Symposium on Advances in Cardiology - III

Diagnosis and Management of Cyanotic Congenital Heart Disease: Part II

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ABSTRACT

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.[1] 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.

TABLE 1. Common Cyanotic Congenital Heart Defects (5 Ts)

1.	Tetralogy of Fallot
2.	Transposition of the great arteries
3.	Tricuspid atresia
4.	Total anomalous pulmonary venous connection
5.	Truncus arteriosus

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[2] 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.[3] 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,

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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.

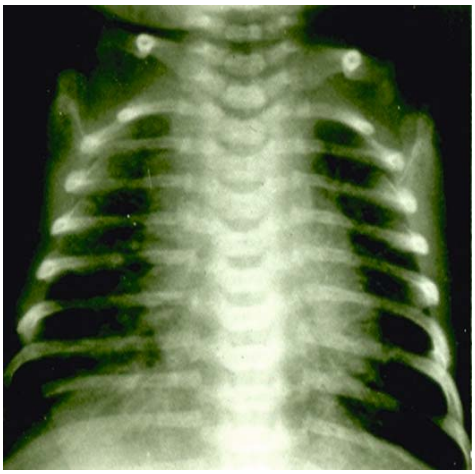

Fig. 1. Postero-anterior view of a chest roentgenogram in a neonate with infradiaphragmatic total anomalous pulmonary venous connection demonstrating severe pulmonary venous congestion.

Management

Management of TAPVC with congestive heart failure is by appropriate inotopic 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.[4] 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 E_1 (PGE_1) infusion may have a beneficial role in decompressing the pulmonary circuit and may even open the ductus venosus,[5] 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.[6] 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.[7] 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,[7] pulmonary blood flow is derived from the ductus arteriosus and/or

multiple aorto-pulmonary collateral vessels from the descending aorta and this entity is no longer considered as a part of truncus arteriosus, but may be described as pulmonary atresia with VSD. The truncal valve leaflets are thickened; they may be tricuspid, bicuspid or quadracuspid; stenosis (5 to 10%) or regurgitation (15 to 20%) of the truncal valve may be present. The pulmonary arteries are not usually stenotic. One of the pulmonary arteries may be absent (16%), usually on the side of the aortic arch or may arise from the descending aorta. In rare cases, one pulmonary artery arises from the ascending aorta and the other comes off the right ventricle, the so called hemi-truncus. Right aortic arch is present in nearly 40% of cases. The association of interrupted aortic arch and DiGeorge syndrome with truncus is well-known.

Clinical Features

Initially the neonate with truncus is not symptomatic because of high pulmonary vascular resistance. Within the next several weeks, the pulmonary vascular resistance drops, increasing the pulmonary flow; eventually signs of congestive heart failure develop. At that point tachypnea, tachycardia, difficulty in feeding and sweating may develop. Because of high pulmonary flow, the cyanosis is minimal. If the pulmonary arteries are stenotic, heart failure may not occur, but the infant becomes cyanotic. In the presence of truncal valve regurgitation, heart failure may occur sooner than those patients without truncal valve disease. Physical examination shows, increased cardiac impulses, bounding pulses and signs of congestive heart failure. Cyanosis, if present is minimal. The first heart sound is usually normal with an ejection systolic click and a single second sound. A holosystolic murmur of VSD is usually present and a mid-diastolic rumble of excessive flow across the mitral valve may also be heard. Classic continuous murmur is not usually present unless there is osteal stenosis of the pulmonary arteries. An early diastolic decrescendo murmur may be present in the presence of truncal valve insufficiency.

Laboratory Data

Chest X-ray demonstrates enlarged cardiac silhouette with increased pulmonary vascular markings. Right aortic arch is present in 30 to 40% patients. The presence of right aortic arch in patients with cardiomegaly and pulmonary plethora who are minimally cyanotic is virtually diagnostic of truncus arteriosus. Biventricular hypertrophy is seen on electrocardiogram. On echocardiogram parasternal long axis and sub-costal views demonstrate a large VSD and a single vessel over-riding the ventricular septum. High parasternal short axis and supra-sternal (Fig 2) views may visualize the origin and branching of the pulmonary artery. Truncal valve may be evaluated in multiple views to document stenosis

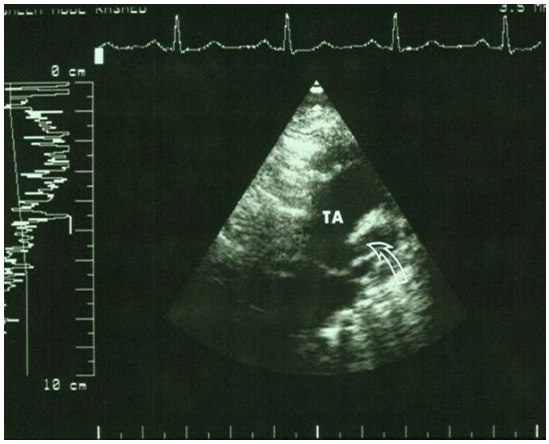

Fig. 2. Selected video frame from a suprasternal notch view demonstrating the truncus arteriosus (TA) giving origin to the pulmonary artery (PA).

or insufficiency. Since the diagnosis may be made with ease on echo-Doppler studies, cardiac catheterization is not usually necessary to make the diagnosis. Some times, catheterization may be necessary to define issues that could not be clearly seen on echo.

