Cardiological diagnosis

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Findings

Please refer to page 74 of the September 2010 issue of the *SAJR* (http://www.sajr.org.za/index.php/sajr/article/view/497) for the clinical details and images. We congratulate Dr Nasreen Mohamed (Department of Radiology, University of the Witwatersrand) for her precise and detailed diagnosis, for which she receives an award of R1 000 from the RSSA. Dr Misser elaborates below on the condition and its radiological signs.

Diagnosis

The patient has a background history of childhood cardiac surgery, which is evident in Fig. 2, by the dense calcification of the inter-atrial repair patch for an atrial septal defect. Pulmonary arterial hypertension is indicated by the dilated pulmonary trunk and branches in Figs 1 and 3. A stomach bubble is slightly displaced from the diaphragm outline owing to intervening bridging liver (Fig. 1). The persistent left-sided SVC (unopacified) and interruption of the IVC, with azygous continuation, is noted (Fig. 3). There is levocardia but with situs anomaly of the subphrenic organs including central, left-dominant liver, and right-sided stomach and spleen (with polysplenia). There is no overt bowel malrotation, and the superior mesenteric vessels are normally positioned (Fig. 4). On the coronal reformat in Fig. 5, mild tricuspid regurgitation is seen, and there is suggestion of mild post-ductal aortic coarctation on the sagittal reformat. An additional abnormality is the dysplastic left kidney with only a small cystic remnant. Diagnosis of heterotaxy syndrome (with polysplenia, azygous continuation of the IVC, repaired atrial septal defect and pulmonary arterial hypertension) was made, with incidental associated mild aortic coarctation and left renal dysplasia.

Discussion

Situs anomalies imply a disorder of the organ arrangement in the chest and abdomen, with abnormality of lateralisation. These are divided broadly into those which are complete (situs inversus totalis) or incomplete (heterotaxy syndromes). The embryologic periods of pregastrulation through gastrulation and somitogenesis have all been identified as possible timing of the failure of lateralisation, especially at the 20 - 30-day embryonic stage when cardiac and venous channel development occurs.[1] Mutations of 9 genes have been identified in humans, accounting for the genetic predisposition; however, a number of cases remain sporadic. Autosomal dominant and recessive as well as X-linked recessive inheritance has been described.[1] The heterotaxy syndromes are quite complex entities that require fairly extensive clinico-radiological evaluation including use of chest radiograph, echocardiography (congenital heart defects), abdominal ultrasound (position of major viscera, IVC, splenic abnormality) and CT/MRI scanning. Angiography, upper GI series and Tc-labelled isotope scanning are less utilised owing to high sensitivity of CT scan. Assessment of the following is mandatory: position of atria and cardiac apex, location of aorta, bi- or trilobed lungs, sub-diaphragmatic venous return, position of stomach and/or malrotation, liver/gallbladder position, and the spleen (present/absent, splenuncules).[1,2]

In patients with heterotaxy (with polysplenia), there is a high incidence of congenital cardiac disease[3] and particularly atrial septal defects, atrio-ventricular canal defects or abnormal pulmonary venous return. Pulmonary hypertension may be due to intracardiac shunt, viscero-vascular configuration anomaly or pulmonary microvascular obstructive disease. Azygous continuation of the interrupted hepatic IVC, multiple splenuncules on the same side as the stomach[4] and left-sided isomerism are classic features. There is, however, no single pathognomonic abnormality for this subtype.[2] Prognosis is slightly better in this group compared with asplenia, where the risk for infection is much higher owing to immune function failure.

Appelgate *et al.* revised the classification of heterotaxy and suggested an individualised approach, as used in the patient described. The simplified method includes all aspects of the anomaly in the individual patient stipulated in parentheses. This is a more universal language to describe the abnormality identified, rather than using the basic terminology of asplenia/polysplenia/ isomerism where the exact constellation of findings will not be communicated to the referring clinician or fellow radiologist.

1. Appelgate KE, Goske MJ, Pierce G, Murphy D. Situs revisited: Imaging of the heterotaxy syndrome. Radiographics 1999;19:837-852.
2. Winer-Muram HT, Tonkin ILD. The spectrum of heterotaxic syndromes. Radiol Clin North Am 1989;27:1147-1170.
3. Peoples WM, Moller JH, Edwards JE. Polysplenia: A review of 146 cases. Pediatr Cardiol 1983;4:129-137.
4. Oleszczuk-Raschke K, Set PAK, von Lengerke HJ, Trojer J. Abdominal sonography in the evaluation of heterotaxia in children. Pediatr Radiol 1995; 25(suppl 1): S150-S156.