A review of the diagnostic accuracy of fetal cardiac anomalies

Abstract

Objectives: In order to assess the diagnostic accuracy of fetal cardiac anomalies in our Department we undertook a retrospective analysis and compared our results with those of the paediatric cardiologists in the same cases.

Methods: Sixty-five patients referred for second and third trimester fetal echocardiographic examinations were identified in our database from November 2005 to February 2014. Of these six scans were found to be normal by ultrasound and not referred on to the paediatric cardiologist. An additional six scans were diagnosed to have complex congenital heart disease (CHD) with or without extra cardiac abnormalities and/or aneuploidy. These six patients opted for termination of pregnancy and were also not seen by the paediatric cardiologist. The remaining 53 cases were referred to the paediatric cardiologist. Our results were analysed and then compared to the cardiologist's findings. There were an additional three cases scanned during the pregnancy in our department and diagnosed as normal but ultimately found in the neonatal period, to have CHD.

Results: The ultrasound findings of the 53 cases scanned in our department were analysed and compared to the findings in the cardiologist's reports. The earlier scans tended to describe the abnormal anatomy but showed a reluctance to name any pathology. As training and confidence levels increased the less complex pathologies were correctly identified by our department and confirmed by the cardiologist.

Conclusion: The skills, training and level of confidence required to diagnose fetal cardiac anomalies in our Department have improved over the eight years of the study period, particularly in regards to some of the less complex cardiac pathologies. However the more complex pathologies remain difficult to assess.

Keywords: fetal cardiac anomalies, paediatric cardiologist, skills and training.

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Introduction

Congenital heart disease (CHD) is the most common birth defect and affects 8–10 per 1000 live births.[1] However the term CHD is broad and includes a spectrum of pathology from the most severe and sometimes lethal anomalies to small ventricular septal defects (VSD) which may not be appreciated prenatally.

While ultrasound has contributed to the prenatal diagnosis of CHD the overall rate of detection remains relatively low at 57%.[2] The reasons for this are varied and include maternal obesity, fetal position, the quality of the equipment and the level of experience and training of those performing the scan.

The quality of equipment has improved over the years and this alone should make imaging easier. However it is a global phenomenon that maternal obesity is becoming increasingly more of a technical challenge. The problem of fetal position can be overcome by waiting for the fetus to change position or by rebooking the scan but this can create scheduling problems and stress for the patients as well as the providers. This leaves the level of experience and training of those performing the scan as the main issue that we can have some control over.

The purpose of this audit was to analyse the fetal echocardiography results found in our Department and compare them with those of the paediatric cardiologist during the study years November 2005 to February 2014. In this article we present eight pathologies which were encountered most frequently in our department during the study years although there are many others not mentioned here. We intended to track our improvement over the years and use the information to modify training if necessary.

Methods

A retrospective review identified 65 patients referred to our Department for second and third trimester fetal echocardiography from Nov 2005 to Feb 2014. Of these, six were found to be normal by ultrasound in our Department and were therefore not referred on to the paediatric

