing sinus of the pulmonary artery. Sometimes the LAD or Cx arises alone. Ectopic location is also possible with any coronary artery, with origin from the anterior left non-facing sinus, the main pulmonary artery, or the pulmonary branches reported.

Before birth, adequate myocardial perfusion in ALCAPA is maintained as the pulmonary artery pressure is similar to that in the aorta. As the pulmonary vascular resistance falls over 4–6 weeks following birth, pressure may initially remain sufficient to maintain adequate LCA perfusion. However, perfusion eventually falls in tandem with the pulmonary artery pressure until flow in the coronary artery reverses. Myocardial blood then drains into the pulmonary artery, resulting in coronary steal and subendocardial ischaemia. It is important to note damage to the myocardium is due to decreased coronary perfusion pressure rather than the coronary artery receiving de-oxygenated blood (as it does in transposition of the great vessels, in which ischaemia is not ordinarily seen). Endocardial fibroelastosis, myocardial fibrosis and infarction lead to ventricular dysfunction and heart failure, usually by the third month of extra-uterine life. Rarely, presentation is delayed until later in life, including adulthood, for a number of reasons. Collaterals may develop between the left and right heart coronary systems, preventing severe ischaemia. Alternatively, a right-dominant coronary supply, in which the inferoposterior left ventricular myocardium is supplied by the RCA system, may prevent critical ischaemia of sufficient myocardium to delay presentation. Similarly, a relatively small proportion of the left ventricle may be supplied by an anomalous vessel if only the LAD or Cx arises from the pulmonary artery. Acquired stenosis of the left coronary artery in older

patients may in fact be protective, by reducing the propensity to coronary steal. However, almost invariably in all patients there will be myocardial fibrosis and mitral regurgitation. This is typically directed posteriorly along the left atrial wall due to infarction of the posterior papillary muscle and consequent retraction and prolapse of the posterior leaflet.

Clinical Scenario

ALCAPA is a rare anomaly, accounting for no more than 1 in 250 cases of congenital heart disease. There is a preponderance in males. As coronary perfusion is maintained during uterine life, ALCAPA is rarely diagnosed antenatally. Typically the child presents in a similar fashion to the case described, though often at a younger age. Failure to thrive and signs of heart failure are usual. The physiologic stress of feeding can induce ischaemia, resulting in sweating, pallor and crying during feeds that may be misdiagnosed as colic. On examination, the child is breathless with hepatomegaly and a laterally displaced apex beat. Frequently, there is an apical, blowing pansystolic murmur of mitral regurgitation in association with a gallop rhythm. Rarely, ischaemia may not develop in infancy for the reasons described, resulting in a delayed presentation with angina pectoris or ventricular dysrhythmias.

A chest x-ray will typically demonstrate cardiomegaly with evidence of pulmonary oedema and pulmonary venous congestion. The ECG is critical. Almost invariably there will be a sinus tachycardia and ST segment changes consistent with either ischaemia or infarction, though this is not universal. Common features include deep (>5 mm) and wide (>0.03 ms) Q waves, ST segment elevation (>1 mm) and T wave inversion in the lateral leads (I, aVL, V₄-V₆ and occasionally II). Whilst there is variability in the T wave morphology in the first week of life, the T wave should be negative in lead V₁ and positive in V₅-V₆ thereafter. ECG changes may be similar to those seen in viral myocarditis, though the latter tends to result in low voltage complexes. Ventricular ectopy may be seen. As sufficient collateralisation may prevent or delay presentation, the absence of classic ECG findings should not necessarily lead to a diagnosis of dilated cardiomyopathy and discounting of ALCAPA; crucially, ALCAPA it is not excluded by a normal ECG. In older patients, the ECG may show increased left ventricular voltages, left axis deviation and poor R wave progression.

Echocardiography

The diagnosis may not be immediately evident. The left ventricle is dilated and poorly contractile. However, this is not discriminatory and is also seen in more common differential diagnoses such as coarctation of the aorta and dilated

cardiomyopathy resulting from other causes. All such diseases will have in common a normally arising left coronary artery with antegrade flow. There may be echogenicity of the myocardium and endocardial fibroelastosis, particularly of the posterior papillary muscle, in association with posteriorly directed mitral regurgitation. The parasternal short-axis view is the most likely to demonstrate the origin of the coronary arteries, which must be clearly visualised. Occasionally, a pericardial fold filled with fluid can be incorrectly mistaken for the origin of the left coronary artery. However, with clockwise rotation of the transducer the bifurcation of the LCA can be visualised. Colour Doppler must be applied to assess filling. A cardinal feature is retrograde flow in the LCA. An enlarged right coronary system can be a marker of ALCAPA, as are multiple collateral flows within the interventricular septum. In the absence of absolute confidence of a normal aortic origin of the LCA and antegrade filling, it is essential that coronary anatomy be demonstrated either on cross-sectional imaging or angiography. High-resolution ECG-gated multi-detector CT angiography usually provides diagnostic certainty, and has more or less replaced invasive angiography, which may be contraindicated in a child with poor ventricular function and haemodynamic compromise. Whilst CT cannot demonstrate the filling pattern of the coronary arteries, this is of lesser importance when anatomic certainty is assured. In contrast, MRI fast cine imaging does allow coronary steal to be demonstrated and is useful in grading mitral valve regurgitation. With delayed gadolinium enhancement areas of myocardial infarction and necrosis are demonstrated.

Management

Definitive management of ALCAPA is surgical, this being undertaken urgently, as soon as heart failure has been stabilised and the patient fit for cardiopulmonary bypass. Positive pressure ventilation is frequently beneficial in resting a compromised left ventricular myocardium. Sometimes stability cannot be achieved before operation, increasing the risk of the procedure. In extreme cases pre-operative ECMO support may be necessary. A careful balance of inotropic therapy, diuretics and afterload reduction is required. Managing a child with ALCAPA and severely reduced myocardial contraction poses a significant challenge in terms of vasoactive haemodynamic support strategy. Whilst the child may be hypotensive, administration of vasoconstrictors or inotropes will impose an increased myocardial oxygen demand and afterload on diseased myocardium. Conversely, leusotropic afterload reduction with the goal of reducing myocardial excursion may reduce diastolic filling of the right coronary artery system that may be providing a critical cross-supply to the left ventricle. Inotropes that are also positive chronotropes such as adrenaline and dobutamine may increase the propensity to ectopy and compromise myocardial diastolic filling. Oxygen therapy, whilst important in improving myocardial oxygen delivery, may theoretically reduce the pulmonary vascular resistance and left coronary driving pressure, inducing ischaemia.

Whilst various surgical techniques have been described, the only practice in widespread use and of proven efficacy is direct reimplantation of the LCA into the aorta. The pulmonary artery is transected, the coronary ostium identified, and a button excised. The LCA is mobilised, the aorta transected, and the button implanted, often into a trapdoor incision. The aorta and pulmonary artery are then repaired, with a patch to the latter at the site of coronary excision. In rare cases such as that described, severe myocardial damage occurs prior to presentation, and myocardial performance does not improve with surgical repair. In these cases, cardiac transplantation offers the only prospect of survival.

Learning Points

- ALCAPA does not present at birth due to an elevated pulmonary artery pressure that preserves coronary perfusion. Presentation is usually delayed by 6–12 weeks.
- In an infant presenting with dilated cardiomyopathy, it is **crucial to demonstrate both the LCA arising from the aorta and antegrade flow**. Any disarray must prompt the use of cross-sectional imaging or invasive angiography. An enlarged right coronary artery might be a pointer to the diagnosis.
- A normal ECG does not exclude ALCAPA.
- Management of the patient with ALCAPA with haemodynamic compromise requires careful balancing of inotropic, leusotropic and afterload reduction therapy with an awareness of the potential pitfalls of each treatment modality.
- The prognosis for a patient presenting late with well-established myocardial dysfunction is poor, irrespective of prompt surgical treatment.

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Chapter 8

The Woes Lie Below

Srinidhi J.V. Rao and Tara Bharucha

Abstract Cardiorespiratory collapse can have a number of causes in the neonate, and echocardiography is an important diagnostic tool in this population. While speed of diagnosis may be important, it is also critically important that the cardiologist is aware of the limitations and potential pitfalls of echocardiography. Echocardiographic evaluation must include a full and accurate assessment of anatomy and haemodynamics, and the cardiologist must also correctly interpret findings. We present a case that highlights the principles of interpretation.

Keywords Critical aortic stenosis • Aortic valve • Pericardial effusion • Left ventricular dysfunction • Collapsed neonate • Left ventricular outflow tract obstruction • Modified Bernoulli equation

Case Description

A neonate presented to the emergency department of a district general hospital at 2 weeks of age, with a few hours' history of poor feeding, lethargy and laboured breathing. Initial assessment revealed an acyanotic infant with respiratory distress and evidence of poor perfusion with a prolonged capillary refill time and reduced pulses. The chest was clear to auscultation and the liver was palpable 5 cm below the right costal margin. There was a marked metabolic acidosis on blood gas analysis, with raised serum lactate levels. In view of the severe cardiorespiratory compromise, the neonate was intubated and ventilated, fluid resuscitated and inotropes

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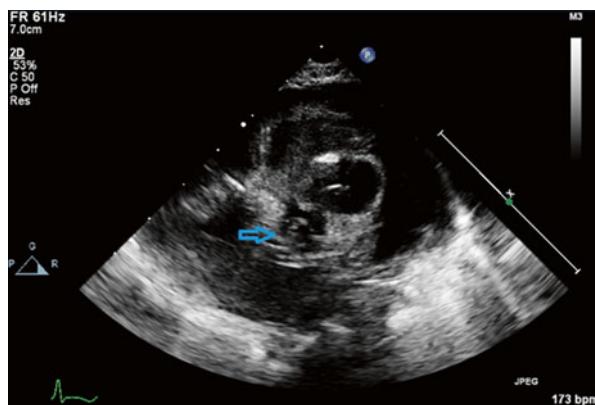
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commenced. Chest x-ray demonstrated cardiomegaly. A sonographer, untrained in congenital heart disease, performed an urgent echocardiogram. This showed a large pericardial effusion and urgent referral was made to a tertiary cardiac centre. Subsequent detailed echocardiography demonstrated a moderate, localised, pericardial effusion (Fig. 8.1) unlikely to be the sole cause of dramatic haemodynamic compromise. The aortic valve was unicuspido with severe stenosis the colour Doppler showing a small effective orifice area with a narrow forward jet (see Figs. 8.1 and 8.2). The Doppler peak velocity across the aortic valve was 2.5 m/s, giving an estimated peak transvalvar gradient of only 24 mmHg, mean gradient 9 mmHg (Fig. 8.3a). There was no aortic regurgitation. The aortic arch was unobstructed and the left ventricle was normal in size. The mitral valve was normal and there were no septal defects. The left ventricular (LV) function was globally severely depressed, with LV ejection fraction less than 20 % when assessed by M mode. The arterial duct was closed. A diagnosis of critical aortic stenosis with poor LV function was made.

Fig. 8.1 Subcostal 5-chamber view showing a doming aortic valve (red arrow) and moderate pericardial effusion (blue arrow)



Fig. 8.2 Parasternal short-axis view of the aortic valve showing a unicuspido aortic valve (blue arrow)



It is easy to underestimate valve gradients in the setting of reduced ventricular function. The presentation of a collapsed neonate, an anatomically abnormal aortic valve on 2D imaging, and poor left ventricular function support the diagnosis of critical aortic stenosis. The pericardial effusion was the result of poor left ventricular function and not the cause of heart failure.

The patient underwent balloon aortic valvoplasty and pericardiocentesis during the same procedure. Valvoplasty achieved a good angiographic result, with substantial improvement in aortic valve opening and forward flow into the ascending aorta. There was significant clinical improvement, and the infant was extubated within a few hours. The post-procedure echocardiogram showed an improvement in LV contractility, but the peak transvalvar gradient was now 88 mm Hg (see Fig. 8.3b). There was mild aortic regurgitation. He remained well, and underwent elective surgical valvotomy at 3 months of age, with an excellent result.

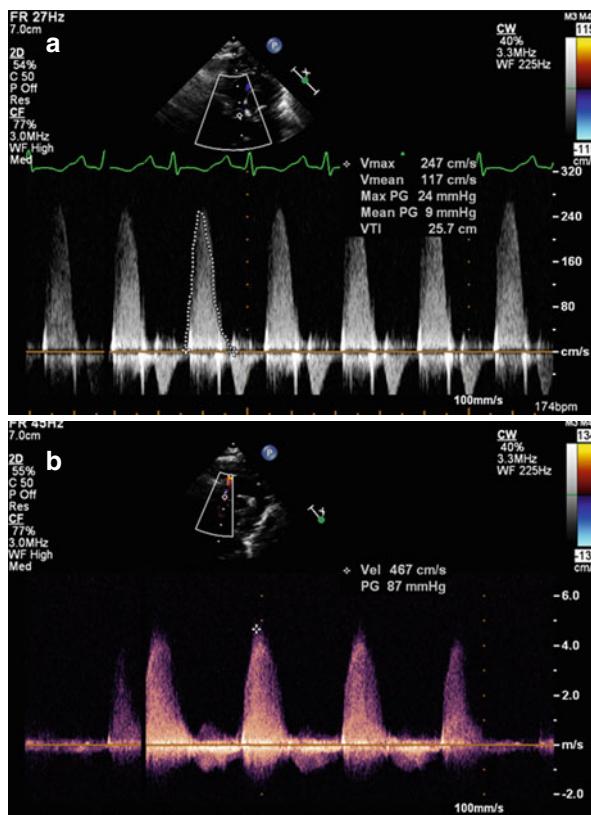


Fig. 8.3 (a) Doppler evaluation of aortic valve from suprasternal angle at presentation showing a peak velocity of 2.47 m/s (peak gradient 24 mmHg, mean gradient 9 mmHg). (b) Post balloon aortic valvotomy continuous wave Doppler evaluation from the suprasternal angle, showing peak velocity of 4.67 m/s. The gradient has “increased” in the setting of improved cardiac output

Discussion

Collapse in the neonatal period may have a number of causes, but cardiac ones are amongst the most common. Critical obstruction to the left heart, such as critical aortic stenosis, hypoplastic left heart syndrome, coarctation of the aorta and interrupted aortic arch, present with acute deterioration at the time of ductal closure. Antenatal diagnosis of congenital heart disease allows appropriate management strategies to be put in place to ensure that the baby stays well from birth and acute collapse is pre-empted. Other cardiac causes of neonatal collapse include myocarditis, tachyarrhythmias and complete heart block.

Echocardiography is the cornerstone of diagnosis of congenital heart disease. In patients with left-sided obstructive lesions, echocardiography gives information regarding structure, function and haemodynamics, and will enable the cardiologist to direct management accordingly. Understanding the limitations of echocardiography is very important for the practitioner in the process of reaching a diagnosis.

A comprehensive sequential segmental analysis must be carried out, and the practitioner should be aware that there are often multiple lesions in the same heart. Multiple left heart lesions often co-exist, and therefore echocardiographic assessment of the left heart must include careful assessment of the mitral valve with its supporting apparatus, the subaortic region, the aortic valve, the ascending aorta, arch and descending aorta, along with assessment of left ventricular volume and function.

Pulsed-wave and continuous-wave Doppler evaluation are routine in most echocardiographic examinations; hence, it is important to be aware of the limitations of these methods. The echo machine automatically uses the Doppler equation to estimate the velocity of blood flow. In order to obtain an accurate estimation of velocity there must be good alignment of the cursor with the colour-flow jet ($<30^\circ$). The simplified Bernoulli equation uses the velocity of blood to estimate the pressure drop across a **discrete** obstruction ($4 V^2$). The Bernoulli equation is not accurate for multiple levels of obstruction, e.g., tunnel-like subaortic stenosis coexisting with valvar aortic stenosis. The findings of pulsed wave and continuous wave Doppler must be interpreted carefully in light of these limitations. In addition, the ventricular function will effect the Doppler findings as outlined below. Ventricular function is assessed routinely in all echocardiographic examinations. Commonly used echocardiographic measures of left ventricular systolic function are assessment of ejection fraction by Simpson's biplane method, or an estimate using M-mode measurement at the level of mitral valve papillary muscles to assess fractional shortening. These methods give us an impression of the global function of the left ventricle; however, obstructive lesions can cause regional wall motion abnormalities. In the presence of global LV dysfunction, the LV cannot generate enough pressure to overcome the stenotic valve and hence the maximum velocity across the valve is low, and the degree of obstruction by this measure alone will be underestimated. The European Association of Echocardiography/American Society of Echocardiography guidelines classify severity of aortic stenosis in adults as follows: mild stenosis if mean gradient <20 mmHg (maximum jet velocity <3 m/s), moderate stenosis if mean gradient 20–40 mmHg (maximum jet velocity 3–4 m/s) and severe stenosis

if mean gradient >40 mmHg (maximum jet velocity >4 m/s) in the absence of LV dysfunction. These gradient values cannot be relied upon in the presence of abnormal LV function or impaired cardiac output. In this case, reliance solely on measurement of the velocity and calculation of the stenotic gradient would be underestimating the importance of the valvar obstruction.

In the presence of severe LV dysfunction, two-dimensional and colour flow imaging of the aortic valve are the key to recognising significant stenosis. Stenotic valves may be unicupid, bicuspid or tricuspid and there may be fusion between cusps. Colour Doppler will show a narrow jet of forward flow across the stenotic valve. Post-stenotic dilatation of the ascending aorta is often seen, and it is important to rule out obstruction at other levels. Measurements should be made at the level of annulus and ascending aorta, and z scores of these structures should be calculated indexed to body surface area. In some cases an indexed diameter of the valve will be too small to support the use of balloon valvoplasty and surgery will be required.

Quantification of effective valve orifice is possible using echocardiography in adults by means of pulse wave Doppler and the continuity equation, but is not routine practice in paediatric cardiology.

An assessment of any aortic regurgitation should also be made using a combination of the jet width on colour Doppler, the pressure half-time of the continuous wave signal and the flow pattern in the descending aorta.

A pericardial effusion in an infant can be due to infection, trauma (e.g., indwelling central venous lines in preterm infants), malignancy, hypoalbuminaemia and congestive heart failure. In this patient, identification of ventricular dysfunction as the cause of the effusion allowed simultaneous treatment of both the cause (outflow tract obstruction leading to ventricular dysfunction) and also the effect (the effusion).

Learning Points

- Obstructive lesions of the left heart may present with heart failure, especially in the newborn period.
- Understanding the limitations of Doppler principles will enable the cardiologist to interpret echocardiographic findings appropriately.
- In the presence of reduced cardiac function, Doppler-derived estimates of severity of valve stenosis may be unreliable.

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Chapter 9

When Not to Intubate Babies Receiving 100 % Oxygen

Shree Vishna Rasiah

Abstract Pulse oximetry screening of neonates is becoming routine practice across most hospitals in the UK. It has 75 % sensitivity and 99 % specificity for identifying babies with significant congenital heart disease. The suggested management of a baby who fails pulse oximetry screening is review by a doctor who should undertake a hyperoxia test. Those babies who fail the test are very likely to have congenital heart disease. Pulse oximetry screening also identifies babies who present with PPHN and hypoxia for non-cardiac reasons. It can be challenging to decide when to intubate and ventilate these babies who have failed pulse oximetry and are ‘requiring 100 % oxygen’. When in doubt, it is best practice to start a prostaglandin infusion and then support the ventilation as required. We illustrate this with two cases that failed the pulse oximetry screening and had increasing oxygen requirements of up to 100 % via 6 L optiflow. Intubation and ventilation were avoided by assessing the baby’s history and clinical signs together with blood gas findings. Echocardiography subsequently confirmed the cardiac diagnoses.

Keywords Pulse oximetry • Screening • Congenital heart disease • PPHN • Persistent pulmonary hypertension of the newborn

Case Description 1

A term baby girl was delivered by spontaneous vaginal delivery. There was meconium-stained liquor at delivery. She was born in a good condition and did not require any resuscitation after delivery. APGARs were 8 at 1 min and 9 at 5 min. She was kept with her mother and monitored closely on the postnatal wards given the presence of meconium stained liquor.

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The paediatrician was asked to review her because she failed pulse oximetry screening on the postnatal ward. Her saturations were 89 % pre-ductal and 88 % post-ductal. She was brought to the neonatal unit, where she was started on 6 L of optiflow in 50 % oxygen. Her oxygen saturations improved on this, with pre-ductal saturations reaching 99 % and post-ductal saturations 98 %. Given the history, she was thought to have meconium aspiration syndrome. She went on to have an infection screen and was started on intravenous first-line antibiotics. Initial capillary blood gas result was pH 7.32, pCO₂ 6.26 kPa and base excess of 1.1.

Over the next 3 hours, her saturations continued to drift downwards in spite of gradually increasing oxygen delivery via optiflow. She eventually reached 100 % oxygen via optiflow and her saturations were high 80s and low 90s. The medical team was informed by the nurse that the baby was in 100 % oxygen and would require ventilation.

On clinical review the baby was very settled with no evidence of respiratory distress. Clinical examination was normal apart from a heart murmur. Her chest x-ray is shown in Fig. 9.1. Her repeat capillary blood gas at the time showed a pH 7.37, pCO₂ 5.17 kPa and base excess of 2.1. Given these findings the consultant suspected a cyanotic duct-dependent lesion given the lack of respiratory symptoms and signs together with a normal capillary blood gas. Prostaglandin was commenced and an echocardiogram requested.

The echocardiogram showed normal situs, atrioventricular and ventricular-arterial connections. The pulmonary veins were seen entering the left atrium. There was right to left shunting across the atrial septum. There was no forward flow across the pulmonary valve and a tortuous duct supplying confluent branch pulmonary arteries. The diagnosis of pulmonary atresia with intact ventricular septum was made and the baby was referred to the regional paediatric cardiac centre.

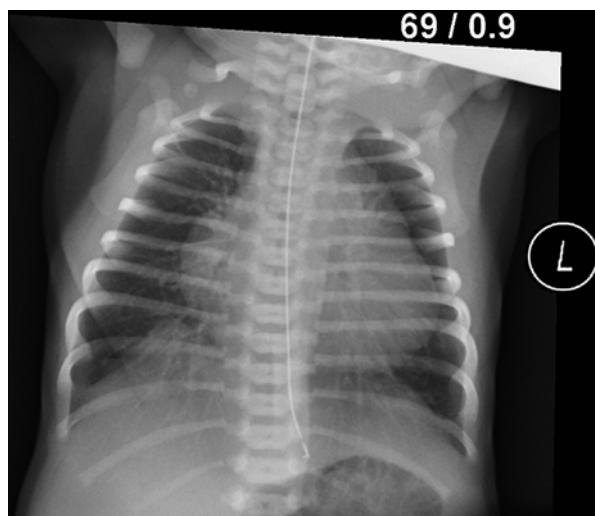


Fig. 9.1 Chest x-ray showing a slightly enlarged cardiac shadow and streaky lung fields in case 1

Case Description 2

A term baby was delivered by emergency caesarean section for a pathological cardiotocogram. There was also a history of prolonged rupture of membranes (PROM). She was born in good condition and did not require resuscitation. There was meconium present, and her APGARs were 7 at 1 min and 9 at 5 min.

The paediatrician was asked to review her because she failed pulse oximetry screening on the postnatal ward. Her saturations were 95 % pre-ductal and 92 % post-ductal. She was brought to the neonatal unit where she was started on 6 L oxygen via optiflow. Her oxygen saturations improved, with pre-ductal reaching 100 % and post-ductal reaching 100 %. Again, given the history of meconium-stained liquor at delivery, she was thought to have meconium aspiration syndrome. She went on to have an infection screen and was started on intravenous first-line antibiotics. Initial capillary blood gas was pH of 7.33, pCO₂ of 5.8 kPa and base excess of 2.6.

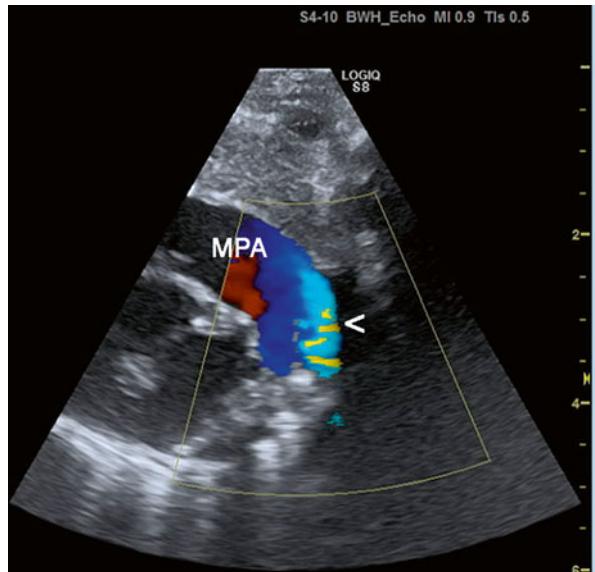
As she was requiring 100 % oxygen, the decision was made to intubate and ventilate her. However, on consultant review she had no signs of respiratory distress and was settled in 6 L of 100 % oxygen via optiflow. Repeat blood gas showed a pH of 7.44, pCO₂ of 4.56 kPa and BE -0.2. Chest x-ray on admission is shown in Fig. 9.2. Given that this was normal, she was suspected of having early PPHN secondary to sepsis, as there was a history of PROM. Intubation was avoided and an echocardiogram showed a structurally normal heart with evidence of mild PPHN. There was predominantly right to left shunt across the PDA and the patent foramen ovale as shown on Fig. 9.3.

She remained clinically well and oxygen requirement was slowly weaned off over the next 3 days. Her initial C-reactive protein (CRP) on admission was elevated at 57 and peaked at 121 before settling. Lumbar puncture and blood cultures were negative but she completed 7 days of antibiotics given the clinical history, presentation and course.



Fig. 9.2 Admission chest x-ray which shows a relatively expiratory chest x-ray and normal lung fields in case 2

Fig. 9.3 Echocardiogram short-axis view showing the right to left shunt across the PDA in case 2 (white arrow). MPA, main pulmonary artery



Discussion

Pulse oximetry screening in neonates is becoming routine practice in most hospitals. The study by Ewer et al. showed the benefits of pre- and post-ductal pulse oximetry screening in identifying not only significant congenital heart disease but also other conditions that can result in PPHN and hypoxia – for example, early onset sepsis and pneumonia. The two cases described highlight how the initial clinical presentations can be very similar between cyanotic congenital heart disease and PPHN and emphasise the importance of history and examination along with bedside investigations in deciding the right course of management and in guiding the need for ventilator support.

Learning Points

- Pulse oximetry screening does not only identify significant congenital heart disease but also other conditions causing PPHN and hypoxia.
- The initial clinical presentation of PPHN and cyanotic congenital cardiac conditions can be very similar.
- Clinical history, signs and clinical examination findings along with bedside investigations should be used to decide the management and need for respiratory support in babies who fail pulse oximetry screening.
- The importance of echocardiography when there is diagnostic uncertainty.

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Chapter 10

A Child with a Long QT?

Elena Montanes and Jan Till

Abstract An asymptomatic 16-year-old boy was found to have a long QT interval on ECG when he was screened for cardiac disease by his GP. On referral to a cardiologist treatment with betablockers, life-style advice and family screening were immediately instituted. However, eventually a blood test was performed and he was discovered to have chronic renal failure with a low calcium, magnesium and a high urea and creatinine which was finally realised as the cause of his long QT. With renal dialysis his QT interval corrected and the ECG abnormality resolved but his renal failure was irreversible and he awaits transplant.

Keywords Screening • QT interval • Renal failure • Diagnosis

Case Description

A 16-year-old boy was referred to the inherited cardiac condition clinic with a prolonged QT interval on 12-lead ECG found during screening due to a family history of “cardiac disease.”