Management

Initial attempts to correct truncus arteriosus by a two stage approach (pulmonary artery banding in infancy followed by complete correction in childhood) have now been replaced with complete correction[8,9] within the first few weeks/months of life. Following stabilization with anti-congestive measures, total correction to include closure of the VSD to direct the left ventricular output into the neo-aorta and connecting the disconnected pulmonary stump (from the truncus) to the right ventricle by a homograft should be undertaken. This appears feasible with relatively low mortality although the homograft conduit may have to be replaced[10] because of growth of the child and/or calcific degeneration of the conduit.

HYPOPLASTIC LEFT HEART SYNDROME

Pathology and Pathophysiology

The term hypoplastic left heart syndrome (HLHS), initially proposed by Noonan and Nadas[11] describes a spectrum of cardiac abnormalities characterized by marked hypoplasia of the left ventricle and ascending aorta. Similar to other congenital heart defects, HLHS also shows a spectrum of severity. In the most severe form aortic and mitral valve are atretic with a diminutive ascending aorta and markedly hypoplastic left ventricle.[12,13] The left atrium is usually smaller than normal, although it may be normal in size or enlarged.

It receives all pulmonary veins. The mitral valve may be atretic, hypoplastic or severely stenotic. The left ventricle is usually a thick-walled, slit-like cavity, especially when there is mitral atresia. When the mitral valve is perforate, the left ventricular cavity is small. Endocardial fibroelastosis is usually present. The aortic valve is either severely stenotic or atretic. The ascending aorta is often severely hypoplastic, measuring 2 to 3 mm in diameter, serving as a conduit to supply blood to both coronary arteries in a retrograde fashion. However, on occasion, the aorta may approach normal dimensions. Coarctation of the aorta may be present in a significant number of patients with HLHS, but aortic arch interruption is rare. The right heart, *i.e.*, right atrium, right ventricle and pulmonary arteries is markedly enlarged. A patent foramen ovale with left-to-right shunt is frequently seen. Some times, it is restrictive. Rarely, the patent foramen ovale is completely closed. Ventricular septal defect is not considered to be an integral part of HLHS, although it may be seen in the syndrome of mitral atresia with normal aortic root. Patent ductus arteriosus is present at birth, but tends constrict with increasing age. Severely hypoplastic left ventricle can be present in hearts with double-outlet right ventricle and common atrio-ventricular canal; in some studies, these variants constitute up to 25% of HLHS cases.

The pathophysiology of HLHS is complex. Fully saturated pulmonary venous blood returns to the left atrium and cannot flow into the left ventricle because of obstructed mitral valve. The pulmonary venous blood crosses the atrial septum and mixes with desaturated systemic venous blood in the right atrium. The right ventricle pumps this mixed blood to both the pulmonary and systemic circulations. Blood exiting the right ventricle flows into the lungs *via* the branch pulmonary arteries and into the body *via* the ductus arteriosus. The amount of blood that flows into each circulation is based on the vascular resistance in each circuit. Following birth, pulmonary vascular resistance decreases which allows a higher percentage of the right ventricular output to go to the lungs instead of the body. While increased pulmonary blood flow results in higher oxygen saturation, systemic blood flow is decreased. Perfusion becomes poor, and metabolic acidosis and oliguria may develop. Alternatively, if pulmonary vascular resistance is significantly higher than systemic vascular resistance, systemic blood flow is increased at the expense of pulmonary blood flow. This may result in hypoxemia. A delicate balance between pulmonary and systemic vascular resistance should be maintained to assure adequate oxygenation and tissue perfusion.

Clinical Features

As alluded to above, the postnatal circulation in HLHS depends upon three major factors, namely, adequacy of interatrial communication, patency of the ductus arteriosus and the level of pulmonary vascular resistance. Consequently, the clinical features also depend upon the same factors. At birth, the infants may be asymptomatic. As the ductus begins to close and the pulmonary resistance falls, tachypnea, tachycardia and mild cyanosis may develop. Full-blown picture of congestive heart failure, pale color and poor pulses develop within hours to days of life. Physical signs are non-specific and are those of congestive heart failure, hyperdynamic precordium, single second heart sound and non-specific grade I-II/VI ejection systolic murmur along the left sternal border.

Laboratory Data

Chest roentgenogram reveals moderately to severely enlarged heart with increased pulmonary vascular markings (Fig. 3). There is evidence for both increased flow and pulmonary venous congestion. Electrocardiogram shows right axis deviation, right ventricular hypertrophy, absent q wave in left chest leads and decreased R waves in left chest leads. However, these finding are not diagnostic of HLHS. Blood gas analysis shows evidence for metabolic acidosis, decreased pH, mild respiratory acidosis and only mild arterial desaturation. Echocardiograms are very useful in diagnosis and demonstrate (Fig. 4) enlarged right ventricle, small left atrium, small and hypoplastic left ventricle, and a small ascending aorta. The size of the left ventricle varies form patient to patient as demonstrated in fig 5. Patency of the ductus arteriosus, restrictiveness of PFO should be examined and careful evaluation for presence of aortic coarctation should be done.

