CASE REPORT

An elusive cause of hypoxaemia in a paediatric patient

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SUMMARY

A 5-year-old girl presented to an ambulatory cardiology clinic with shortness of breath, described by the patient's mother and chest discomfort on exertion. At this visit, she was noted to have low baseline room air oxygen saturations of 89%. Subsequent cardiac, respiratory and haematological investigations including contrast echocardiography, CT scan of the chest, polysomnography and haemoglobin electrophoresis were unhelpful in yielding a diagnosis. A CT scan of the chest with contrast was performed which revealed a persistent left-sided superior vena cava that appeared to be draining into the left atrium, creating a right-to-left shunt. This was not revealed on contrast echocardiography as contrast (micro bubbles) had been injected into the right arm. This case report highlights the nuances of diagnostic investigations, which without a high index of suspicion may exclude a timely diagnosis and therapeutic intervention.

BACKGROUND

Hypoxaemia, often recognised by low oxygen saturation, is a common presenting concern in paediatrics. Common acute conditions associated with hypoxaemia include respiratory tract infections, asthma, aspiration, sedative use and fluid overload. The diagnosis in these acute situations is often clearly based on history and physical exam. Causes of chronic hypoxaemia, however, are often more challenging to diagnose based on history and physical exam alone and usually require focused investigations. Investigating for hypoventilation, right-to-left shunt, ventilation-perfusion mismatch and increased barrier to diffusion provides a very useful physiological framework to guide investigations. This case highlights the approach to hypoxaemia and the importance of understanding the limitations and nuances of investigations that may thwart a timely diagnosis.

CASE PRESENTATION

A previously well 5-year-old girl presented to a paediatric cardiology clinic in a tertiary referral centre. History revealed 1 month of intermittent and poorly characterised chest discomfort, particularly on exertion and worsening exercise tolerance. She was otherwise well.

Physical examination demonstrated a room air oxygen saturation of 89%. A preliminary cardiac evaluation consisting of physical exam, a 12-lead ECG, routine echocardiography and contrast echocardiography were non-contributory to a diagnosis. Respiratory medicine was thus consulted.

The patient's mother reported that the child described subjective 'shortness of breath' on

exertion, but this was not substantiated by any history of signs consistent with increased work of breathing or laboured breathing (persistent or episodic). There was no history of cough, wheeze, haemoptysis, epistaxis immunodeficiency, asthma, cystic fibrosis or ciliopathy. Medical history was remarkable for a Dandy-Walker malformation, psoriasis, H1N1 influenza infection and a single hospital admission for respiratory syncytial virus bronchiolitis at age 18 months. Family history was non-contributory.

On examination, the child had a heart rate of 130 bpm, respiratory rate of 20 breaths/min, room air oxygen saturation of 89% and a grade 2/6 innocent systolic murmur at the left sternal border. The child's height was below the third centile. Oxygen was administered via nasal prongs at 3.5 L/min, and this resulted in an increase in oxygen saturation from 89% to 94%. Pertinent negatives included no distress, clubbing, telangiectasia or stigmata of chronic liver disease.

Further investigations were undertaken to try and identify the cause for the child's apparent hypoxaemia.

INVESTIGATIONS

The physiological framework to approach hypoxaemia presented above was used clinically to guide investigations to assess for hypoventilation, ventilation-perfusion mismatch, barriers to diffusion and right-to-left shunt. To assess for hypoventilation, a capillary blood gas was performed and was normal with a pH of 7.35, PCO₂ of 35 mm Hg and bicarbonate of 24 mmol/L. The patient's mother refused for her child to have an arterial blood gas. A polysomnogram showed no evidence of significant nocturnal hypoventilation. Spirometry and lung volumes were performed and were normal ruling out obstructive or restrictive lung diseases that could cause ventilation-perfusion mismatch or increase the barrier to diffusion. Unfortunately, the child was unable to perform the technique for diffusing capacity of the lung for carbon monoxide testing to allow us to assess diffusion capacity. Lung diseases which may contribute to ventilation-perfusion mismatch and increased barrier to diffusion were excluded with a chest X-ray and CT scan of the chest which demonstrated normal pulmonary parenchyma, interstitium and air spaces.

