

## Case Report

# Unusual variant of scimitar syndrome associated with an absent right pulmonary artery, stenosis of the inferior vena cava, hemi-azygous continuation and the VACTERL association

FARIRAI F TAKAWIRA, FAREED OMAR

### Abstract

**We report on a two-month-old infant with an unusual form of scimitar syndrome, associated with an absent right pulmonary artery, obstructed inferior vena cava, hemi-azygous continuation and the VACTERL association. The infant posed a major management problem and eventually died from a lower respiratory tract infection.**

**Keywords:** scimitar syndrome, absent pulmonary artery, hemi-azygous, stenosis, inferior vena cava, VACTERL association

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Scimitar syndrome is a rare and complex congenital anomaly that often involves hypoplasia of the right lung, partial anomalous pulmonary venous connection (PAPVC) of the right lung draining into the inferior vena cava (IVC) and a systemic collateral artery from the descending aorta supplying the right lower lobe (lobar sequestration). The right pulmonary artery (RPA) is often hypoplastic but may be completely absent.<sup>1-3</sup> Scimitar syndrome may be associated with other congenital heart defects or occur in isolation.

We describe a young infant with scimitar syndrome associated with an absent RPA, an obstructed IVC and the VACTERL association (Vertebral defects, Anal atresia, Cardiac anomalies, Tracheo-oesophageal fistula, Renal abnormalities and Limb anomalies). These conditions have not previously been described together in the English literature.

The infant posed a major management problem and eventually died from a lower respiratory tract infection.

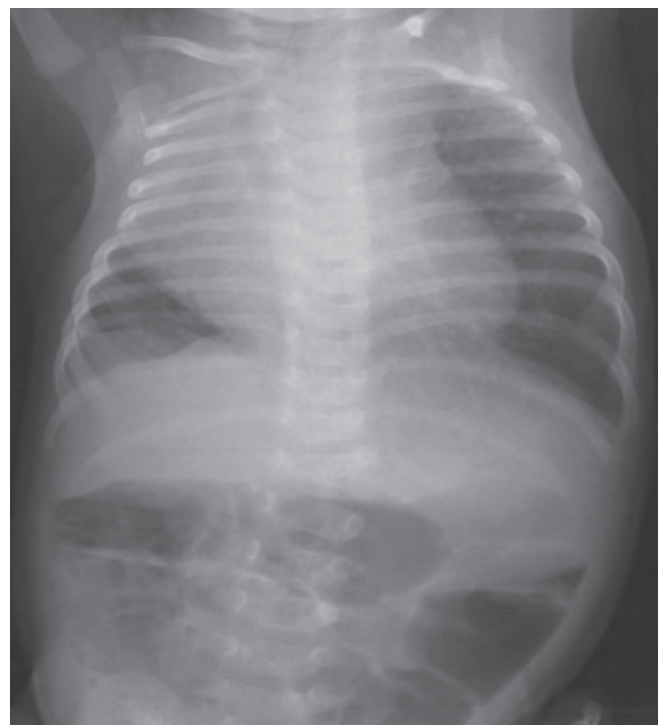
### Case report

A female infant was noted soon after birth to have anal atresia, a recto-vaginal fistula and hypoplasia of the right thumb. She was the product of the first pregnancy of a 19-year-old mother.

She was transferred to our institution at two months of age for elective surgical repair of the fistula.

She was noted to be tachypnoeic and on chest radiography was suspected to have 'dextrocardia'. She was therefore referred to our cardiology service for evaluation.

On clinical examination she weighed 3.3 kg and was not thriving. She was not cyanosed but had saturations of 90% on 2 l/min of nasal oxygen. Her pulse was 160 beats/min and the blood pressure 75/44 mmHg. The trachea deviated to the right due to a right mediastinal shift. The apex beat was felt in the fourth right intercostal space/mid-clavicular line. The second heart sound was loud and a soft 2/6 ejection systolic murmur was audible along the upper sternal border. She was in congestive cardiac failure, as evidenced by tachypnoea, tachycardia, a gallop rhythm and hepatomegaly. There was dullness to percussion of the right hemithorax with decreased air entry.

