ASD

Atrial septal defect (ASD) is the commonest form of congenital heart disease seen in adults. The commonest form of defect is the **secundum ASD**, accounting for two thirds of cases, in which the fossa ovalis is absent, leaving a defect in the centre of the interatrial septum. **Primum ASD** is rarer and causes a defect in the inferior interatrial septum, often associated with a cleft anterior mitral valve leaflet. **Sinus venosus ASD** is also rare and is found near to where the superior or inferior vena cava joins the right atrium (RA). It is associated with partial anomalous pulmonary venous drainage, in which one or more pulmonary veins drain directly into the RA (or one of the vena cavae) instead of the LA.

In the apical view, it is not unusual to see areas of 'apparent' dropout in the IAS, which is quite a long way from the probe, so be careful not to report dropout as an ASD unless you can also see it in other views and/or you also have further supporting evidence.

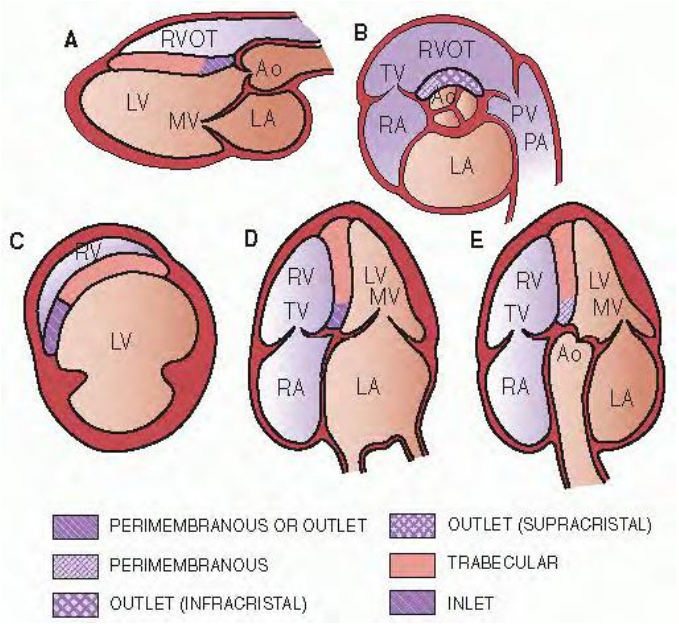
Echo assessment of ASD:

- Use colour Doppler to check for the presence of flow across the defect. Flow across an ASD is normally from left to right, mainly during diastole, and also in systole
- In the subcostal view, use pulsed-wave (PW) Doppler to assess flow across the defect
- Dilated RA & RV as a consequence of a left-to-right shunt. Remember: no LV volume overload or dilatation.
- Evidence of right heart volume overload (paradoxical motion of the IVS)
- Shunt ratio (Qp/Qs) is calculated using Bernoulli equation and continuity equation. $Qp/Qs = SV_{RVOT} / SV_{LVOT} = (CSA_{RVOT} \times VTI_{RVOT}) / (CSA_{LVOT} \times VTI_{LVOT})$. Use PW Doppler to measure the VTI_{RVOT} and VTI_{LVOT}. A significant limitation to shunt calculations is that they are heavily dependent on an accurate measurement of RVOT and LVOT diameter – as the calculation involves squaring these measurements

Atrial septal aneurysm is a congenital cardiac abnormality that is characterized by saccular formation of the IAS, generally at the level of the fossa ovale. Redundant atrial septal tissue results in bulging of the septum into either or both atria during the cardiac cycle, reflecting the relative pressure gradient between LA and RA. The diagnosis is best established with TOE since it can be easily missed with transthoracic echocardiography. It can be seen in all age groups and are often a coincidental finding. A standardized definition of atrial septal aneurysm requires maximal deviation of the aneurysmal tissue of at least 10 mm from the plane of the septum. Thrombi may form in the pouches created by the septum on either the left or right side and have been associated with thromboembolic events. Atrial septal aneurysms are associated with either a PFO or an ASD in as many as 75% of cases. The combination of an atrial septal aneurysm and a PFO has recently been associated with substantial risk of thromboembolism. When an atrial septal aneurysm is detected, it is often appropriate to perform a venous saline contrast injection to search for an associated PFO because its presence may alter management.