Thus far, our results were negative with regards to a diagnosis for this child and we sought advice from the haematology team to ascertain if the child had any underlying haemoglobinopathies to account for the low oxygen saturation, particularly as oximetry can be misleading for delineating the true arterial oxygen saturations and presence of

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hypoxaemia in certain haemoglobinopathies.¹ A complete assessment for haemoglobinopathies, including methaemoglobinemia was negative. Interestingly, the haematologist felt that the child had subtle non-specific dysmorphic features that were not further characterised. This prompted a karyotype to be sent and revealed a diagnosis of Turner's syndrome.

After further deliberation, a second chest CT scan with contrast was performed (figure 1). This revealed that the child had a persistent left-sided superior vena cava (L-SVC) that appeared to be draining into the left atrium, creating a right-to-left shunt which would account for her hypoxaemia. Importantly, it was then noted that during the original contrast echocardiography the contrast was injected into the right arm of this child.

DIFFERENTIAL DIAGNOSIS

We outlined above a physiological approach to hypoxaemia and these categories can be used to generate the following differential diagnosis:

Hypoventilation:

- A. pulmonary hypoplasia
- B. central causes of hypoventilation (eg, congenital central hypoventilation syndrome)
- C. neuromuscular disease
- D. chest wall restriction
- E. sedative agents.

Ventilation-perfusion mismatch:

- A. airways obstruction (asthma, bronchiolitis obliterans)
- B. bronchiectasis
- C. pulmonary oedema/haemorrhage
- D. pulmonary parenchymal disease
- E. pulmonary hypertension.

Right-to-left shunt:

- A. Intracardiac shunt, which includes congenital cyanotic heart lesions (transposition of the great arteries, tricuspid atresia, total/partial anomalous pulmonary venous drainage, truncus arteriosus, tetralogy of Fallot) and acyanotic lesions that lead to elevated pulmonary circulation with resultant Eisenmenger's syndrome and subsequent right-to-left shunt.

Figure 1 Contrast-enhanced CT scan of the chest, demonstrating a persistent left-sided superior vena cava (L-SVC) draining into the left atrium. The arrow towards the top of the image is identifying the persistent L-SVC and the arrow towards the bottom of the image is identifying the left atrium.

- B. Intrapulmonary shunt, which includes pulmonary arteriovenous malformations, either in isolation or in conditions such as hereditary haemorrhagic telangiectasia with or without hepatic/cerebral arteriovenous malformations and hepatopulmonary syndrome.

Widened barrier to diffusion:

- A. Interstitial lung disease
- B. Interstitial oedema
- C. Pulmonary haemorrhage
- D. Hepatopulmonary syndrome

'False' hypoxaemia:

- A. Haemoglobinopathies

TREATMENT

Treatment for a persistent L-SVC is most often performed by an open surgical procedure.

Multiple techniques can be used including: (1) ligation of the SVC and over-sewing the atrial side, (2) the L-SVC can be transposed to the right atrium if it is of sufficient length, (3) the L-SVC can be connected to the right atrium via the coronary sinus by unroofing the coronary sinus, (4) the L-SVC can be transposed to the left pulmonary artery and (5) recent case reports describe an endovascular approach for occlusion of the persistent L-SVC.²

OUTCOME AND FOLLOW-UP

Our patient has been referred to the interventional cardiology and cardiac surgical teams for assessment of repair of this anatomic lesion. They will assess the patient further and decide on the optimal technique for management.