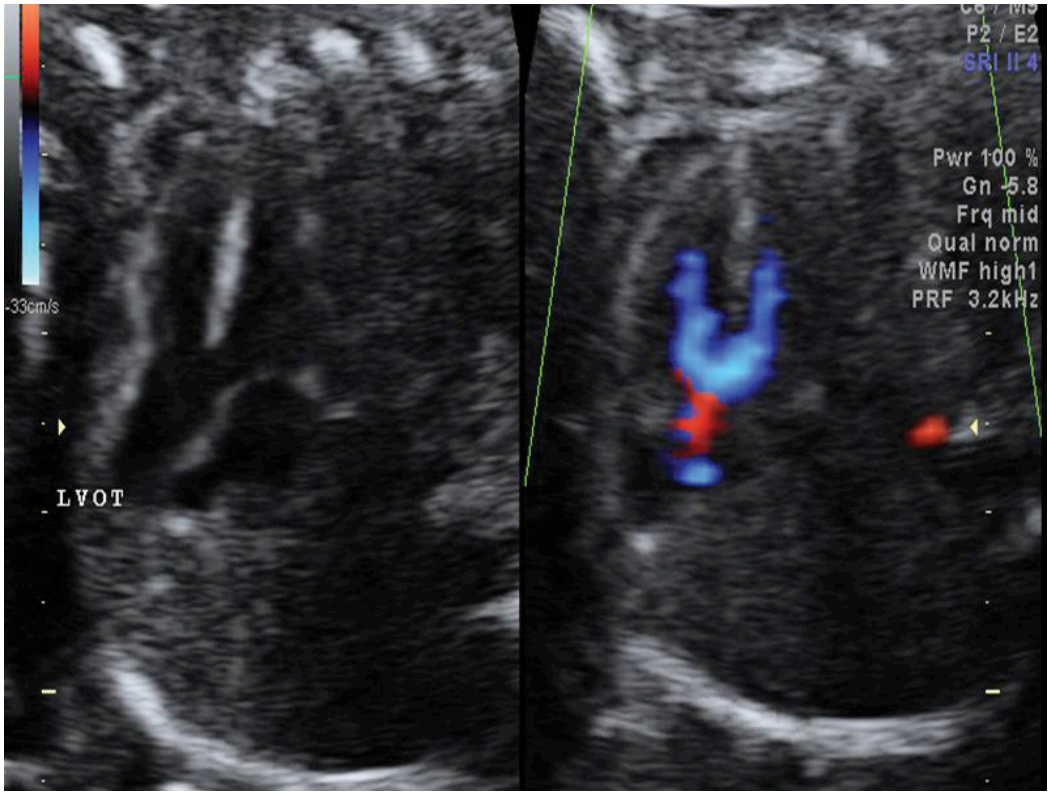

Figure 1: Tetralogy of Fallot.

cardiologist. An additional six fetuses were diagnosed to have major complex congenital heart disease with or without extra cardiac abnormalities and/or aneuploidy. These patients opted for termination of pregnancy and were also not seen by the paediatric cardiologist. The remaining 53 cases were referred to the paediatric cardiologist. Our results were analysed and then compared to the cardiologist's findings.

The data collected in the study were: gestational age at the time of fetal echocardiography scan, the findings at scan in our Department, the findings in the pediatric cardiologist's report, karyotype result and post natal findings if available.

There were an additional three cases scanned during the pregnancy in our Department which appeared structurally normal at the time of the scan but ultimately were found in the neonatal period, to have CHD. As these scans were felt to be normal these patients did not have fetal echocardiography examinations and were not included in the study.

The first of these cases was found to have a hypoplastic aortic outflow, suspected coarctation of the aorta, patent ductus arteriosus (PDA), 3–4 mm VSD, post natal right to left shunt and a large nodular mass on the mitral valve. The second case was diagnosed with Tetralogy of Fallot (TOF). The third case was a 41-year-old obese patient who had a high risk combined nuchal translucency serum screening test. She declined all invasive testing and had a normal fetal anatomy scan (FAS) in the second trimester. In the third trimester she was diagnosed with polyhydramnios although no reason for this could be identified on ultrasound.

Postnatally this baby was diagnosed with double outlet right ventricle (DORV), oesophageal atresia, trachea-oesophageal fistula and had a normal karyotype.

Discussion

Our greatest improvement was shown in the ability to recognise and identify Tetralogy of Fallot (TOF), hypoplastic left heart syndrome (HLHS) and coarctation of the aorta.

Tetralogy of Fallot (Figure 1) has four main features, three of which can be seen prenatally.

These are a ventricular septal defect (VSD), an aorta which overrides the VSD and right ventricular outflow tract (RVOT) narrowing which can range from mild pulmonary stenosis to atresia. The fourth feature is hypertrophy of the right ventricular wall which develops over time and is often not appreciated until the neonatal scan.

It is common practice to image the outflow tracts at an 18–20 week fetal anatomy scan (FAS).

When the RVOT is difficult to image TOF should be considered as a possible diagnosis. With this information in mind the sonographers in our Department have become increasingly aware to examine the interventricular septum (IVS) for a VSD and/or an overriding aorta. As the experience and confidence level increased, the staff in our Department progressed from describing a "narrow RVOT" to recognizing the features of and correctly diagnosing TOF.

Hypoplastic left heart syndrome (Figure 2) is actually hypoplasia of most if not all of the left heart, involving the mitral valve, left ventricle, left ventricular outflow tract (LVOT) and aortic valve. It is caused by compromised blood flow to one or more of the parts of the left heart and results in hypoplasia of the whole.

