Child with ventricular septal defect, absent left upper limb pulses and dysmorphism

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We report a case of a five-year-old boy who presented with recurrent lower respiratory infections since infancy. Examination revealed dysmorphic features in the form of shortening of neck on left side, shortening of left arm by 2 cm, left scapular deformity with prominent spine and under developed wing (Figure 1). On cardiovascular system examination left brachial and radial pulses were absent and a pan systolic murmur of grade IV/VI was audible in left 3rd space. Echocardiography demonstrated a large malaligned non-restrictive ventricular septal defect (VSD) with 40% aortic override. A hypertrophic muscle bundle was present in the middle of right ventricle which produced a gradient of 80 mm recorded on echocardiography. Infundibulum and pulmonary valves were normal. Patient also had a right aortic arch. Association of these dysmorphic features with VSD and double chambered right ventricle (DCRV) was not available in searched literature. Cardiac catheterisation done to look for cause of absent left upper limb pulses demonstrated anomalous origin of left brachiocephalic artery from descending aorta at level of 4th thoracic vertebrae, which further formed a tortuous loop before taking course as normal left subclavian and left carotid artery (Figure 2). Barium swallow examination for pressure effects of anomalous artery on oesophagus did not demonstrate any indentation.

Abnormal body features and absent left upper limb pulsations can be attributed to the abnormality of blood supply to the affected region. A case of right aortic arch, non-communicating double aortic arch, aberrant left brachiocephalic artery causing mid tracheal compression and a small restrictive VSD was reported by Calcagni et al in 2008 from France.¹ Another report by Moes et al² described a case of right aortic arch, restrictive perimembranous VSD with retro oesophageal innominate artery forming a vascular ring with pulmonary artery resulting in tracheal compression and stridor. Dysmorphism was not reported in any of these cases of anomalous left brachiocephalic artery. Association of DCRV, aberrant origin of left brachiocephalic artery at the level of 4th vertebrae, anomalous course with a loop formation resulting in marked dysmorphism and unilateral upper limb pulselessness were the interesting aspects of our case. This is probably the first report of such combination

Figure 1 Photograph showing deformed left scapular spine and wing with shortening of left side of neck.

Figure 2 Angiogram showing anomalous left brachio-cephalic artery which is forming a loop before taking course as left carotid and left sub-clavian artery.

of features in literature. Diagnostic angiography highlighted that the dysmorphism could be related to abnormal blood flow pattern thus widening the spectrum of dysmorphisms secondary to abnormalities in blood flow dynamics.

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ANSWER TO RADIOLOGICAL QUIZ

Chest radiograph (Figure 1) shows smaller right hemithorax with mediastinal shift to right side and a vertically oriented large anomalous curvilinear vessel coursing down from right mid zone and directed inferomedially to the right cardio-phrenic angle parallel to the heart. Contrast enhanced CT scan of chest at the levels of superior and inferior pulmonary veins (Figure 2) shows absence of right superior pulmonary vein and both inferior pulmonary veins are draining normally into the left atrium. Aberrant pulmonary vein is seen alongside of heart which is draining the right upper and middle lobes into the inferior vena cava. Fibreoptic bronchoscopy showed two segmental bronchi in right upper lobe, hypoplastic right middle lobe bronchi and lower lobe bronchi originating high up just below RUL bronchi. Two-dimensional echo cardiograph was normal.

DISCUSSION

The scimitar syndrome or pulmonary venolobar syndrome is a rare, complex and variable malformation of the right lung characterized by an abnormal right sided pulmonary venous drainage into the inferior vena cava, hypoplasia of the right lung, abnormal arterial supply and occasionally cardiac malformation,^{1,2} the appearance of scimitar syndrome is often characteristic. The hypoplastic lung is recognized because of dextraposition of the heart and mediastinal shift to right as seen in our patient.^{3,4} The abnormal venous return is the main component of scimitar syndrome which gives the characteristic abnormal curvilinear radiographic opacities descending along the right cardiac border resembling a scimitar (Curved Turkish sword)⁵ or a women's leg.⁶ The abnormal scimitar vein may drain the entire lung or only a lobe.³⁻⁶ In our case the CT angiography has clearly delineated the scimitar vein which was seen draining the right upper and middle lobe (Figure 3). The usual drainage of the anomalous vein is into the inferior vena cava below or just above the diaphragm.⁷ In our patient it was seen entering the suprahepatic portion of inferior vena cava just below the diaphragm.

The incidence of aberrant systemic arterial supply arising from the aorta varies. Other than conventional angiography, they can be evaluated by CT angiography^{7,8} or MR angiography.^{9,10} Moreover CT scan has the added advantage of demonstrating any associated pulmonary or tracheo-bronchial architectural abnormalities. In our case the pulmonary arterial branches on the right side were smaller with hypoplastic right middle lobe artery and right upper lobe was supplied by two separate arterial branches taking origin directly from right main pulmonary artery.

The clinical presentation of the patients can vary widely.¹¹ About half the patients are asymptomatic despite varying degree

of hypoplasia and pulmonary arterial hypertension. Infants who present with scimitar syndrome are almost always very ill, with evidence of congestive heart failure, and severe pulmonary hypertension. Their symptoms are secondary to the associated congenital cardiac lesions, the presence of systemic collateral arterial supply and the obligatory left-to-right shunt from the anomalous pulmonary venous drainage. Adults on the other hand generally present with minor symptoms, or none at all and the condition is often discovered accidentally. Symptoms may be due to an associated cardiac malformation or to problems in the abnormal lung. Surgery may be needed for patients having associated tracheobronchial anomalies (such as bronchiectasis or pulmonary sequestration), associated cardiac anomalies or left-to-right shunts greater than 2:1. In these cases, identification of a systemic arterial supply is important in order to avoid inadvertent transaction during surgery.

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