DISCUSSION

Persistent L-SVC is a common anomaly seen in 0.3–0.5% of the general population but may occur in up to 12% of patients with other documented congenital cardiac abnormalities.³ These associated anomalies are most commonly: atrial septal defects, ventricular septal defects, coarctation of the aorta, transposition of the great vessels and tetralogy of Fallot.³ It is also seen in up to 13% of patients with Turners syndrome.⁴ In 80–90% of cases, a persistent L-SVC will drain into the right atrium resulting in no pathological or clinical consequences.³ However, in 10–20% of cases persistent L-SVC drains into the left atrium, which may result in clinically significant hypoxaemia and/or embolic phenomena.^{2 3} Embolic complications resulting in stroke, cerebral abscess and renal/mesenteric infarcts have been described in this condition.^{2 3} This highlights the importance of awareness, recognition and early intervention in persistent L-SVC.

Routine echocardiography performed by inexperienced or uninformed users may miss the diagnosis of persistent L-SVC.⁵ Contrast echocardiography is a sensitive marker for both intracardiac and intrapulmonary shunting.⁶ The shunting in a persistent L-SVC, however, is extracardiac and extrapulmonary and will certainly be missed if contrast is injected in the right arm as was the case in our patient. Diagnosis of persistent L-SVC thus requires a high index of suspicion, informing the echocardiography technician of this concern and ensuring contrast injection into the left arm. Treatment of persistent L-SVC involves endovascular or surgical intervention, as outlined above, depending on patient anatomy and centre-specific expertise.²

In conclusion, persistent L-SVC draining into the left atrium must be considered in the differential diagnosis for hypoxaemia, especially in the context of Turners syndrome or other congenital heart diseases with a higher incidence of this anomaly. Given

the potential serious complications, clinicians need to be aware of this condition and the nuances in diagnosis to facilitate appropriate identification and management.

Learning points

- ▶ Persistent left superior vena cava (L-SVC) draining into the right atrium is common, but much less commonly it drains into the left atrium and may result in hypoxaemia and/or embolic phenomena.
- ▶ Diagnosis of persistent L-SVC requires a high index of suspicion and informative contrast echocardiography requires contrast injection into the left arm of a patient.
- ▶ Identification of this anomaly is important so therapeutic measures can be taken to reduce/eliminate the risk of complications.

Contributors SSB first saw patient in consultation with IN. SSB organised initial investigations and follow-up after first consultation, conducted literature review and prepared manuscript. IN supervised SSB during initial consultation. IN followed up

the patient and provided editorial support in drafting of manuscript. SSB and IN are guarantors.

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REFERENCES

- 1 Bruns CM, Thet LA, Woodson RD, *et al.* Hemoglobinopathy case finding by pulse oximetry. *Am J Hematol* 2003;74:142–3.
- 2 Kougias P, Peden EK, Lin P, *et al.* Endovascular occlusion of right to left arteriovenous shunt associated with persistent left superior vena cava. *J Vasc Surg* 2006;44:875–8.
- 3 Povoski SP, Khabiri H. Persistent left superior vena cava: review of the literature, clinical implications, and relevance of alterations in thoracic central venous anatomy as pertaining to the general principles of central venous access device placement and venography in cancer patients. *World J Surg Oncol* 2011;9:173. <http://www.wjso.com/content/9/1/173> (accessed 2 Jul 2013).
- 4 Ho VB, Bakalov VK, Cooley M, *et al.* Major vascular anomalies in turner syndrome: prevalence and magnetic resonance angiographic features. *Circulation* 2004;110:1694–700.
- 5 Haas C, Doernberger V. LETTER TO THE EDITOR: Persistent left superior vena cava in a patient with congenital heart disease. *Open Cardiovasc Thorac Surg J*. [eLetter] 2009;2:1–2. <http://www.benthamscience.com/open/toctsj/articles/V002/1TOCTSJ.pdf>
- 6 Soliman OII, Geleijnse ML, Meijboom FJ, *et al.* The use of contrast echocardiography for the detection of cardiac shunts. *Eur J Echocardiogr* 2007;8:S2–12.

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