CASE REPORT

An unusual constellation of congenital malformations in a single patient including partial anomalous pulmonary venous return, persistent left superior vena cava, aberrant pulmonary fissure, anomalous aortic arch, tracheal diverticulum and annular pancreas

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Accepted 15 October 2014

SUMMARY

We report a case of a male patient with a constellation of rare congenital anomalies consisting of: partial anomalous pulmonary venous return (PAPVR), persistent left superior vena cava, 'bovine arch' aortic branching, tracheal diverticulum, aberrant lung fissure anatomy and an annular pancreas. He had presented with a history of worsening dyspnoea. To the best of our knowledge, this is the first reported case of a single patient with such a vast constellation of anomalies. The radiological evaluations, epidemiology, embryology and clinical features of the anomalies are discussed. It is important for radiologists to be aware of each of these anomalies as distinct entities; detection of a single anomaly should alert to the possibility that further anatomic aberrancies may be present.

BACKGROUND

Although the precise mechanisms of their development are not fully understood, there is evidence that a patient presenting with one congenital vascular or pulmonary anomaly may have further undetected but clinically relevant anomalies.¹⁻⁴ Partial anomalous pulmonary venous return (PAPVR) is a rare congenital anomaly of the venous connections in the lung. The vein draining the lobe, most commonly the right lower lobe, drains into the inferior vena cava or portal vein, rather than to the left atrium. While PAPVR has previously been reported together with a persistent left superior vena cava (PLSVC), it has never before been seen with a series of other rare congenital malformations comprising aberrant fissure anatomy, a common origin of the brachiocephalic trunk and left common carotid, a tracheal diverticulum and an annular pancreas. This case report describes the radiological evaluations, epidemiology, embryology and clinical implications of these anomalies.

CASE PRESENTATION

A 49-year-old man presented to the accident and emergency department with gradually worsening exertional dyspnoea. Intermittent recent chest pain prior to presentation was also reported. A chest radiograph showed abnormal opacification at the

right hilum ([figure 1](#)). A postcontrast CT scan was performed through the chest and upper abdomen, which showed a number of previously undiagnosed rare congenital malformations. The patient had previously undergone surgery as a newborn for oesophageal atresia. There was no history of haemoptysis, and his initial blood gas, full blood count and electrolytes were unremarkable, although mildly elevated troponin T level and ST depression were noted on ECG.

INVESTIGATIONS

A distended pulmonary vein was seen on chest radiography as a prominence at the right hilum (arrow, [figure 1](#)). A subsequent CT scan revealed an anomalous vertical vein crossing the right lower lobe and draining into the inferior vena cava (horizontal arrow [figure 2](#)). A 'bovine arch' anomaly of the aortic arch was present (vertical arrow [figure 2](#)). The horizontal fissure was also absent and the right lung consisted of only two lobes; an incomplete fissure was seen coursing through the inferior right lower lobe ([figure 3](#) arrow). A left-sided superior vena cava was evident ([figure 4](#)). There was evidence of minor right lung hypoplasia; however, the bronchial tree of the lower lobe was in communication with the trachea and there was no evidence of systemic arterial supply to the affected lower lobe.

Figure 1 Chest X-ray on admission.

To cite: Lapa T, Vedelago J, Kim H, et al. *BMJ Case Rep* Published online: [please include Day Month Year] doi:10.1136/bcr-2012-008014

Figure 2 CT scan performed to investigate abnormality detected on the chest X-ray.

A small diverticulum arising from the posterior aspect of the trachea at the level of the aortic arch was also present (figure 5 arrow). Within the abdomen (figure 6), an annular pancreas was demonstrated (short arrow), encircling the second part of the duodenum with evidence of chronic distention of the first and proximal second parts (long arrow).

OUTCOME AND FOLLOW-UP

The patient was admitted and treated for a non-ST elevation myocardial infarct, several cardiovascular risk factors and early signs of suspected failure. Echocardiography did not

Figure 3 Lower section of the chest CT scan.

Figure 4 Three-dimensional reconstruction of the chest CT scan.

demonstrate evidence of pulmonary arterial hypertension. The patient underwent cardiac catheterisation and placement of two coronary arterial stents at high-grade left coronary arterial circulation stenoses. Surgical therapy was not considered as necessary for the annular pancreas.

DISCUSSION

A case of all of these congenital anomalies occurring in the same patient has never been reported. Scimitar syndrome was first reported by Cooper¹ in 1836 and later named by Neill *et al*² in 1960 as a syndrome consisting of partial anomalous pulmonary venous drainage of the right lung into the inferior vena cava (IVC), partial systemic arterial blood supply and hypoplasia of the affected lung, with bronchial abnormalities and abnormal lobation. Scimitar syndrome is said to share some features with bronchopulmonary sequestration, except that the lung is normally connected to the bronchial tree.