Clinically, he reported that he was completely well without symptoms, in particular, he had never experienced palpitations or chest pain and had no history of syncope. On closer questioning the patient reported daily leg cramps in the last couple of years. Otherwise, he was fit and healthy, with good exercise tolerance. He was not on any medication, and his diet was described as normal. The screening had

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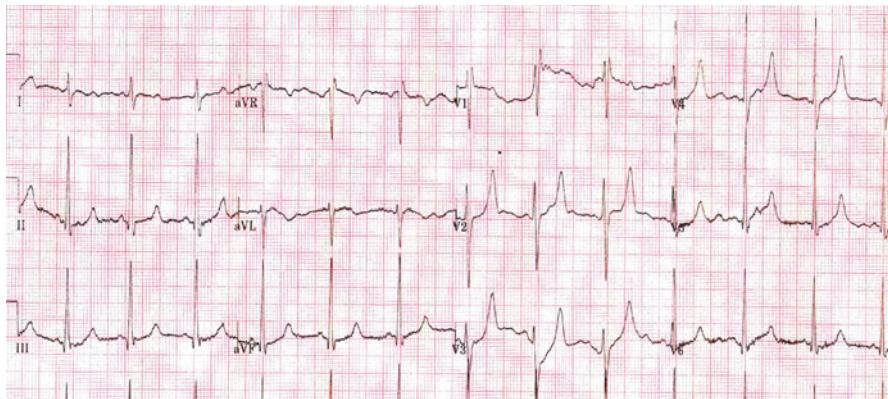


Fig. 10.1 Twelve-lead ECG showing peaked T waves. The QT interval is prolonged. Paper speed 25 mm/s, 10 mm/mV

been initiated by his GP at his parents request because his father had coronary artery disease and his mother had had SVT. On finding a long QT interval the boy had been referred to a general cardiologist and an electrophysiologist before the referral to the inherited cardiac conditions service.

As a result of the long QT seen, screening had been initiated in other family members. His 18-year-old sister was completely well and had a normal ECG with a QTc within normal limits and was awaiting an exercise test and 24-h tape. His mother was already under the care of an adult electrophysiologist for an “arrhythmia” and had had an ablation. She and her husband, the patient’s father, both had a QT interval within normal limits. In the extended family history, his paternal grandfather had died suddenly at the age of 40 years, assumed secondary to a myocardial infarct.

The 12-lead ECG of our patient at the time of the consultation showed tall and peaked T waves with a prolonged QT interval (480 ms in lead V5). There was an incomplete right bundle branch block (Fig. 10.1).

A 24-h tape and an exercise test were performed and confirmed a persistently prolonged QT interval. The QTc remained fixed with no adaptation to change in rate (Fig. 10.2).

The patient had been informed about the suspected diagnosis and had been given lifestyle advice that included limiting exercise. He was advised to stop playing 5-a-side football. He was commenced on beta-blocker therapy with bisoprolol.

Blood analysis had been said to have been done early on in his screening, but blood was taken in the inherited cardiac conditions clinic for genetic analysis and also sent for urea and electrolytes and thyroid function.

Surprisingly, his blood test was returned reporting very abnormal parameters with hypomagnesaemia, hypocalcaemia and hyperphosphatemia and high urea and creatinine (Table 10.1). Blood results were initially disbelief as the patient was so well, but the results were rapidly checked and returned as similar.

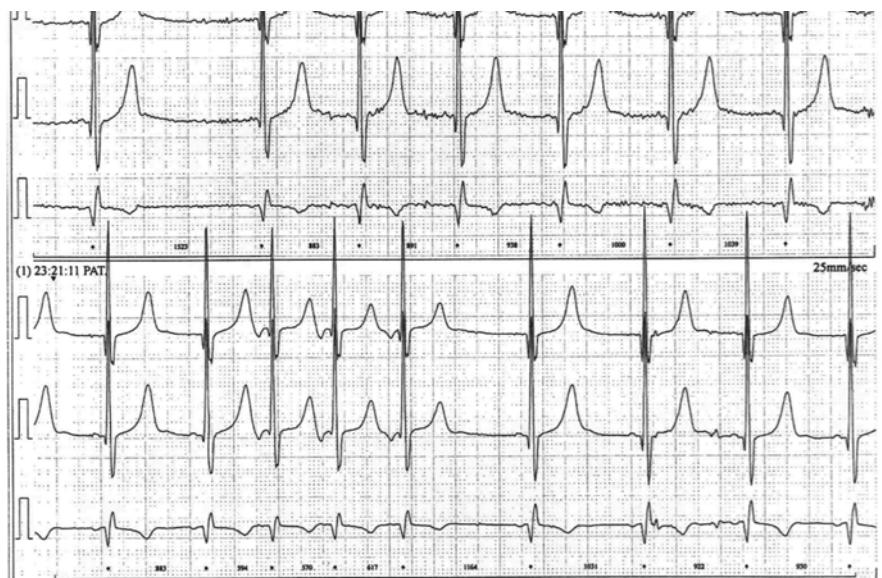


Fig. 10.2 Section of 24-h recording showing no adaptation to rate change in QT interval

Table 10.1 Blood results from patients with a long QT

| | Patient value | Normal range |
|---------------------|---------------|--------------|
| Sodium | 140 | (133–146) |
| Potassium | 3.8 | (3.5–5.3) |
| Urea | 35.2 | (2.5–7.8) |
| Creatinine | 924 | (60–120) |
| Corrected Calcium | 1.09 | (2.20–2.60) |
| Inorganic Phosphate | 2.43 | (0.80–1.50) |
| Magnesium | 0.55 | (0.7–1.0) |

Despite looking remarkably well, a diagnosis of renal failure was raised and the patient was urgently referred to a nephrologist. He was subsequently found to have bilateral hydronephrosis with presumed chronic urinary retention. Further investigation revealed that he suffered from undiagnosed posterior urethral valves. Immediate dialysis was started but unfortunately damage proved irreversible, and he is currently awaiting renal transplant. On further investigation it was realised that initial blood tests some 2 months beforehand had been abnormal but not acted upon. The repeated ECGs once started on dialysis showed improvement of the QT interval duration with completely normalization of the measurement.

Discussion

Congenital long QT syndrome is an important cause of cardiac death in young people. It is one of the channelopathies that can present with syncope, aborted cardiac arrest or sudden death in otherwise seemingly completely well young people.

The exact prevalence is unknown but an incidence of 1:10,000 has been proposed. The condition may be lethal and in approximately 13 % of cases sudden death is the first manifestation. As a consequence screening of first degree relatives of anyone diagnosed is strongly advised so that asymptomatic people may be identified and protected with betablockers before their first symptom.

In some countries all sportsmen and women undergo compulsory screening before they are allowed to compete. In the UK, many professional clubs in different sports now insist on screening. Screening of the general population, however, has not been adopted and remains controversial. Screening for long QT syndrome is complicated by the lability of the QT interval, the absence of a long QT in some people who have the genetic mutation, the difficulty in measuring and correcting the interval and the problem of the interpretation of borderline findings.

Congenital long QT syndrome may be inherited as it can be caused by a genetic mutation. There are 13 different types now recognised. The exact mutation can be identified in up to 75 % of cases with good genetic testing with 90 % of the cases genetically identified being long QT 1, 2 or 3. Once found, the mutation allows more specific treatment and lifestyle advice for the index case and allows genetic screening to be offered to relatives.

If the genetic mutation cannot be found, or, prior to genetic analysis, screening is performed clinically by ECG, exercise testing ±24-h tape. It is important to remember that a significant minority (25 %) of the patients with long QT syndrome confirmed by the presence of a mutation of one of the long QT genes may have a normal QTc interval on the ECG.

Some cases of long QT syndrome arise “de novo,” meaning that the mistake in the DNA has arisen for the first time in that individual and that person will be the first affected in a family. It is useful to bear in mind that when assessing a patient without a family history the pre test probability for long QT syndrome is 1 in 2000, whereas for a relative of someone with confirmed long QT syndrome the probability can be 50 %.

Making a diagnosis of long QT syndrome can be difficult and challenging even for experienced physicians. The QT interval is labile and can be challenging to measure.

The normal QT interval adapts with RR interval (rate). To account for this a mathematical formula has been adopted – Bazetts’ formula. Although known not to totally adjust at higher and lower heart rates it has been universally accepted and used by physicians worldwide. Whilst the upper limit of the QTc interval is often quoted as 440 in males and 460 in females the recent guidelines for diagnosis for long QT syndrome recommend at least two ECGs with a corrected QT interval of 480 or greater. Not only is the duration of the QT interval important but also the

Table 10.2 1993–2012 diagnostic criteria for diagnosing long QT syndrome. Score: ≤1 point: low probability of LQTS. 1.5–3 points: intermediate probability of PQTS. ≥3.5 points: high probability

| Electrocardiographic findings ^a | | | |
|--|--|-------------------|-----|
| | | Points | |
| A | QTc ^b | ≥480 ms | 3 |
| | | 460–479 ms | 2 |
| | | 450–459 (male) ms | 1 |
| B | QTc ^b 4th minute of recovery from Exercise stress test ≥480 ms | | 1 |
| C | Torsade de pointes ^c | | 2 |
| D | T-wave alterans | | 1 |
| E | Notched T-wave in three leads | | 1 |
| F | Low heart rate for age ^d | | 0.5 |
| Clinical history | | | |
| A | Syncope ^e | With stress | 2 |
| | | Without stress | 1 |
| B | Congenital deafness | | 0.5 |
| Family history | | | |
| A | Family members with definite LQTS ^e | | 1 |
| B | Unexplained sudden cardiac death below Age 30 among immediate family members ^e | | 0.5 |

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^aIn the absence of medications or disorders known to affect these electrocardiographic features

^bQTc calculated by Bazett's formula where $QTc = QT/\sqrt{RR}$

^cMutually exclusive

^dResting heart rate below the 2nd percentile for age

^eThe same family member cannot be counted in A and B

morphology of the T wave. It has been reported that there are different shapes of T wave that are characteristic of the different types of long QT. A broad-based T wave may be seen in long QT 1, whereas T waves are typically low amplitude or bifid in long QT2, and in the long QT3 late-onset T waves are characteristic. In retrospect our patient had very tall peaked T waves characteristic of a metabolic abnormality.

Because of the difficulty in diagnosis, Schwartz in 1993 designed a diagnostic score to try to quantify the probability of having long QT syndrome. The latest version of the score is shown in Table 10.2. Scoring more than 3.5 points means that that patient has a high probability of having long QT syndrome.

Additional diagnostic help can be gained from evaluating how the QT interval adapts with change in rate during an exercise test or 24-h tape. Others have advocated the adrenaline challenge but this remains controversial.

Before establishing a diagnosis of congenital long QT it is vital to exclude other causes of a prolonged QT on a resting ECG. This is particularly prudent when considering a person without characteristic symptoms of LQT and no family history as in our case. Known causes of acquired long QT are listed in Table 10.3.

Table 10.3 Secondary causes of a long QT

| Drugs: Listed on LQT drugs.org |
|---|
| High T4 and hyperthyroidism |
| Low calcium |
| Low potassium |
| Anorexia |
| Low protein diets |
| Post cardiac arrest |
| Head injury |
| Subarachnoid haemorrhage/raised intracranial pressure |

Acquired causes of long QT are extremely important to identify as they are not only potentially reversible but may need medical intervention.

The most common cause of acquired long QT is medication as listed www.QTdrugs.org. These drugs result in a long QT interval indistinguishable from congenital long QT syndrome by blocking the delayed rectifier potassium current (IKr). This current is encoded by the KCNH2 gene and in some people a genetic mutation is first identified when one such drug is given and a long QT interval is recognised. As many as 35–40 % of drug induced long QT syndrome patients have been found to have mutations associated with congenital long QT syndrome.

Patients with long QT syndrome need to avoid such drugs as they will exacerbate their condition and it is well known that concurrent administration of QT prolonging drugs along with non compliance of betablocker treatment represent the most important reasons for breakthrough events in established patients with long QT syndrome.

Metabolic and electrolyte disturbance, especially hypokalaemia, hypocalcemia or hypomagnesaemia that are a well-known cause of acquired long QT. Hypokalemia can result in a decreased T wave amplitude, T wave inversion, prominence of the U wave and prolongation of the QT interval. Ventricular tachycardia, often in the form of Torsades de Pointes can ensue. Prolongation of the QT interval has also been reported in cases of hypocalcemia and hypomagnesemia. The use of diuretics is known to produce electrolyte disturbance and thereby indirectly prolong the QT interval.

Another less frequent cause of a long QT interval is end-stage renal disease (ESRD) patients. This appears to be mainly in relation to rapid changes in electrolyte plasma concentrations. In fact, ventricular arrhythmia is a major cause of death in ESRD patients (up to 25 % in some series). Several mechanisms for QTc interval prolongation in these patients are: increased renin angiotensin system activity that can remodel cardiac ion channels, which results in prolonged repolarization. Also increased LV mass, typical at this stage of the disease, creates LV systolic and diastolic dysfunction that is well correlated with QTc interval prolongation.

Learning Points

- Diagnosis of LQTS can be difficult and challenging.
- Secondary causes of a long QT should always be thought about and excluded if possible. This is particularly pertinent in an isolated case without a family history. A blood test for thyroid and electrolytes disturbance should always be performed and checked.
- As a result of the high lethality of long QT syndrome, screening should be performed in all first-degree relatives.

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Chapter 11

Breathlessness in an Ex-Prem When All Is Not What It Seems

Anna N. Seale

Abstract Premature infants often suffer chronic lung disease. We present a child born at 28 weeks gestation, whose deterioration in respiratory function was initially attributed to chronic lung disease. When assessed by respiratory specialists the child's symptoms were felt to be over and above that expected for his lung disease, and a subsequent cardiac review revealed the rare condition of isolated pulmonary vein stenosis. This case illustrates the difficulties in making this diagnosis.

Keywords Pulmonary vein stenosis • Pulmonary hypertension • Prematurity

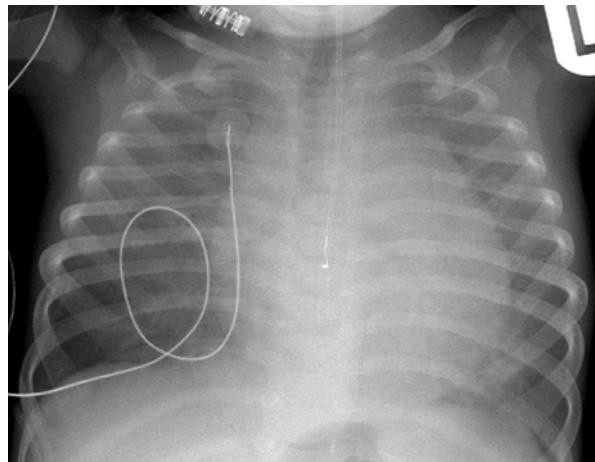
Case Description

A 1-year-old child presented with breathlessness, recurrent chest infections and failure to thrive. He had been born at 28 weeks gestation weighing 700 g; he received surfactant and required mechanical ventilation for 3 days and nasal CPAP for a further 6 days. He subsequently received respiratory support on two further occasions for infection and necrotising enterocolitis. Altogether he had two episodes of necrotising enterocolitis, both treated conservatively, and received indomethacin for a patent arterial duct. He was weaned off nasal cannula oxygen by 3 months of age. He was also known to have a grade II intraventricular haemorrhage, hypothyroidism and retinopathy of prematurity.

Despite his initial improvement in respiratory status, he began to deteriorate with a recurrent cough, increased respiratory effort, frequent admissions to

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Fig. 11.1 Chest radiograph in 1-year-old child, showing cardiomegaly and pulmonary venous congestion

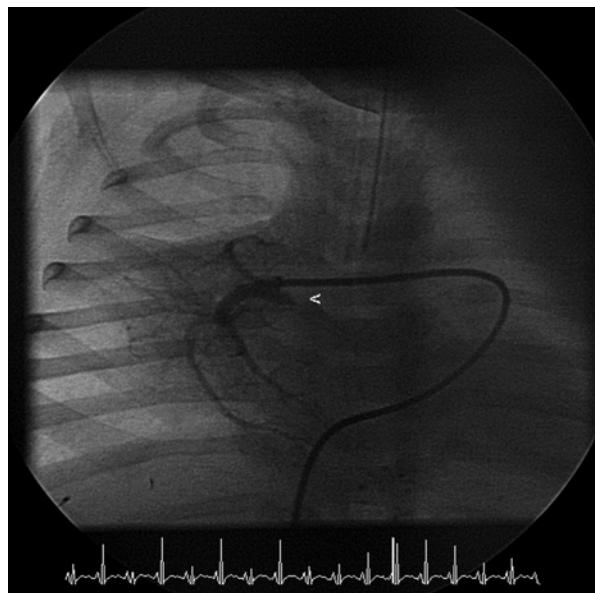


hospital for chest infections and an oxygen requirement. He was therefore admitted to the paediatric ward for investigation by the respiratory physicians. On examination he had marked subcostal recession with Harrison's sulci and diffuse crackles on chest auscultation. He had an accentuated pulmonary component to the second heart sound with no heart murmurs. There was 3 cm hepatomegaly and normal femoral pulses. Chest radiograph showed cardiomegaly with increased pulmonary venous markings (Fig. 11.1) and ECG showed right ventricular hypertrophy.

In view of the abnormal chest radiograph and ECG, cardiac review and echocardiogram were arranged. Echocardiogram showed a normally connected heart. The right atrium and right ventricle were dilated, with mild tricuspid valve regurgitation giving an estimated right ventricular pressure of 50 mmHg by continuous-wave Doppler interrogation. There was no evidence of right ventricular outflow tract obstruction; the branch pulmonary arteries were confluent and unobstructed, the left pulmonary artery appearing smaller than the right. Flow was seen returning to the left atrium from all four pulmonary veins however in all but the right lower pulmonary vein, the flow was turbulent on colour-flow. Pulsed-wave Doppler interrogation showed high-velocity flow signal (2 m/s) not returning to the baseline. There were no intra-cardiac shunts.

In view of the findings on echocardiogram, pulmonary vein stenosis was suspected and cardiac catheterisation performed. This showed pulmonary hypertension with the pulmonary pressure at two-thirds of systemic blood pressure. There was a gradient of 18–20 mmHg across the left upper, left lower and right upper pulmonary veins. Angiography showed discrete narrowing of these three vessels as they entered the left atrium (Fig. 11.2). The distal veins were of normal size. The child subsequently underwent repeated catheter interventions to balloon dilate the stenoses with conventional and cutting balloons and obtained good symptomatic relief.

Fig. 11.2 Angiogram showing discrete right upper pulmonary vein stenosis (arrow)



Discussion

This case highlights the importance of assessing the pulmonary veins in infants that have chronic lung disease of prematurity but whose respiratory symptoms are more severe than would be expected from the degree of prematurity and early course.

Individual stenosis of one or more of the pulmonary veins was first described by Reye in 1951 in an 8-year-old girl with normal intra-cardiac anatomy. The lower pulmonary veins were stenotic, the left upper pulmonary vein was atretic and the right upper pulmonary vein ended blindly. The girl died of heart failure.

Pulmonary vein stenosis is rare, with two main categories found in children:

1. Isolated pulmonary vein stenosis in the setting of a normally connected heart;
2. Pulmonary vein stenosis in association with other forms of congenital heart disease and where the pulmonary veins return directly to the morphological left atrium. The incidence of associated cardiac defects is reported to range from 30 % to 80 %.

In 1988 Fong described three types of stenosis: bilateral tubular hypoplasia extending from the venoatrial junction for a variable length, intimal hourglass constriction at the venoatrial junction, and bilateral multiple short pulmonary veins that are hypoplastic for their entire extra-pulmonary course. Serial angiography has documented progression of disease with a discrete stenosis progressing to tubular and then more generalised narrowing. In some cases, the vein can become critically stenosed or atretic. Collateral pulmonary venous vessels may form, diverting pulmonary venous blood to either the systemic circulation or other lesser affected lobes of the ipsilateral pulmonary venous circulation.

The pathological mechanisms responsible for pulmonary vein stenosis remain unclear. Histological studies have shown a variable manifestation of intimal and medial interstitial fibromuscular proliferation, together with fibrotic displacement of the muscle bundles in the venous wall. This eventually results in occlusion of the lumen of one or more of the pulmonary veins connecting the lungs to the left atrium. Most commonly, the thickening of intimal tissue begins at the junction of the left atrium and pulmonary vein; the narrowing then extends outwards along the pulmonary vein towards the hilum of the lung.

Unilateral pulmonary vein stenosis leads to hypoperfusion of the affected lung, and patients are susceptible to haemoptysis. Untreated patients with severe stenosis of all the pulmonary veins have a very poor prognosis with progressive pulmonary hypertension and death, and even with early treatment the overall results remain disappointing.

Presentation

There seems to be a dichotomy of age at presentation, with some patients presenting in the first few weeks of life, and others presenting later. Pulmonary vein stenosis does seem to be linked with prematurity although infants born prematurely tend to present relatively late at approximately 7 months with many labelled as having chronic lung disease of prematurity.

Investigations

Diagnosis of pulmonary vein stenosis can be difficult. The chest radiograph shows pulmonary venous congestion, which can appear similar to the interstitial lung changes of chronic lung disease. It is important that neonatologists are aware of this, as it is possible that the true incidence of pulmonary vein stenosis is higher than is currently appreciated due to misdiagnosis. Echocardiography can also be difficult with studies showing that 26 % of pulmonary vein stenoses are missed by echocardiography and subsequently diagnosed either by angiography or at surgery. If there is a critical stenosis, a colour flow jet may not be present and lack of flow is more difficult to appreciate than a turbulent jet. In patients with unilateral disease and pulmonary hypertension, a disparity between branch pulmonary artery sizes is a good indicator of pulmonary vein stenosis and should trigger further investigation (Fig. 11.3). Lung perfusion scans and magnetic resonance imaging can also be used to assess differential blood flow.

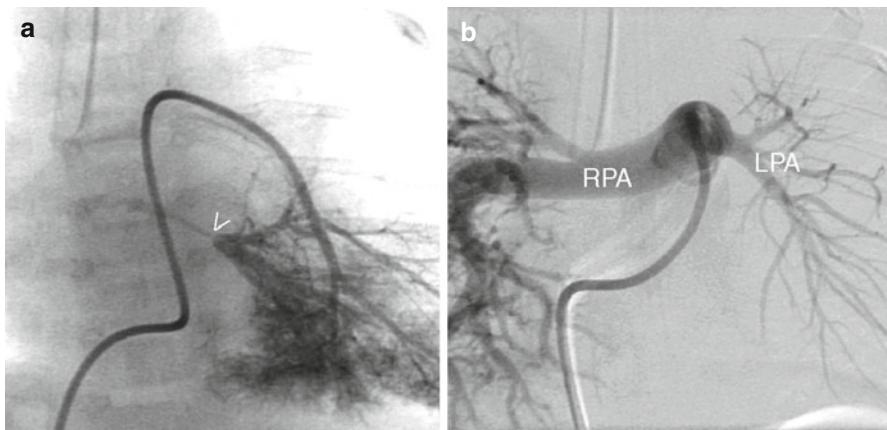


Fig. 11.3 (a) Angiogram in a different patient with left pulmonary vein stenosis (arrow). (b) Note the disparity in size of the branch pulmonary arteries with a diffusely small left pulmonary artery

Learning Points

- Isolated pulmonary vein stenosis is a rare disease that is associated with prematurity.
- Pulmonary vein stenosis should be particularly considered when the respiratory symptoms seem more severe than the degree of chronic lung disease.
- Echocardiography can be misleading, and if there is suspicion of pulmonary vein stenosis further imaging is required, particularly if there is evidence of unexplained pulmonary hypertension and disparity in branch pulmonary artery size.

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Chapter 12

Think Outside the Chest

Srinidhi J.V. Rao

Abstract Heart failure in the neonate can be due to extracardiac conditions, with the only features on echocardiogram being a dilated right heart with or without pulmonary hypertension. This should prompt a search for extracardiac left to right shunts due to arteriovenous malformations such as those present in the brain, liver or kidneys.

Keywords Vein of Galen malformation • Pulmonary hypertension • Right ventricular dilatation • Congestive heart failure • Neonate

Case Description

An infant born at term with a birthweight of 2.9 kg developed respiratory distress soon after birth. There had been no antenatal complications and no perinatal risk factors for sepsis. The delivery was uncomplicated. The baby was intubated and ventilated and a significant difference was noted between pre-ductal and post-ductal oxygen saturations, with post-ductal saturations being lower. There was also a wide pulse pressure. Clinical suspicion was of persistent pulmonary hypertension of the newborn. The baby was commenced on inotropes and prostaglandin prior to transfer to a cardiac center. Echocardiogram on arrival showed normal cardiac anatomy with a dilated right heart and evidence of severe pulmonary hypertension (Fig. 12.1). There was very prominent venous return in the superior vena cava with normal pulmonary venous drainage (Fig. 12.2). The innominate artery was significantly dilated (Fig. 12.3), and diastolic run off was seen in the transverse arch and descending aorta. Auscultation of the cranium revealed a cranial bruit. Cranial ultrasound scan followed by MRI brain scan confirmed the

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Fig. 12.1 Apical 4-chamber view demonstrating the dilated right atrium and right ventricle. *RA* right atrium, *RV* right ventricle, *LA* left atrium, *LV* left ventricle

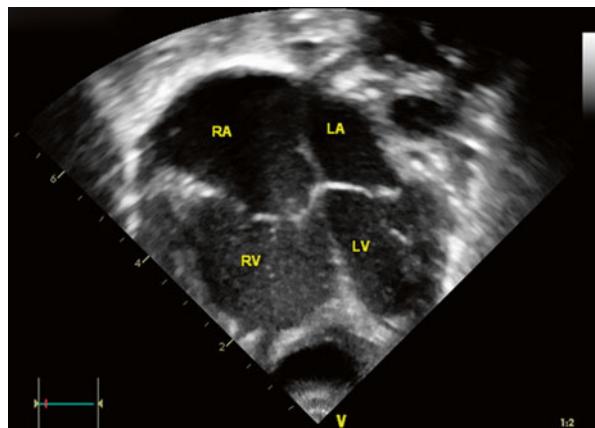


Fig. 12.2 Suprasternal long-axis view showing dilated superior vena cava and innominate vein resulting from increased venous return. Also seen is the transverse arch inferior to innominate vein. *SVC* superior vena cava, *Innom* innominate vein

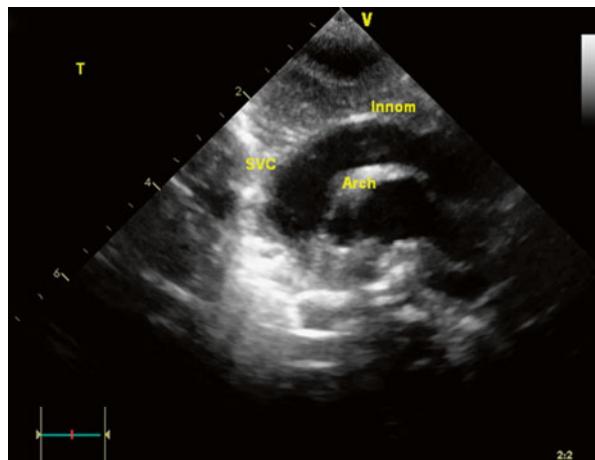
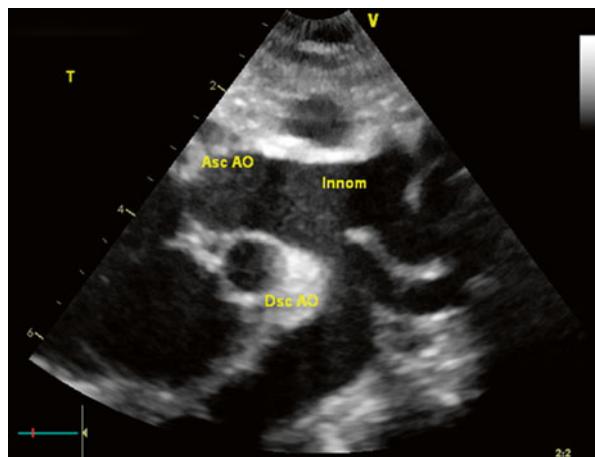


Fig. 12.3 Sagittal image from the suprasternal notch of the aortic arch. There is a dilated proximal innominate artery and the left common carotid. Note narrowing of the transverse arch and aortic isthmus. *Asc AO* ascending aorta; *Innom*, brachiocephalic trunk (also giving rise to left common carotid artery in this patient); *Dsc AO* descending aorta



presence of a vein of Galen aneurysm. The newborn underwent a series of interventional procedures to manage the aneurysm but sadly died within a few days due to complications.

Discussion

Arteriovenous malformations are high-flow communications due to absence of a developed capillary bed. They can occur in various organs, and presenting features can vary greatly depending on patient age, lesion size and location. Vein of Galen malformations are the most common form of symptomatic cerebrovascular malformation in neonates and infants. They account for less than 1–2 % of all intracranial vascular malformations but are the cause of 30 % of cerebral vascular malformations presenting in the pediatric age group. It is also the most common antenatally diagnosed intracranial vascular malformation. The malformation is due to a cerebral arteriovenous fistula of the median prosencephalic vein (MPV) (a precursor of the vein of Galen) occurring at 6–11 weeks of gestation, which fails to regress and becomes aneurysmal.