Management

Previously considered inoperable, there are two reasonable options at this time:[12,13] cardiac

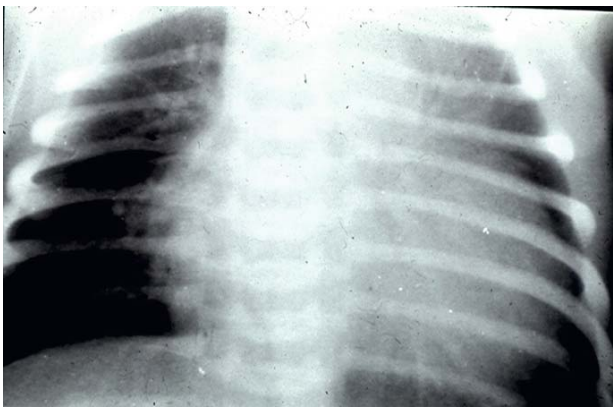

Fig. 3. Postero-anterior view of a chest roentgenogram in a neonate with hypoplastic left heart syndrome demonstrating severe cardiomegaly and increased pulmonary vascular markings.

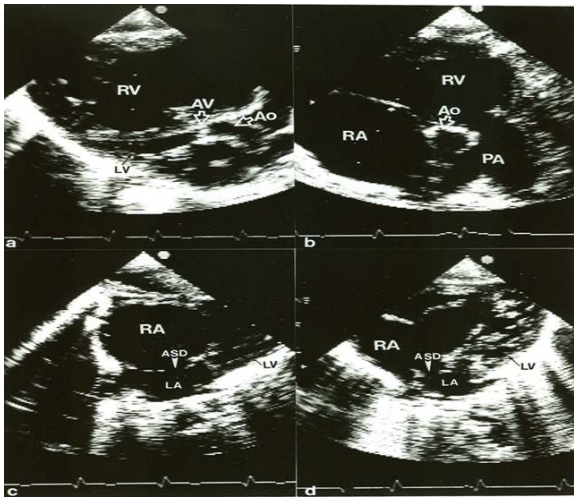

Fig. 4. Selected video frame from a two-dimensional echocardiographic study in a neonate with hypoplastic left heart syndrome demonstrating: a. a very small left ventricle (LV), severely stenotic aortic valve (AV) and a small aorta (Ao). Note the large right ventricle (RV). b. short axis view demonstrating a small Ao, large right atrium (RA), RV and pulmonary artery (PA). c. Subcostal view showing a large RA, small left atrium (LA) and a small LV. The atrial defect (ASD) is show with an arrowhead. d. Another subcostal view, again demonstrated large RA, small LA, small LV and a large RV (not labeled) and an ASD (Arrowhead).

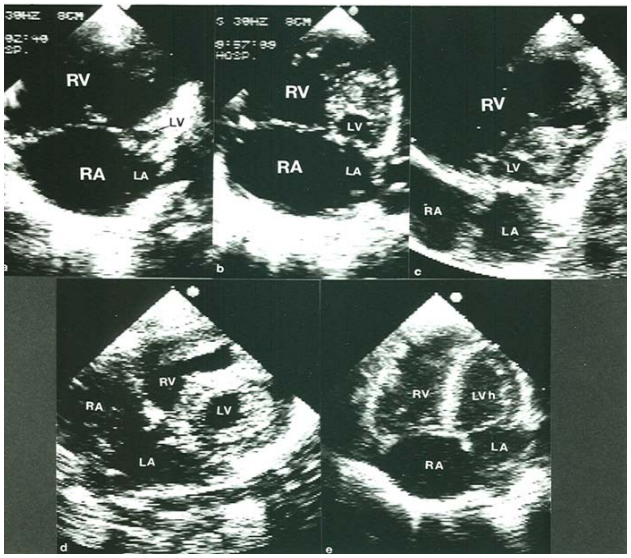

Fig. 5. Selected video frame from four-chamber two-dimensional echocardiographic views in five different neonates with varying sizes of left ventricle (LV). Other abbreviations are as in figure 4.

transplantation[14] and multistage Norwood correction.[15] Because of scarcity of cardiac donors, the majority of centers are performing Norwood procedure initially followed by two-stage Fontan (bidirectional Glenn and Fontan conversion).

Stabilization with medical management including PGE_1 infusion and anti-congestive measures is instituted while decision on final treatment plans is made. The options of supportive care, multistage Norwood procedure and heart transplant are explained to parents in detail. Most parents appear to prefer multistage Norwood procedure. Administration of PGE_1 to maintain ductus open, ensure adequacy of interatrial septal opening, no hyperventilation (maintain $PaCO_2$ at 40 mmHg), and no supplemental oxygen is recommended. Careful balancing of systemic and pulmonary circulations to avoid systemic hypoperfusion is needed.[12,13] Most of the time ambient oxygen concentration less than room air (14 to 18%) to increase pulmonary vascular resistance is necessary to maintain good systemic perfusion while waiting for surgery.