VSD

The interventricular septum has two parts: the muscular septum and the thinner, fibrous membranous septum (which lies just below the aortic valve). VSD is the commonest congenital heart defect and can be acquired as a complication of myocardial infarction. VSDs can be categorized according to their location as:



- **(Peri-)membranous VSD** – the commonest type, located in the membranous part of the septum below the aortic valve. It is well seen in the PLAX.
- **Muscular VSD** – found in the muscular part of the septum. Muscular VSDs can be multiple ('Swiss cheese septum').
- **Inlet VSD** – also known as canal-type or posterior VSD, this is found posterior to the tricuspid septal leaflet and may be associated with an AV canal defect. It is well seen in the apical 4C.
- **Subpulmonary VSD** – also known as supracristal, outlet or doubly committed VSD, this type is uncommon, and lies just below the aortic and pulmonary valves. It is well seen on the PSAX. This type of VSD is commonly associated with AR due to prolapse of the right coronary cusp of the AV.

With a left-to-right shunting VSD, blood is shunted from the LV to the RV in systole whilst both are contracting, and hence blood is shunted directly from LV to RV to PA without pooling in the RV, causing PA, LA, and LV volume overload, but not RV volume overload or dilatation. Presence of RV volume overload should prompt a search for another cause such as TR or ASD.

The left-to-right shunting of blood can lead to pulmonary hypertension which can cause reversal of the shunt (Eisenmenger's syndrome)

The Gerbode defect usually involves a shunt from the LV to the RA

Echo assessment of VSD:

- Multiple views are required to examine the entire septal region and a single imaging plane will neither interrogate the complete structure nor detect every defect.
- **Perimembranous** defects are visible in the PLAX and PSAX. Slight medial angulation of the long-axis plane is required to record this area.
- The SAX view further permits classification of **outlet defects** as being either above or below the crista supraventricularis. Supracristal defects are optimally detected from a high parasternal long-axis or parasternal short-axis view.
- The apical 4C permits visualization of both the **inlet and trabecular** ventricular septum. By tilting the scanning plane inferiorly, the inlet portion of the septum is imaged in the area between the atrioventricular valves.
- In the subcostal view, use CW and PW Doppler to assess flow across the defect. There is usually a high-velocity jet from left to right ventricle during systole, with lower-velocity flow during diastole. If you identify a VSD, check for any associated abnormalities (e.g. aortic cusp prolapse, AR). Check also for the presence of tricuspid and/or pulmonary regurgitation and, where possible, assess pulmonary artery pressure in case pulmonary hypertension has developed. Shunt ratio (Qp/Qs) is calculated using Bernoulli equation and continuity equation. $Qp/Qs = SV_{RVOT} / SV_{LVOT} = (CSA_{RVOT} \times VTI_{RVOT}) / (CSA_{LVOT} \times VTI_{LVOT})$.

ENDOCARDIAL CUSHION DEFECT

This is a defect at the A-V junction. Failure of division of the common A-V canal into left and right sides results in an **atrioventricular septal defect** with various combinations of ostium primum ASD, inlet VSD, and structural abnormalities of the A-V valves. Thus, an endocardial cushion defect is a spectrum of lesions including partial A-V canal (implying separate atrioventricular orifices), complete A-V canal (a common A-V orifice), and isolated inlet VSD. A common A-V valve is common. The lack of normal offsetting of septal leaflet of atrioventricular valve is best demonstrated in the apical 4C.