The pathophysiology involves low systemic vascular resistance and high cardiac output resulting in increased venous return to the right heart. In the neonate, with high pulmonary vascular resistance the right ventricle is hypertrophied and has low compliance. These factors in turn lead to right to left shunting at atrial and ductal levels. In the presence of a significant sized malformation, high-output cardiac failure rapidly develops. There may also be cerebral ischaemia due to steal.

Presenting Features

Typically, neonates and young infants with a symptomatic vein of Galen malformation present with intractable heart failure, which is the most common cause of death. Other features in children include a large head due to hydrocephalus, persistent pulmonary hypertension of the newborn, and seizures. Clinical examination will reveal a breathless neonate with increased pulses and often with poor skin perfusion. Cardiac auscultation may be unremarkable or one might hear the systolic murmur of tricuspid regurgitation or the arterial duct. The liver is generally enlarged. The key feature on auscultation is the presence of a cranial bruit. Vein of Galen malformations can be associated with congenital heart diseases such as sinus venosus atrial septal defect, aortic coarctation, secundum atrial septal defect, partial anomalous pulmonary venous return, ventricular septal defect, atrioventricular septal defect and transposition of great arteries.

Imaging

If there is a significant shunt, the echocardiogram will show features of severe pulmonary hypertension. The right atrium and ventricle will be dilated and the right ventricle hypertrophied (see Fig. 12.1). The atrial septum may bulge to the left with a right to left shunt across the foramen ovale. Right ventricular pressure estimated by tricuspid regurgitation velocity may show suprasystemic right ventricular pressure. The arterial duct, when present, may shunt from right to left. Important specific clues to the diagnosis are dilatation of the innominate artery, brachiocephalic vein and superior vena cava (see Fig. 12.2) as well as diastolic run off in the descending aortic. Figure 12.3 shows the aortic arch of the patient described. Dilatation of the innominate artery can give the impression that the aortic arch is narrowed and of aortic arch pathology; however, this is not the case and is an important pitfall to avoid. Prompt referral for further cranial imaging is important. Cranial ultrasound scan is a useful screening tool, and cerebral MRI or computed tomography confirms the diagnosis.

Treatment

A detailed discussion of treatment modalities is beyond the scope of this chapter. Treatment involves supportive management of cardiac failure followed by percutaneous embolisation using coils and acrylic glue. Prior to endovascular treatment, outcomes in neonates were dismal without treatment, and mortality was 90 % following surgery.

Learning Points

- High-output conditions secondary to non-cardiac conditions include severe anaemia, thyrotoxicosis and the presence of arteriovenous malformations. Right ventricular volume overload in the absence of a cardiac lesion should prompt a careful evaluation of non-cardiac causes.
- Arteriovenous malformations can occur anywhere but are most common in the brain, followed by kidneys and liver.

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Chapter 13

The Fontan Circulation: Never Forget the Atrial Septum

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Abstract The Fontan circulation has become the standard palliation for patients with non-correctable congenital heart disease. Complete bypass of a sub-pulmonary ventricular pump makes systemic venous pressure the sole driver of trans-pulmonary flow. In such a low pressure circuit, even very minor degrees of obstruction may constitute important resistance for flow into the systemic ventricle. Typically, resistance across the pulmonary vascular bed is the predominant limiting factor, largely dictating cardiac output after Fontan. However, if the entire cardiac output must then pass through the atrial communication, (such as in a hypoplastic left heart post-Fontan) restriction at atrial level creates additional resistance. As systemic venous pressures rise, driving flow across the combined resistance, the clinical manifestations of ‘the failing Fontan’ may begin to emerge. Within the context of a clinical case and an overview of Fontan physiology, the diagnosis and management of this rare but important pitfall are discussed.

Keywords Fontan • Restrictive atrial septum • Atrial communication • Protein losing enteropathy • Total cavopulmonary connection • Norwood • Glenn • Single ventricle

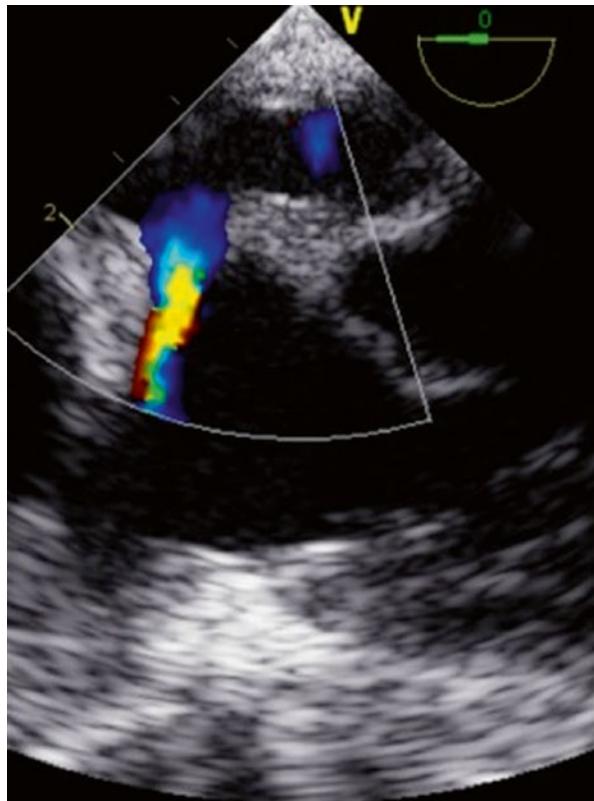
Case Description

A female patient with double-inlet left ventricle, ventriculo-arterial discordance and pulmonary stenosis underwent uneventful superior cavopulmonary connection in infancy. There was mild hypoplasia of the left atrioventricular valve (mitral valve) and a small central atrial septal defect.

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Fig. 13.1 Transeosophageal echocardiographic still frame shows evidence of small atrial communication on 2D and turbulent flow on colour mapping



Pre-Fontan echocardiography and invasive haemodynamics were satisfactory, with no gradient demonstrated across the atrial septum or mitral valve. The patient was referred for surgery at age 6.2 years and underwent fenestrated Fontan with a 20-mm GoreTex conduit. No atrial septectomy was performed because the mitral valve was believed to be unobstructed.

Postoperatively, there was severe, prolonged pleural drainage. Transthoracic echocardiography windows were limited. Transoesophageal echocardiogram showed a small native atrial communication with turbulence on color flow mapping (Fig. 13.1) and slightly increased flow velocities across the mitral valve. Cardiac catheter revealed a trans-atrial gradient of 5 mmHg and Fontan pressures of 18 mmHg (Fig. 13.2). There was no stenosis of the Fontan anastomoses, pulmonary arteries or pulmonary veins.

The fenestration was crossed from the femoral vein and a stable wire position was secured in the left lower pulmonary vein. A large Genesis stent mounted onto a 16-mm balloon was placed via a 10 French long sheath, with abolition of the trans-atrial gradient (Fig. 13.3). The fenestration was also enlarged by a separate, short stent. Final Fontan pressure was 15 mmHg and saturations were 89 %. Pleural drainage stopped over the next 48 h, the patient was discharge day 6 post catheterization. At 16 month follow-up, saturations were 90 % and exercise tolerance was satisfactory.

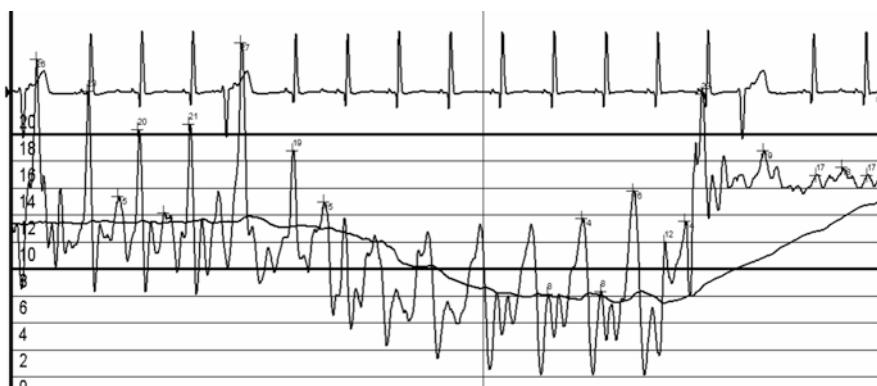
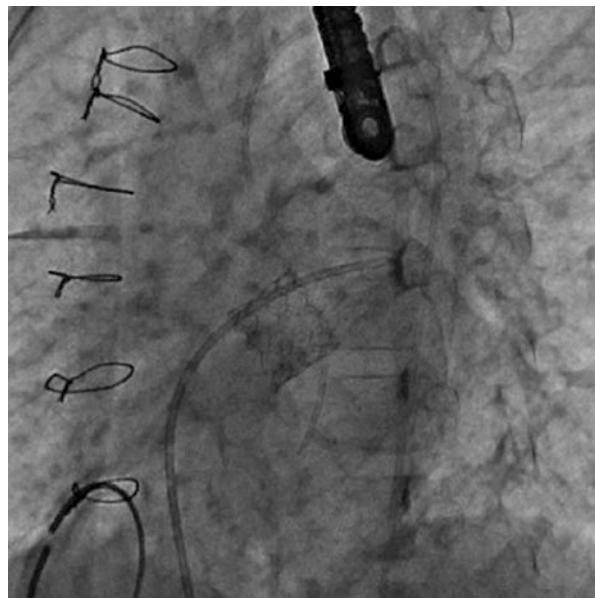


Fig. 13.2 Haemodynamic pressure tracing shows a reproducible pressure gradient during pull-back of a multitrack catheter over a wire (*left to right* on figure) from pulmonary vein to left atrium to right atrium then through the fenestration into the Fontan conduit

Fig. 13.3 Fluoroscopic still shows the large Genesis stent in place across the atrial septum



The Fontan Circulation

The Fontan circulation results in a ‘neo-portal’ system; systemic venous blood is re-routed directly to the pulmonary arteries, bypassing the ventricular pump. Trans-pulmonary flow is driven solely by the pressure gradient between systemic veins and receiving ventricle. For the majority, this gradient is dictated by relatively fixed resistance across the pulmonary capillary bed. Due to the infant’s pulmonary vascular resistance (PVR) being relatively high, successful total cavopulmonary connection must be performed in a staged fashion.

Initial surgical palliation secures an adequate outlet for systemic arterial flow and a systemic to pulmonary arterial shunt for pulmonary blood flow. At this stage, ventricle pre-load is high due to shunt return, and complete mixing means saturations are low. Superior caval venous return is surgically connected to the pulmonary arteries in the first 4–12 months of life with the ‘bidirectional cavopulmonary shunt’ (a modification on the original ‘Glenn procedure’,) and the shunt is taken down. Cyanosis persists at this stage, but ventricular pre-load is restored to near physiological levels. Inferior caval blood is later connected to the pulmonary arteries with a conduit or baffle, completing the Fontan circuit. (See Fig. 13.4 for a schematic overview of the staged circulations). Once the Fontan circuit is complete, no systemic venous blood enters the heart, achieving normal systemic oxygen saturation.

In 1977, Choussat described ten criteria for the ideal Fontan candidate (Table 13.1). Though these remain desirable, most of these ‘commandments’ have since been broken. However, it is crucial that the pulmonary arteries must be large enough to receive a conduit adequate for adulthood and the PVR must have fallen adequately. Completion is typically performed between the ages of 3 and 5 years. Operating centre experience and clinical criteria, such as oxygen saturation and exercise tolerance, usually govern timing.

Pre-load is entirely provided by trans-pulmonary flow, thus cardiac output becomes largely fixed after Fontan completion. With this in mind, medical management of post-Fontan low cardiac output state must be rationalised; traditional measures such as inotropes may worsen the situation by raising PVR or increasing heart rate thus lowering filling time. Low total intra-thoracic pressure is desirable and spontaneous ventilation generates a ‘negative pressure,’ facilitating forward flow into the vascular bed. Once blood has crossed the pulmonary vascular bed, it must also then overcome any resistance across the pulmonary veins, atrial septum, inlet valves and ventricular end-diastolic pressure. With only systemic venous pressure to drive flow, an added gradient of even 1 mmHg may be enough to precipitate Fontan failure.

In high risk patients, the conduit connecting the IVC to the pulmonary artery may be fenestrated at the time of surgery; a 3- to 5-mm hole in the conduit anastomosed to the nearest atrium permits a small amount of systemic venous return directly to the heart. This acts to augment cardiac output with additional preload, but does so at the expense of oxygen saturation. The resulting reduced trans-pulmonary flow, in turn, permits lower systemic venous pressure. An increasing body of evidence shows fenestration protects from acute mortality and ‘failing Fontan’ morbidities.

Assessment Prior to Fontan Completion

Pre-Fontan assessment is still based on Choussat’s original criteria (see table), Assessment seeks to identify rhythm or conduction abnormality, pulmonary artery anatomy requiring surgical augmentation, estimation of PVR, and quantification of ventricular function and sub-systemic atrioventricular valve competence.

The indexed PVR desirable for completion of Fontan is <2.5 Wood U/m², though up to 4 U/m² may be tolerated. Accurate PVR is difficult to obtain via catheter because SVC flow can only be estimated as a portion of total cardiac output although flow

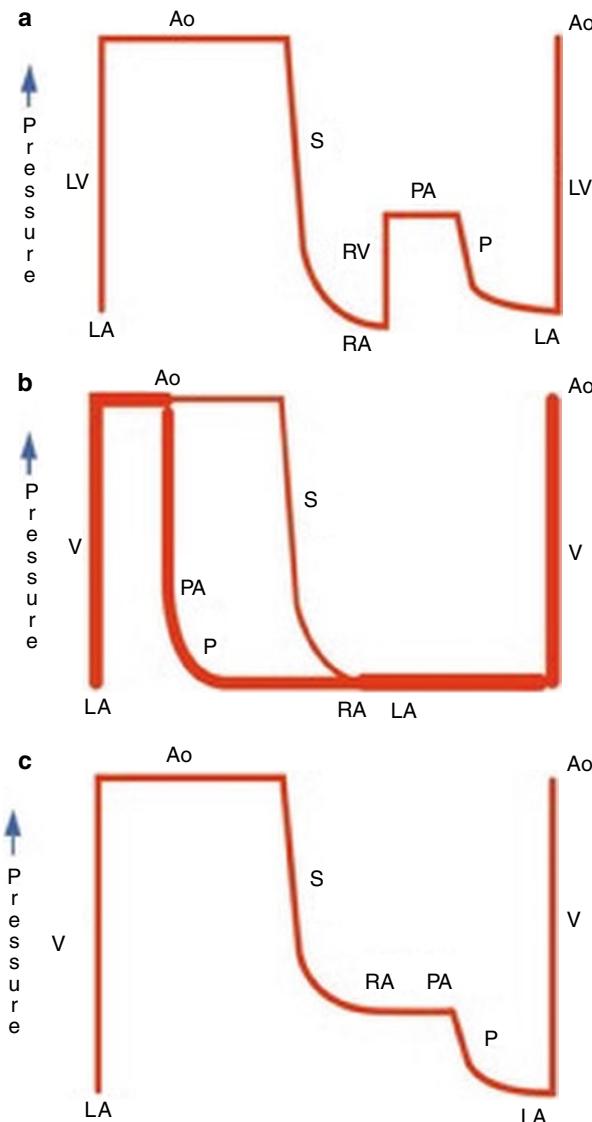


Fig. 13.4 (a) Normal biventricular circulation. Pulmonary circulation (*P*) is connected with the systemic circulation (*S*) via the right ventricle. The right ventricle maintains right atrial pressure lower than left atrial pressure, and provides enough energy for the blood to pass through the pulmonary resistance. (b) Arterially shunted palliation. The systemic (*S*) and pulmonary (*P*) circuits are connected in parallel, with a considerable volume overload to the single ventricle. There is complete admixture of systemic and pulmonary venous blood, causing arterial oxygen desaturation. (c) Fontan circuit. Systemic veins (*V*) are connected to the pulmonary artery, without a sub-pulmonary ventricle or systemic atrium. The lungs are thereby converted into a neo-portal system, which limits flow to the ventricle. In the absence of a fenestration, there is no admixture of systemic and pulmonary venous blood. *Ao* aorta, *CV* caval veins, *F* fenestration, *LA* left atrium, *LV* left ventricle, *PA* pulmonary artery, *RA* right atrium, *RV* right ventricle, *V* single ventricle. Line thickness reflects output (Reproduced from Heart, The Fontan circulation, Gewillig M, 91(6), 839–846, Copyright 2005, with permission from BMJ Publishing Group Ltd.)

Table 13.1 Choussat's 1977 selection criteria for identifying the ideal Fontan candidate (frequently referred to as 'The Ten Fontan Commandments'^a)

| |
|--|
| Age >4 years |
| Sinus rhythm |
| Normal systemic venous return |
| Normal right atrial volume |
| Mean pulmonary artery pressure <15 mmHg |
| Pulmonary vascular resistance <4 Wood U/m ² |
| Pulmonary artery-aorta ratio >0.75 |
| Left ventricular ejection fraction >60 % |
| Competent mitral valve |
| Absence of pulmonary artery distortion |

^aChoussat's 'Ten Fontan Commandments' published in 1977 originally intended for patients with tricuspid atresia

measurement using magnetic resonance imaging is now possible. Pre-Fontan trans-pulmonary pressure gradient is often used as a substitute at catheter. A trans-pulmonary gradient of 3 to 5 mmHg is desirable, >10 mmHg would generally preclude Fontan. Downstream pressures in the atrium and ventricle must also be assessed.

Once the additional flow of inferior caval blood is connected to the pulmonary arteries, the trans-pulmonary gradient increases. Simultaneously, cardiac output is affected by the acute reduction in ventricular pre-load. So, despite best efforts with thorough investigation, it remains difficult to predict individual post-Fontan performance.

Fontan Failure

Acute failure is not always predicted by the systemic venous pressure, as there is no current evidence defining the pressure at which a circulation will fail. Early studies performed in patients with lateral tunnel Fontan showed that systemic venous pressures in excess of 18 mmHg lead to a risk of death or Fontan take-down of around 50 %. Mid- to late-term clinical manifestations, such as effusions, ascites and protein-losing enteropathy may appear at any time in any patient with systemic venous hypertension (>10 mmHg). Wide variation is noted between individuals, probably due to interplay between ventricular function, lymphatic performance, vascular resistance, and secondary inflammation. Late failure of the Fontan is a separate topic not covered here but is described by Gewillig et al. for those seeking further reading.

Restriction of the Atrial Communication

If blood arriving into the left atrium can flow directly into the systemic ventricle without restriction (such as in double-inlet left ventricle with adequate mitral valve), an atrial communication may not even be required for cavopulmonary connection. More typically, a widely patent atrial communication is essential. Most Fontan

palliated hearts feature at least one atretic or stenotic inlet valve, or an atrium that connects to an inadequate, or obstructed ventricle. Examples include hypoplastic left heart, severe mitral stenosis, borderline left ventricle, tricuspid atresia, double-inlet left ventricle with inadequate mitral valve, or double-outlet right ventricle with restrictive ventricular septal defect.

The atrial septum is usually resected at the initial surgical palliation or may be performed at the time of the superior cavopulmonary anastomosis. Additional unroofing of the coronary sinus may be required to create an adequate orifice when the left atrium is small. Less common options for creating adequate atrial communications include trans-catheter stenting of the atrial septum, balloon/blade septostomy or balloon dilatation, though surgical septectomy is often performed eventually after these approaches.

Restrictive atrial communication despite previous septectomy in patients between first palliative surgery and superior caval connection is a recognised phenomenon. Delaying intervention to relieve a restrictive atrial septum at this stage is not recommended; the resulting raised pulmonary venous pressures may lead to irreversible change to the vascular bed and result in increased PVR, potentially precluding Fontan. The high preload to the left atrium in a patient following an arterial shunt (e.g., Blalock-Taussig shunt) provides high flow across the atrial septum, permitting straightforward diagnosis with transthoracic echocardiogram or cardiac catheter. Such patients are destined for further surgery; repeat septectomy can usually be carried out at the time of superior cavopulmonary connection.

In contrast, identifying an inadequate atrial communication in the interval between superior caval connection and Fontan completion may be difficult. If the atrial communication is an adequate size for trans-pulmonary SVC return, no pressure gradient will be apparent at pre-Fontan assessment. However, the increased flow of total cavopulmonary return may unmask the restriction, as a pressure gradient arises. Occurrence of restriction at the level of the atrial septum *after* Fontan is a rare finding and a potential contributor to Fontan circuit failure.

In the case described above, prior to Fontan completion the mitral valve and the atrial septum provided an adequate composite orifice for trans-pulmonary flow. A pressure gradient became apparent only once all cardiac output was directed back to the left atrium. Systemic venous pressures rose in order to overcome the obstruction resulting in prolonged pleural effusions.

Assessment of a Failing Fontan Circulation

In addition to routine echocardiography, patients demonstrating ‘failing Fontan’ morbidity require repeat haemodynamic assessment to identify reversible obstruction to flow. Pressure gradients obtained during haemodynamic assessment are always lower in the anaesthetised patient than that of the awake and active. It must be considered that cardiac output, intra-thoracic pressure and vascular resistance are all altered during anaesthetic. It is routine practice to record inspired oxygen

concentration and arterial CO₂. Because of the potential confounding factors, haemodynamic assessment with pharmacologic stress testing may be warranted. A dobutamine infusion administered to increase heart rate to 85 % predicted for age provides adequate ‘stress’ to unmask important gradients.

Advanced imaging such as 3D echocardiography and cardiac MRI may be utilised, according to the level of suspected obstruction and cardiac centre expertise. Assessment of respiratory physiology must not be neglected and may identify reversible causes of increased PVR, such as pneumonia, restrictive lung disease or post-operative paralysis of the hemi-diaphragm. Arrhythmias such as atrial tachycardia or junctional rhythm may significantly compromise the efficiency of the Fontan circuit and must also be excluded and managed.

Management of a Restrictive Atrial Communication

Decisions regarding management of a restrictive atrial communication must be led by symptoms, haemodynamic assessment, morphology and individual risk for further cardiac surgery. Redo surgery in Fontan patients is strongly associated with significant mortality and morbidity.

Trans-catheter stenting of the atrial septum may provide a lower risk option than surgery for some and is well described in pre-Fontan single ventricle patients. A conduit fenestration must be present, or else created at time of procedure. The left atrium must be adequate in volume to accommodate the length of stent and the fenestration must be an adequate distance from the atrial septum to avoid inadvertent stenting of the fenestration during deployment. Large-diameter stents that flare and shorten during deployment are preferred, short delivery balloons may be essential for the placement of large caliber stents within the confines of the atria. Regurgitation caused by damage to the systemic inlet valve by an embolised stent would result in significant compromise in a single ventricle circulation.

Fenestrations can be occluded with devices, or crimped by snaring at a later date, if pressures are acceptable but saturations low.

Learning Points

- Good Fontan physiology relies mainly upon a low trans-pulmonary gradient, with PVR the chief predictor of cardiac output. It must be remembered that ventricular end-diastolic pressure, the gradient across pulmonary veins, atrial septum and inlet valves all contribute to the eventual systemic venous pressure required to return blood to the ventricle.
- The perfect predictor of suitability for Fontan completion does not exist; cautious clinical interpretation of pre-Fontan haemodynamic data, derived from any method, is always required.

- A failing Fontan circulation requires thorough haemodynamic, respiratory and electrophysiological investigation. Sub-clinical and rare causes of compromise must be actively sought out and managed.
- Restrictive atrial septum is a rare, but reversible cause of raised Fontan pressures. A pressure gradient may only become manifest once inferior caval blood flow joins the circuit.
- Patients with double-inlet left ventricle require a widely open atrial communication if there is any suspicion that the left atrio-ventricular valve is too small to receive the entire cardiac output.

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Chapter 14

Is This Really Bronchiolitis?

Paraskevi Mikrou and Pavanasm Ramesh

Abstract Total anomalous pulmonary venous connection (TAPVC) is a rare type of congenital heart disease in which all four pulmonary veins connect to the systemic venous circulation, the right atrium or the coronary sinus. Signs and symptoms of TAPVC can sometimes mimic primary pulmonary pathology such as bronchiolitis, resulting in delay of diagnosis, especially during winter months when bronchiolitis is prevalent. Prompt diagnosis is vital as obstructed TAPVC is a true cardiac surgical emergency. We present the case of a 3-week-old female, who was admitted to our Paediatric Intensive Care Unit with presumed bronchiolitis, but was then found to have infracardiac TAPVC.

Keywords Infant • Total anomalous pulmonary venous connection (TAPVC) • Bronchiolitis • Cyanosis • Congenital heart disease • Hyperoxia test • Pulmonary venous confluence

Case Description

A 3-week-old female infant presented to her local hospital, referred by her GP because of poor weight gain. Her birth weight was 3 kg and at presentation she weighed 2.96 kg. Pregnancy was uneventful and antenatal scans were normal. She was born at term by normal delivery, there were no risk factors for sepsis and she was breast fed. Apart from mild gastroesophageal reflux, her mother had no other concerns in terms of feeding or breathing difficulties.

On arrival to hospital she was noted to be hypoxic with percutaneous oxygen saturations of 82 % in air. She was mildly tachypnoeic and had mild subcostal recession. Her oxygen saturations improved to 100 % in 45–50 % of headbox

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oxygen. There was no history of fever, cough or coryzal symptoms. Clinical examination revealed equal air entry bilaterally with inspiratory crackles, normal heart sounds with no murmurs and normal femoral pulses. Liver was palpable 3 cm below the right costal margin. Chest radiograph showed normal heart size, widespread coarse markings in both the perihilar regions with streaky opacification extending into the upper and lower lobes. These findings were reported as being consistent with bronchiolitis (Fig. 14.1a).

It was mid-October and the beginning of annual winter bronchiolitis epidemic in the UK. She was reviewed by a Consultant Paediatrician and a diagnosis of bronchiolitis was made. She was managed with headbox oxygen, antibiotics and continuation of breast feeds. Over the next 5 h she became increasingly tachypnoeic (respiratory rate 80 and 100 breaths/min) and tachycardic (heart rate 160–170/min). During a breast-feed she suddenly became pale and dropped her oxygen saturations to 40 %. Trial of continuous positive airway pressure (CPAP) failed; she was therefore intubated and ventilated and transferred to the Paediatric Intensive Care Unit (PICU).

On admission to PICU, she was noted to have normal heart sounds, no murmurs and good volume femoral pulses with a heart rate of 140–150/min and a blood pressure of 70/55 mmHg. Auscultation of chest revealed bilateral equal air entry with no

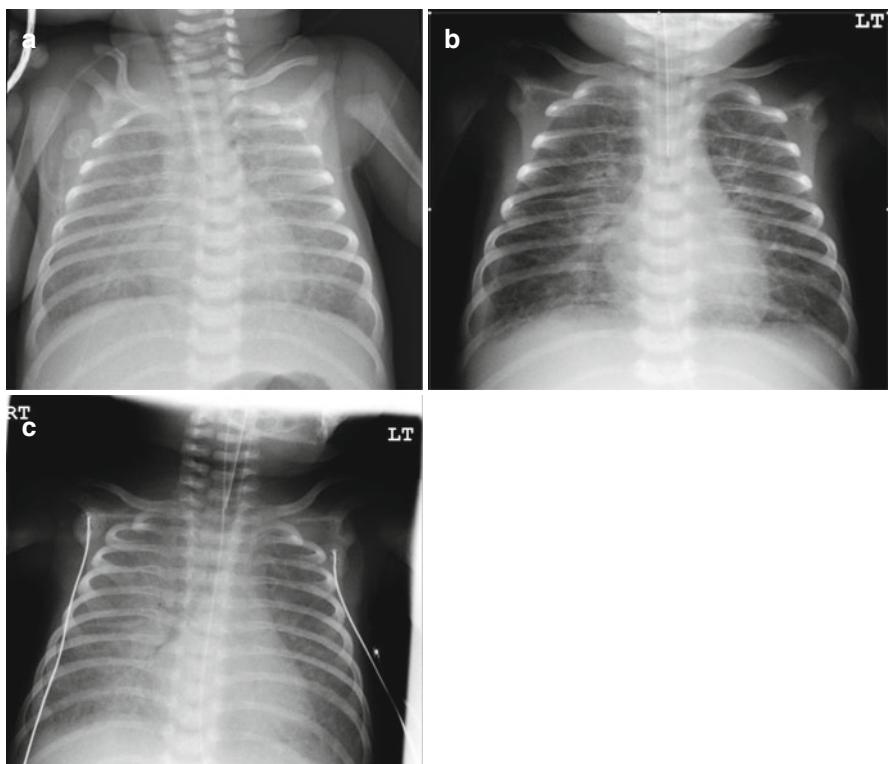


Fig. 14.1 (a, b, c) Serial chest radiographs showing a relatively normal cardiothoracic ratio and pulmonary venous congestion

additional wheeze or crackles. Liver was palpable 4 cm below the right costal margin. Endotracheal suction yielded thick white secretions. She was afebrile and her admission CRP was <4, with slightly elevated WCC ($17.6 \times 10^9/l$) and neutrophil count ($9.5 \times 10^9/l$). Radiology again reported the chest radiograph as showing ‘bilateral perihilar ground glass appearance with streaky opacification consistent with bronchiolitis’ (Fig. 14.1b).