Because of relatively high mortality associated with stage I Norwood operation, alternative procedures such as Sano modification[16] and hybrid procedures[17,18] (stenting the ductus, banding the branch pulmonary arteries and opening the atrial septum by stenting it) are under active investigation.

PULMONARY ATRESIA WITH INTACT VENTRICULAR SEPTUM

Pathology and Pathophysiology

Pulmonary atresia with intact ventricular septum is a complex cyanotic congenital heart defect characterized by complete obstruction of the pulmonary valve, two distinct ventricles, a patent tricuspid valve and no ventricular septal defect.[19] It is a rare disease accounting for 1% of all CHDs. The right ventricle is usually, but not invariably, small and hypoplastic. The main and branch pulmonary arteries are usually of good size; this is in contradistinction to pulmonary atresia with VSD where the pulmonary arteries are usually small and hypoplastic. There is usually a PFO, allowing right to left shunt, decompressing the right atrium. However, in 5 to 10% patients it may be obstructive. Coronary arteiovenous fistulae may be present in 10 to 50% patients. Some of these patients may have right ventricular dependent coronary circulation; these patients are not candidates for relieving the right ventricular outflow obstruction.

Clinical Features

Since there is complete blockage of the pulmonary valve, the pulmonary blood flow is entirely dependent upon the patency of the ductus. Initially (at birth) the ductus is patent and the pulmonary blood flow may be adequate with reasonable arterial PO_2. However, as the ductus begins to close during the natural process of

closure, marked hypoxemia will ensue. At this stage the infants present with severe cyanosis and tachypnea. On examination, the cardiac impulses are quiet, the second heart sound is single and either no murmur or a holosystolic murmur of tricuspid insufficiency may be heard. Occasionally a continuous murmur of PDA may be present. If the atrial septum is obstructive, increased hepatic pulsation and jugular venous distention may be seen.

Laboratory Data

On chest X-ray the heart size may be normal or mildly enlarged with prominent right atrial shadow. The pulmonary vascular markings are decreased. Electrocardiogram shows a normal axis (0 to 90^0) with decreased right ventricular forces (Fig. 6). In the setting of neonatal cyanosis with pulmonary oligemia, this ECG pattern is virtually diagnostic of pulmonary atresia with intact ventricular septum.[20] Echocardiogram will demonstrate small right ventricle without demonstrable forward flow from the right ventricle into the pulmonary artery. Tricuspid regurgitation may be present. Tricuspid valve z scores are largely predictive of potential growth of the right ventricle. Right-to-left shunting across the PFO and left to right shunting across the ductus may be demonstrated. Cardiac catheterization and angiography (Fig. 7) is not usually necessary for diagnosis, but are needed if transcatheter perforation of the pulmonary valve[21] is being considered.

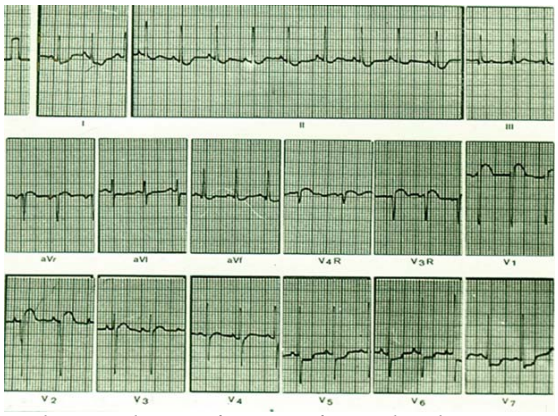

Fig. 6. Electrocardiogram from an infant with pulmonary atresia with intact ventricular septum demonstrating normal axis and diminished right ventricular electrical forces (see text for details).

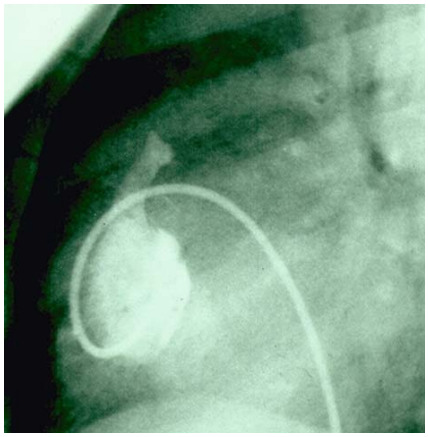

Fig. 7. Selected cine frames in anterio-posterior and lateral views of a right ventricular cine angiogram demonstrating a small and hypoplastic right ventricle. Note that there is no forward flow from the right ventricle, indicating pulmonary atresia.

Management

The prognosis for patients with pulmonary atresia with intact ventricular septum is poor with and without conventional surgical treatment. Because of this reason, a comprehensive program of medical, transcatheter and surgical treatment is necessary to improve long-term outlook of these infants. The principles of such a comprehensive program are: a. to relieve hypoxemia and acidosis by a timely and appropriate procedure to increase pulmonary blood flow at initial presentation, usually in the neonatal period. This is best provided by establishing continuity between the right ventricle and pulmonary artery either by a transcatheter or surgical methodology. If this is not feasible, surgical aortopulmonary shunt or stenting the ductus arteriosus may have to be undertaken. b. to facilitate adequate egress of blood from the right atrium. c. to stimulate growth of the right ventricle so that it could eventually sustain pulmonary circulation, and d. to eventually separate right and left heart circulations, *i.e.*, closure of interatrial and aortopulmonary connections.