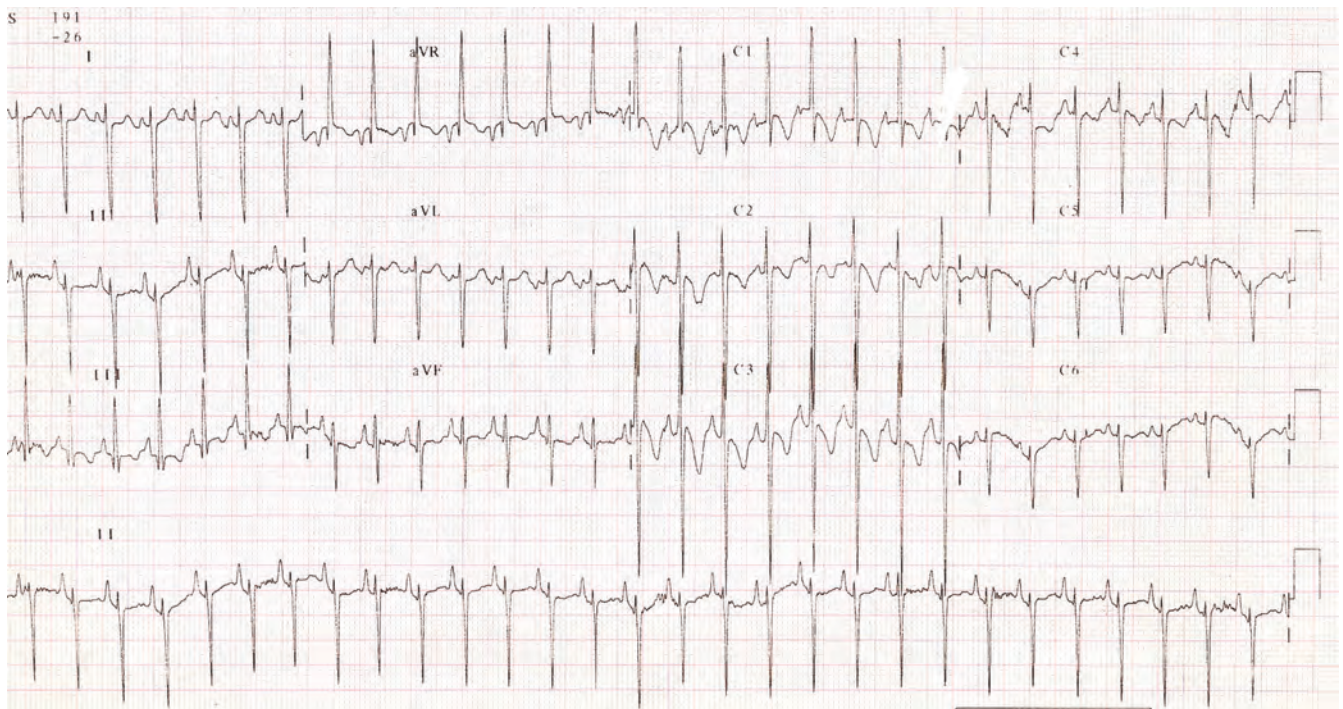


**Fig. 1. Chest radiography demonstrating dextrocardia and right mediastinal shift. Hemivertebrae and scoliosis are also evident.**

Steve Biko Academic Hospital and Department of Paediatric Cardiology, University of Pretoria, South Africa

FARIRAI F TAKAWIRA, FCPaed (SA), fari.takawira@up.ac.za

FAREED OMAR, MBCHB, FCPaed (SA)