During her stay, she required moderate ventilatory pressures (PIP 24, PEEP 6, rate 35/min) and the FiO₂ decreased from 60 % to 35 % maintaining her oxygen saturations greater than 95 %. Her arterial blood gases were satisfactory (pH 7.44, PCO₂ 4.12 kPa, PO₂ 9.2 kPa, BE R-3, Lactate 0.4). She was sedated and paralysed with morphine and rocuronium infusions. She passed an adequate volume of urine and nasogastric feeds were well tolerated. All these features reassured the team that the clinical diagnosis was most likely bronchiolitis. She did, however, become significantly bradycardic and hypoxic with any stimulation, such as suction or changing her position. It was believed that these episodes could be due to vagal stimulation, and the endotracheal tube was changed from oral to nasal in an attempt to resolve this issue.

Over the following 48 h, her ventilator settings remained more or less the same. However, she continued to have episodes of bradycardia, with a mottled appearance and profound desaturations during any intervention. Hyperoxia test (100 % FiO₂ for 10 min) yielded a PaO₂ of 31 kPa. She continued to have thick white secretions requiring regular saline washouts. In the meantime her respiratory secretions did not grow any bacteria or viruses. Repeat chest radiograph showed increased airspace shadowing, especially on the right side (Fig. 14.1c).

In view of negative microbiology with low inflammatory markers, persistent desaturations with handling, static clinical course and worsening chest x-ray picture, an echocardiogram was requested on day 3 of admission. This showed infracardiac total anomalous pulmonary venous connection (TAPVC) with no obvious obstruction and an atrial septal defect (ASD) with right to left flow. A 12-lead ECG showed normal sinus rhythm, right axis deviation (QRS +120), tall R-waves in V₁/V₂ and inverted T wave, consistent with right ventricular hypertrophy (Fig. 14.2).

Following the diagnosis, she was referred to the local cardiology centre, and arrangements made for transfer. In the next 24 h, she deteriorated and behaved as though the pulmonary veins were obstructed, requiring increasing respiratory and cardiovascular support. She was transferred to the cardiac surgical centre on the fourth day following her admission to hospital. A CT scan was performed to define the pulmonary venous anatomy and confirmed infracardiac TAPVC.

She was operated 5 days after her initial presentation to hospital. Intraoperative findings confirmed an infracardiac TAPVC, with a vertical connecting vein formed by the confluence of right and left pulmonary veins. This connecting vein drained to inferior vena cava via the portal vein. An anastomosis was performed between the connecting vein and left atrium, and the ASD was closed. Her post-operative period was complicated with recurrent pleural effusions. She was eventually discharged home 1 month after her operation. Follow-up 6 weeks later revealed a thriving baby

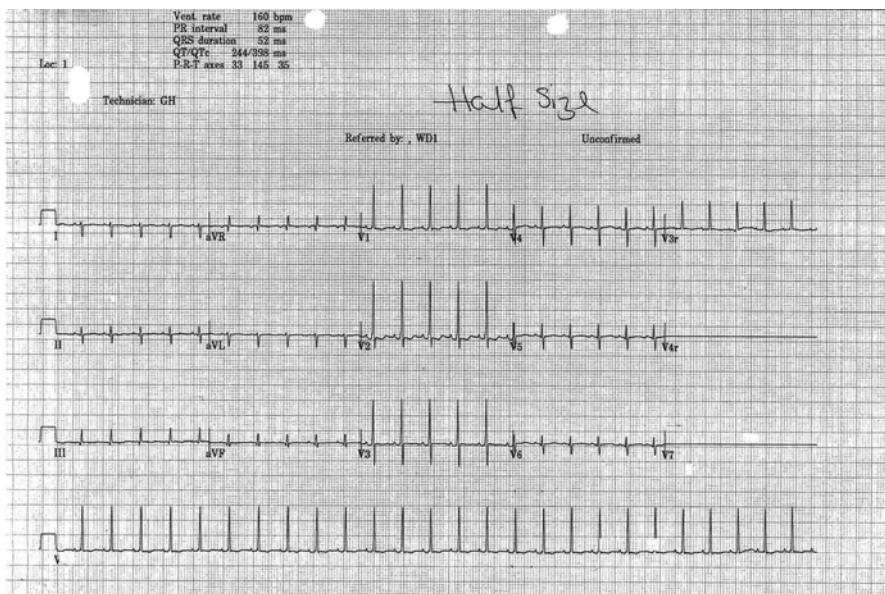


Fig. 14.2 Twelve-lead ECG, half standard, illustrating right ventricular hypertrophy

who was feeding and growing well. Repeat echocardiogram confirmed a good surgical result with a widely patent pulmonary venous anastomosis. She is now 2½ years old and is doing well.

Discussion

Total anomalous pulmonary venous connection (TAPVC) is a rare form of congenital heart disease in which all pulmonary veins connect to the systemic veins, right atrium or coronary sinus. In order to survive, there is an obligate interatrial communication. TAPVC can occur in conjunction with a wide variety of other cardiac malformations, in particular atrial isomerism. The disease was first described by Wilson in 1798.

There are different types of TAPVC defined by the site of connection of the pulmonary venous pathways. The pulmonary veins usually join to form a common chamber (confluence), but this does not connect to the morphological left atrium, instead joining the systemic veins above the heart via a vertical vein (supracardiac TAPVC), or systemic veins below the heart (infracardiac TAPVC), or directly to other parts of the heart (cardiac TAPVC). Mixed TAPVC occurs when some of the pulmonary veins connect at one site and some to another. The confluence can vary in shape. It is usually posterior to the pericardium with the long axis orientated transversely; however, in infracardiac connection the pulmonary veins join the vertical vein to form a tree-shaped confluence. In mixed TAPVC, there may not be a common confluence.

The pulmonary venous pathways in TAPVC can be obstructed by several mechanisms: (1) localised stenosis can occur at the junction between the vertical vein and the systemic circulation or coronary sinus (2) the vertical vein can be relatively narrow (3) in supracardiac TAPVC, the left vertical vein may pass posterior rather than anterior to the left pulmonary artery and is compressed between it and the left main bronchus (4) stenoses of the individual pulmonary veins, and (5) by a restrictive interatrial communication.

Before the advent of cardiac surgery, almost all these children died in the first few months of life. Muller reported the first successful surgical repair of TAPVC in 1951; however, survival rates in infancy were extremely poor until the late 1960s. The principle of operative repair is to establish an unobstructed communication between the pulmonary veins and the left atrium, close the atrial septal defect and usually to interrupt the connections with the systemic venous circulation. The specific repair is dependent on the type of anomalous connection and there is variation in techniques used. Whichever technique is used, it is essential that no kinking or twisting occurs at the anastomosis. With improvement in intraoperative techniques there has been continued improvement in the mortality and morbidity of isolated TAPVC, with surgical mortalities less than 5 % in more recent series. This cannot only be attributed to progress in surgical expertise, but also to developments in intensive care such as use of nitric oxide and extracorporeal membrane oxygenation that have led to salvage of the sickest neonates. Pulmonary hypertensive crises are a major contributor to postoperative morbidity and mortality. Earlier diagnosis and improvements in transthoracic echocardiography have also limited the use of risky preoperative angiography. Despite the improvement over time, there is an ongoing late mortality in patients with TAPVC frequently associated with postoperative pulmonary venous occlusion. Many studies have shown that postoperative pulmonary venous obstruction occurs with an incidence of 0–17 %. This presents a major hurdle for improving ultimate outcome of TAPVC.

Presentation

Prenatal diagnosis is rare, with most reported series including patients with atrial isomerism and/or functionally univentricular hearts. Most patients present in early infancy or childhood, however, there have been occasional reports of TAPVC presenting in adulthood. Timing and mode of postnatal presentation of TAPVC depend upon whether the pulmonary venous pathways are obstructed and the presence of coexisting intracardiac malformations. Coexisting intracardiac malformations that reduce pulmonary blood flow, such as pulmonary stenosis or atresia, may mask clinical recognition of severe pulmonary venous obstruction.

Preoperative pulmonary venous obstruction results in raised pulmonary venous pressure and subsequent pulmonary oedema. Neonates with obstructed TAPVC present early cyanosed and breathless, sometimes being misdiagnosed as persistent pulmonary hypertension of the newborn. Brown found that some neonates referred

for extracorporeal membrane oxygenation with presumed persistent pulmonary hypertension did, in fact, have TAPVC illustrating the importance of early accurate echocardiography.

If no obstruction is present, there is increased pulmonary blood flow often with presentation at a later age with non-specific symptoms such as failure to thrive, mild cyanosis or features similar to a large left to right shunt such as dyspnoea and tachypnoea.

Diagnosis

Clinical picture varies depending on whether the TAPVC is obstructed or not. ECG will typically show signs of right ventricular hypertrophy and right axis deviation. Chest radiograph will demonstrate pulmonary venous congestion with a relatively normal cardio-thoracic ratio in obstructed TAPVC; as with unobstructed TAPVC there are moderate to marked cardiomegaly and pulmonary plethora.

Both obstructed and unobstructed forms of TAPVC can pose a true diagnostic challenge, as initial clinical presentation can resemble a respiratory illness. In our patient the clinical picture was consistent with bronchiolitis, which is the commonest cause of respiratory distress in young infants, especially during winter months. The initial chest radiograph appearances, the moderate ventilatory requirement and the blood gas results were all typical of a baby with bronchiolitis. Oxygen saturations of up to 100 % and the results of the hyperoxia test gave false reassurance that there was no cyanotic congenital heart disease. However, it has been described in the literature that infants with TAPVC can have a normal hyperoxia test. It is important to remember the assessment of a hyperoxia test should be purely based on arterial PO₂ values and not percutaneous oxygen saturation as fetal haemoglobin has higher affinity to oxygen (i.e., left shift of the oxygen-haemoglobin dissociation curve).

In our case, there were an unusually high number of significant desaturation episodes with handling; this is not routinely encountered in infants with respiratory infections. Several explanations have been suggested to explain the pathophysiology of such episodes in patients with TAPVC: increased pressure in the pulmonary veins produces reflex vasoconstriction of the pulmonary arterioles and raises the pulmonary artery pressure. In addition, an elevated pulmonary venous pressure may narrow or close small airways, causing alveolar hypoxia and contributing further to vasoconstriction. To support an underlying cardiac defect, our patient had signs of right ventricular hypertrophy on the ECG and worsening pulmonary venous congestion on subsequent chest radiographs; the latter is a particularly important “red flag” for diagnosis of TAPVC. Negative viral screen and normal inflammatory markers were also supportive of an alternative cause for her symptoms other than a respiratory infection. All of the above justified the clinical decision to perform an echocardiogram.

Definitive postnatal diagnosis of TAPVC is usually by echocardiography, with angiography being reserved for cases where the diagnosis is not clear. Even in the hands of an expert echocardiographer this is a difficult diagnosis to make, so there should be a low threshold to get a second opinion if the pulmonary veins cannot be clearly evaluated. Cardiac magnetic resonance or CT imaging can also be used to evaluate the pulmonary veins as in this case.

Learning Points

- Every child presenting with symptoms of bronchiolitis must have thorough history and examination to rule out underlying cardiac pathology even during peak of bronchiolitis season.
- Sometimes babies with congenital heart disease present to hospital because of associated bronchiolitis; therefore, even in confirmed cases of bronchiolitis, an underlying cardiac condition must be suspected if the course of illness is not typical.
- Clinical diagnosis of TAPVC can sometimes be extremely difficult.
- A hyperoxia test suggestive of lung pathology and oxygen saturations that rise to 100 %, do not exclude a diagnosis of TAPVC.
- Infracardiac TAPVC usually becomes obstructed with clinical deterioration and transfer to a cardiac surgical centre for intervention should not be delayed.

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Chapter 15

A Neonatal Dilemma

Shree Vishna Rasiah

Abstract Persistent pulmonary hypertension of the newborn (PPHN) is not an unusual presentation in term babies. This challenging condition can mimic the presentation of cyanotic congenital heart disease. We present a case of a term baby with antenatal diagnosis of an exomphalos who deteriorated following delivery and was managed as PPHN and treatment was escalated. The initial echocardiographic findings were suggestive of PPHN. There was no clinical improvement after 72 h and repeat echocardiography showed TAPVC (total anomalous pulmonary venous connection). The management was reoriented and the baby was transferred to the regional cardiology centre for surgical management. We would like to emphasise that the echocardiographic findings of TAPVC can be very similar to severe PPHN. It is therefore paramount to ensure that all the pulmonary veins are seen returning to the left atrium. Finally, we should always reassess both clinically and echocardiographically when there is not the expected clinical improvement in babies with PPHN.

Keywords Persistent pulmonary hypertension of the newborn • PPHN
• Echocardiography • Total anomalous pulmonary venous connection

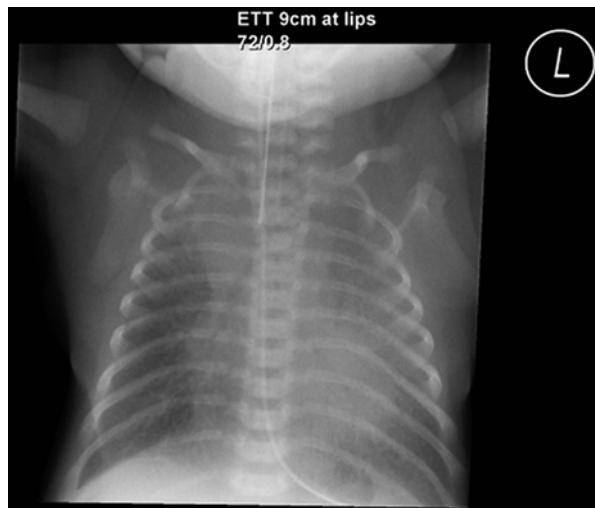
Case Description

On a Friday morning a term baby was delivered by elective section following an antenatal diagnosis of large exomphalos. This was the mother's first pregnancy and there were no other antenatal concerns apart from the exomphalos. They were white British in origin and were not consanguineous. The baby was born in good condition but quickly developed signs of respiratory distress. Her APGARs were 8 at 1 min, 7 at 5 min and 7 at 10 min.

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Fig. 15.1 First chest x-ray soon after intubation and ventilation. This was thought to be in keeping with surfactant deficient respiratory distress syndrome

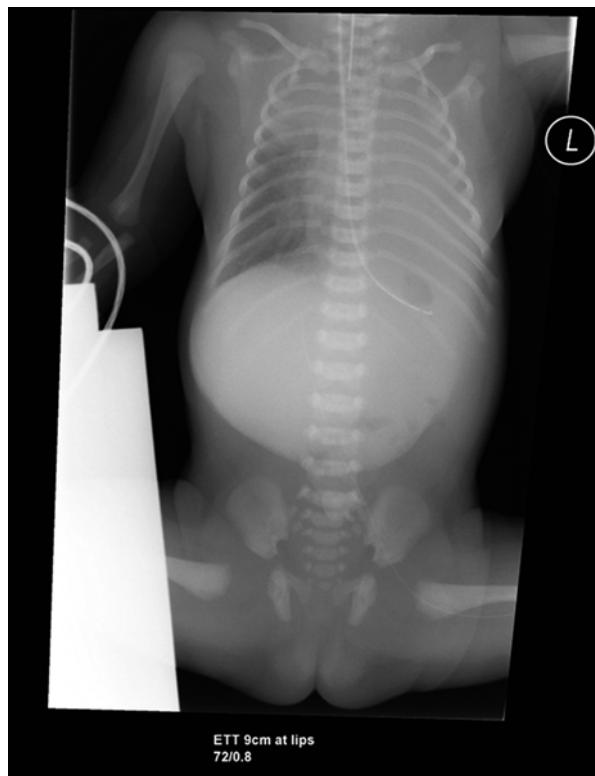


On admission to the neonatal unit, she had worsening respiratory distress and her first capillary blood gas was poor. In view of this she was electively intubated and ventilated. An infection screen was performed and she was started on intravenous antibiotics to cover for infection as a possible cause for her poor condition. She had very poor oxygenation despite ventilator support with a significant pre- and post-ductal saturation difference suggesting persistent pulmonary hypertension of the newborn (PPHN). Her management was escalated to support a diagnosis of PPHN. She was paralysed and sedated and oscillated on the ventilator. An echocardiogram was performed because of the high oxygen requirement and poor saturations. The echocardiogram showed a moderate-sized ventricular septal defect with bidirectional shunt across it, tricuspid regurgitation jet of 3 m/s on continuous wave Doppler interrogation and a large patent ductus arteriosus with bidirectional shunting. The aortic arch was slightly small but there was no obvious aortic coarctation in the presence of a large PDA. The rest of the heart was thought to be structurally normal. These findings were thought to be in keeping with PPHN. Therefore, inhaled nitric oxide (iNO) was commenced for the management of severe PPHN.

Her first chest x-ray following intubation and ventilation is shown on Fig. 15.1. The endotracheal tube was in a good position. The lung fields were generally hazy on both sides. The left lung fields appeared more affected than the right. These findings were thought to be in keeping with respiratory distress syndrome. Figure 15.2 shows the chest and abdominal x-ray. This was done to check the position of the long line which was inserted for the infusion of the inotropes. The inotropes were started to support the cardiovascular system. She required dopamine, dobutamine and adrenaline infusions. She was also on regular hydrocortisone and had magnesium sulphate to help manage PPHN.

Over the next 3 days, she continued to be managed with high-frequency ventilation, inhaled nitric oxide and inotropic infusions, in line with the presumed diagnosis of PPHN. Over this weekend, she remained clinically stable on this support but there

Fig. 15.2 Chest and abdominal x-ray showing the exomphalos and the position of the long line inserted for the infusion of the inotropes to support her cardiovascular system



was no opportunity to wean the inspired oxygen, which remained at 100 % on 20 ppm of iNO. Her arterial blood gases were satisfactory during this period. Therefore, little change was made with the hope that with supportive management the pulmonary hypertension would improve.

On Monday, day 4 of life, she was assessed by a different team who were starting their week on service. On review it became clear that, although her clinical parameters and blood gases were satisfactory on the support she was receiving, there was no evidence of improvement of her oxygenation. The presumptive diagnosis of PPHN was therefore challenged and it was noted that her oxygen saturations did not deteriorate when the oxygen was reduced. The question of possible cyanotic congenital heart disease was raised, given the lack of response to the management of PPHN and supplemental oxygen.

An echocardiogram was immediately repeated which showed evidence of a supra-cardiac total anomalous pulmonary venous connection (TAPVC) (Fig. 15.3). The findings were discussed with the paediatric cardiologist at the regional paediatric cardiac centre who agreed with the findings. Following this, the support was slowly weaned, and the baby came off all inotropes and iNO and was managed with ventilation alone. She was transferred to the paediatric intensive care unit at day 7 of life for the surgical management of her TAPVC.

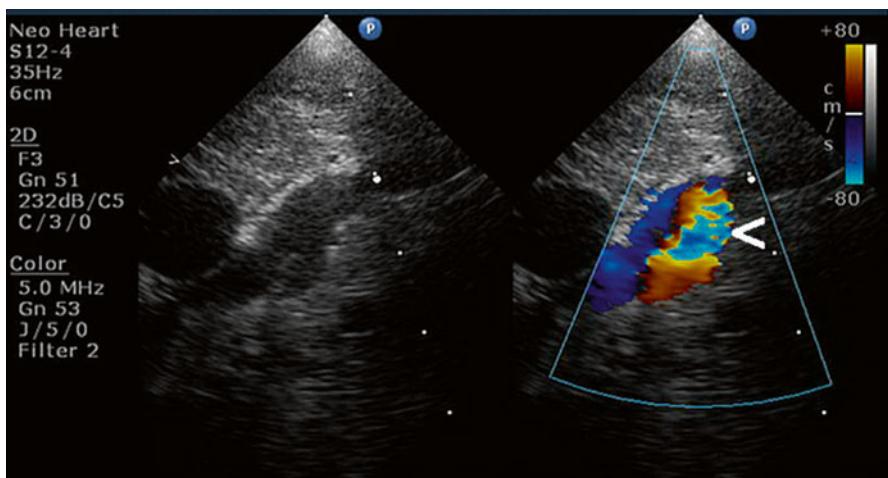


Fig. 15.3 Echocardiogram image showing the ascending vessel (*white arrow*) connecting to the left superior vena cava

Discussion

Persistent pulmonary hypertension of the newborn (PPHN) is not an unusual presentation in term babies for various reasons. It occurs because the fetal circulation has failed to make the transition to the neonatal circulation after delivery and there is a persistently raised pulmonary vascular pressure. It is a medical emergency in the neonatal period as babies with PPHN have an increased risk of morbidity and mortality if not managed appropriately.

The clinical presentation of a baby with PPHN includes poor oxygen saturations and the presence of a pre- and post-ductal saturation difference despite being in 100 % oxygen. Babies with severe PPHN can fail the hyperoxia or nitrogen wash-out test; hence, they can be challenging to differentiate from a cyanotic congenital heart condition. In some cases there are factors that put babies at risk of PPHN, e.g., meconium aspiration, sepsis, pneumonia, hypoxic ischaemic encephalopathy or pulmonary hypoplasia secondary to congenital diaphragmatic hernia, oligohydramnios or pleural effusions.

Echocardiography helps differentiate congenital heart disease from PPHN. The echocardiographic findings of severe PPHN may include right to left shunt across the patent foramen ovale (PFO), right to left shunt through the patent ductus arteriosus, significant tricuspid regurgitation that has high velocity when interrogated with continuous wave Doppler, and right heart dilatation sometimes with reduced right ventricular systolic function. These echocardiographic findings in the presence of a structurally normal heart and clinical presentation would suggest PPHN. All these echocardiographic findings, especially the shunt across the PFO, are seen on echocardiogram of babies with TAPVC. The only difference being that in patients with TAPVC the individual pulmonary veins do not connect to the left atrium correctly.

It can be challenging for the echocardiographer to distinguish these from cases with PPHN.

The primary role of echocardiography in the assessment of a cyanotic baby with suspected PPHN is to confirm a structurally normal heart. The pitfalls for the neonatologist, paediatrician with a special interest in cardiology or trainees performing and interpreting echocardiography in these settings stem from the fact that they have failed to demonstrate a structurally normal heart. In my experience, the diagnoses which have been missed in these settings are TAPVC, where the echocardiographer has not demonstrated all the pulmonary veins draining into the left atrium, and TGA, where they have not demonstrated the normal spatial relationship of the major arteries. In these settings the management of these babies have been compromised with the escalation of treatment to presumably treat a suspected PPHN. Fortunately, in this case the diagnosis had come to light when there was no clinical response to the escalation of management for PPHN and a further assessment by another echocardiographer had taken place.

Failure to progress in a patient with PPHN and a worsening picture on chest x-ray should always lead to prompt reconsideration of the diagnosis of PPHN. Further assessment of the heart is necessary particularly to assess the connection of the pulmonary veins. If echocardiography is not definitive, further imaging, such as computerised tomography, magnetic resonance imaging, or angiography, should be considered.

Learning Points

- The clinical presentation of a baby with signs of PPHN can mimic the presentation of cyanotic congenital heart disease.
- Echocardiography findings of TAPVC can look similar to PPHN.
- Primary role of echocardiography in the assessment of a cyanotic baby suspected of having PPHN is to confirm a structurally normal heart. Particular attention should be taken to ensure that the individual pulmonary veins are connected to the left atrium.
- There should be clinical evidence of improvement of babies managed with PPHN.
- Lack of improvement following escalation of management for PPHN should warrant a reassessment of the initial diagnosis with further imaging of the heart.

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Chapter 16

The Collapsing Teenager

Andrew B. Ho and James P. Gnanapragasam

Abstract In this chapter we describe a teenager with exertional collapse. He was found to have an aberrant left coronary artery arising from the right coronary sinus and running between the great arteries. Although this lesion is uncommon, there is a strong association with sudden cardiac death, particularly during or immediately after strenuous exercise. Diagnosis requires careful echocardiographic evaluation of the coronary arteries often supported by CT. We discuss the presentation, diagnosis and management of this lesion.

Keywords Paediatrics • Cardiology • Exercise • Sudden death • Coronary artery • Echocardiography • Computed tomography • Exercise testing

Case Description

A 13-year-old, previously well boy presented after two episodes of exertional collapse within two months. On each episode he had been at maximal exertion (the first while running and the second while playing football) before developing an abnormal awareness of his heart beating, shortness of breath followed by acute loss of consciousness. When he came round he was aware of palpitations for up to an hour. There was no history of chest pain or nausea.

ECG in the accident and emergency department following his second collapse (Fig. 16.1) demonstrated ST-segment depression in the inferolateral leads which normalised over the next few hours. An urgent referral was made for cardiology review.

Echocardiogram demonstrated mild left ventricular hypertrophy (interventricular septum in diastole 1.7 cm, posterior wall 1.2 cm). Inspection of his coronary arteries demonstrated a left coronary artery arising aberrantly from the right

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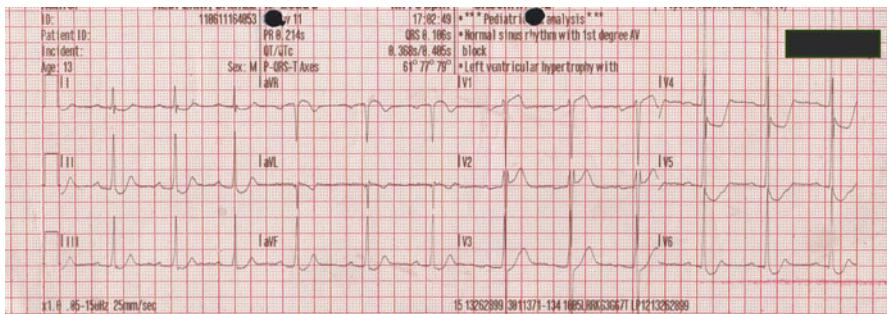


Fig. 16.1 Admission ECG demonstrating widespread ST segment changes

coronary sinus and coursing between the aorta and pulmonary artery (Fig. 16.2a–c). There were no regional wall motion abnormalities and function was normal with a fractional shortening of 37 %.

A computed tomogram (CT) of his chest was performed to further assess coronary anatomy (Fig. 16.3a, b). This demonstrated the left main stem arising from the right coronary sinus separate from the right coronary artery with a hooded orifice and narrowing through its inter-arterial portion with no intramural course. Cardiac function was normal.

He was discussed at the multidisciplinary meeting and the decision made for surgical repair. At surgery, he was found to have a slit-like orifice of his left main stem from the right aortic sinus with an inter-arterial course. His left coronary artery was reimplanted into the left sinus. He came off bypass easily and made an uneventful recovery.

Follow-up investigations included an exercise tolerance test and cardiac magnetic resonance imaging. On exercise testing he has completed 12 min of the Bruce protocol with no ischaemic changes. MRI scan has shown an unobstructed course of the left coronary artery and good left ventricular function with no evidence of myocardial scarring. He has remained perfectly well in the 3 years since his operation.

Discussion

Normal Coronary Anatomy

The right coronary artery normally arises from the right coronary sinus, before coursing between the right atrial appendage and right ventricular infundibulum through the right atrioventricular groove. A high conal branch emerges in 50 % followed by a sinoatrial nodal branch in 50 %. Marginal vessels then emerge to supply the right ventricular free wall. The vessel then continues posteriorly in the right atrioventricular groove to the posterior interventricular groove, where in 90 %, the posterior descending coronary artery emerges to run in the posterior AV groove and supply the posterior third of the interventricular septum.