The objective of any treatment plan is to achieve a four-chamber, biventricular, completely separated pulmonary and systemic circulations and has been reviewed time-to-time by the author.[19, 21-23] This aim may be achieved in the absence of a. right ventricular dependent coronary circulation, b. severe right ventricular hypoplasia and c. infundibular atresia. If biventricular repair can not be achieved, a one and one-half ventricular repair or a one ventricle (*i.e.*, Fontan) repair should be sought. Algorithms of management plans should be developed based on presence of right ventricular dependent coronary circulation as well as size and morphology of the right ventricle.

In a tripartite or bipartite right ventricle transcatheter radiofrequency perforation is preferable. Alternatively, surgical valvotomy may be performed. Augmentation of pulmonary blood flow by prolonged infusion of PGE_1, stenting the ductus or a surgical modified Blalock-Taussig shunt may be necessary in some of these patients. In patients with a unipartite or very small right ventricle (Tricuspid valve Z score <-2.5) or a right ventricular dependent coronary circulation, augmentation of pulmonary flow along

with atrial septostomy should be undertaken.

Follow-up studies to determine the feasibility of biventricular repair should be undertaken and if feasible, surgical and/or transcatheter methods may be utilized to achieve the goals. If not suitable for biventricular repair, one-ventricle (Fontan) or one and one-half ventricular repair should be considered. Comprehensive and well-planned treatment algorithms may help improve survival rate.

DOUBLE-OUTLET RIGHT VENTRICLE (DORV)

Pathology and Pathophysiology

DORV is a rare cardiac abnormality where both the pulmonary artery and aorta come off of the morphologic right ventricle,[24] comprising less than 1% of all CHDs. Varying definitions have been used: both great vessels coming off of the right ventricle with neither great vessel is in continuity with the atrioventricular valve or one complete great vessel and more than one half of the other great vessel arise from the right ventricle. A large VSD is usually present and is the only exit for the left ventricular output and determines the pathophysiology. In the most common form (50%) the VSD is perimembraneous and subaortic directing the left ventricular output into the aorta. If there is no pulmonary stenosis, the clinical features are those of regular VSD. If there is significant pulmonary stenosis, the clinical picture is that of tetralogy of Fallot. If the VSD is sub-pulmonary (25%), the left ventricular output is largely directed into the pulmonary artery; the physiology is that of transposition of the great arteries and is commonly referred to as Taussig-Bing malformation. In the remaining 25% cases, the VSD is either doubly committed to both vessels or non-committed to either vessel.

The VSD is usually large, but may be congenitally absent causing left ventricular hypoplasia or it may spontaneously close after birth causing severe left ventricular obstruction; these VSDs are termed physiologically advantageous VSDs.[25-27] Pulmonary stenosis, usually subvalvar (rarely pulmonary atresia) may be present in subjects with sub-aortic VSD while in patients with sub-pulmonary VSD, sub-aortic stenosis is present; the latter may be associated with coarctation of the aorta. Most usually the great vessels are abnormally related: side-by-side in 2/3rds, d-malposition in 25% and l-malposition in a small minority. A few patients, particularly with sub-aortic VSD may have normal great artery relationship.

Clinical Features

The clinical features are largely determined by the location of the VSD and presence of pulmonary stenosis. If the VSD is subaortic and there is no pulmonary stenosis, the clinical features are those of regular VSD. If there is significant pulmonary stenosis, the clinical picture is that of tetralogy of Fallot. If the VSD is sub-pulmonary (25%), the clinical picture is that of transposition of the great arteries.

Laboratory Data

Features on chest X-ray, again, largely depend upon the VSD position and degree of right ventricular outflow obstruction, demonstrating cardiomegaly and increased pulmonary blood flow in patients with subaortic VSD and no pulmonary stenosis and mild or no cardiomegaly and decreased pulmonary vascular markings in the patients with pulmonary stenosis. Sub-pulmonary VSD patients simulate transposition with cardiomegaly and increased pulmonary markings. There is no pathagnomonic ECG pattern, but right ventricular or biventricular hypertrophy is usual. Severe left ventricular hypertrophy may be seen if the VSD is obstructive. Echocardiogram is useful in evaluation of the size and function of the ventricles, size and location of the VSD, relative positions of the great arteries and degree of pulmonary stenosis, if present. Sub-aortic obstruction and coarctation of the aorta should be scrutinized in patients with sub-pulmonary VSD. Cardiac catheterization and selective cineangiography is needed in most cases to define the various issues alluded to, unless the echocardiographic studies clearly demonstrate the anatomy.