As the four chamber heart view (4CH) is the original gold standard view in fetal echocardiography an inability to image this should alert the scanner to a potential problem. In earlier scans a

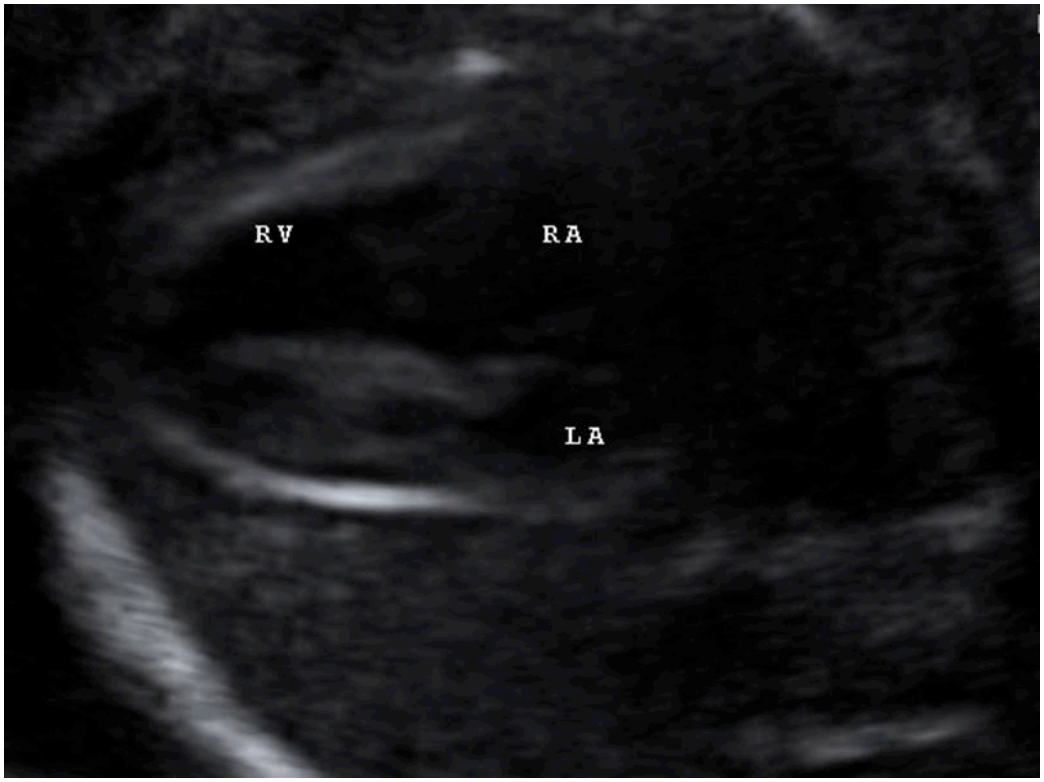

Figure 2: Hypoplastic Left Heart.

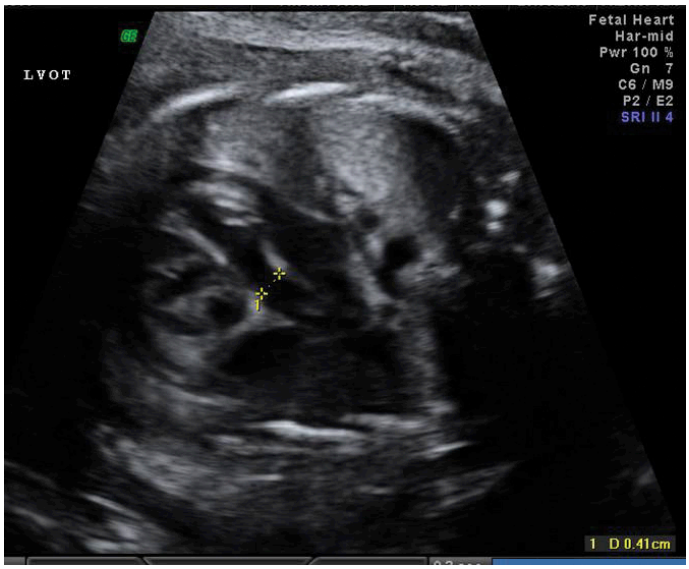

Figure 3a: Coarctation of the Aorta, LVOT.