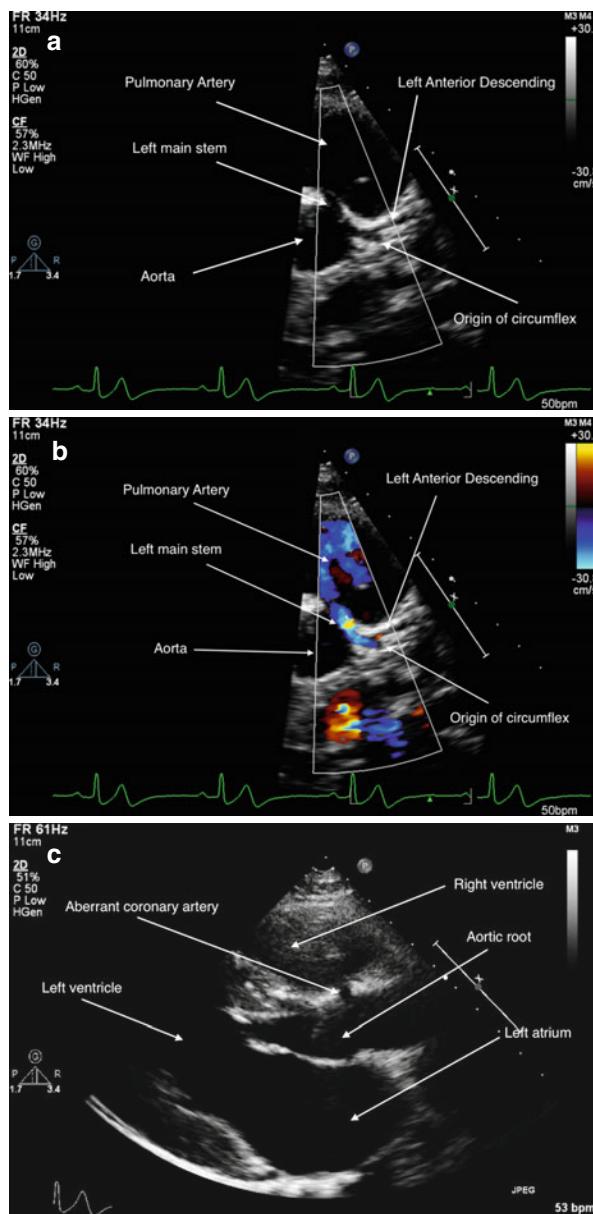


Fig. 16.2 (a) Echocardiographic 2D image of left coronary artery demonstrating interarterial segment of LCA followed by normal bifurcation. (b) Colour flow image of LCA demonstrating interarterial segment. (c) 2D image in the parasternal long axis showing the aberrant coronary artery

The left coronary artery (LCA) emerges from the left coronary sinus and covers a short distance under the orifice of the left atrial appendage before dividing into the left circumflex (LCx) and left anterior descending (LAD). The LCx runs in the left

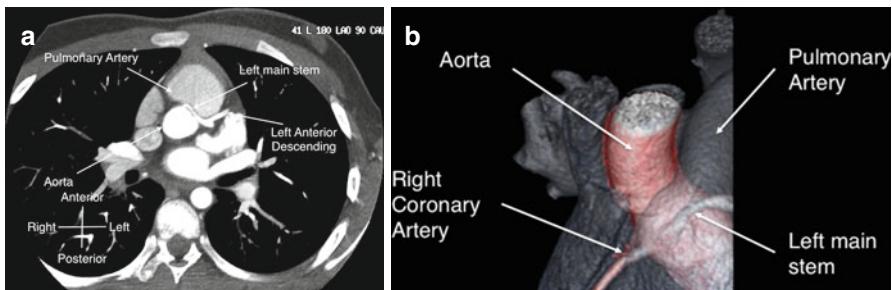


Fig. 16.3 (a) Transverse slice from CT demonstrating left main coronary artery arising from anterior (right coronary) sinus and coursing between aorta and pulmonary artery. (b) CT reconstruction of aortic root. The aorta is coloured red and the pulmonary artery grey. The left coronary is seen emerging acutely from the right sinus and then coursing between the great vessels

atrioventricular groove, giving off obtuse marginal branches which supply the free wall of the left ventricle. In the majority of patients, the circumflex ends towards the posterior interventricular groove, although in 10 % the vessel continues into the posterior interventricular groove as the posterior descending artery. The LAD passes down the anterior interventricular groove, giving a left conal branch, diagonal branches to left and right ventricular free walls and septal perforator vessels to feed the anterior two-thirds of the interventricular septum.

The atrioventricular nodal artery is given by the right coronary artery in 50 %, and in the remainder from the left coronary. A similar division of frequencies is seen for the origin of the sinoatrial artery.

Variants of Coronary Anatomy

As is clear from the preceding description of “normal” anatomy, a plethora of variants of coronary anatomy can be seen, from the benign (e.g., conal branch from the right coronary sinus, separate origins of LCx and LAD from the left coronary sinus), through lower risk lesions (e.g., right coronary from left coronary sinus) to high-risk lesions (e.g., anomalous left coronary artery from pulmonary artery).

With aberrant origin of the left coronary from the right coronary sinus (ALCA-R), pathogenicity is related to the proximal course of the vessel before it bifurcates. There are three possibilities: rightward and posterior to the aorta, running inferiorly into the ventricular mass before emerging adjacent to the left coronary sinus, or an interarterial course between the two great vessels.

This interarterial course (which can be either with or without an intramural portion inside the aorta) has a well-described association with sudden cardiac death. Previously unidentified coronary lesions have been found in approximately 15 % of sudden unexpected deaths in young athletes, and a 25-year study of American military recruits who suffered non-traumatic deaths found a third of those with

identifiable causes had ALCA-R. The majority of deaths have occurred between 15 and 30 years of age.

The overall incidence of ALCA-R has been estimated from screening otherwise normal teenagers at between 0.1 % and 0.15 %. Estimation of the annual risk of death is difficult as there are no prospective cohorts from screening data, although the estimated risk is between 0.1 % and 0.4 % per annum.

The pathological effect is related to the course of the vessel and the effect of the aberrant course on the shape of the ostium. The strong association with exercise suggests that increased pressures in the aorta during exercise leads to coronary compression of the through its interarterial course. This has been observed in patients with ALCA-R investigated with intravenous ultrasound. The coronary ostium is often oval in shape and emerging from the sinus at acute angle, which may further deform during exercise, thereby limiting flow. There is also the risk of late arrhythmias secondary to scarring from recurrent episodes of subclinical ischaemia.

Presentation

Precise definition of symptomatology is difficult, as collapse may be the primary event. Patients have also presented with aborted sudden death but overall only half of patients will have experienced prior symptoms which may include nonspecific chest pain or palpitations, which are common in the general population. While these symptoms have limited predictive power, any symptoms related to exertion must always be taken seriously

Investigations

The key investigation is the echocardiogram. It is important to identify the coronary origins, the flow direction and exclude any vessel running in between the two great arteries. Other features may include regional wall motion abnormalities or a bright appearance of the myocardium, although these are likely to be late findings.

The images of the origins and proximal courses of the coronary vessels are usually obtained in the parasternal short-axis views, often with the probe rotated clockwise towards the 3 o'clock position. Increasing transducer frequency will often improve diagnostic power, and reducing sector size will improve temporal resolution. Colour-scale adjustments should be made to optimise the balance between demonstration of flow in the coronaries and avoiding colour bleed from nearby structures. A specific search should be made between the great vessels for an intra-arterial coronary. The abnormal vessel may also be seen in the parasternal long-axis views, as demonstrated in Fig. 16.2c.

Echocardiographic examination of the coronaries can be difficult. Suboptimal windows can make delineation of the coronaries difficult, nearby coronary veins

can be mistaken for arterial vessels as can the echo-free spaces in relation to the pericardial sinuses. An abnormal vessel may also easily be missed, as in ALCA-R the vessel turns sharply leftward away from the aorta where the ‘normal’ left coronary artery would emerge, giving appearances very similar to a normal ostium (see Fig. 16.2a, b).

Electrocardiograms during pain or immediately following a collapse may demonstrate ST changes (see Fig. 16.1), ventricular arrhythmias or may be entirely normal. Late changes may include pathological Q waves as evidence of infarction.

Although one would expect exercise testing to be diagnostic in ALCA-R, there is documentation of sudden death in athletes with undiagnosed ALCA-R who have previously undergone maximal exercise testing. Indeed, some studies have demonstrated normal exercise test results prior to surgery and evidence of ischaemia postoperatively.

Although invasive coronary angiography has long been considered the gold standard investigation for delineation of coronary arteries, computed tomography (CT) and magnetic resonance imaging (MRI) are increasingly finding a role.

Both MRI and CT offer high-quality imaging of the proximal coronaries, with CT in particular offering superb spatial resolution with short scan times, removing the need sedation/anaesthesia, which is required for MRI scanning in children. MRI, however, does have the ability to analyse cardiac function and can detect scar formation using late gadolinium enhancement.

Management

Although there may be controversy regarding the management of some variants of coronary anatomy, ALCA-R will always require surgical repair. Pending surgery it is prudent to try limit exercise or at least competitive sports.

There are several options for surgical repair:

- Deroofing of the intramural segment, which involves opening the intramural course of the aberrant vessel into the aorta out to the sinuses of Valsalva
- Creation of a neo-ostium by local deroofing of the left coronary where it crosses the left coronary sinus
- Disconnection of the aberrant vessel and reanastomosis to the correct sinus
- Pulmonary artery translocation
- Coronary artery grafts surgery may be performed as a definitive procedure or in addition to the above.

There has been no early operative mortality in any of the case series published to date, although it should be noted that aortic regurgitation can develop with procedures involving re-suspension of an aortic valve cusp.

From the limited data available due to the rarity of this condition, the vast majority of patients appear to have good long-term outcomes, with most returning to full participation in sports. A handful of studies have reported changes on exercise

testing post procedure (not present pre-op) and late symptoms, so long-term follow-up is warranted.

Learning Points

- Patients presenting with exertional collapse require careful evaluation.
- Exercise testing may be normal in patients with pathological variants of coronary artery anatomy.
- Echocardiographic evaluation of the proximal coronary arteries requires careful examination of the regions close to the aortic sinuses.

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Chapter 17

Dilated Cardiomyopathy: Think of the Diet

David F.A. Lloyd

Abstract A term, breast-fed infant with a known inherited bile acid transport defect and cholestatic jaundice presented collapsed with signs of congestive heart failure at 6 months of age. He was diagnosed with severe dilated cardiomyopathy (DCM) and stabilised with medical treatment.

As part of diagnostic screening for DCM he was found to be severely vitamin D deficient, due to a combination of cholestatic liver disease and maternal vitamin D deficiency. Once appropriate supplementation was instituted he made a complete recovery over the following 12 months with restitution of normal cardiac function.

Whilst vitamin D deficiency is relatively prevalent, presentation with vitamin D dependent cardiomyopathy is rare. Cardiac function is often severely impaired at presentation, and the disease course can be fulminant and rapidly life-threatening without urgent treatment, including mechanical circulatory support and/or primary cardiac transplantation. Despite the high early mortality, however, vitamin D deficient cardiomyopathy is one of the few treatable causes of dilated cardiomyopathy with an excellent long-term prognosis in those who survive their initial presentation.

This chapter highlights a rare, life-threatening, and potentially fully reversible cause of dilated cardiomyopathy, and the importance of rigorous and comprehensive screening for patients with unexplained DCM.

Keywords Cardiology • Congenital heart diseases • Paediatrics • Cardiomyopathy • Dilated cardiomyopathy • Heart failure • Vitamin D • Cholestasis • Hepatology

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Case Description

A 6-month-old exclusively breast-fed infant of Asian background presented to the Emergency Department in cardiogenic shock with signs of congestive heart failure, on the background of a 4-week history of poor feeding and reduced weight gain. Transthoracic echocardiography showed a structurally normal heart with normal coronary artery origins. The left ventricle was dilated with severely impaired global systolic function (ejection fraction 14 % by Simpson biplane method) (Fig. 17.1a, b).

The child had been previously investigated for cholestatic jaundice, diagnosed at 3 weeks of age. An inherited disorder of bile transport was suspected and medical management had been instituted, including oral multivitamin supplements.

A diagnosis of dilated cardiomyopathy was made and the child was stabilised with respiratory support and diuretic therapy. Due to the severity of the presentation, the parents were counselled that the disease course was not easy to predict, and he was referred for urgent cardiac transplant assessment with the caveat that this may be contraindicated due to the history of liver disease. A full cardiomyopathy

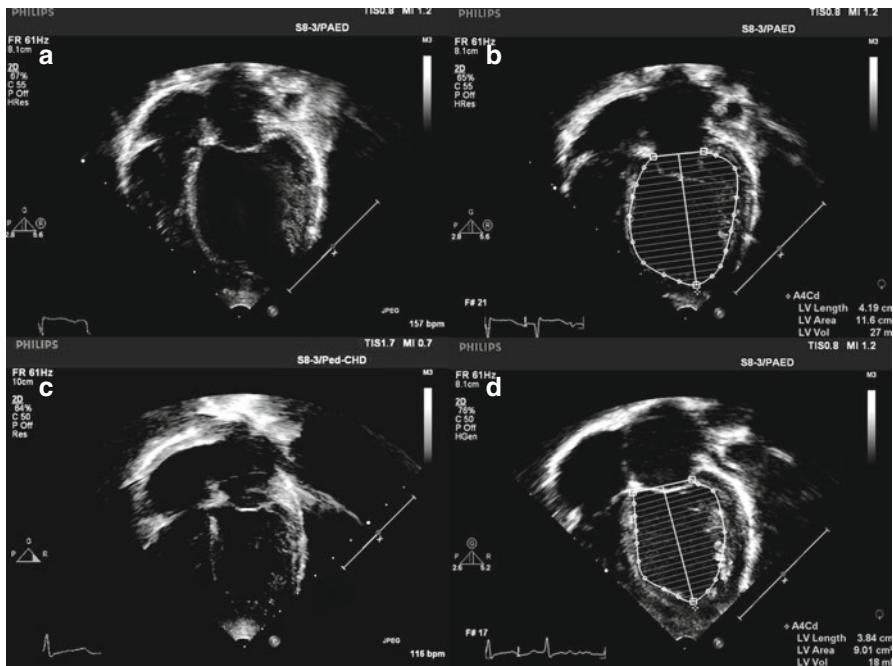


Fig. 17.1 (a) Apical four-chamber view at presentation showed a dilated left ventricle with impaired systolic function and (b) Simpson's biplane calculation showing estimated LV diastolic volume of 27 mls (90 mls/m²). (c) Apical four-chamber view 10 months after presentation showing a reduction in LV volume and improved systolic function and (d) Simpson's biplane calculation showing estimated LV diastolic volume of 18 mls (60 mls/m²)

screen was performed that was unremarkable aside from low serum hydroxylated vitamin D [25(OH)D] levels (4 nmol/l, normal range >50 nmol/l) and high parathyroid hormone (PTH) (38.6 pmol/l, normal range <6.1 pmol/l). Vitamin D levels from the child's mother were also tested and found to be significantly reduced.

The child made excellent progress with medical treatment over the following 72 h. He was eventually discharged home on oral diuretics, an ACE inhibitor, digoxin and aspirin. Vitamin D supplementation was optimised with oral hydroxylated vitamin D (alfacalcidol). At follow-up 3 weeks later the child remained well with some improvement in cardiac function (EF 21 %), and carvedilol was introduced. Serum vitamin D and PTH levels were re-checked, and both were now within normal limits.

Around 1 month later the child was readmitted emergently to intensive care after contracting a viral respiratory infection, accompanied by acute haemodynamic collapse. He required intensive cardiorespiratory support including inotropic therapy with dopamine, milrinone and adrenaline. He recovered and was discharged on his pre-existing regime 1 week later.

During serial outpatient follow-up over the following 9 months his cardiac function returned to normal range (EF 58 % at 10 months of age) (see Fig. 17.1c, d) without any further admissions. He remained asymptomatic and has re-established normal growth.

Discussion

Vitamin D deficiency is relatively prevalent in most Western societies, particularly in breast-fed infants of African or Indian subcontinent origin due to a combination of reduced exposure to ultraviolet, maternal vitamin D deficiency, and genetic factors. Despite the relatively high prevalence of vitamin D deficiency, associated cardiomyopathy is rare, although it has been speculated that this may be due to under diagnosis.

Serum vitamin D levels are generally measured in the 1a-hydroxylated form, 25-hydroxyvitamin D [25(OH)D]. In most tissues and cells of the body this is then further hydroxylated to the most active form, 1,25-dihydroxyvitamin D [1,25(OH)2D]. This has a number of direct and indirect effects on the myocardium, including promoting differentiation and proliferation of cardiomyocytes, regulation of myocardial contractility and intracellular calcium handling. In patients for whom deficiency results in important cardiomyopathy, presentation in infancy with signs of cardiac failure is typical. The disease course in this group is often fulminant and can be rapidly fatal without intensive cardiorespiratory support, including access to mechanical circulatory support and/or primary transplantation if required. However, in those that survive initial presentation, vitamin D deficient cardiomyopathy is one of the few readily treatable causes of DCM with an excellent long-term prognosis once appropriate support and nutritional supplementation are put in place. A list of other potentially reversible causes of DCM in the paediatric population is shown in Table 17.1.

Table 17.1 Reversible causes of cardiomyopathy in the paediatric population

| | Diagnosis | Diagnostic tests | Potential | Associated features | Treatment | Cardiac prognosis |
|-----------|---|---|---|---|---|---|
| Cardiac | Tachyarrhythmia (incessant) | ECG Electrophysiological studies | PJRT Junctional tachycardia Atrial tachycardias | Palpitations Structural heart disease | Restore sinus rhythm/rate control | Generally good |
| | Ischaemia and coronary abnormalities | ECG Cardiac enzymes ± Multi-modality imaging | Cardiac surgery Embolic events ALCAPA Kawasaki disease | Chest pain Arrhythmia Regional myocardial dysfunction | Improve coronary perfusion | Varies depending on underlying diagnosis and presentation |
| | Nutritional | Vitamin D deficiency | Serum vitamin D, calcium, parathyroid hormone | Nutritional deficiency Liver disease Renal disease | Poor growth Impaired bone mineralisation Supplementation ± Underlying cause | Generally good |
| Toxic | Selenium deficiency (<i>Keshan disease</i>) | Serum selenium level | Nutritional deficiency | Ketogenic diet (cf epilepsy) Hypothyroidism | Supplementation | Generally good |
| | Thiamine deficiency (<i>Beri-Beri</i>) | Serum thiamine levels | Nutritional deficiency | Neuropathy Encephalopathy | Supplementation | Generally good |
| Endocrine | Anthracycline toxicity (chemotherapy) | Regular cardiac screening ?Serum cardiac markers | Cardiotoxicity leading to apoptosis/cellular dysfunction | Radiotherapy to chest Trisomy 21 | Withdraw anthracycline (preferred) or reduce dose | Peri-treatment: good Late presentation: poor |
| | Hypothyroidism | Thyroid stimulating hormone, free T ₄ | Congenital Autoimmune Iodine deficiency | Other autoimmune disease Conduction disturbances | Thyroid hormone replacement therapy | Generally good |
| | Hyperthyroidism | Thyroid stimulating hormone, free T ₄ | Rare in children – usually autoimmune | Other autoimmune disease Arrhythmias | Drug treatment Surgery | Generally good |

| | | | | | | |
|------------------------------|--|--|--|---|--|-----------------------|
| Metabolic | Tyrosinaemia type 1 Urine organic acids | Serum amino acids Urine organic acids | Autosomal recessive | Liver and renal failure Encephalopathy Hyperammonemia | Dietary restrictions Nitisinone (NTBC) | Generally good |
| Propionic aciduria | Urinary organic acids Acylcarnitine profile | Autosomal recessive | | Low protein diet, carnitine Liver transplant | Reported improvement following liver transplant | |
| Fatty-acid oxidation defects | Free fatty acids Acylcarnitine profile | Autosomal recessive | | Hypoketotic hypoglycaemia Peripheral myopathy | Dietary restrictions Avoid fasting ±Carnitine | Variable; can improve |
| Carnitine deficiency | Acylcarnitine profile | Autosomal recessive | | Peripheral myopathy | Carnitine supplementation | Generally good |
| Storage disorders | Urinary glycosaminoglycans Enzymology | Most autosomal recessive | Characteristic physical features Organomegaly Sodium valproate therapy | Enzyme replacement therapy (ERT) | Cardiomyopathy may improve with ERT (generally more so than peripheral myopathy) | |

Abbreviations: *ECG* electrocardiogram, *PJRT* permanent junctional reciprocating tachycardia, *ALCAPA* anomalous left coronary arising from the pulmonary artery

The absorption and metabolism of vitamin D is also particularly dependent on normal hepatic synthetic and exocrine function and renal function. In this case, cholestatic jaundice has led to reduced intestinal absorption of fat-soluble vitamins, which in combination with being exclusively breast fed by a vitamin D deficient mother has resulted in severe nutritional deficiency. Whilst the child was already being supplemented at presentation, this was as part of a multivitamin package with a relatively lower total dose of vitamin D. Serum 25(OH)D levels recovered rapidly with appropriate oral high-dose supplementation.

Even once normal levels are re-established, myocardial function can take around a year to fully recover following the diagnosis of vitamin D deficient cardiomyopathy. Whilst this child initially responded well to medical support and nutritional supplementation, his cardiac function was showing only limited signs of recovery when he was readmitted for a second time a month after diagnosis with a life-threatening lung infection. He survived this episode and went on to make a full recovery with normal cardiac function, somatic growth and development.

This important and potentially fully reversible cause of cardiomyopathy must be considered in any infant presenting with a DCM-type picture, particularly those with established risk factors for vitamin D deficiency.

Learning Points

- Vitamin D deficiency is common, particularly in established at-risk groups; however certain conditions and co-morbidities can predispose some children to particularly severe deficiency.
- Vitamin D deficiency can rarely lead to a profound cardiomyopathy that can be fulminant and rapidly fatal without intensive cardiorespiratory support, particularly in infancy.
- In those who survive initial presentation, vitamin D deficient cardiomyopathy is one of the few treatable causes of DCM with good long-term outcome. Rigorous attention should be paid to achieving normal serum vitamin D levels, with aggressive nutritional supplementation and regular monitoring from the point of diagnosis.

Acknowledgements Thanks to Dr. Roshni Vara, Inherited Metabolic Disease Service, Evelina Children's Hospital, for her kind support.

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Chapter 18

A T-Wave Tight Spot

Luke D. Starling and Jan Till

Abstract Presyncopal symptoms are common and usually benign in children. Suspicion should be increased when these symptoms occur in relation to exertion as this may represent significant underlying pathology, including an inherited arrhythmia or cardiomyopathy. Arrhythmogenic right ventricular cardiomyopathy (ARVC) is an uncommon inherited cardiomyopathy associated with mutations in genes encoding desmosomal proteins. It is characterised by fibro-fatty replacement of myocardial tissue, cardiac failure, ventricular arrhythmias and sudden cardiac death. Diagnostic criteria include resting ECG abnormalities and demonstration of abnormal myocardial tissue and impaired ventricular function by means of diagnostic imaging; however, detection can be very difficult in the early stages and echocardiography may not be sufficiently sensitive to identify subtle abnormalities. We describe the case of a child presenting with exertional-related presyncope and an abnormal resting 12-lead ECG in whom the diagnosis was missed until he presented with a broad complex tachycardia.

Keywords Electrocardiogram • Exertional syncope • Ventricular tachycardia • Arrhythmogenic right ventricular cardiomyopathy • Signal-average electrocardiogram

Case Description

A 13-year-old Caucasian boy who was a very keen and competitive swimmer presented to his local paediatric outpatient clinic following two short-lived episodes of dizziness, the first after climbing out of the swimming pool and the second

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following micturition. An ambulance was called during this latter episode, but his symptoms had fully resolved prior to arrival and he was not taken to hospital. He was subsequently referred to the Paediatric Cardiology clinic on a routine basis.

He was seen in the Paediatric Cardiology clinic 9 months later. In the interim, he had continued to swim as normal and had remained entirely well. On further questioning, both episodes occurred immediately after swimming and he experienced no associated palpitations or chest pain during either event with no additional symptoms, such as visual disturbance, which may have suggested a vasovagal origin. He had never experienced a previous episode of frank syncope or previous palpitations or chest pain. Though he had experienced febrile convulsions as a baby, he had no further episodes beyond infancy and was otherwise entirely well and took no regular medications. There was no known history of sudden or unexplained death in the family.

Cardiovascular examination was unremarkable, and he underwent a routine screening echocardiogram and 12-lead ECG. His echocardiogram demonstrated a structurally and functionally normal heart, including normal coronary artery origins and proximal course, and no overt myocardial disease. His 12-lead ECG was reported to be normal, demonstrating sinus rhythm with no ventricular pre-excitation and a manually calculated QTc of 417 ms. Believing these symptoms were vasovagal presyncope, he was reassured and discharged with advice to maintain a good fluid and salt intake.

He remained entirely asymptomatic and continued to train in the swimming pool for around 12 hours per week until 2 years later, when he presented to his local Emergency Department, now aged 15 years. Following a 3-day history of symptoms consistent with a viral upper-respiratory tract infection he began to feel more unwell and went to bed early. He then began to experience palpitations and had several episodes of vomiting. His parents took him to Accident & Emergency, where he was noted to be markedly tachycardic (220 bpm), albeit with an adequate blood pressure (106/72 mmHg) and no evidence of impaired peripheral perfusion.

His 12-lead ECG at this time is presented in Fig. 18.1.

The doctors in the Emergency Department identified a ‘broad-complex tachycardia’. Perhaps in view of the patient’s age and the absence of a history of congenital heart disease, they felt that this was most likely to be a form of supraventricular tachycardia (SVT) with aberrant conduction accounting for the broad QRS complexes. With this in mind, he was treated with intravenous adenosine at a dose of 10 mg. It is reported that the tachycardia reverted to sinus rhythm shortly after the administration of adenosine; however, there was no contemporaneous ECG recording during the administration of adenosine and precise timings were uncertain.

The ECG following cardioversion is shown in Fig. 18.2.

The doctors in the Emergency Department made contact with their local Paediatric Cardiology service. The 12-lead ECGs were then reviewed with the Paediatric Electrophysiology team, who felt that the broad-complex tachycardia was likely to be a ventricular tachycardia (VT). The QRS complexes are of left bundle branch block morphology, suggesting a right ventricular origin, and the QRS axis is directed superiorly (positive in lead aVL and negative in leads II, III, and

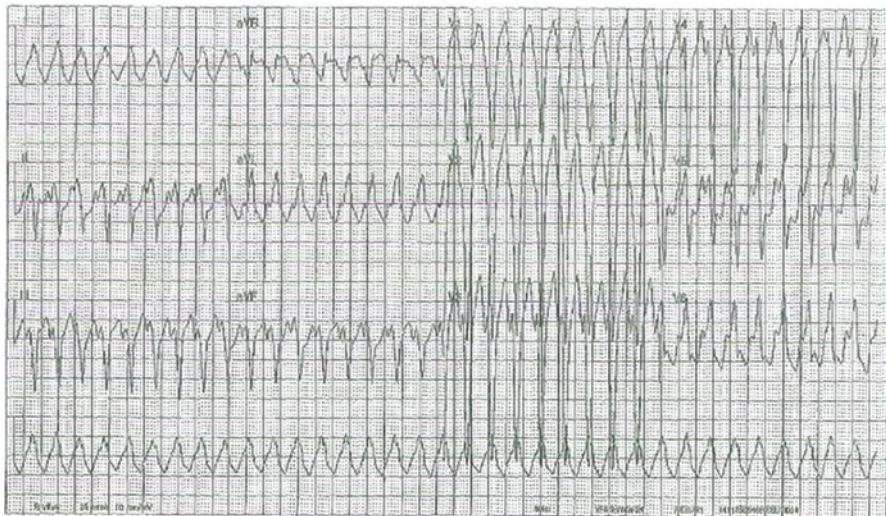


Fig. 18.1 Broad-complex tachycardia at presentation to the local emergency department

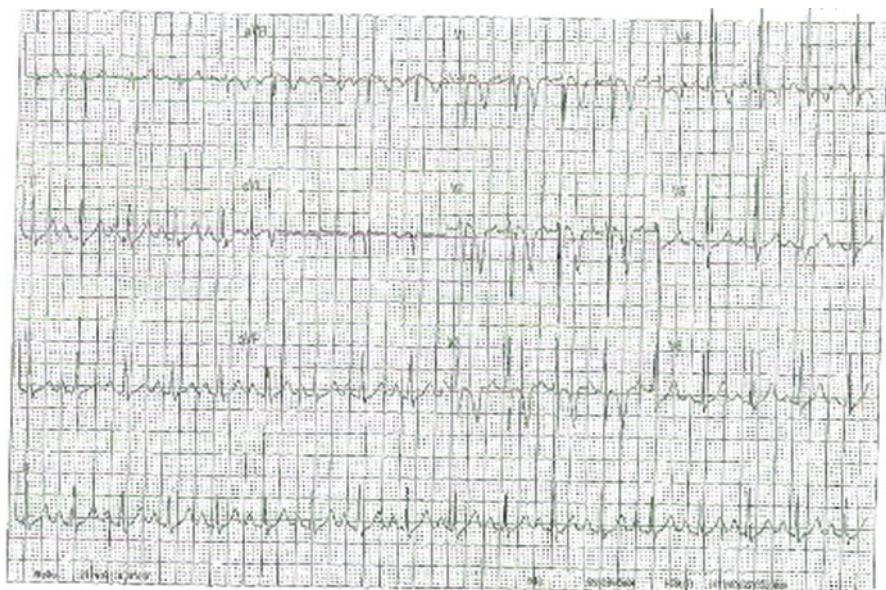


Fig. 18.2 ECG following reversion to sinus rhythm

aVF), suggesting an apical origin. That the tachycardia cardioverted to sinus rhythm following adenosine administration sparked debate, as the ECG demonstrating broad-complex tachycardia fulfilled many of the conventional criteria applied for identifying VT as well as those criteria for identifying SVT with aberrancy. In terms

of VT, features such as ventriculo-atrial dissociation, capture beats, and fusion beats were not obviously present. The absence of a contemporaneous ECG during adenosine administration made it impossible to understand the mode by which tachycardia was terminated, which may have permitted a more confident diagnosis of the tachyarrhythmia. Thus it remained equivocal as to whether adenosine terminated the arrhythmia or whether cardioversion occurred spontaneously, shortly after adenosine was administered.