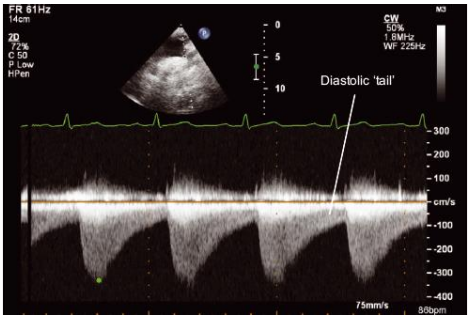
PDA

Persistent ductus arteriosus (PDA) is also sometimes referred to as *patent* ductus arteriosus. In the foetus, the ductus arteriosus acts as a shunt connecting the PA (at the junction of the main and left pulmonary arteries) to the aortic arch (just after the origin of the left subclavian artery). This allows most (90%) of the blood pumped by the RV to reach the systemic circulation directly, bypassing the lungs. The ductus arteriosus normally starts to close immediately after birth, and is normally fully closed within a few days, leaving behind just a cord-like remnant (the ligamentum arteriosum). Failure of the ductus arteriosus to close means that a L-R shunt persists between the aortic arch and the pulmonary artery, with blood flow from the high pressure aorta to the lower-pressure PA. This leads to excessive blood flow through the pulmonary circulation and, in the longer term, can cause pulmonary hypertension. The increased pulmonary blood flow and therefore pulmonary venous return to the LA causes LA and LV dilatation due to volume overload.

EISENMENGER SYNDROME

The presence of a L-R shunt (such as an ASD, VSD or PDA) allows blood to pass directly from the left side of the circulation to the right, increasing the volume of blood flowing through the pulmonary circulation. This leads to an increased pressure within the pulmonary vessels (pulmonary hypertension) and, over time, the vessels develop an increasing resistance to blood flow. This leads to a back pressure on the right heart and the development of right ventricular hypertrophy. Gradually, the right-sided pressures rise and begin to equal and then exceed the pressures found in the left heart. As this occurs, the L-R shunt reverses, causing R-L shunt instead. At this point, the patient is said to have developed **Eisenmenger syndrome** (or reaction). This means that a portion of the venous (deoxygenated) blood entering the right heart starts crossing directly into the left heart, bypassing the lungs, and reducing the overall oxygen content in the arterial circulation. Clinically, the patient develops **cyanosis**, a blue discoloration of the skin and tongue, together with breathlessness and a fall in exercise capacity.

AORTIC COARCTATION

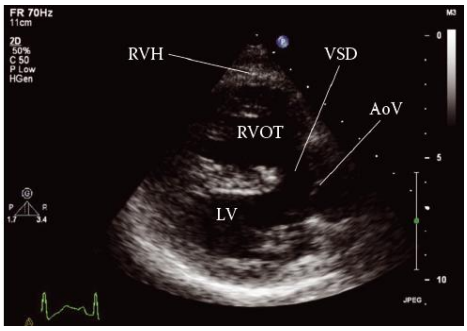


Aortic coarctation is a narrowing of the aorta that most commonly occurs just distal to the origin of the left subclavian artery. It accounts for 5–8% of congenital heart disease (more commonly in males). Patients may have the clinical features of an associated condition such as bicuspid aortic valve (**> 50% of cases of aortic coarctation** are associated with bicuspid AV) or Turner syndrome. It is also associated with subvalvar, valvular and supravalvar aortic stenosis and with parachute mitral valve. There may be dilatation of the aorta on either side of the coarctation. Use CW Doppler to assess flow in the descending aorta, looking for evidence of increased flow velocity through the coarctation. However, be aware that high flow velocities measured with CW Doppler are *not* a reliable guide to coarctation severity. A better guide is the presence of sustained anterograde diastolic flow in the aorta (diastolic 'tail' or 'run-off'), the presence of which suggests a significant coarctation.

TETRALOGY OF FALLOT

Anatomy of Tetralogy of Fallot:

A. Antero-cephalad deviation of the outlet septum result in 4 defining features:
   1) PS (RVOT obstruction)
   2) VSD (non restrictive)
   3) Aortic override of the ventricular septum (best demonstrated from the apical 4C)
   4) RVH
B. Associated defects:
   1) Anomalous origin of the LAD from the RCA (5%)
   2) Prominent conal branch from the RCA
   3) Right aortic arch (25%)
   4) Secundum ASD (15%- Pentalogy of Fallot)
   5) Persistent left SVC (5%)