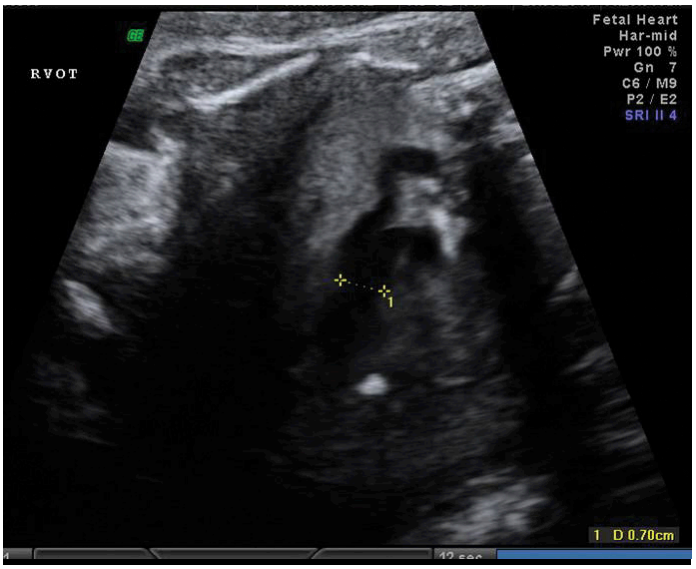

Figure 3b: Coarctation of the Aorta, RVOT.

"small left ventricle" may have been observed and described. Once again as the experience and confidence level increased the staff in our Department progressed from this description. The mitral valve, LVOT and aortic valve are now examined for stenosis or atresia and the diagnosis of HLHS is confidently made.

Coarctation of the aorta (Figure 3a and 3b) is a narrowing of the aorta either pre or post ductal. It can be a very difficult diagnosis to make prenatally. The reason for this is that it evolves over time and is rarely appreciated at the 18–20 week anatomy scan. In fact the actual coarctation or narrowing is rarely seen even by the paediatric cardiologist. However in the third trimester the left heart structures may appear small compared to the right without any specific abnormality identified. With this in mind our Department has updated our policy and third trimester scans now include a limited review of the heart. If a discrepancy in the sizes of the left and right heart is noted it may by diagnostic of an evolving coarctation of the aorta.

During the study years there was also an increased recognition of normal variants such as isolated right sided aortic arch and persistent left superior vena cava (SVC).

Right sided aortic arch (Figure 4) is diagnosed in the RVOT view. In the normal situation the main pulmonary artery and aorta are seen to come together in a 'V' shaped confluence anterior to the trachea and superior to the three vessel view. In