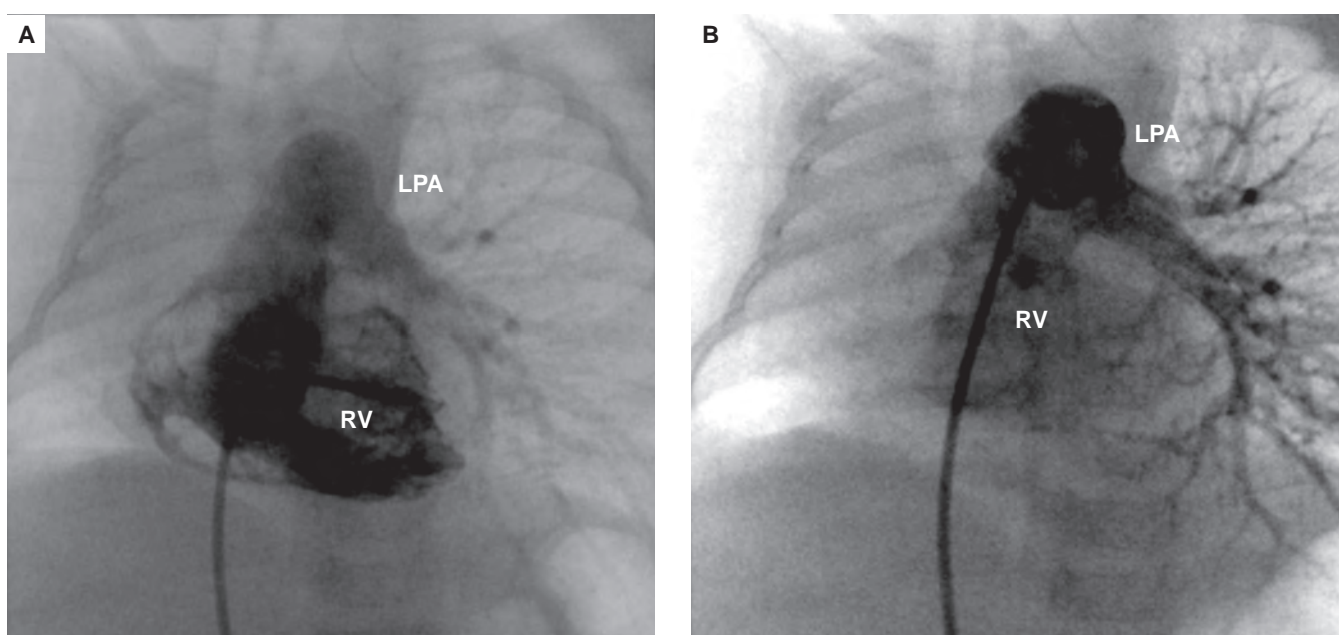


**Fig. 2. Electrocardiogram demonstrates tachycardia, extreme right-axis deviation, right atrial enlargement and right ventricular hypertrophy.**

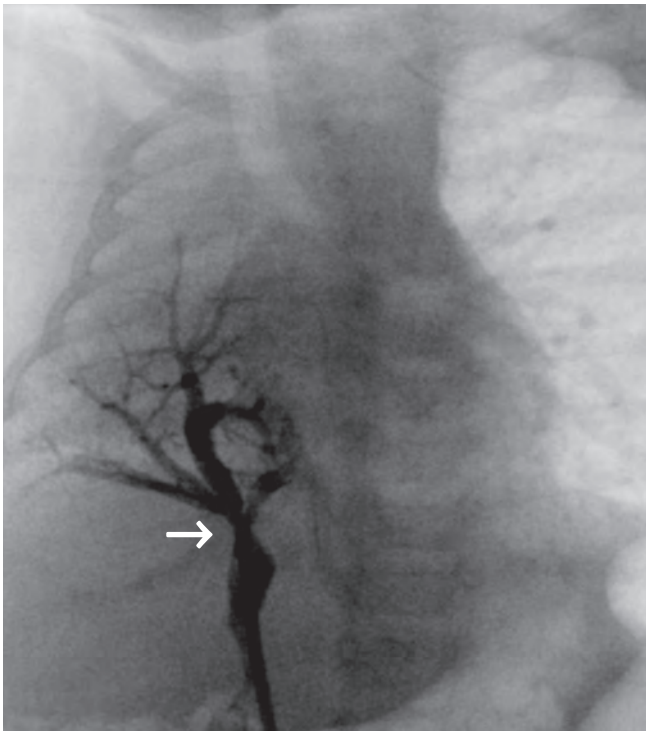
Chest radiography (Fig. 1) demonstrated hypoplasia of the right lung, with a right mediastinal shift. There were hemi-vertebrae and a mild scoliosis present.

Electrocardiography (Fig. 2) demonstrated a heart rate of 170 beats/min, extreme right-axis deviation, right atrial enlargement and marked right ventricular hypertrophy. On echocardiography the heart was shifted over to the right hemithorax and standard views could be obtained by moving the probe towards the right chest.

There was situs solitus and no atrial or ventricular inversion. The right atrium (RA), right ventricle (RV) and main pulmonary artery (MPA) segment were dilated. The dilated right heart compressed the left, with both the interventricular and interatrial septa deviated to the left. The estimated pulmonary pressure from the tricuspid regurgitation gradient was 70 mmHg. A small patent foramen ovale with bidirectional shunting was noted. The right pulmonary artery (RPA) could not be visualised. The left pulmonary veins drained normally into the left atrium, but the



**Fig. 3. RV (A) and MPA (B) angiograms demonstrating RV hypertrophy and an absent RPA. Tracheal deviation to the right is also shown (arrows). (RV, right ventricle; MPA, main pulmonary artery; LPA, left pulmonary artery; RPA, right pulmonary artery).**



**Fig. 4. Abnormal right pulmonary vein draining into the stenosed inferior vena cava (arrow).**

right pulmonary vein was draining into a stenosed inferior vena cava, just before it entered the RA. A gradient of 15 mmHg at this junction was demonstrated.