The 12-lead ECG following termination of tachycardia was of particular interest. Sinus rhythm is clearly demonstrated and there is no manifest pre-excitation. The QRS duration is normal with no bundle branch block at baseline (therefore, any aberrancy displayed during tachycardia would have to have been rate-related) and with age-appropriate R-wave progression, which has an adult pattern in this instance. The pattern of repolarisation, however, is clearly abnormal. Though a negative T-wave in lead V1 is acceptable at this age, the deep negative T-waves in leads V1 to V4, in the absence of bundle branch block, is abnormal in a Caucasian without early repolarisation. In particular, evidence of VT of right ventricular origin in the context of a resting ECG with inverted T-waves in the anterior chest leads raises specific concerns about arrhythmogenic right ventricular cardiomyopathy (ARVC), and warrants further investigation.

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC)

- Inherited cardiomyopathy characterised by:
 - Fibro-fatty replacement of myocytes, predominantly affecting the right ventricle
 - Ventricular arrhythmias and risk of sudden cardiac death
 - Congestive cardiac failure
- Prevalence of 1:2500–1:5000 (compared to 1:500 adults for hypertrophic cardiomyopathy)
- Associated with mutations in genes encoding desmosomal proteins (plakoglobin (JUP), desmoplakin (DSP), plakophilin-2 (PKP2), desmoglein-2 (DSG2), desmocollin-2 (DSC2), transforming growth factor- β 3 (TGF β 3) and TMEM-43)
- Diagnosis based on criteria proposed by International Task Force of the Working Group of Myocardial and Pericardial Disease of the European Society of Cardiology – includes:
 - Evidence of fibro-fatty replacement of myocardial tissue – detected via imaging (echocardiography and cardiac MRI) and histology
 - ECG evidence of depolarisation and repolarisation abnormalities
 - Arrhythmia of right ventricular origin (though LV disease exists in ~30 %)
 - Positive family history including presence of known pathogenic mutations

- ECG changes include:
 - Inverted T-waves in leads V1-V3 in the absence of RBBB (in those aged >12 years)
 - QRS prolongation (>110 ms) in leads V1-V3
 - Terminal slurring of QRS complexes in V1-V3 (≥ 55 ms from S-wave nadir to isoelectric line)
 - Epsilon waves – low amplitude deflection between QRS and T-wave
 - Poor R-wave progression in right praecordial leads
 - Low-voltage QRS complexes in the limb leads \pm praecordial leads
- Overall mortality rates are estimated to be 1–3 %/year and are associated with sudden cardiac death (1–2 %/year) and progressive cardiac failure
- Risk factors for sudden cardiac death include: previous cardiac arrest, VT with haemodynamic compromise, significant RV dilatation/dysfunction, presence of LV disease

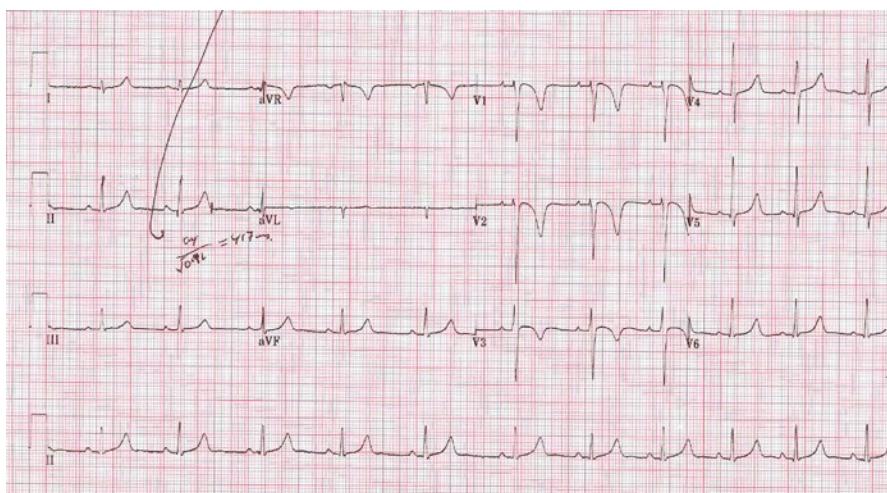


Fig. 18.3 Twelve-lead ECG from initial paediatric cardiology outpatient clinic (aged 13 years)

Having revisited the history of his previous episodes when aged 13 years, the concern was that these earlier events may have represented self-limiting runs of VT. When his 12-lead ECG from 2 years ago was reviewed (Fig. 18.3), it was noted to be very similar in appearance to his current resting ECG with evidence of markedly abnormal repolarisation, manifest as deeply negative T-waves in leads V1 to V3.

Whilst in sinus rhythm, the local paediatrician with expertise in cardiology was able to perform a focussed echocardiogram, again confirming a structurally normal heart with no chamber dilatation and with good biventricular systolic function. A beta-blocker (bisoprolol) was commenced and the dose titrated upwards.

Arrangements were made for him to be seen at the Paediatric Cardiology centre on an urgent basis for review with the electrophysiology team, including an exercise test, signal-averaged ECG (SAECG) and cardiac MRI.

When he attended for review, he had remained well with no further episodes suggestive of arrhythmia. A Holter monitor performed at the admitting hospital demonstrated evidence of ventricular ectopy, manifest as isolated ventricular ectopics of a single morphology and runs of slow ventricular bigeminy and trigeminy, again of single morphology. There were no runs of non-sustained VT, and there were no symptoms coinciding with ectopy. He was now on bisoprolol 3.75 mg once a day, which he was tolerating well. His 12-lead ECG was unchanged and his echocardiogram was unremarkable, with no impairment of ventricular function and with no overt evidence of myocardial disease.

He underwent a cardiac MRI immediately followed by SAECG and exercise test whilst the MRI report was pending. The electrophysiology team were summoned to the exercise laboratory in response to development of the following ECG changes shown in Fig. 18.4 during the early recovery phase.

He remained entirely asymptomatic with an adequate blood pressure during this tachyarrhythmia, which spontaneously terminated shortly after onset. This broad-complex tachycardia differs from the previously recorded tachyarrhythmia – it is faster (almost 300 bpm) and is once again of likely right ventricular origin (QRS morphology of left bundle branch block), though on this occasion, the

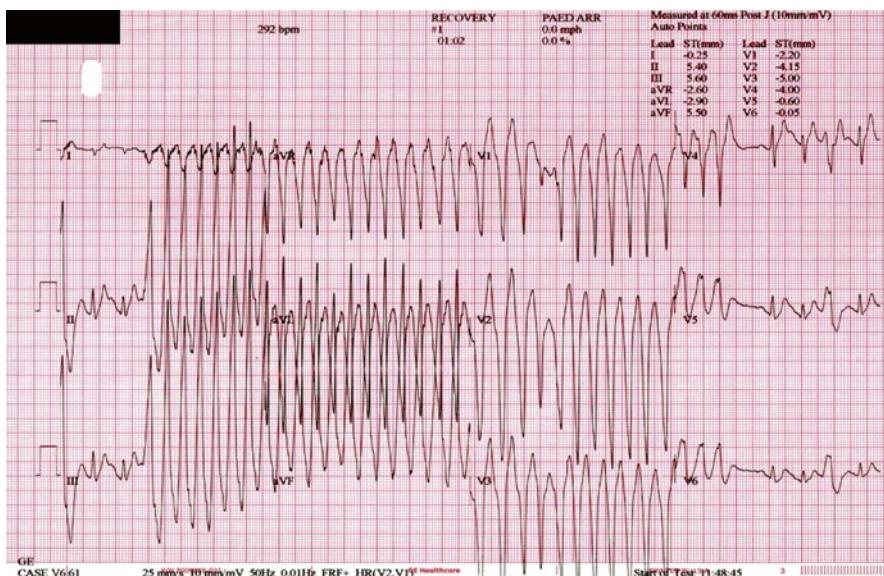


Fig. 18.4 Ventricular tachycardia of right ventricular origin during early recovery phase of exercise test

axis is inferior (positive in leads II, III, and aVF), suggesting a right ventricular outflow tract origin.

A SAEKG was performed prior to the exercise test (Fig. 18.5). Though no accepted 'normal' limits exist for the SAEKG assessment of the paediatric population, when judged by adult criteria, the SAEKG was abnormal. The SAEKG is utilised to demonstrate the presence of late potentials, representing fragmented ventricular depolarisation. This is relevant because if neighbouring regions of ventricular myocardium are depolarised at different times or at different rates (perhaps due to myocardial disease, such as fibrosis), it follows that these regions are also refractory at different times, thereby forming the substrate for a micro-re-entry circuit and ventricular tachycardia. These late potentials are usually of such low amplitude that they are seldom evident on a standard 12-lead ECG, however, by recording a larger number of QRS complexes over a longer period (usually 250 complexes over approximately 10 min), an overall average signal can be displayed with random artefactual noise (i.e. from skeletal muscle) filtered out. This allows low amplitude potentials occurring at the end of the QRS complex (late potentials) to be demonstrated. As such, late potentials prolong the overall QRS duration (QRSd) by slurring the terminal aspect with low amplitude signals. A SAEKG is therefore considered 'positive' (or abnormal) if it fulfils two of the following three criteria:

- A QRSd >114 ms
- A low amplitude signal (LAS) of <38 μ V persisting for >40 ms of the QRS complex

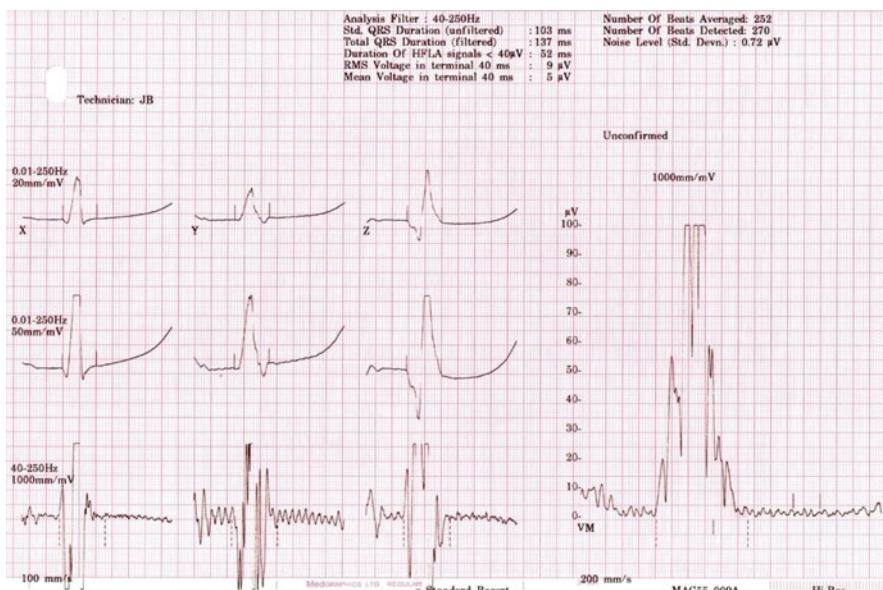


Fig. 18.5 Signal averaged ECG (SAEKG) demonstrating significant late potentials and therefore a potential substrate for VT

- A root means squared (RMS) of voltages measured during the terminal portion (last 40 ms) of the QRS complex of <20 µV

The SAECG in this instance satisfied all three of these criteria, reflecting an increased risk of VT.

Following his abnormal exercise test and SAECG, the result of the cardiac MRI became available. There was evidence of dilatation of the right ventricle (RV) with a globally hypokinetic RV free wall and a low-normal RV ejection fraction. Late gadolinium enhancement confirmed prominent fibrosis at the meeting point of the inferior interventricular septum and the RV apex with a suspicion of further fibrosis of the RV free wall, with no evidence of aneurysm formation. There was no evidence of left ventricular disease, which was non-dilated with a normal ejection fraction. These findings were consistent with a diagnosis of ARVC. In light of the above with an overt propensity towards fast VT, a cardioverter-defibrillator (ICD) was implanted. He was discharged having increased his bisoprolol to 7.5 mg bd with a view to him abstaining from exercise until he returned for further assessment on the exercise treadmill to determine if his tendency to arrhythmia had been suppressed. Further episodes of VT despite bisoprolol precipitated discussions about ablation and he is currently awaiting this procedure. There is no known family history of significant arrhythmia or sudden death dating back three generations and genetic assessment is underway.

Discussion

Presyncopal and syncopal episodes are relatively common in children and are frequently vasovagally-mediated. Vasovagal syncope is often associated with prodromal or coexisting autonomic disturbance, resulting in visual or auditory symptoms. Autonomic disturbance with associated dizziness but in the absence of actual collapse (frank syncope) is described as presyncope.

Certain activities, such as micturition, alter vagal tone and may precipitate a vasovagal episode. Similarly, exercise and in particular, the phase immediately following exercise, stimulates vagal tone and may result in presyncope or even frank syncope. However, presyncopal or syncopal episodes in the context of exertion always warrant further cardiovascular assessment as they may herald a potentially sinister underlying pathology. Pathologies that may present as such include structural congenital cardiac lesions, and in particular those with obstruction to left ventricular outflow (i.e. aortic stenosis) where the patient is unable to successfully increase their cardiac output in response to increased demand. Primary myocardial disease (cardiomyopathy), such as hypertrophic cardiomyopathy, may manifest syncope in a similar haemodynamic fashion, whilst certain cardiomyopathies may also be associated with a tendency towards arrhythmia. Furthermore, certain arrhythmias may be induced by the increase in circulating catecholamines seen with stress and exercise. These include primary arrhythmia syndromes, such as inherited ion channel disorders (channelopathies), examples being

catecholaminergic polymorphic ventricular tachycardia (CPVT) and certain subtypes of long QT syndrome, which are associated with a tendency towards sudden cardiac death.

These examples highlight the importance of a thorough, systematic cardiovascular assessment of episodes of syncope and presyncope associated with exertion. Vasovagal syncope should be considered a diagnosis of exclusion, having sought evidence of more sinister pathologies. A reasonable approach includes history, examination, 12-lead ECG and echocardiogram in the first instance, following which, one can proceed to a 24-hour tape (or longer period of monitoring) and a formal exercise test. Abnormalities noted during any of these baseline screening investigations warrant further targeted evaluation.

In the case described, the initial history was compatible with vasovagal syncope as the symptoms occurred soon after exercise. An unremarkable clinical examination and a normal Echocardiogram excluded any structural heart disease but cannot exclude subtle or early primary myocardial disease. Whilst negative T-waves in the right praecordial leads are a non-pathological finding in children from the neonatal period (beyond two to three days of life) until into their teens, the pattern of repolarisation in this instance was abnormal, with deep negative T-waves extending as far as V4. Documentation of VT of right ventricular origin in the context of this abnormal right ventricular repolarisation was highly suggestive of ARVC. The cardiac MRI confirmed this and whilst the SAEKG suggests a tendency towards ventricular arrhythmia, the exercise test further suggested this risk to be heightened on exertion.

This case emphasises the importance of maintaining a high index of suspicion when children present with presyncope or syncope, particularly in relation to exercise. It highlights a need for vigilance when analysing routine investigations and the importance of awareness regarding uncommon but potentially lethal diagnoses, which may masquerade as benign phenomena in children.

Learning Points

- Vague presyncopal symptoms are common in children. The 12-lead ECG is a vital screening tool and one should be vigilant for features suggestive of significant pathology.
- It can be very difficult to distinguish SVT with aberrant conduction (bundle branch block) from VT using the ECG during tachycardia.
- Always record an ECG whilst administering adenosine. The mode by which the arrhythmia terminates can allow you to make a diagnosis.
- The ECG during sinus rhythm can provide vital clues as to the underlying arrhythmia mechanism.
- Be mindful of inherited cardiac conditions when confronted with arrhythmia in children, particularly when associated with exercise and/or where abnormalities exist on the resting ECG.
- Be aware of tools such as the SAEKG, their utilisation, interpretation, and limitations.

- ARVC is an inherited cardiac condition associated with abnormal depolarisation and repolarisation and a tendency towards ventricular arrhythmia and sudden cardiac death.

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Chapter 19

Don't Forget the Head and Neck Vessels

Norah Y.S. Yap, Stephen Harden, and Tara Bharucha

Abstract An 8-week-old baby girl was diagnosed with aortopulmonary (AP) window after presenting to her local hospital with failure to thrive and respiratory distress. Echocardiogram also identified a right-sided aortic arch. Unfortunately, there was incomplete imaging of the arch branching pattern prior to surgery. Her early post-operative recovery period was complicated by two cardiac arrests, she remained in hospital for a long time, and after discharge continued to fail to thrive with frequent respiratory infections. Further investigations revealed a right-sided aortic arch with aberrant left subclavian artery forming a vascular ring. A vascular ring is a rare congenital anomaly of the great vessels, and the severity of symptoms is related to the degree of compression of the trachea and oesophagus. Some varieties of vascular ring are highly associated with other congenital cardiac defects. Currently there is no gold standard diagnostic imaging for vascular ring; however, computed tomography angiography (CTA) and cardiac magnetic resonance imaging (MRI) are superseding echocardiography because of their ability to provide detailed anatomy and relationship to other structures. A symptomatic untreated vascular ring carries significant risk of mortality and diagnosis is often delayed due to attribution of the respiratory symptoms to more common paediatric disorders.

Keywords Vascular ring • Aberrant left subclavian artery • Right-sided aortic arch • Aortopulmonary window • Double aortic arch • Respiratory distress • Dysphagia

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Case Description

An 8-week-old baby girl, born at term with a birth weight of 3.3 kg (50th centile), presented to her local hospital with failure to thrive and mild respiratory distress since birth. At presentation, she weighed 3.8 kg (2nd centile) and had a grade 2/6 systolic murmur. Echocardiography showed a 7-mm aortopulmonary (AP) window (Fig. 19.1a), a right-sided aortic arch (see Fig. 19.1b) and estimated right ventricular pressure at systemic levels. She was transferred to a tertiary paediatric cardiology centre and repeat echocardiography confirmed these findings. Surgical repair of the AP window was uneventful, but the early post-operative recovery period was complicated by two unexpected cardiac arrests within the first 4 days, and she required extra-corporeal membranous oxygenation (ECMO) support for 2 days. Re-opening and re-exploration of her chest showed good surgical repair of the AP window, with no new cardiac findings. In light of her pre-operative pulmonary hypertension, a pulmonary hypertensive crisis was suspected to have caused collapse, and she was treated with n.g. sildenafil. She was successfully extubated 10 days after surgery and was discharged home after a month.

On follow-up she had persistently poor feeding and recurrent respiratory infections, and was re-admitted to hospital with stridor and mild respiratory distress. Bronchoscopy showed severe tracheomalacia in the mid-portion of the trachea. CT angiogram showed a vascular ring as a result of a right aortic arch with an aberrant left subclavian artery (Fig. 19.2a, b, c, d). It was noted that the initial echocardiogram demonstrated a right aortic arch but showed only three head and neck branches; the left subclavian artery had not been demonstrated.

The patient underwent surgical repair of the vascular ring with transection and resection of Kommerell's diverticulum and re-implantation of the left subclavian artery to the left common carotid artery. Recovery was uneventful and is now completely asymptomatic.

Vascular Ring

Background

Vascular ring is a congenital anomaly of the aortic arch and pulmonary artery that occurs early in development of the embryonic branchial arches. Due to their close spatial relationships with the trachea and oesophagus, abnormalities in the size, position and/or branching pattern of the aortic arch and pulmonary arteries can result in compression of these adjacent structures. This can lead to life-threatening respiratory distress signs, with or without oesophageal compression symptoms.

Dr Robert Gross first described a vascular ring in 1945 after he performed the first successful division of a double aortic arch. Incidence is only 1–3 % of all

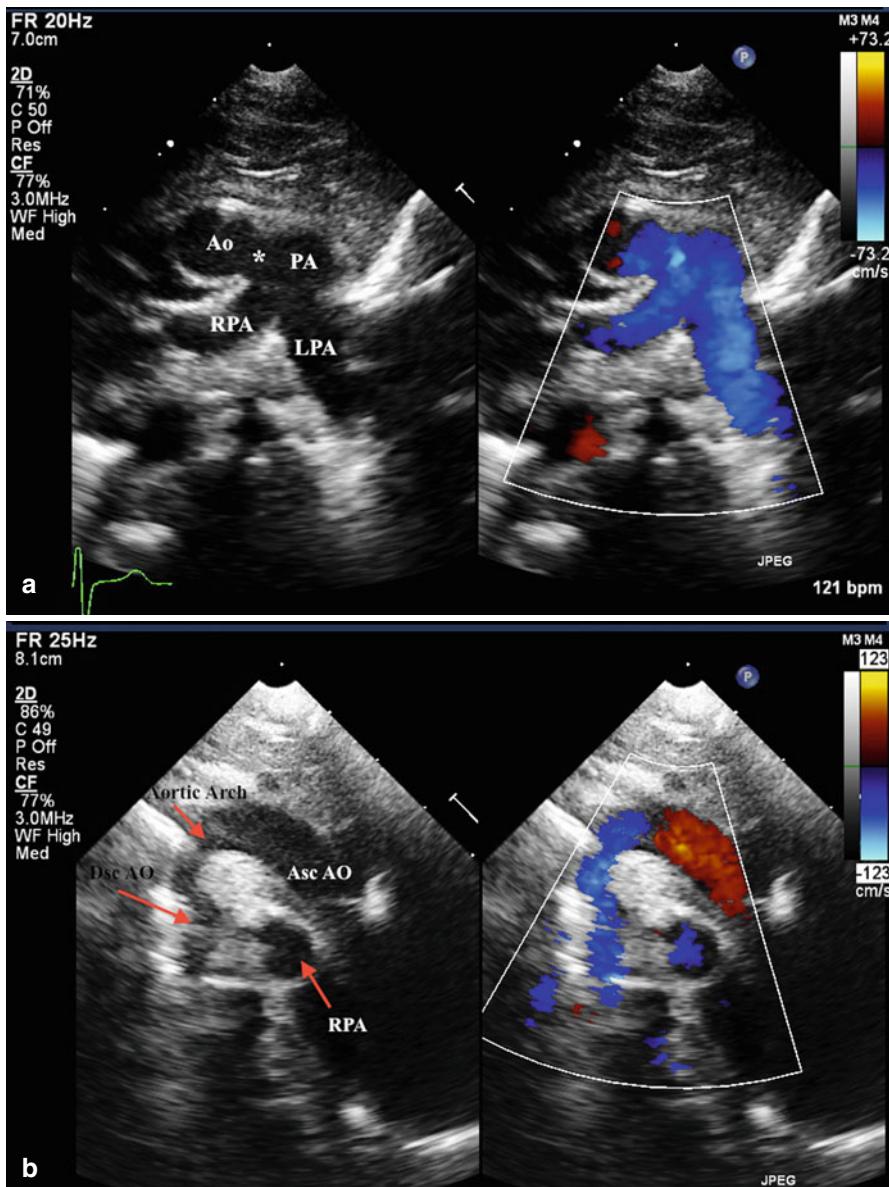


Fig. 19.1 (a) 2D echocardiogram and colour Doppler imaging of a high parasternal short-axis view with colour Doppler showing the aortopulmonary window (*) between the ascending aorta (Ao) and main pulmonary artery (PA). *RPA* right pulmonary artery; *LPA* left pulmonary artery. (b) 2D echocardiogram and colour Doppler imaging of a suprasternal long axis of a right aortic arch with inadequate view of the aortic arch branching pattern. *Asc Ao* ascending aorta; *Dsc AO* descending aorta; *RPA* right pulmonary artery

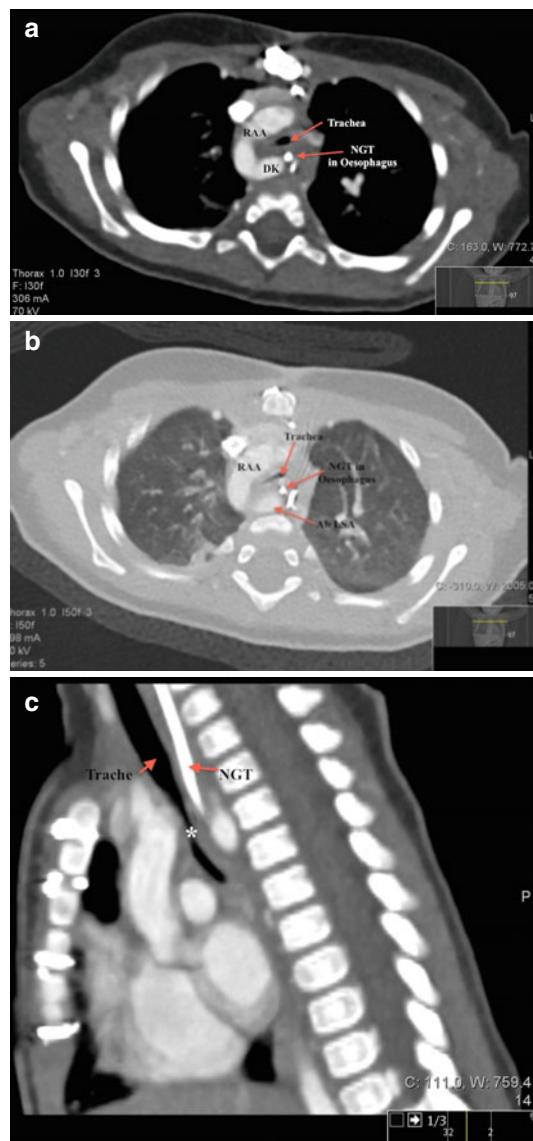


Fig. 19.2 (a) Axial image from the patient's CT angiogram on a mediastinal window showing right aortic arch (RAA) with diverticulum of Kommerell (DK) and NGT inside the oesophagus. Significant compression of the trachea can be seen. (b) Axial image from the patient's CT angiogram on a lung window showing the right aortic arch (RAA) and aberrant left subclavian artery (Ab LSA) compressing on the trachea and oesophagus. The trachea is severely narrowed. (c) Sagittal view of the patient's CT angiogram showed that the trachea and oesophagus are compressed by the vascular ring causing severe narrowing (*). (d) 3D reconstruction of the CT angiogram from the posterior aspect to show the aberrant left subclavian artery (Ab LSA) and right aortic arch impinging on the oesophagus and trachea (oesophagus and trachea not shown; nasogastric tube (NGT) indicates position of oesophagus)

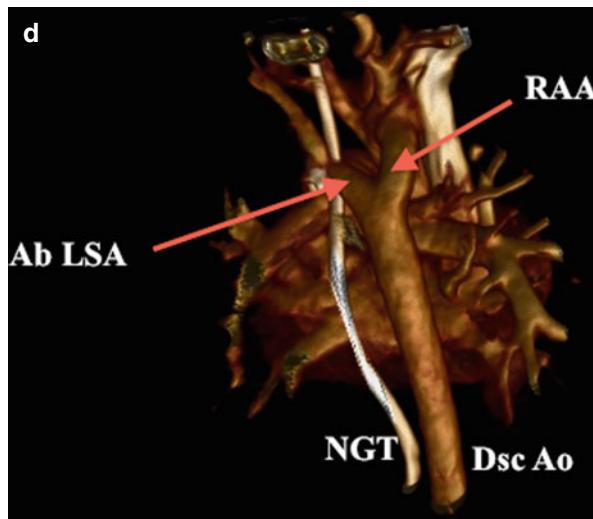


Fig. 19.2 (continued)

congenital heart disease. Given the risk of airway compromise, a high level of suspicion of these malformations must be maintained, as prompt diagnosis and treatment can be lifesaving.