The basic anatomical malformation in ToF is anterior deviation of the outlet septum. It's very rare to see adults with untreated ToF, as fewer than 10 per cent of patients with untreated ToF survive to the age of 20 years. As a result, almost all the adults seen with a history of ToF will have undergone surgical correction. ToF is usually treated with primary repair (closing the VSD and relieving the RVOT obstruction) before the age of 1 year. However, where necessary it is possible to perform a modified Blalock–Taussig shunt procedure (placing a graft between the subclavian artery and the pulmonary artery) as a palliative measure – this does not fully correct the ToF, but it does increase blood flow to the pulmonary circulation. The key problems to assess during echo follow-up include:

- Pulmonary regurgitation
- Severity of any residual RVOT obstruction
- Any shunting across a residual VSD.

Note that although echo plays an important role in follow-up after ToF repair, cardiac MRI can provide more detailed information about residual right heart abnormalities.

TRANSPOSITION OF THE GREAT ARTERIES (TGA or TGV)

In the normal heart with concordant connections, the morphologic left ventricle gives rise to the aorta and the PA serves as the outlet of the right ventricle. Discordant ventriculo-arterial connections occur when the great arteries arise from the opposite ventricle. Two forms of transposition exist.

- **D-TGA** (commonly referred to as **TGA**)- there is atrioventricular concordance and ventriculo-arterial discordance. Ventricular relationship is normal, with the morphologic RV located to the right of the morphologic LV. The pulmonary and systemic circulations are separate unless there is a communication at atrial, ventricular or arterial level. Early surgical corrections involved creating an atrial septostomy to allow mixing of systemic and pulmonary blood at the atrial level. As the two circulations would be unconnected without this, patients would otherwise die soon after birth.
- **L-TGA** (commonly referred to as **congenitally corrected** transposition)- there is both atrioventricular and ventriculo–arterial discordance (*circulation is normal but ventricles, along with their valves, are swapped)*. Atrioventricular discordance is present because of formation of an L-loop during embryogenesis so that the morphologic RV lies to the left and connects to the aorta, whereas the morphological LV lies to the right and connects to the PA. The RA connects to the morphological LV and LA connects to the morphological RV. The atrioventricular valves are always attached to the corresponding ventricle, thus the MV is always with the morphological LV

Commonly associated lesions are a VSD and PS

Surgical corrections of D-transposition of the great arteries (ventriculo-arterial discordance):

- **Rashkind**: atrial balloon septostomy to create mixing of systemic and pulmonary circulation
- **Blalock-Hanlon**: atrial surgical septostomy
- **Mustard or Senning (atrial switch)**: baffle material (Mustard) or native atrial tissue (Senning) used to direct pulmonary venous blood → RV → aorta; systemic venous blood → LV → pulmonary artery.
- **Jatene (arterial switch)**: great arteries are transected and re-anastomosed to the appropriate ventricle. Coronary arteries are removed with a button of surrounding tissue and re-implanted to the appropriate sinuses.
- **Rastelli**: for D-TGA with VSD and pulmonary outflow tract obstruction. VSD patch closure that directs LV blood across the VSD to the aorta. Pulmonary valve is over-sewn. Valved conduit from the RV to the PA to create RV outflow