At cardiac catheterisation, there was a step-up in the high IVC

saturation. Severe pulmonary hypertension of 80/22 mmHg was demonstrated. Angiography demonstrated a hypertrophied and dilated RV, a large MPA and an absent RPA (Fig. 3). PAPVC of the right pulmonary vein draining into the IVC was also noted (Fig. 4). The stenosed IVC was clearly demonstrated before its entry into the RA (Fig. 5). There was a hemi-azygous continuation which drained into the superior vena cava (SVC) and the RA (Fig. 5).

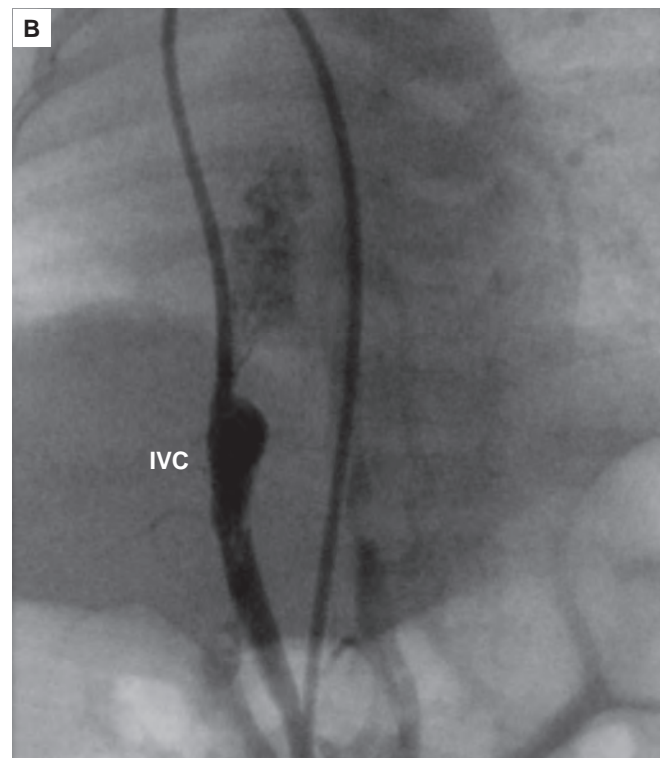
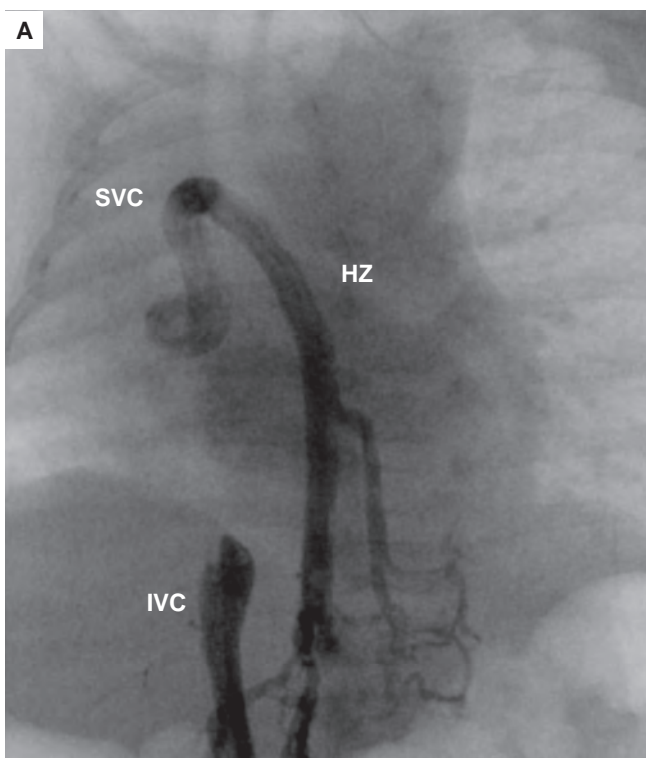
A systemic collateral from the descending aorta supplying the lower lobe of the right lung was visible during the venous phase of the pulmonary angiogram. A diagnosis of scimitar syndrome with an absent RPA and an obstructed IVC was made. In view of the anal atresia, recto-vaginal fistula, hemi-vertebrae with scoliosis, right thumb hypoplasia and scimitar syndrome, the criteria for diagnosis of the VACTERL association were fulfilled.