Embryology

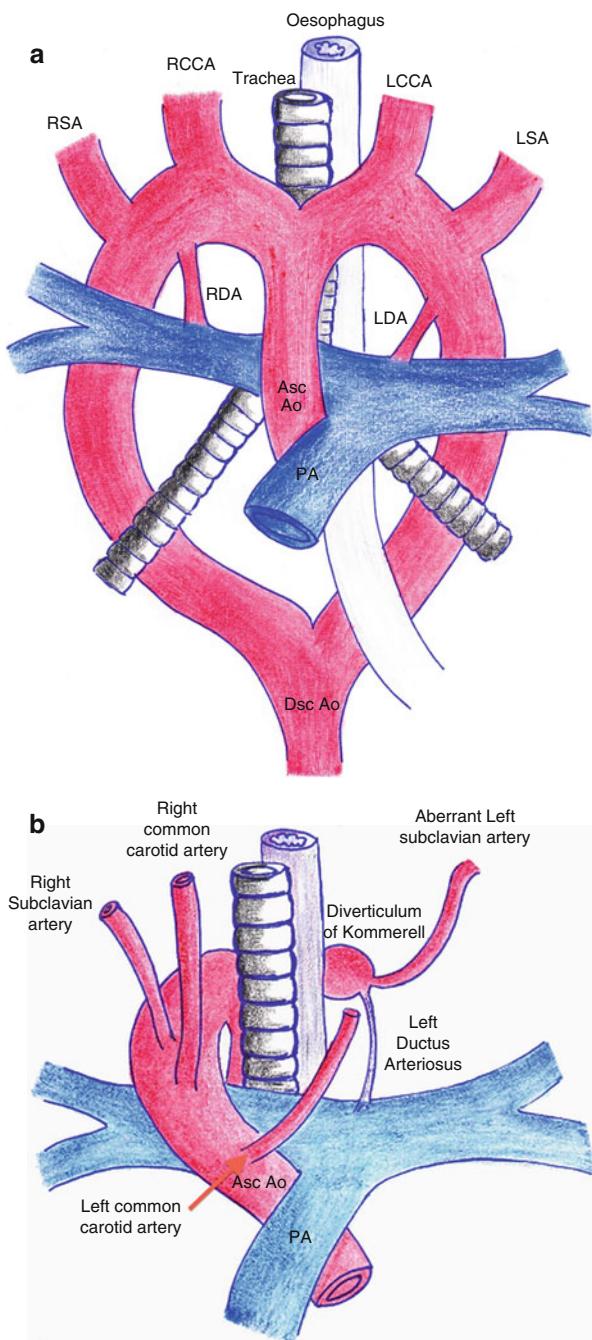
By the fourth week of embryogenesis, six symmetrical paired aortic arches and a pair of dorsal aortae are present in the embryo. Sequential fusion and regression of this complex structure results in the formation of a normal left aortic arch (i.e., an aortic arch which crosses over the left mainstem bronchus), descending on the left side of the spine. A normal left aortic arch gives rise to three branching vessels:

- Innominate (brachiocephalic) artery, bifurcating into the right common carotid artery and the right subclavian artery
- Left common carotid artery
- Left subclavian artery

Persistence of the double arch results in the commonest form of vascular ring, which can be explained by the hypothetical double aortic arch model (Fig. 19.3a) proposed by Dr Jesse E. Edwards in 1948. According to his model, the ascending and descending aorta are connected by symmetrical arches on each side, forming a complete vascular ring around the trachea and oesophagus. Each aortic arch gives origin to common carotid and subclavian arteries. On each side, the corresponding ductus arteriosus connects the pulmonary artery and subclavian artery, forming an additional vascular ring.

Fig. 19.3 (a) Schematic diagram of Edward's hypothetical double aortic arch model. RSA right subclavian artery; RCCA right common carotid artery; LCCA left common carotid artery; LSA left subclavian artery; *Asc Ao* ascending aorta; *Dsc Ao* descending aorta; PA pulmonary artery.

(b) Schematic diagram of a right aortic arch with aberrant left subclavian artery and left ductus arteriosus. The aberrant left subclavian artery can be seen arising from the diverticulum of Kommerell. *Asc Ao* ascending aorta; PA pulmonary artery



In normal development, the left arch and ductus persist, whereas the right aortic arch, distal to the right ductus, regresses. The proximal part of the embryological right arch forms the brachiocephalic artery, which bifurcates into the right common carotid artery and right subclavian artery. The embryological left aortic arch gives rise, in sequence, to the left common carotid artery and left subclavian artery.

A right aortic arch is seen in 0.5 % of the population. It is commonly seen in association with tetralogy of Fallot and truncus arteriosus. In addition, right aortic arch is also frequently associated with 22q11 deletion, occurring either in isolation or with a congenital cardiac anomaly.

A right aortic arch crosses the right mainstem bronchus and either occupies a retro-oesophageal position, or remains at its ipsilateral position on the right, without crossing the oesophagus posteriorly.

A right aortic arch with a retro-oesophageal position increases the risk of vascular ring, whereas a right aortic arch without a retro-oesophageal position only does so if the left ductus arteriosus is present. Although a right aortic arch can be an isolated and inconsequential finding, it is highly associated with congenital cardiac anomalies, and the cardiac structures should be carefully examined for other malformations.

Right Aortic Arch with Aberrant Left Subclavian Artery and Left Ductus Arteriosus

This is the second most common form of vascular ring, accounting for 30 % of all cases, and 10 % of this variety are associated with another cardiac anomaly.

This type of malformation is a result of abnormal persistence of right aortic arch and regression of the left aortic arch between the left common carotid artery and subclavian artery. The distal remnant of the left aortic arch persists as an outpouching, known as diverticulum of Kommerell, from which the left subclavian artery and also usually the left ductus arteriosus arise. The ductus arteriosus has its other end inserted into the proximal left pulmonary artery and pulls the aorta and the diverticulum forward so compressing the oesophagus and trachea.

As the aorta ascends from the heart, it gives off the first branch, the left common carotid artery, which crosses to the left and anterior to the trachea. The ascending aorta continues to ascend on the right and arch over the right main bronchus before descending posteriorly to the trachea and oesophagus. Therefore, the trachea and oesophagus are encircled by the ascending aorta and left common carotid artery anteriorly, the aortic arch on the right, the descending aorta posteriorly and the ductus arteriosus and the left pulmonary artery on the left (see Fig. 19.3b)

Other Common Vascular Rings

- Right aortic arch with mirror-image branching and retroesophageal ductus arteriosus
- Left aortic arch with an aberrant right subclavian and a right ductus arteriosus
- Right aortic arch with a left descending aorta and a left ductus arteriosus

Clinical Presentation

The presentation of vascular ring varies from life-threatening airway compression to an incidental finding in an asymptomatic adult. However, the vast majority of patients with a vascular ring present early in childhood or infancy with signs and symptoms consistent with tracheobronchial compression. It is estimated that 70–95 % present with mainly respiratory symptoms, and only 5–15 % present with late presentation of oesophageal compression.

The degree of narrowing of the trachea and oesophagus determines the age and severity of symptoms at presentation. Double aortic arch normally presents in early infancy with noisy breathing within the first few weeks of life, whereas left aortic arch with aberrant right subclavian artery may be clinically silent, as this type of vascular ring is often incomplete.

Common respiratory symptoms:

- Stridor, often worsen on feeding or activity
- Recurrent respiratory infection
- Respiratory distress
- Wheeze
- Barking cough
- Apnoea

Common gastrointestinal symptoms:

- Feeding difficulty
- Vomiting
- Gastro-oesophageal reflux
- Dysphagia – may be subtle until solid food is introduced
- Aspiration pneumonia

Diagnostic Work-Up

As illustrated by the above case, the diagnosis of vascular ring is challenging and requires a high index of suspicion. Detection of right aortic arch and/or abnormal arch branching patterns should highlight the need for further evaluation to avoid

overlooking a vascular ring. In addition, one should always be mindful of the association of vascular ring and other congenital heart defects.

There remains no gold standard evaluation for assessment of vascular ring. It is vital to have comprehensive preoperative imaging to confirm the diagnosis and assessment of vascular patency to aid surgical planning.

Thorough echocardiography assessment using a sequential segmental approach is adequate to provide sufficient information for diagnosis and preoperative planning. It also has the additional benefit of defining associated intracardiac anomalies. If a left aortic arch with normal branching pattern of arch vessels is demonstrated on echocardiogram, a vascular ring can be confidently excluded. However, merely showing a right aortic arch with mirror image branching cannot exclude vascular ring for reasons mentioned above. The limitations of echocardiography include poor visualisation of the airway and oesophagus, and part of the distal aortic arch may be obscured by the trachea and posterior vessels, so aberrant subclavian arteries may be difficult to visualise.

Additional imaging using computed tomogram (CT) or cardiac magnetic resonance imaging (MRI) is used for confirmation of diagnosis. CT and MRI clearly delineate the effect of the ring on adjacent structures, depict the patent vascular structures and vascular branching pattern to facilitate surgical planning.

CT has better contrast and spatial resolution for the airway, shorter scanning time, requiring only a breath-hold at end inspiration and minimal sedation, avoiding the need for general anaesthesia. Modern CT scanners with high temporal resolution can reduce the effective radiation dose yet maintain high diagnostic accuracy. CT also allows multiplanar and 3D reconstructions off-line and detailed pulmonary parenchymal evaluation.

Although cardiac MRI has the added advantage of avoiding ionizing radiation it requires anaesthesia in the paediatric population. This poses an additional significant risk to an already compromised airway. Intubation alters the tracheal dimension and shape and therefore interferes with the assessment of the airway structures.

The choice between these two imaging tools should be tailored according to the patient's age and co-morbidities, local expertise and the information required.

Management

The only definitive treatment for a vascular ring is surgical correction. All symptomatic patients should be repaired surgically as, untreated, the mortality approaches 90 %. Aortic aneurysm and dissection are potential complications of repair.

The timing of surgery is dictated by the severity of the symptoms.

The conventional method of vascular ring division is by open thoracotomy but video-assisted thoracoscopic surgical (VATS) and endoscopic robotic-assisted techniques are now available.

Prognosis

Clinical outcome after surgical division of the vascular ring is usually excellent. Symptoms from tracheal and oesophageal compression may persist after surgery but these usually improve and resolve with time.

Learning Points

- The degree of narrowing of the trachea and/or oesophagus determines the severity of symptoms and age of presentation of vascular ring.
- A high level of suspicion is required to make or exclude the diagnosis of vascular ring and should be considered in the presence of unexplained respiratory and/or gastrointestinal symptoms.
- Vascular rings may co-exist with other cardiac malformations. A complete echocardiographic study should be carried out on every patient, and include an assessment of the branching pattern of the aortic arch.
- In many centres, CT and cardiac MRI are the preferred imaging modalities for preoperative planning as they provide anatomical, structural and spatial information.
- Not all vascular rings require surgical division but a symptomatic vascular ring carries a significant mortality risk if left untreated.
- Post-operatively, airway and oesophageal symptoms may persist but usually improve with time.

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Chapter 20

The Test That Gets Forgotten

Michael Harris

Abstract Systemic hypertension in the context of a dilated, poorly functioning heart is not commonly seen in the paediatric population. Patients with poor left ventricular function are usually hypotensive; however, there is a subset in whom heart failure appears to present secondary to systemic hypertension (increased afterload). This is a well-described phenomenon in hypertensive adult patients. In the paediatric age group, the cause of systemic hypertension needs to be established. Causes of systemic hypertension include aortic coarctation, kidney disease, tumours or endocrine abnormalities.

Keywords Left ventricular dysfunction • Hypertension • Neurofibromatosis • Aortic coarctation • Renal failure • Renal artery stenosis

Case Description

A 9-month-old girl of Asian ethnicity born to non-consanguineous parents was admitted for assessment to a tertiary cardiac unit. She had been seen in her local hospital due to concerns regarding poor weight gain and eczema. Parental concerns included breathlessness and sweatiness over a 4-month period. The referring paediatrician was concerned by her degree of breathlessness, the amount of hepatomegaly and evidence of cardiomegaly on a chest radiograph. He felt that these clinical features were in keeping with heart failure, and referred the child for urgent cardiological assessment.

On examination, the child had approximately 10 café-au-lait spots. She was emaciated, but pink and well perfused. Saturation were 100 % in air, heart rate 130 beats per minute and blood pressure, 130/90 mmHg. Both the systolic and diastolic

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pressures were above the 99.6th percentile for blood pressure according to age. She had normal heart sounds with an additional heart sound (gallop rhythm). She had a soft systolic murmur maximal at the cardiac apex. Brachial and femoral pulses were easily palpable and without brachio-femoral delay. Her chest was clear to auscultation, but her respiratory rate was mildly elevated with mild sub- and intercostal recession. Her liver was enlarged and palpable 3 cm below the right costal margin.

Initial echocardiographic examination confirmed a structurally normal heart. The left ventricle was dilated (left ventricular internal dimension in diastole, LVIDd 3.8 cm, $z=4.13$) and poorly contractile (fractional shortening 11 %). The left ventricular posterior wall dimension in diastole (LVPWd) was 0.5 cm, $z=1.77$ and inter ventricular septal dimension (IVSD) in diastole was 0.8 cm, $z=2.94$. There was no left ventricular outflow tract obstruction or coarctation of the aorta and the left coronary artery origin was normal. There was at least moderate mitral regurgitation.

The child was established on anti-failure medications for a presumptive diagnosis of dilated cardiomyopathy, including diuretics, ACE inhibition and digoxin. Aspirin was started. Standard investigations as part of a cardiomyopathy screen were undertaken.

It became apparent during the course of the child's admission that her blood pressure remained persistently elevated for age. Heart rate was in the normal range and respiratory status improved. The hypertension was repeatedly brought to the attention of the attending ward doctors and finally triggered a re-investigation of the presentation and investigations.

An ECG at the local hospital included with the transfer paperwork showed large R waves in the left chest leads with deep S waves in the right chest leads. This was initially missed, as the ECG had been done at half scale deflection (i.e., 5 mm/mV). A subsequent ECG confirmed left ventricular hypertrophy (Fig. 20.1).

Of note, there was a family history of Neurofibromatosis Type 1 (NF1) in the paternal grandfather, although the child's mother and father were unaffected. The child herself was noted to have multiple café-au-lait spots. These findings triggered reconsideration and broadening of the differential diagnosis, including consideration of hypertension, which can be associated with NF1, as a cause of the dilated cardiomyopathy.

Further investigations were arranged. An ultrasound examination of the kidneys and renal arteries confirmed that there was a size discrepancy between the left and right kidneys. The flow velocity in left main renal artery at the level of the hilum appeared of lower velocity than the contralateral side and had a *parvus tardus* type waveform (literally, a 'small, slow' waveform, which is indicative of proximal stenosis in an artery, in which there is a low amplitude waveform followed by a slow systolic upstroke). These findings raised the significant possibility of a proximal left renal artery stenosis.

The child underwent a CT angiogram, which confirmed proximal left renal artery stenosis, with a short, stenosed segment of the left renal artery ostium extending for approximately 5 mm. This stenosed segment measured approximately 1.5 mm in diameter compared to 3.5 mm distally. Proximal, rather than distal, renal artery stenosis is typical of that found in renal artery stenosis in NF1 patients.

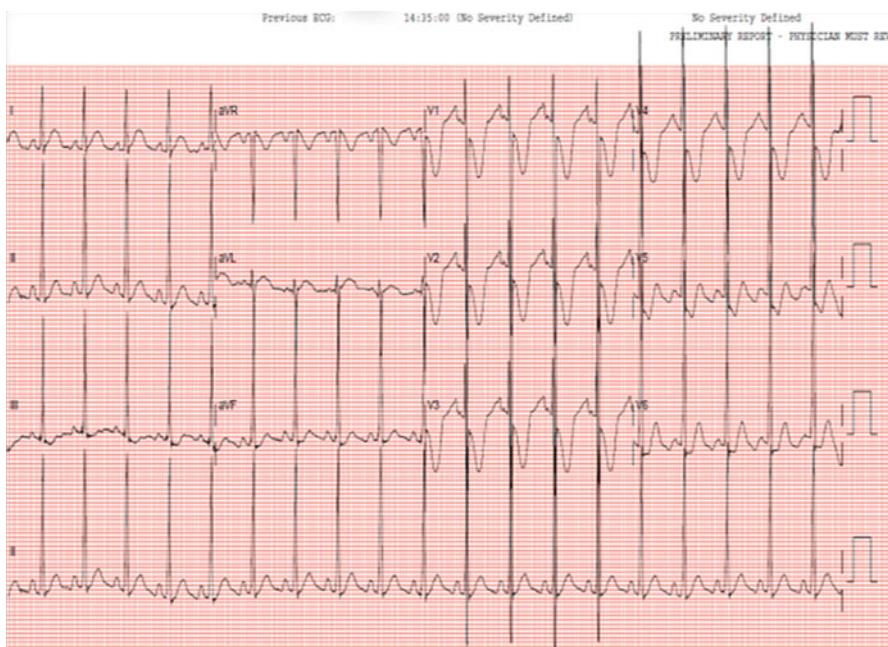


Fig. 20.1 ECG demonstrating left ventricular hypertrophy

The patient was transferred to the care of the renal physicians for commencement of antihypertensive treatment and further appropriate investigations. She subsequently underwent renal artery angioplasty and is under expectant management. Follow-up continues under cardiology.

Discussion: Children with Left Ventricular Failure; Keeping an Open Mind

In the case of children who present with acquired cardiac failure – that is, not secondary to operated or unoperated structural congenital heart disease – and in the context of an echocardiogram that demonstrates a dilated and poorly contractile left ventricle, the usual diagnosis is of dilated cardiomyopathy (DCM). These patients then embark on a well-worn management and investigation pathway.

There are several causes for dilated cardiomyopathy in the paediatric population, although a large proportion are still idiopathic (Wilkinson et al. 2010). Known causes account for approximately one-third of DCM cases, and include myocarditis, vitamin D deficiency, neuromuscular conditions, chronic arrhythmia, familial causes, inborn errors of metabolism and malformation syndromes.

When faced with a child with poor left ventricular function it is important to exclude structural causes, which in the paediatric population include:

- Left ventricular outflow tract obstruction
- Aortic stenosis
- Co-arctation of the aorta
- Anomalous origin of the left coronary artery from the pulmonary artery

Severe coarctation of the aorta often presents in the neonatal or early infant period, usually within 1 to 2 weeks of birth following closure of the arterial duct. These babies have a duct-dependent systemic circulation and present collapsed with severe metabolic acidosis as the arterial duct closes. If the coarctation is not duct-dependent, babies can present with breathlessness, sweatiness, restlessness, and irritability and they may feed poorly. Blood pressure in the arms may be mildly to moderately elevated. Often, blood pressure in the lower limbs is unrecordable or is significantly lower than that in the upper limbs. In older infants, it is possible for much higher blood pressures to be recorded in the upper limbs only, as neurohormonal mechanisms (renin-aldosterone-angiotensin and sympathetic nervous systems) are activated to maintain perfusion pressure to the kidneys (distal to the coarctation) at the expense of hypertension in the vasculature proximal to the coarctation. In circumstances where the obstruction develops over time, sometimes there is compensatory left ventricular hypertrophy and the development of collateral vessels bypassing the aortic obstruction. Such patients may be relatively asymptomatic, presenting later in life either incidentally or with systemic hypertension or its complications (e.g., stroke). In some infants, however, the left ventricle fails to compensate and dilates with poor systolic function. The left ventricular failure may progress, and infants will present with persistent tachypnoea, sweating, poor feeding and other signs of cardiac failure. It is important to note that aortic coarctation is the only pathology in the preceding list where the patient may have systemic hypertension on presentation, in the upper limbs only.

Our patient was hypertensive in both upper and lower limbs, and echocardiography excluded any structural heart disease. Therefore, other causes of systemic hypertension in children needed to be considered. Amongst adults systemic hypertension is a known cause of dilated cardiomyopathy, presumably via a progression from a hypertrophied ventricle secondary to pressure loading, to a dilated, ‘burnt out’ left ventricle (Drazner 2011). It seems likely that a similar mechanism operates in children who have systemic hypertension.

Assessing Hypertensive Children: Measuring Blood Pressure

Accurate blood pressure measurement in children can be difficult. Oscillometric sphygmomanometers use proprietary algorithms. Consequently, hypertension should really be confirmed with the use of a manual sphygmomanometer on three separate occasions. Even better is to use ambulatory blood pressure monitoring, but

this is sometimes poorly tolerated, and hence inaccurate, in the younger paediatric population. There is much documentation about how best to obtain accurate blood pressure measurements in children (Kapur and Baracco [2013](#)).

How Is Hypertension Defined?

Children are normotensive if their blood pressure is less than the 90th centile for age and height. Centile charts are available from various sources in the UK and USA (Jackson et al. [2007](#)). If the average of the systolic or diastolic blood pressure measurements on three separate occasions is greater than the 95th centile, then the child is hypertensive. There are further subdivisions into ‘pre-hypertension’ and ‘stage 1’ and ‘stage 2’ hypertension, depending on how far outside of the normal range the blood pressure lies (Gauer et al. [2014](#)).

How to Investigate Hypertension

Once hypertension has been confirmed, it is important to confirm or exclude secondary causes of hypertension (Gauer et al. [2014](#); Kapur and Baracco [2013](#)).

Those at increased risk of secondary hypertension include those under 10 years of age at presentation, or who have a very high blood pressure or symptomatic hypertension – including cardiac failure and encephalopathy – or who have hypertension that is uncontrolled on two agents. Table 20.1 outlines the secondary causes of systemic hypertension.

Table 20.1 Causes of secondary hypertension

| Renal parenchymal diseases (~68 %) | Cardiovascular |
|--|--|
| Acute and chronic renal failure | Coarctation of the aorta |
| Glomerulonephritides | Mid-aortic hypoplasia |
| Systemic vasculitides | Syndromes associated with a risk of aortic hypoplasia, including William's, Turner's |
| Congenital malformations | |
| Renovascular causes (~10 %) | Medications |
| Renal artery/vein stenosis | Steroids |
| Syndromes predisposing to stenosis, e.g., NF1, William's, Turner's | NSAIDs |
| Arteritis, e.g., Kawasaki disease, Moyamoya | Immunosuppressants |
| | Cocaine |
| Endocrine (~10 %) | Miscellaneous |
| Phaeochromacytoma | Raised intra-cranial pressure |
| Hyperthyroidism | Post-ECMO |
| Congenital adrenal hyperplasia | Obstructive sleep apnoea |
| Cushing syndrome | Monogenic disorders, e.g., NF1 |
| | Obesity |
| | Tumours, e.g., neuroblastoma, Wilms |

After a thorough history and examination, initial investigations should include blood tests and further imaging of the kidneys, vessels and other end organs. Suggested baseline investigations for a patient with heart failure and systemic hypertension are outlined in Table 20.2. In general this is a very rare cause of cardiomyopathy. Investigation should be individualised and specialist advice from renal and endocrine specialists is needed.

Our patient was strongly suspected to have Neurofibromatosis. It is known that Neurofibromatosis Type 1 is associated with hypertension in a proportion of patients, and indeed, guidelines on the management of NF1 (Ferner et al. 2007) suggest at least annual blood pressure checks on NF1 patients. Causes of hypertension in this group include:

- Uni- or bilateral renal artery stenosis
- Phaeochromacytoma
- Coarctation of the aorta

Investigation showed that she had renal artery stenosis, which, when treated, resulted in lowering of her blood pressure, reduction in afterload and subsequent improvement in ventricular function. The ACE inhibitor, which is standard treatment for dilated cardiomyopathy, was stopped as it is contraindicated in renal artery stenosis.

Summary

This case illustrates the importance of not ignoring clinical signs that do not fit with the initial differential diagnosis. In cases of paediatric dilated cardiomyopathy, patients are usually hypotensive. However, in cases where children are hypertensive and unexpectedly so, then alternative diagnoses should be actively excluded.

Table 20.2 Investigating hypertension

| Investigation | Reason |
|---------------------------------|---|
| Electrolytes, urea, creatinine | Assess kidney function |
| Thyroid function | Assess for hyperthyroidism |
| Plasma renin and aldosterone | Renal causes of hypertension |
| Serum and urine metanephhrines | Phaeochromacytoma |
| Steroid profiles | Congenital adrenal hyperplasia |
| Urinalysis | Proteinuria suggests renal parenchymal disease |
| Renal ultrasound | Delineates kidney size, consistency, Doppler waveforms of veins and arteries, pointer to further, more definitive imaging |
| Echocardiography | Assess left ventricular hypertrophy and coarctation of the aorta |
| Fundoscopy | To exclude hypertensive encephalopathy |
| CT/MR angiograms | Assess renal vascular disease and middle aortic pathology |
| Digital subtraction angiography | Assess renovascular causes of hypertension |
| CT/MRI or whole body MIBG scan | Assess for tumours, e.g., Phaeochromacytoma |

Learning Points

- Don't ignore clinical signs, symptoms and measurements that don't fit with the diagnosis.
- Beware of ECGs recorded at half scale deflection.
- Keep an open mind in infants presenting with heart failure.
- Record blood pressures in children.

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Chapter 21

Don't Ignore Reverse Differential Cyanosis

Andrew James McArdle and Anna N. Seale

Abstract Reverse differential cyanosis (RDC) is an uncommon but potentially significant finding, as it may be associated with duct-dependent congenital heart disease and cause post-natal collapse if not detected and managed appropriately. We present the case of an apparently well term baby who was found to have an innocent-sounding heart murmur at initial examination with RDC. Echocardiography identified double-outlet right ventricle (DORV) with a sub-pulmonary ventricular septal defect and coarctation of the aorta (CoA). Urgent transfer and surgical intervention was required. There was the potential for cardiac assessment to be delayed as the importance of RDC in a clinically well baby was not initially recognised. In this case presentation we review the physiology and causes of RDC and its frequency in neonatal pulse oximetry screening. We propose that in the absence of screening, RDC is at risk of being under-recognised as a sign of duct-dependent congenital heart disease.

Keywords Congenital heart disease • Cyanosis • Reverse differential cyanosis • Neonatology • Cardiology

Case Description

An apparently well term baby of African origin was seen for routine neonatal examination at 24 h of age by a neonatal junior doctor. The baby was considered normal on examination, except for a grade 2–3 systolic murmur heard loudest at the left lower sternal edge and an oral mucous cyst. Femoral pulses were palpable. Oxygen

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saturations, measured as per unit protocol, were 80–85 % pre-ductally and 95 % post-ductally. These measurements were confirmed using a Masimo pulse oximeter (California, USA).

The junior doctor was surprised by the differential saturations but had once encountered this in the treatment of a sick infant with persistent pulmonary hypertension of the newborn and transposition of the great arteries (TGA). A middle-grade colleague suggested the reading was most likely artefactual, though the trace on the oximeter was good. The infant was brought to the neonatal unit for measurement of four-limb blood pressure readings, which showed no gradient. A decision was made to observe and reassess the next day.

The next day the baby remained well, though the differential saturations persisted. The neonatal doctors remained puzzled but were not particularly concerned by the infant's clinical findings, and advised outpatient echocardiography. Despite this the junior doctor remained uneasy with the plan to discharge the infant and arranged a review by the visiting paediatric cardiologist who was conducting a weekly clinic. She was concerned by the presentation and on examination found weak femoral pulses. Echocardiogram identified double-outlet right ventricle (DORV) with sub-pulmonary ventricular septal defect (anterior aorta) and coarctation of the aorta (CoA). The infant was admitted to the neonatal unit to commence prostaglandin E₁ while transfer was arranged to the paediatric cardiac surgery unit.

