In this review, the clinical features and management of less commonly encountered cyanotic cardiac lesions are reviewed. Pathophysiology, clinical features, laboratory studies and management are discussed. The clinical and non-invasive laboratory features of these cardiac defects are sufficiently characteristic for the diagnosis and invasive cardiac catheterization and angiographic studies are not routinely required. Such studies may be needed either to define features that could not be clearly defined by non-invasive studies or prior to performing trans-catheter interventions. Surgical correction or effective palliation is possible at relatively low risk. But, residual defects, some requiring repeat catheter or surgical intervention, may be seen in a significant percentage of patients and consequently, continued follow-up after surgery is recommended. [Indian J Pediatr 2009; 76 (3) : 297-308] E-mail: P.Syamasundar.Rao@uth.tmc.edu

Key Words: Cyanotic congenital heart defects; Total anomalous pulmonary venous connection; Truncus arteriosus; Hypoplastic left heart syndrome; Pulmonary atresia; Single ventricle

The most common cyanotic congenital heart lesions (CHDs) are what are called “5 Ts” (Table I). In the first part of this series, the more common cyanotic CHDs namely, tetralogy of Fallot, transposition of the great arteries and tricuspid atresia were discussed. In the second part, the remaining 5 Ts, namely total anomalous pulmonary venous connection (TAPVC) and truncus arteriosus will be reviewed followed by a brief discussion of the other selected cyanotic CHD.

TOTAL ANOMALOUS PULMONARY VENOUS CONNECTION

Pathology and Pathophysiology

In this entity, all the pulmonary veins drain into systemic veins, most commonly they drain into a common pulmonary vein which is then connected to the left innominate vein, superior vena cava, coronary sinus, portal vein or other rare sites. Occasionally individual veins drain directly into the right atrium. Irrespective of the type, all pulmonary venous blood eventually gets back into right atrium, mixes with systemic venous return and gets redistributed to the systemic (via patent foramen ovale) and pulmonary (via tricuspid valve) circulations. TAPVC is rare, occurring in 2 to 3% of CHDs presenting in infancy.

The TAPVC is classified based on the anatomic location to which the connecting veins drain, namely, supra-diaphragmatic (supra-cardiac) or infra-diaphragmatic and physiologic based on obstruction to the pulmonary venous return, namely, obstructive or non-obstructive. The supra-diaphragmatic forms are generally non-obstructive although obstruction can occur in these, as reviewed elsewhere. However; the infra-diaphragmatic forms are almost always obstructive. Connection to the left innominate vein is the most common type of TAPVC. Infra-diaphragmatic type is most common form in the neonate.

The right atrium, right ventricle and pulmonary arteries are enlarged. The left ventricle is of normal size while the left atrium is smaller than normal,
presumably related to lack of pulmonary venous contribution.

Clinical Features
The non-obstructive TAPVC patients usually present with signs of congestive heart failure at about 4 to 6 weeks of life. On examination, they have very mild or no visible cyanosis and may have clinical signs of heart failure. Other clinical features are similar to those seen in patients with secundum atrial septal defect (ASD) in that there is hyperdynamic right ventricular impulse, widely split, fixed second heart sound, a grade II to III/VI ejection systolic murmur at the left upper sternal border and a grade I to II/VI mid-diastolic flow rumble at the left lower sternal border. The obstructive types, on the other hand present within the first few hours to days of life with signs of severe pulmonary venous congestion and manifest severe tachypnea, tachycardia and cyanosis. High degree of suspicion is necessary to rapidly indentify these babies. Sometimes the clinical features are indistinguishable from severe respiratory distress syndrome and group B streptococcal infection.

Laboratory Data
In the non-obstructive type, cardiomegaly and increased pulmonary vascular markings on chest X-ray and right ventricular hypertrophy on an electrocardiogram are seen. In the obstructive type, the heart size is small or normal with evidence for severe pulmonary venous congestion (Fig. 1). Electrocardiogram reveals right ventricular hypertrophy. Echocardiogram shows evidence for right ventricular enlargement and a patent foramen ovale (PFO) with right-to-left shunt. Careful color flow imaging usually demonstrates the site of drainage of pulmonary venous return. Cardiac catheterization is not usually necessary to confirm the diagnosis.

Management
Management of TAPVC with congestive heart failure is by appropriate inotropic support and diuretic administration. The entire systemic flow must pass through the PFO. Consequently, restrictive PFO may cause decreased systemic perfusion. Some patients with supra-diaphragmatic types of TAPVC may have restrictive PFO and in such patients balloon atrial septostomy is beneficial. However, by and large, the management is by surgical correction by anastomosis of the common pulmonary vein with the left atrium. Ligation of the connecting vein and closure of the patent foramen ovale are usually performed, although some surgeon may not opt to close the PFO. In the non-obstructive type, control of congestive heart failure and stabilization of the patient, followed by elective or semi-elective surgery is recommended. In the obstructive type, initial stabilization by intubation and ventilation with high airway pressure should be undertaken. Prostaglandin E1 (PGE1) infusion may have a beneficial role in decompressing the pulmonary circuit and may even open the ductus venosus, thus reducing pulmonary venous obstruction. Following initial stabilization, emergent surgical correction by anastomosis of the common pulmonary vein with the left atrium is mandatory. High mortality associated with surgery has decreased over the years. Clinical and echocardiographic follow-up is recommended to detect development of pulmonary venous obstruction.

TRUNCUS ARTERIOSUS

Pathology and Pathophysiology
In truncus arteriosus, one large vessel (truncus) arises from the heart which overrides a large outlet ventricular septal defect (VSD). The coronary, pulmonary and systemic arteries arise from this single vessel. The atria and ventricles are usually normally formed. The pulmonary artery arises from the truncus and forms the basis of classification. In Type I, the main pulmonary artery (usually short) arises from the side of the truncus (ascending aorta) and divides into right and left pulmonary arteries; this is the most common type of truncus (50 to 70%). In Type II, the right and left pulmonary arteries arise from the posterior aspect of the truncus, most commonly as separate vessels; this is the second most common variety (30 to 50%). In Type III, the pulmonary arteries arise from the lateral aspect of the truncus; this is least common (6 to 10%). In type IV, as described by Collett and Edwards, pulmonary blood flow is derived from the ductus arteriosus and/or