Due to the absent RPA and right lung hypoplasia, surgical repair of the PAPVC and re-routing of the anomalous right pulmonary vein into the left atrium (LA) were considered of little haemodynamic benefit. Right pneumonectomy was deemed the best surgical option, but could not be undertaken in early infancy.

The patient initially improved with the administration of anti-failure treatment (furosemide and digoxin) and could be discharged from hospital. Unfortunately, she succumbed a few weeks later to a lower respiratory tract infection while at home and an autopsy could not be performed.

## Discussion

Scimitar syndrome is a rare form of PAPVC involving the right lung. There is associated hypoplasia of the right lung and RPA with a right mediastinal shift creating dextroposition of the heart. The RPA may be completely absent,<sup>3,5</sup> as was the



**Fig. 5. (A) Obstructed IVC with hemi-azygous continuation draining into the SVC. (B) Catheter advanced from the femoral vein into the hemi-azygous continuation, SVC, RA, and into the IVC (IVC, inferior vena cava; SVC, superior vena cava; HZ, hemi-zygous, RA, right atrium).**

case in our patient. The abnormal pulmonary vein draining into the IVC can often be seen as a classic curvilinear density on the right chest on plain radiography and is shaped like a Turkish sword or scimitar. This may not always be evident. It is also known as congenital pulmonary veno-lobar syndrome, hypogenic lung syndrome, mirror image lung syndrome, vena cava bronchovascular syndrome, epibronchial right pulmonary artery syndrome or Halasz's syndrome.<sup>1-3</sup>

Halasz first used the term 'scimitar' in 1956,<sup>3</sup> but Neill coined the term 'scimitar syndrome' in 1960.<sup>1-3,6</sup> The aetiology remains unknown. It occurs more frequently in females and may be familial.<sup>1,6</sup> It remains unclear why it predominantly involves the right lung, although there have been a few case reports involving the left lung.<sup>7</sup>

There are several associations described with the scimitar syndrome, including the horse-shoe lung and absence of the RPA. It has rarely been described with a hypoplastic or interrupted IVC. Agnoletti *et al.* described a similar case with scimitar syndrome, absent RPA and a persistent primitive hepatic venous plexus.<sup>8,9</sup> The IVC was also stenosed in their case.

The pathophysiology in scimitar syndrome resembles that of PAPVC or left-to-right shunting at atrial level with volume overload and dilatation of the RA, RV and pulmonary arteries. Pulmonary hypertension may develop, especially where there is associated obstruction of the scimitar vein. The systemic collateral supply from the aorta to the right lung also functions as a left-to-right shunt, increasing the flow through the scimitar vein and worsening the pulmonary hypertension. In our patient, the left lung received the whole cardiac output and this exacerbated the pulmonary hypertension. The abnormal systemic supply leads to sequestration of the left lung. This may lead to recurrent chest infections and bronchiectasis.

There is a wide clinical spectrum in scimitar syndrome, ranging from severely ill infants to asymptomatic adults. The infantile type is the severe form and presents in the first three months of life with respiratory and cardiac failure and often has a poor long-term outcome.<sup>10,11</sup> This group usually has other associated cardiac defects, commonly atrial septal and ventricular septal defects and tends to have severe pulmonary hypertension. The adult type is often asymptomatic, presenting later in life as an incidental finding on chest radiography and is usually benign.

Treatment for symptomatic patients with scimitar syndrome consists of surgical repair of the PAPVC, with the anomalous scimitar vein being redirected to the LA and ligation or coil embolisation of the systemic collateral supply to the right lung.<sup>12-15</sup> Obstruction by thrombosis and fibrosis of the redirected pulmonary vein is a common post-operative complication.<sup>12</sup> Lobectomy, and in some cases, pneumonectomy of the affected lung has been performed.<sup>3,12</sup>

VACTERL association is an acronym used to describe a series of non-randomly associated birth defects (vertebral defects, anal

atresia, cardiac anomalies, tracheo-esophageal fistula, renal abnormalities and limb anomalies). To our knowledge, this rare association of scimitar syndrome, absent RPA, stenosis of the IVC with hemi-azygous continuation and the VACTERL association has not been previously described in the English literature.

Scimitar syndrome as the cardiac component of the VACTERL association is very uncommon. In their series of 32 patients with scimitar syndrome over 20 years, Najm and colleagues describe only one patient with the VACTERL association.<sup>16</sup>

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