Management

In patients with subaortic VSD and no pulmonary stenosis, surgical closure of the VSD to divert the left ventricular outflow into the aorta is recommended once the patient is stabilized by appropriate anti-congestive measures. In patients with subaortic VSD and pulmonary stenosis, surgical correction is similar that used for tetralogy of Fallot repair including patch augmentation of the subpulmonary obstruction. Alternatively, initially a modified Blalock-Taussig shunt may be performed if the anatomy is not suitable with later total correction planned. In patients with subpulmonary VSD (Taussig-Bing), arterial switch along with VSD closure is recommended. If there is associated pulmonary stenosis, Rastelli type of surgery may be required. In the presence severe subaortic obstruction, Damu-Kaye-Stansel procedure may be required.

UNIVENTRICULAR HEARTS

Pathology and Pathophysiology

A number of entities are included in univentricular hearts and include double-inlet left ventricle (DILV), single ventricle, common ventricle and univentricular

atrio-ventricular (AV) connection. These hearts may have both AV valves empting into the ventricle (DILV), may have one AV valve empting into the ventricle (tricuspid and mitral atresia) or there may be a common AV valve emptying into the ventricle. DILV is the most common form of univentricular atrio-ventricular (AV) connection and will be dealt in this section. The management of the other entities is similar.

In DILV, the ventricle most commonly has left ventricular morphology, although right ventricular, mixed ventricular and indeterminate or undifferentiated morphologies can occur. In the most common form, the main ventricle has left ventricular morphology with an outlet chamber attached to it which has a right ventricular morphology. The AV valves may be normal, stenotic, hypoplastic or atretic. The great vessels are most usually transposed, the aorta arising from the hypoplastic right ventricular chamber and the pulmonary artery from the main left ventricular chamber. L-transposition occurs more frequently than d-transposition. Normally related great arteries are found in less than 30% cases. Double-outlet right ventricle can also occur where both great arteries come off the rudimentary right ventricular chamber. Pulmonary stenosis occurs in 2/3rds of the patients and such obstruction is present in all forms of great artery relationship. The narrowing may be valvar or subvalvar. Pulmonary atresia may also present. Subaortic obstruction occurs in patients with transposition and is due to the stenosis of the bulbovnetricular foramen (VSD). These patients often have associated coarctation of the aorta.

Clinical Features

The clinical presentation is largely determined by the degree of pulmonary outflow obstruction. In patients without pulmonary stenosis, symptoms of heart failure develop within the first few months of life. There is minimal cyanosis because of high pulmonary blood flow. The heart sounds are normal with a nonspecific ejection murmur (I to II/VI) along the left sternal border and a mid-diastolic rumble at the apex. In patients with pulmonary stenosis, cyanosis and hypoxemia are the presenting symptoms. If the pulmonary stenosis is severe, they may present in the neonatal period. Single second heart sound and a long ejection systolic murmur with a thrill at the left upper sternal border may be present.

Laboratory Data

The chest X-ray appearance is variable depending upon the type of the anatomy and magnitude of pulmonary flow. In patients with increased pulmonary flow, moderate cardiomegaly and increased pulmonary vascular markings are seen. In patients with decreased pulmonary flow, mild cardiomegaly and decreased pulmonary vascular markings are seen. In patients with l-transposition, prominent left heart border may be seen. Electrocardiogram may show abnormal initial QRS forces with qR pattern in the right chest leads and Rs pattern in the left chest leads. Right, left or bi-ventricular hypertrophy may be seen depending upon the anatomic type. Echocardiogram is very useful in making the diagnosis. The absence of the ventricular septum can be demonstrated (Fig. 8). The AV connections, valves, ventriculo-arterial connections and presence of pulmonary stenosis can be ascertained. Sometimes, cardiac catheterization and angiography may become necessary to define all the issues associated with single ventricle.

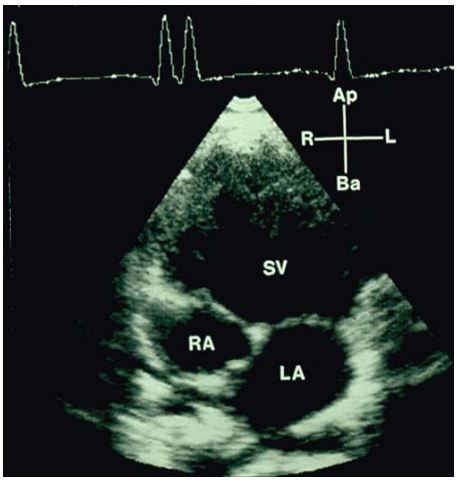

Fig. 8. Selected video frame from four-chamber two-dimensional echocardiographic views from two children: Left panel is from a normal child demonstrating normal position of the right atrium (RA), right ventricle (RV), left atrium (LA) and left ventricle (LV). The atrial and ventricular septae are seen and appear intact. Right panel is from a child with double-inlet left ventricle demonstrating normal atria and atrioventricular valves and a single ventricle (SV). Note the intact atrial septum and no ventricular septum.