Figure 5 Sagittal reconstruction of the trachea.

Figure 6 CT image of the upper abdomen.

Scimitar syndrome is one of a variety of congenital disorders known as partial anomalous pulmonary venous connection (PAPVC). PAPVC is present in 0.4–0.7% of adults at autopsy⁵ and scimitar syndrome makes up about 3–5% of all cases of PAPVC.^{6–7} The incidence may in fact be higher than this as most cases of scimitar syndrome present in early childhood with large left-to-right shunts and a clinical picture of heart failure and respiratory distress.

Those cases of PAPVC who go unnoticed into adulthood have a small left-to-right shunt and hence may have few or no symptoms.³ Presentation in later life tends to be with shortness of breath on exertion or as an incidental finding on radiological imaging. The characteristic appearance of the aberrant vein on a chest radiograph is of a small ipsilateral lung with mediastinal shift towards the affected side. An abnormal vessel is sometimes seen draining down and enlarging as it curves towards the diaphragm in the shape of a ‘scimitar’, a Turkish sword that it is said to resemble.

Scimitar syndrome is often present with other congenital anomalies. Several authors have reported cases of the syndrome in association with a PLSVC.^{7–8} PLSVC is a remnant of a vessel which drains into the right atrium via the coronary sinus and is the most common thoracic vein anomaly reported in the literature. It is found in approximately 0.5% of the general population⁸ but more commonly in those with congenital heart disease.

A failure in obliteration of the left anterior cardinal vein results in the PLSVC anomaly.⁸ Similarly, a failure in obliteration of the connections between the pulmonary and splanchnic venous plexi results in a persistent pulmonary venous connection with the IVC.⁸ Both venous anomalies are the result of

failure in obliteration of vessels during embryonic development. Sun *et al*⁷ therefore believe that both venous variations can be justified by the same cause.

Scimitar syndrome and PLSVC both have the potential to go undiagnosed for many years until the individual undergoes radiological imaging or surgery. In isolation, neither the ‘adult’ form of scimitar syndrome nor PLSVC carry much clinical significance. However, their association with complex and serious congenital cardiac anomalies and penchant for presenting insidiously with recurrent chest infections, shortness of breath and fatigue, make awareness and radiological diagnosis of these anomalies of particular importance.

Learning points

- ▶ Congenital anomalies of pulmonary venous drainage may present in the adult patient as dyspnoea.
- ▶ A variety of additional congenital anomalies in the chest and abdomen may also be present simultaneously.
- ▶ This patient demonstrated an annular pancreas, in addition to tracheal diverticulum, anomalous aortic arch branch pattern, aberrant pulmonary fissure and pulmonary venous drainage and a persistent left superior vena cava. Radiologists and clinicians should be aware of these anomalies, their clinical implications and the possibility of their co-existence.

Competing interests None.

Patient consent Obtained.

Provenance and peer review Not commissioned; externally peer reviewed.

REFERENCES

- 1 Cooper G. Case of malformation of the thoracic viscera consisting of imperfect development of the right lung and transposition of the heart. *London Med Gaz* 1836;18:600–1.
- 2 Neill CA, Ferencs C, Sabiston DC. The familial occurrence of hypoplastic right lung with systemic arterial supply and venous return, “scimitar syndrome”. *Bull Johns Hopkins Hosp* 1960;107:1–21.
- 3 Kissner DG, Sorkin RP. Anomalous pulmonary venous connection, medical therapy. *Chest* 1986;89:752–4.
- 4 Mathey J, Galey JJ, Logeais Y, *et al*. Anomalous pulmonary venous return to inferior vena cava and associated bronchovascular anomalies (the scimitar syndrome): report of three cases and review of the literature. *Thorax* 1968;23:398–407.
- 5 Snellen HA, van Ingen HC, Hoefsmits EC. Patterns of anomalous pulmonary venous drainage. *Circulation* 1968;38:45–63.
- 6 Tagaki S, Naito M, Kuwabara T, *et al*. Case of scimitar syndrome with persistent left superior vena cava. *Kokyu To Junkan* 1973;21:663–7.
- 7 Sun J, Zhang S, Jiang D, *et al*. Scimitar syndrome with the left persistent superior vena cava. *Surg Radiol Anat* 2009;31:307–9.
- 8 Steinberg I, Dubilier W, Lukas DS. Persistence of left superior vena cava. *Chest* 1953;24:479–88.

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