EBSTEIN'S ANOMALY

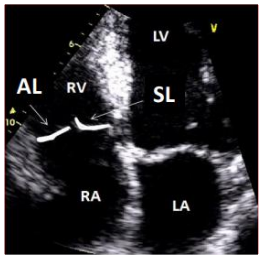
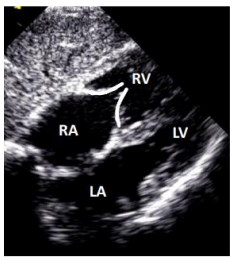
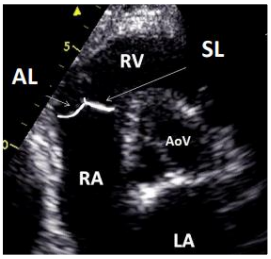
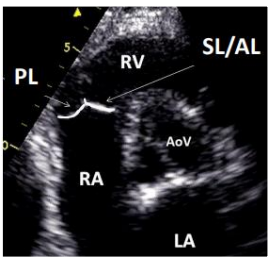
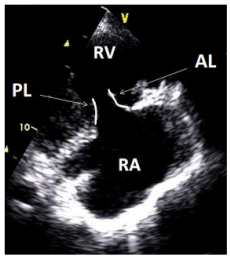
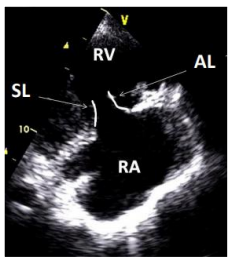
In Ebstein's anomaly the tricuspid valve (specifically the septal and posterior leaflets) is displaced towards the RV apex. As a result, part of the RV becomes 'atrialized' – although it becomes part of the RA, it still contracts with the RV, which impairs the haemodynamic function of the right heart and tends to exacerbate the **tricuspid regurgitation** that is usually present.

There are several associated conditions:

- ASD and VSD
- Pulmonary stenosis
- Accessory pathway (Wolff–Parkinson–White syndrome)

Eco assessment of Ebstein's anomaly

- **The tricuspid valve can best be seen in:**
  - Anterior leaflet: apical 4C, subcostal 4C, PSAX (AV level), PLAX RV inflow view
  - Septal leaflet: apical 4C, subcostal 4C, PSAX (AV level), ?PLAX RV inflow view
  - Posterior leaflet: PLAX RV inflow view, ?PSAX (AV level)

Apical 4C **anterior & septal** leaflets	**Subcostal** 4C **anterior & septal** leaflets	PSAX (**AV level**) **anterior & septal** leaflets ? posterior	PLAX **RV inflow** view **anterior** & **posterior** leaflets ? septal
		 	 

- Use colour Doppler to:
  - assess the severity of tricuspid regurgitation
  - look for shunts (see 'Associated features').
- Use CW Doppler to obtain a trace of regurgitant flow through the tricuspid valve.

HYPOPLASTIC LEFT HEART SYNDROME

Hypoplastic left heart syndrome is thought to result from premature closure of the foramen ovale in utero leading to an underdeveloped LV and its inflow and outflow components. There is a spectrum of severity that can include any or all of the complications mentioned.

DEFICIENT PULMONARY ARTERY OR RVOT

Conditions in which there is deficient pulmonary artery or RVOT:
1. Pulmonary atresia
2. TOF with hypoplastic pulmonary arteries

Most of these patients will have undergone surgical repair with or without a prior palliative procedure. Palliative procedure is a systemic-to-pulmonary shunt that is performed to supplement the deficiency of pulmonary blood flow and is taken down at the time of complete repair:
1. Classic Blalock-Taussing: directly connects the subclavian artery to the PA. Advantage: the subclavian artery small diameter prevents excessive blood flow and, hence, congestive cardiac failure. For the same reason, this shunt is unlikely to cause pulmonary vascular disease. Disadvantage: high risk of thrombosis of the shunt due to its smallness.
2. Modified Blalock-Taussing: connects the subclavian artery to the PA via Gore-Tex
3. Pott: connects the descending aorta to the LPA. Advantage: does not involve vessels of small calibre, such as the subclavian artery. Hence it is easier to perform and carries less risk of shunt thrombosis and occlusion. Disadvantage: high incidence of subsequent pulmonary hypertension.
4. ***W***aterston: connects the ***a***scending aorta to the ***R***PA. Waterston shunt is no longer used because it has similar disadvantages to the Potts shunt (excessive pulmonary blood flow and risk of pulmonary hypertension).
5. Glenn: connects the SVC to the PA. Advantage: does not increase the volume load on the ventricle. Disadvantage: not suitable for patients with elevated pulmonary vascular resistance, e.g. children younger than 3-6 months.

PLSVC

PLSVC results from failure of obliteration of the left common cardinal vein, and it typically drains the left jugular and subclavian veins into the RA via the coronary sinus and, in such case, it has no hemodynamic significance and is often an incidental finding, during echocardiography, as an apparent cystic cavity in the LA in the PLAX in otherwise normal hearts. PLSVC is the most common congenital anomaly involving the systemic veins.