The next day he became acutely unwell, requiring intubation, ventilation and inotropic support. Coarctation repair and pulmonary artery banding were performed, and after a few days he was discharged from intensive care. Subsequently, he underwent successful complete repair of his cardiac defect.

Discussion

The junior doctor performing the neonatal examination was surprised by the finding of reverse differential cyanosis in an apparently well infant, but members of the team were tempted to dismiss these findings lacking a physiological explanation in an apparently well infant. Importantly, the doctor sought a cardiology consultation prior to discharge. In retrospect, the examining doctor recalled the femoral pulses had not been easy to palpate.

In normally connected hearts, differential cyanosis – where the pre-ductal saturations are higher than the post-ductal saturations – occurs due to pulmonary artery-to-aorta shunting across the arterial duct and is a well-known sign of the most common duct-dependent systemic circulations (e.g., coarctation of the aorta and hypoplastic left heart syndrome) but can also occur in infants with a normally connected heart and persistent pulmonary hypertension of the newborn (PPHN).

Reverse differential cyanosis – where the pre-ductal saturations are lower than the post-ductal saturations – is a less well-recognised sign of important congenital

heart disease and thought to be less common . However, a recent study by de-Wahl Granelli found that 5 of 14 infants with more than 3 % difference between upper- and lower-limb percutaneous oxygen saturations had a reverse differential.

Reverse differential cyanosis can occur when the great arteries are transposed and there is pulmonary artery-to-aorta shunting in the arterial duct. The oxygenated blood in the pulmonary artery passes through the duct to the descending aorta (Fig. 21.1). This occurs in transposition of the great arteries (TGA) along with PPHN or a significant obstruction to the aortic arch (e.g., interrupted aortic arch or pre-ductal aortic coarctation). In our case, there was a double outlet right ventricle with subpulmonary VSD giving the same physiology as TGA. The aorta is anterior and streaming of blood is the same as that in transposition of the great arteries. Other reported causes of reverse differential cyanosis include supracardiac total anomalous pulmonary venous drainage. Of the five infants in the study by de-Wahl Granelli, two of these had TGA with double inlet left ventricle; one had TGA with a ventricular septal defect and CoA; and two had pulmonary atresia with VSD.

Reverse differential cyanosis may be an under-recognised sign of critical heart disease. Depending on the underlying pathology, neonates may initially appear well with no murmur and with normal femoral pulses.

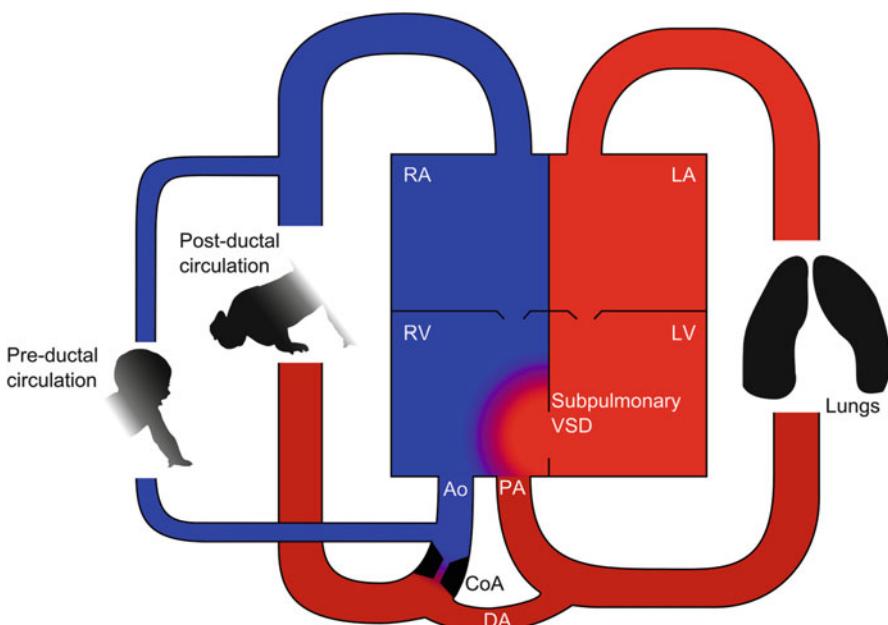


Fig. 21.1 Schematic showing the structural anomalies in this infant leading to reverse differential cyanosis. Blue represents deoxygenated blood, and red oxygenated blood. RA right atrium, RV right ventricle, LA left atrium, LV left ventricle, Ao aorta, PA pulmonary artery, DA ductus arteriosus, CoA coarctation of the aorta, VSD ventricular septal defect

Learning Points

- Reverse differential cyanosis may be commoner than recognised. It should not be ignored, and critical cardiac lesions should be ruled out as a priority.
- Screening programmes which measure only post-ductal saturations may miss many of these cases
- Normal four-limb blood pressure measurements cannot rule out coarctation of the aorta
- Assessment of femoral pulse strength is subjective. Clinicians should have a high index of suspicion when femoral pulses are difficult to identify.
- Dismissing findings that cannot be immediately explained is risky.

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Chapter 22

Pulmonary Resistance: How Best to Measure?

James Wong and Mohammed Tarique Hussain

Abstract Assessing the pulmonary vascular resistance (PVR) is important in subjects with intra-cardiac shunts who present later in life or those with functionally univentricular physiology where there is suspicion of raised pulmonary arterial pressures. Methods to measure PVR include catheter or combined catheter and magnetic resonance imaging (XMR). Whilst catheter alone is more easily performed and available, XMR techniques offer improved accuracy particularly in the presence of large shunts or when using vasodilator agents. We describe two cases where PVR was calculated and was crucial in planning further management.

Keywords Pulmonary vascular resistance • Pulmonary hypertension • Cardiac catheterisation • Fick method • Cardiac magnetic resonance imaging • Shunts • Single ventricle physiology • Atrio-ventricular septal defect • Trisomy 21

Case Description 1

A 15-year-old female with trisomy 21 and complete atrio-ventricular septal defect (AVSD) was referred to our service for assessment. She was breathless and unable to walk up one flight of stairs. The patient had been lost to follow-up at an early age, as her parents had not wished for her to have cardiac surgery due to fear of potential risks. At presentation her transcutaneous oxygen saturations were 88 % in air. The diagnosis of AVSD was confirmed on echocardiography with the additional finding of possible pulmonary hypertension due to near systemic RV pressures based on continuous wave Doppler interrogation of the tricuspid regurgitant jet by

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echocardiography. She was referred to the cardiac magnetic resonance imaging service for a PVR study to determine whether she would be suitable to undergo a complete repair.

She underwent a combined cardiac catheter with magnetic resonance imaging (XMR) under general anaesthesia. A diagnostic right heart catheter was performed via femoral venous access including a pulmonary wedge pressure as an approximation of left atrial pressure. The patient was then transferred for magnetic resonance imaging (MRI) and simultaneous pressures and flow data were recorded both at baseline conditions and after administration of 100 % oxygen and inhaled nitric oxide at 20 parts per million. MRI sequences included phase contrast flow imaging in the branch pulmonary arteries and aorta enabling pulmonary flow (Q_p) and systemic flow (Q_s) to be measured. Balanced steady state free precession (bSSFP) cine imaging was used to assess function and anatomical structures. PVR was calculated; results are illustrated in Table 22.1. Sadly, the high fixed PVR meant that she was unsuitable for surgical repair. A referral was made to a specialist pulmonary hypertension team for further palliative management.

Case Description 2

A 7-year-old female with trisomy 21 was recently discovered to have a murmur on auscultation. She was asymptomatic with transcutaneous oxygen saturations of 98 % in air. Her haemoglobin measured 13 g/dL. A transthoracic echocardiogram showed a large 8-mm patent ductus arteriosus (PDA) with bidirectional shunting. Flow was right to left in early systole and left to right through diastole. In addition there was a very small 2-mm VSD with a gradient of 35 mmHg across the defect. This child proceeded to conventional cardiac catheterisation for haemodynamic assessment and potential device closure of the PDA if PVR was found to be suitable.

Table 22.1 Haemodynamic assessment of both patients

| | Patient 1 (AVSD) (XMR) | Patient 2 (PDA) (conventional catheter) |
|-------------------------------------|---------------------------|---|
| PA pressure at baseline | Systemic | Systemic |
| Qp:QS at baseline | 1.9:1 | 1.8:1 |
| PVR at baseline | 10 WU.m ² | 8.1–12.4 wu.m ² (based on estimated oxygen consumption values of 100–150 ml/min/m ²) |
| PA pressure with iNO & 100 % oxygen | Systemic | Systemic |
| Qp:Qs with iNO & 100 % oxygen | 1.9:1 | 2.1:1 |
| PVR with iNO & 100 % oxygen | 9.7 wu.m ² | 6.0–9.0 wu.m ² (based on estimated oxygen consumption values of 100–150 ml/min/m ²) |

Venous and arterial access was achieved; pressures, blood gas and saturations were measured in the right atrium, the pulmonary artery and the aorta. Measurements were taken at baseline and after administration of 100 % oxygen and inhaled nitric oxide at 20 parts per million. Additional selective angiography of the duct was performed. PVR was calculated; results are illustrated in Table 22.1.

In view of the relatively high PVR which displayed some variability, the patient was referred to the ear, nose and throat (ENT) and respiratory teams for assessment of her airway. She was also commenced on sildenafil. A repeat PVR study was planned 3 months later to reassess suitability for PDA closure.

Discussion

Clearly in both cases accurate measurement of PVR was crucial. The presence of large left to right shunts is commonplace in the setting of congenital heart disease. If the shunt is large, early diagnosis leads to surgical closure once the patient is of an appropriate size or earlier if the patient develops symptoms that cannot be controlled with medication. Occasionally late diagnosis of large left to right shunt is made and in such circumstances this can result in pulmonary vascular changes (increased resistance), elevation of right ventricular pressures and irreversible pulmonary hypertension. Accurate assessment of the PVR and whether it is reversible is essential to determine whether the shunt can be repaired. If there is irreversible high PVR, repairing the shunt may have a detrimental effect resulting in right heart failure due to the increased afterload.

In other situations, such as univentricular physiology, a low PVR is essential prior to completion of the Fontan circulation as subjects are reliant on low PVR for venous return. Assessing PVR in such patients is difficult by conventional cardiac catheterisation and a high pulmonary artery pressure, trans pulmonary gradient and high filling pressures are used as surrogates.

PVR is dependent upon blood flow and the trans-pulmonary gradient. A high PAP might well be normal in the presence of a large shunt. It is therefore essential that patients congenital heart disease and suspicion of pulmonary hypertension a study is performed to assess PVR reversibility to help plan treatment strategies.

PVR can be determined either by conventional cardiac catheterisation or the newer technique of cardiac catheter combined with MRI (XMR). PVR (Woods units metres²) is the mean trans pulmonary gradient (mmHg) divided by the indexed flow to lungs l/min/m².

Method of Catheter-Derived Calculation of PVR

This relies on the Fick method of deriving pulmonary blood flow. A cardiac catheterisation is performed with pressures measured in the pulmonary arteries and left atrium or pulmonary capillary wedge pressure (PCWP). Blood saturations and

oxygen content are measured in the pulmonary arteries and pulmonary veins or equivalent. PCWP involves inflating a balloon catheter in the distal pulmonary artery. The wedged pressure gives an estimation of the pressure across a capillary bed.

- Pulmonary blood flow (Q_p) = $\dot{V}O_2/C_{PV} - C_{PA}$
- Systemic blood flow (Q_s) = $\dot{V}O_2/C_{AO} - C_{MV}$

$\dot{V}O_2$ = indexed oxygen consumption and can be measured directly or is often estimated at 125 ml/min/m²

C = oxygen content at various sites

PV = pulmonary veins

PA = pulmonary artery

Ao = Aorta

MV = mixed venous

- C = Oxygen content (ml/100 ml blood) = oxygen capacity x % saturations
- Oxygen capacity (ml/100 mL blood)

= oxygen carried by a haemoglobin when fully saturated

= $1.36 \times Hb$ (in g/dL)

Despite using a lot of assumptions, particularly oxygen consumption, these calculations appear to be accurate for patients with low PVR at baseline, but deviate from XMR measurements when there is high PVR, larger shunts or with pulmonary vasodilatation drugs.

XMR Derived Method of PVR

A cardiac catheterisation is used to measure branch pulmonary artery pressures. A pulmonary capillary wedge pressure is used to approximate mean left atrial pressure. The difference between the two values is the mean trans pulmonary gradient. A MRI compatible catheter is then left in situ and the patient undergoes a MRI scan. Anatomical and functional data can be attained and flows can be measured to each lung using phase contrast flow imaging. Flows are indexed to body surface area and using the above equation differential lung pulmonary vascular resistance can be defined. Simultaneous pressure and flow measurements can be recorded under stress conditions or with administration of inhaled oxygen and nitric oxide to attempt to maximally vasodilate the pulmonary vasculature to assess for reversibility. Calculation of $Q_p:Q_s$ is non-invasive and can be measured alone from flow based phase contrast imaging.

The benefits of XMR include that radiation exposure is comparable to conventional techniques or reduced. Accurate reproducible values are obtained that are not affected by left to right shunting or additional sources of pulmonary blood flow. Differential PVR to each lung can be calculated. Additional information such as the

size and function of ventricles, assessment of outflow tract and orientation and exclusion of associated defects can be obtained. Unfortunately, however, this technique is expensive, time consuming, requires expertise not as readily available.

When a patient is found to have raised PVR, the cause needs to be explored and any treatments that may help lower the PVR need to be explored. Unlike patient 1, patient 2 displayed some variability in PVR with administration of oxygen and nitric oxide the patient was therefore referred to ENT and respiratory teams for assessment of her airway. Optimising the airway, for instance by removal of large adenoids, can alleviate chronic hypercapnia which may exacerbate pulmonary vascular tone. The same is true of an airway that is compromised during sleep by obstructive sleep apnoea which may be common in larger patients or those with reduced tone common in trisomy 21. Patients with trisomy 21 require early aggressive management of underlying shunts as they have a tendency to develop pulmonary hypertension at an earlier age than individual's normal karyotype and the same isolated lesion. In a recently reported series, almost 13 % of 362 individuals with pulmonary hypertension had a chromosomal abnormality usually trisomy 21. In these instances, non-invasive nocturnal ventilation may be required.

Learning Points

- Pulmonary artery pressure is an imprecise marker of PVR in subjects with congenital heart disease where the presence of a shunt can affect the volume of flow to the lungs.
- Estimation of PVR and its reversibility is crucial when assessing patients who present late with left to right shunts and those patients who are going down the Fontan series of procedures where a low PVR is essential for the circulation to be successful.
- Conventional catheter techniques give an estimation of PVR; however, combined cardiac catheter and MRI may be more accurate and have other benefits.

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Chapter 23

Cardiomyopathy in Infants: Look at the Rhythm, Then Look Again

Alan G. Magee

Abstract A 17-month-old girl was admitted with acute left ventricular failure after a short history of lethargy, poor feeding and increasing shortness of breath. She required intubation and ventilation as well as administration of diuretics and inotropes to control her heart failure. Initially, her 12-lead ECG was thought to show sinus tachycardia and the underlying cause of heart failure to be either acute myocarditis or idiopathic dilated cardiomyopathy. On review the ECG clearly demonstrated evidence of persistent junctional reciprocating tachycardia (PJRT) and anti-arrhythmic drugs commenced. Rate control was established with improvement in clinical and echocardiographic findings.

Keywords Left ventricular failure • Dilated cardiomyopathy • Persistent junctional reciprocating tachycardia

Case Description

A 17-month-old girl presented to her local hospital acutely unwell with signs of incipient cardiorespiratory failure. She had initially attended her GP 5 days before with some difficulty breathing. At that time her GP found some signs of infection in the right lung and noted a heart rate of 190/min. She deteriorated in spite of antibiotics with increasing lethargy, poor feeding, cough, shortness of breath and vomiting. On arrival in the emergency room her heart rate was 220/min, pulses were difficult to palpate, capillary refill time was 4 seconds, respiratory rate was 65/min and liver was enlarged to 4 cm below the right costal margin. Initial venous blood gas revealed a normal pH but a lactate of 4.4 mmol/l. Intravenous access was established, she was intubated, mechanically ventilated, given frusemide and commenced on

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infusions of adrenaline, dopamine and milrinone. Urgent retrieval was arranged to a tertiary cardiology centre. Chest x-ray showed a large heart and pulmonary oedema (Fig. 23.1). ECG was thought to show sinus rhythm at a rate of 133/min with inferior and lateral p wave inversion (Fig. 23.2). Echocardiogram showed globally poor left ventricular function (Fig. 23.a, b) with mild mitral regurgitation and normal origins of the coronary arteries. A provisional diagnosis of acute myocarditis or dilated cardiomyopathy was made and appropriate screening investigations sent.

The next afternoon her ECG was reviewed, she was given adenosine 50 mcg/kg with slowing of her heart rate from 170 to 105/min and temporary normalisation of P wave morphology. A diagnosis of permanent reciprocating junctional tachycardia was made and digoxin was commenced. Eventually heart rate control was achieved with amiodarone and a reduced digoxin dose. She was extubated 3 days after admission and repeat echocardiogram showed improvement in left ventricular function with an increase in the ejection fraction by Simpson's rule from 8 % to 25 % over a 7-day period (see Fig. 23.c, d).

Discussion

Permanent or persistent junctional reciprocating tachycardia is a rare tachycardia accounting for only 5 % of all cases of supraventricular tachycardia (SVT) in children. It was first described by Coumel and colleagues in 1967 and is an incessant tachycardia with antegrade conduction through the atrioventricular node (orthodromic) and retrograde conduction via an accessory pathway which is located in the postero-septal region between the atrioventricular valves and exhibits slow and decremental (i.e., rate dependant slowing) conduction. Because of the slow conduction through the accessory pathway, the RP interval is long, giving the impression that the P wave is appearing at the correct time in the cardiac cycle; however, abnormal atrial activation means that the P wave is inverted in the inferior and lateral leads. The tachycardia is

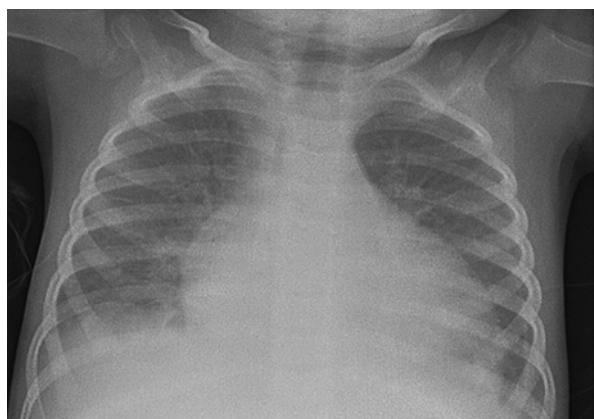


Fig. 23.1 Chest x-ray at presentation showing enlarged heart and pulmonary oedema

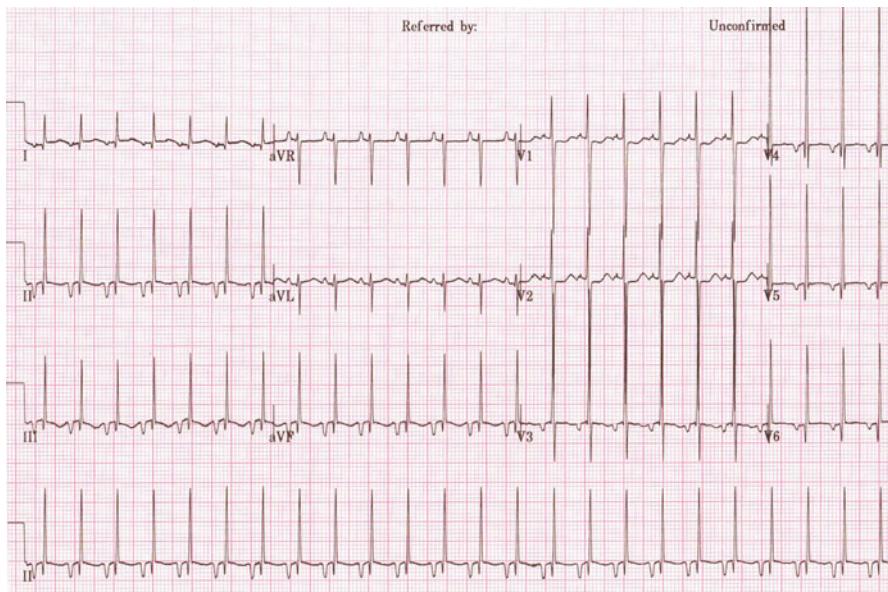


Fig. 23.2 ECG at presentation with inferior and lateral P wave inversion. The heart rate was 163/min

frequently incessant from birth or infancy and can lead to a heart rate – induced dilated cardiomyopathy which resolves after the rate is controlled. Decompensated dilated cardiomyopathy will lead to heart failure as occurred in this case. Older children and adults who present with PJRT presumably had an unrecognised slower tachycardia that was less sustained during their early childhood which would explain why it is tolerated and therefore presents later. The re-entry circuit in PJRT has a long excitable gap which means that although it can be readily terminated with adenosine bolus, it quickly restarts itself. In spite of this, in some patients it completely resolves spontaneously but in most pharmacological rate control is required. PJRT is very rarely associated with underlying structural heart disease, unlike other forms of SVT in children.

Tachycardia-induced cardiomyopathy is defined as systolic and/or diastolic ventricular dysfunction caused by a prolonged elevated heart rate which is reversible on control of the arrhythmia or the heart rate. The exact mechanism by which chronic tachycardia produces myocardial dysfunction is not entirely clear; however, abnormal calcium handling, reduced cellular energy storing and abnormal energy use have been proposed. An animal model of biventricular systolic dysfunction was first established in 1962, and it is known that rapid pacing produces a marked depression in left ventricular ejection fraction, increased filling pressures, lower cardiac output and increased systemic vascular resistance. Usually these changes are reversible with cessation of the tachycardia. Adults who have a sustained heart rate over 100 beats/min may be at risk of developing cardiomyopathy depending on age, drugs and other medical conditions. Supraventricular causes of tachycardia-induced

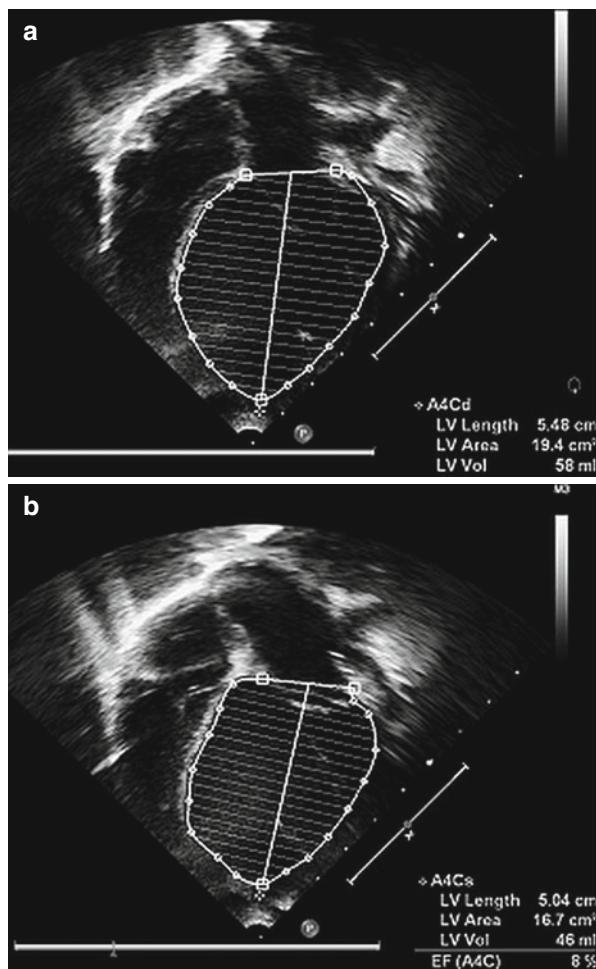


Fig. 23.3 (a, b, c, d) apical four-chamber views in diastole and systole before and after rate control was established, with improvement in ejection fraction by Simpson's rule from 8 % to 25 %

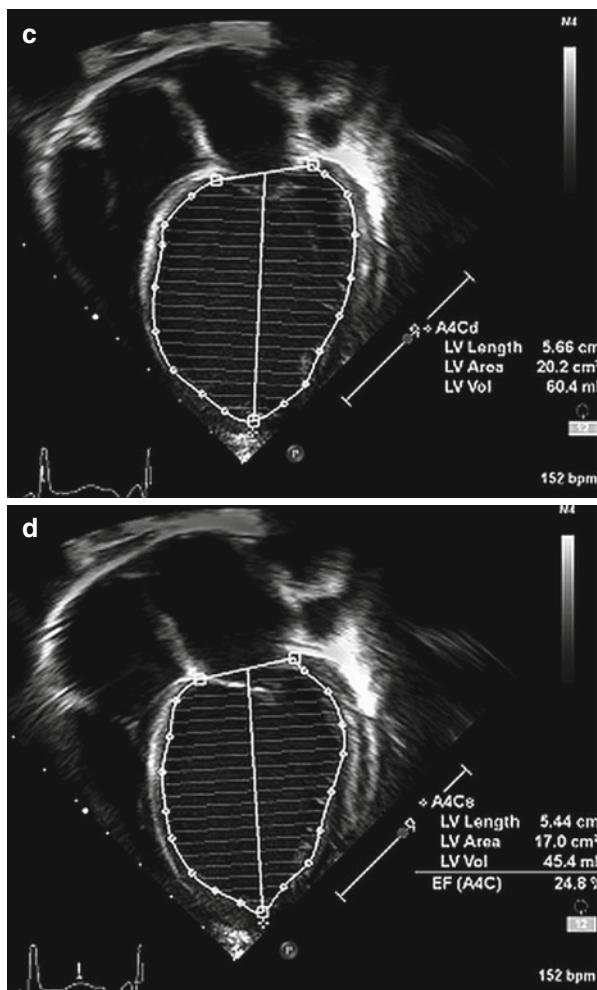


Fig. 23.3 (continued)

cardiomyopathy in all ages include atrial flutter, atrial fibrillation, atrioventricular nodal reentry tachycardia, atrioventricular tachycardia (the usual type of SVT in children), PJRT, ectopic atrial tachycardia and prolonged atrial pacing at high rates. Ventricular causes include frequent premature ventricular complexes, right ventricular outflow tachycardia (which is a type of ventricular tachycardia), idiopathic left ventricular tachycardia, bundle-branch reentry VT as well as ventricular pacing at high rates.

PJRT is easily missed, as the heart rates are often much slower than those found in children with SVT, where heart rates are usually over 200 beats/min. In addition, P waves are clearly seen, albeit with an unusual morphology, and the PR interval appears to be normal. Therefore, identification requires a high index of suspicion and keeping an open mind.

Treatment of PJRT

The large French multicentre study on drug treatment in children gives some indication of which drugs are likely to be useful. Digoxin alone was successful in 52 % of 43 patients in whom it was attempted, but the highest initial success rate was for amiodarone and verapamil either alone or in combination with digoxin, with a success rate of between 84 % and 94 % of children. Based on this it would seem reasonable to attempt digoxin alone as a first-line therapy and then proceed to a combination of digoxin with either amiodarone and verapamil. As digoxin either orally or intravenously takes some time to have an effect and as verapamil is a negative inotrope, it might be worth considering commencing amiodarone infusion alone for patients with very poor ventricular function. Treatment success could either be defined as conversion to sinus rhythm or predominant sinus rhythm with short episodes of low-rate PJRT and normal function on echo. Obviously supportive drugs are often also required in infants and small children depending on the degree of heart failure and circulatory compromise.

While radiofrequency ablation of the accessory pathway is effective in older children and adults, it carries a greater chance of causing damage to adjacent structures in smaller children. In addition, the lesions created during ablation can enlarge with time in the developing heart. It is therefore reserved for those young children in whom drug treatment fails, especially in the setting of poor ventricular function.

Learning Points

- Always consider treatable causes of dilated cardiomyopathy.
- Persistent junctional reciprocating tachycardia is slower than other forms of supraventricular tachycardia in children and can be difficult to recognize (see also Chap. 1).

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