Management

Following initial description by Fontan and Kreutzer[28,29] of physiologically corrective operation for tricuspid atresia, it was widely adapted and the concept extended to treat other cardiac defects with a functionally single ventricle. A number of modifications of these procedures were undertaken by these and other workers in the field, as reviewed by elsewhere.[30-32] At the present time, staged cavopulmonary connection[33] is the Fontan procedure of choice *i.e.*, a bidirectional Glenn initially, followed later by final conversion to Fontan. Most surgeons opine that extra-cardiac conduit is preferable to lateral tunnel for diversion of inferior vena caval blood into the pulmonary artery. Creation of fenestration at the time of Fontan[34-36] for high risk patients was found beneficial. However, some surgeons perform fenestrated Fontan irrespective of the risk

factors. Because the fenestrations produce hypoxemia (by design) and provide an avenue for paradoxical embolism, potentially causing cerebrovascular accidents, there was some hesitation in fenestrating all patients. Some workers have demonstrated better results after fenestrated Fontan than without fenestration for all groups of patients.

The age and weight of the patient and anatomic and physiologic status of the patient determine the types of surgery needed. The overall objective is to achieve a total cavopulmonary connection. In the neonates and young infants with pulmonary oligemia, a modified Blalock-Taussig shunt is undertaken to improve the pulmonary oligemia. In patients with pulmonary plethora, early pulmonary artery banding and relief of aortic coarctation, if present, should be promptly performed. Development of subaortic obstruction following banding has been of concern, but the author believes that development of subaortic stenosis is more likely related to time-related natural history of the disease rather than a direct relationship to banding.[37] The neonatal palliations should be followed by bidirectional Glenn and Fontan as alluded to above. Bypassing (by Damus-Kaye-Stansel) or resection of subaortic obstruction should be incorporated into the management plan.

PULMONARY ATRESIA WITHOUT INTACT VENTRICULAR SEPTUM

Pulmonary atresia can also occur in patients with tetralogy of Fallot and many other complex congenital heart defects such as double-inlet left ventricle, transposition of the great arteries, double outlet right ventricle, corrected transposition and tricuspid atresia. Some of the patients with tetralogy of Fallot may have multiple aortopulmonary collateral arteries (MAPCAS). Initial palliation with PGE_1 infusion followed by a modified Blalock-Taussig shunt is undertaken. Patients with two functioning ventricle should undergo bi-ventricular repair with closure of the VSD and right ventricle to pulmonary artery conduit. Unifocalization or other forms of augmentation of pulmonary arteries may be required in patients with MAPCAS. Patients with one functioning ventricle should have Fontan palliation. The readers are referred to standard textbooks on Pediatric Cardiology for further discussion of these entities.

INTERRUPTED AORTIC ARCH

Pathology and Pathophysiology

Interrupted aortic arch (IAA) may be defined as a complete loss of luminal communication between the ascending and descending aorta; this may occur at various levels in the aortic arch.[38] IAA comprises less than 1% of all congenital heart disease. Three types of IAA are identified:[39] type A, type B, and type C, each with subtypes depending on the origin and/or course of the subclavian arteries. In type A, the arch discontinuity is distal to left subclavian artery. Type B is discontinuity between the left common carotid artery and the left subclavian artery. In type C, the discontinuity is between the right innominate artery and the left common carotid artery. The interruption can occur both with left and right aortic arches. Type B interruption is the most common,[40] comprising approximately 52% of all interrupted aortic arch cases. Type A interruption comprises 44% of cases. Type C interruption is least common and occurs with a frequency of 4% of cases.

IAA is not usually seen as an isolated lesion. It is almost certainly associated with a patent ductus arteriosus which establishes continuity between the main pulmonary artery and the descending aorta. A VSD is present in 50% of patients with type A interruption, and close to 80% of cases with type B defects. Aortic stenosis, both subvalvar and valvar, truncus arteriosus, double outlet right ventricle (Taussig-Bing anomaly), single ventricle, transposition of the great arteries, and aorto-pulmonary septal defects are also commonly found in association with interrupted arch. There is a strong association with DiGeorge's syndrome, chromosome 22q11 microdeletion; this is most common in the type B interruption.

Clinical Features

In the immediate neonatal period, the patient is asymptomatic, and remains so until ductal constriction ensues or pulmonary vascular resistance drops. Even though the lower half of the body is supplied by the right ventricle by way of the patent ductus arteriosus, there is no deferential cyanosis (upper extremity pink, lower extremity cyanotic) because of shunting of oxygenated blood into the right ventricle and pulmonary artery across the VSD. There is no blood pressure differential in the setting of an unrestricted PDA. As the PDA constricts, the blood flow to the descending aorta is compromised and cardiovascular collapse becomes apparent from metabolic acidosis and pulmonary edema.

Laboratory Data

Chest radiography shows cardiomegaly and increased pulmonary blood flow or pulmonary edema. Electrocardiogram may show right, left or bi-ventricular hypertrophy. The corrected QTc may be prolonged due to hypocalcaemia if the patient has DiGeoge's syndrome. Diagnosis rests on clinical intuition and is

usually confirmed by two dimensional echocardiography.[41] Cardiac catheterization is rarely necessary to establish a diagnosis.