In less than 10% of cases, PLSVC drains directly into the LA or into a pulmonary vein and, in such case, it results in R-L shunt and is almost always associated with other congenital anomalies.

PLSVC should be suspected when a dilated coronary sinus is identified in parasternal long axis view.

Performing a bilateral ‘bubble study’ from both antecubital veins can help establish the diagnosis of PLSVC. In PLSVC bubbles from left arm opacify the coronary sinus first, whereas bubbles from right arm show a normal sequence of opacification starting from the RA followed by the RV, with no contrast in the coronary sinus. In the rare cases of LA drainage, bubbles injected from the left arm opacify the LA.

In general, PLSVC may cause a dilated coronary sinus and possibly RA but not biatrial enlargement.

Differential diagnosis of coronary sinus dilatation include:
- Persistent LSVC
- Any cause of elevated RA pressure
- Partial APVD
- Coronary A-V fistula
- Unroofed coronary sinus with shunt flow between left atrium and coronary sinus (coronary sinus ASD)

CONGENITAL ASSOCIATIONS

- **The commonest cardiac defect in Down’s syndrome** is a primum ASD or AVSD.
- Noonan’s syndrome is associated with PS, ASD and cardiomyopathy.
- In Turner’s syndrome, the AV and root are mainly involved, with bicuspid valves/aortic coarctation being seen in approximately 10–15% of patients. Other abnormalities can include partial anomalous pulmonary venous drainage.

ROSS PROCEDURE

The Ross Procedure is a type of specialized **aortic valve surgery** where the patient's diseased aortic valve is replaced with his own pulmonary valve (autograft). The pulmonary valve is then replaced with a xenograft (porcine or bovine) or homograft (cadaveric) valve. Homografts are also used in valved conduits but are rarely used to replace a mitral or tricuspid valve.

FONTAN PROCEDURE

A univentricular (UV) heart is found when both atria drain mainly into one ventricle; while if either ventricle cannot sustain the systemic or the pulmonary circulation, a *functionally* UV heart emerges. A functionally univentricular heart (single ventricle) is doing nearly twice the expected amount of work because it has to pump blood for the body and lungs. The **Fontan procedure** is the final procedure in staged **palliation** for patients with **functionally univentricular heart** in the following situations:

- A complex congenital heart where a bi-ventricular repair is impossible or inadvisable.
- Lack of a heart valve (e.g. tricuspid or mitral atresia)
- An abnormality of the pumping ability of the heart (e.g. hypoplastic left heart syndrome or hypoplastic right heart syndrome)

The first stage, also called a *Bidirectional Glenn procedure* or *Hemi-Fontan*, involves redirecting blood from the SVC to the lungs. That is, (1) pulmonary arteries are disconnected from their existing blood supply (e.g. a shunt created during a Norwood procedure, a PDA, etc.) and (2) SVC is disconnected from the heart and redirected into the pulmonary arteries.

The second stage, also called *Fontan completion*, involves redirecting the blood from the IVC to the lungs. At this point, the blood from SVC & IVC flows through the lungs without being pumped (driven only by the pressure that builds up in the veins). This corrects the hypoxia and leaves the single ventricle responsible only for supplying blood to the body.

So, in general, all patients with single functional ventricle will undergo staged reconstructive procedures ultimately resulting in a Fontan circulation:

1) Norwood: connects ascending aorta to main PA (one of which may be hypoplastic or atretic) to produce “neoaorta” for the single ventricle. The main PA is transected from the heart and the pulmonary flow is maintained with BT shunt. Atrial septectomy is often performed to allow complete mixing at the atrial level.
2) Glenn: performed at 4-6 months. Connects SVC to PA, usually with takedown of a previously placed systemic to pulmonary shunt
3) Fontan: performed at 1-5 years. Connects IVC to PA. pulmonary blood flow is achieved passively, without the assistance of a ventricular pumping chamber