Management

If left untreated, IAA is fatal within the first week or two of life. Prior to the advent of PGE_1, these patients would present in severe cardiovascular collapse with renal and hepatic dysfunction, and would have to undergo surgical intervention under suboptimal conditions. Intravenous administration of PGE_1 should start as soon as the diagnosis is made and the infant should stabilized prior to surgical intervention, allowing a return to normalcy of end organ function. Surgical correction is aimed at a complete primary correction with arch repair and closure of the VSD. Of the several options available, direct anastomosis with patch augmentation is preferable.[42] The use of interposition grafts should be avoided as they will not grow with the baby. The mortality is high and the higher risk is seen with associated complex heart disease and poor condition at presentation. In cases, with sever left ventricular outflow tract obstruction/ hypoplasia, Norwood or Damus-Kaye-Stansel approaches may become necessary. Re-intervention is required in 28% patients.[42] The prognosis has improved during the last decade because of advances in neonatal, medical, anesthetic and surgical management of these sick babies.

SYNDROME OF ABSENT PULMONARY VALVE

Pathology and Pathophysiology

The pathologic features of absent pulmonary valve syndrome are absent or rudimentary pulmonary valve cusps causing pulmonary insufficiency, pulmonary valve ring hypoplasia producing pulmonary stenosis and massive dilatation of the main and major branch pulmonary arteries resulting in varying degrees of compression of the tracheobronchial tree. This is a rare congenital heart defect and constitutes 3 to 5% of tetralogy of Fallot cases. This syndrome is usually associated with ventricular septal defect and pulmonary stenosis although it may be occasionally seen as an isolated malformation or with other defects, namely, atrial septal defect, ventricular septal defect, patient ductus arteriosus, endocardial cushion defect and double outlet right ventricle.

Clinical Features

Two types of clinical presentation are recognized: presentation in early infancy with severe cardio-respiratory distress (because of tracheobronchial compression) and those who present beyond infancy with milder respiratory difficulty. Apart from respiratory difficulty, the infants may have signs of heart failure and are mildly cyanotic. Hyperdynamic cardiac impulses, single second sound, and a to-and-fro murmur of pulmonary stenosis and insufficiency at the left upper sternal border are the other physical findings.

Laboratory Data

Chest roentgenogram shows mild cardiomegaly, slightly increased pulmonary vascular markings and most importantly prominent right and left pulmonary arteries. Electrocardiogram is suggestive of right ventricular hypertrophy. Echo-Doppler studies demonstrate a large ventricular septal defect, markedly dilated main and branch pulmonary arteries, absent or rudimentary pulmonary valve leaflets and Doppler evidence for pulmonary stenosis and pulmonary insufficiency. Cardiac catheterization data are suggestive of left-to-right shunt at ventricular level, mild systemic arterial desaturation (secondary to both pulmonary venous desaturation and small interventricular right-to-left shunting), significant peak-to-peak systolic pressure gradient across the pulmonary valve and an increased ratio of systemic-to-pulmonary flow. Selective cineangiography demonstrates a large subaortic perimembraneous ventricular septal defect, moderate-to-severe pulmonary valve ring stenosis and impressive dilatation of main, right and left pulmonary arteries (Fig 9).

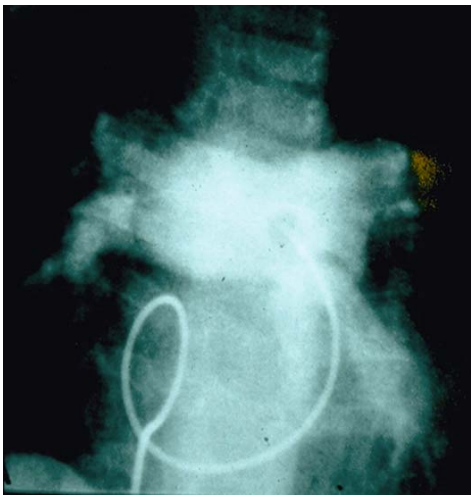

Fig. 9. Selected right ventricular outflow tract cineangiographic frame in posterior-anterior view demonstrating markedly dilated main and branch pulmonary arteries.

Management

Treatment consists of anticongestive measures, chest physiotherapy and ventilatory support to stabilize the patient followed by total surgical correction (under cardiopulmonary bypass), including closure of the ventricular septal defect, relief of pulmonary stenosis by

a transannular pericardial patch as necessary and partial resection and plastic repair of aneurysmally dilated pulmonary arteries.[43] There is some controversy with regard to prosthetic replacement of the pulmonary valve at the time of primary repair[44] but, valve replacement may not be necessary at the time of primary repair.[43]

CONCLUSION

In this review, total anomalous pulmonary venous connection, truncus arteriosus, hypoplastic left heart syndrome, pulmonary atresia with intact ventricular septum, double outlet right ventricle, univentricular hearts (double-inlet left ventricle), pulmonary atresia associated other complex cardiac defects, interrupted aortic arch and syndrome of absent pulmonary valve are discussed. The pathophysiologic features, clinical presentation, laboratory findings and management options are described. These defects have sufficiently distinctive features such that they can be diagnosed with relative ease. Upon diagnosis, some require immediate treatment for stabilization and all require subsequent corrective and/or palliative surgical therapy. Significant post-operative residual abnormalities may occur, some may requiring catheter interventional and/or repeat surgical procedures. Periodic follow-up to detect and address these abnormalities is important.

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