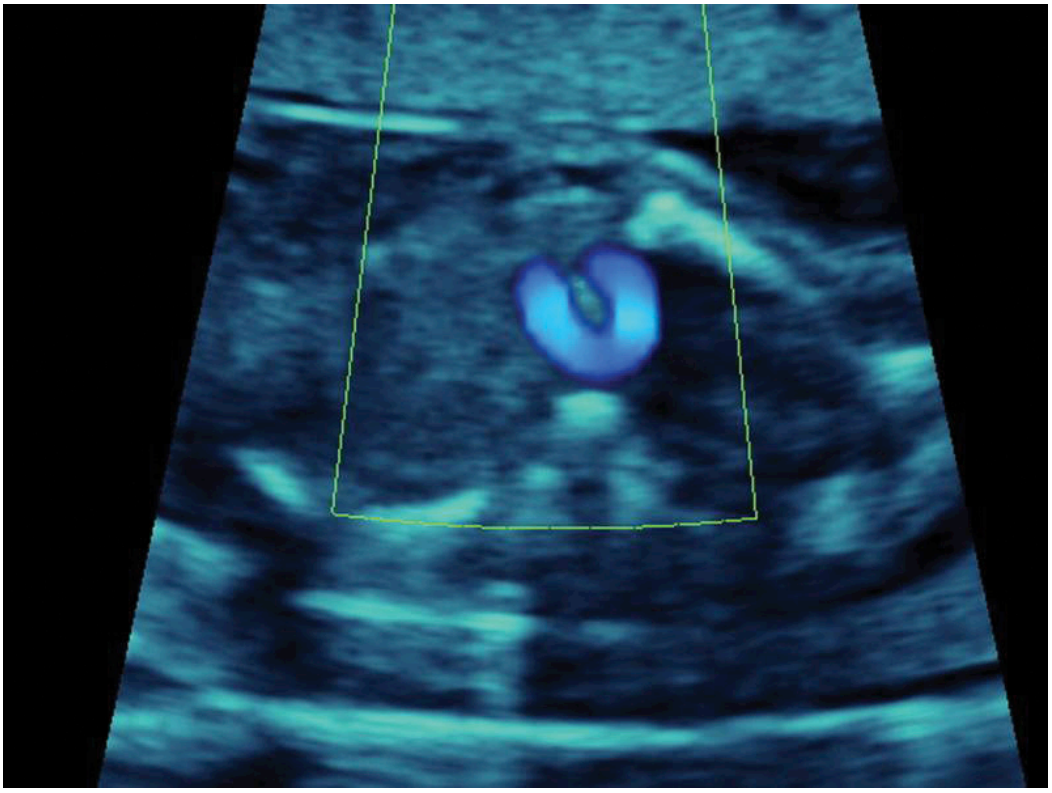

Figure 4: Right Sided Aortic Arch.

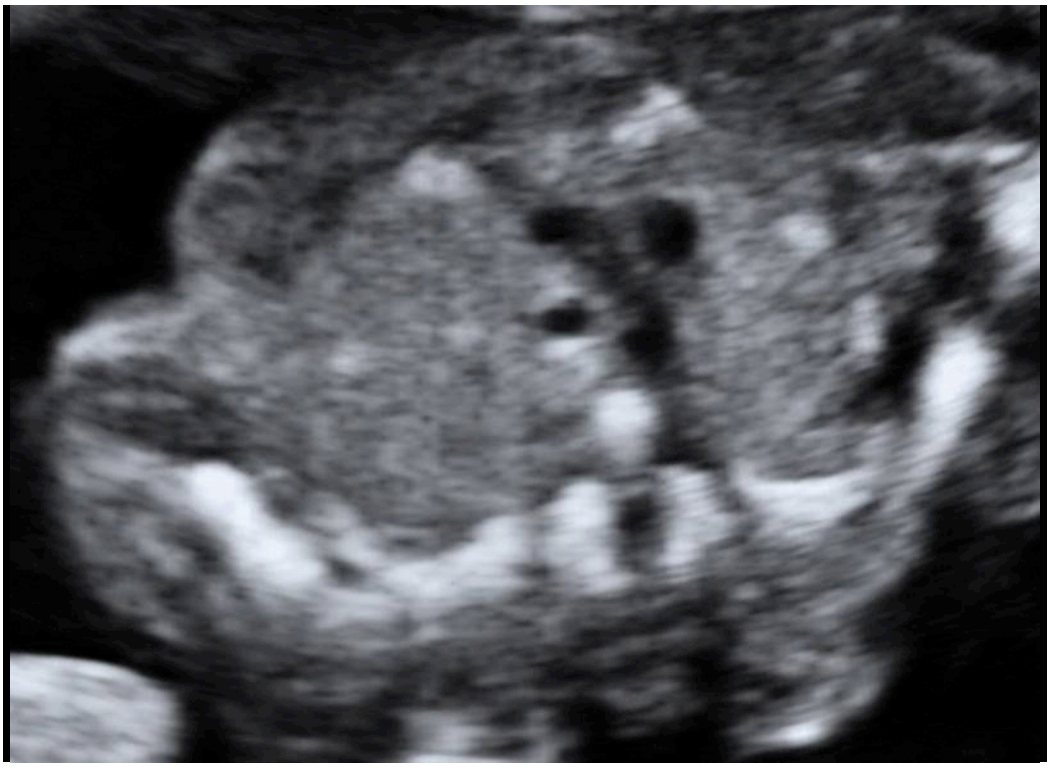

Figure 5: Persistent Left SVC.

right sided aortic arch the confluence is 'U' shaped posterior to the trachea. With experience this is now an easy diagnosis and as an isolated finding this is a normal variant.[3]

Persistent left SVC (Figure 5) is one of the most common systemic venous abnormalities.[4] It is also diagnosed in the RVOT view when a vessel is seen to the left of the main pulmonary artery as well as or instead of the normal right SVC seen to the right of the aorta. It is also often associated with a dilated coronary sinus in the posterior left atrioventricular groove.

Recognition of this vessel in the usual three vessel/right outflow view has allowed us to correctly identify this. As an isolated finding this is also a normal variant.[3]

Some of the more complex pathologies remain a challenge to recognise and diagnose.

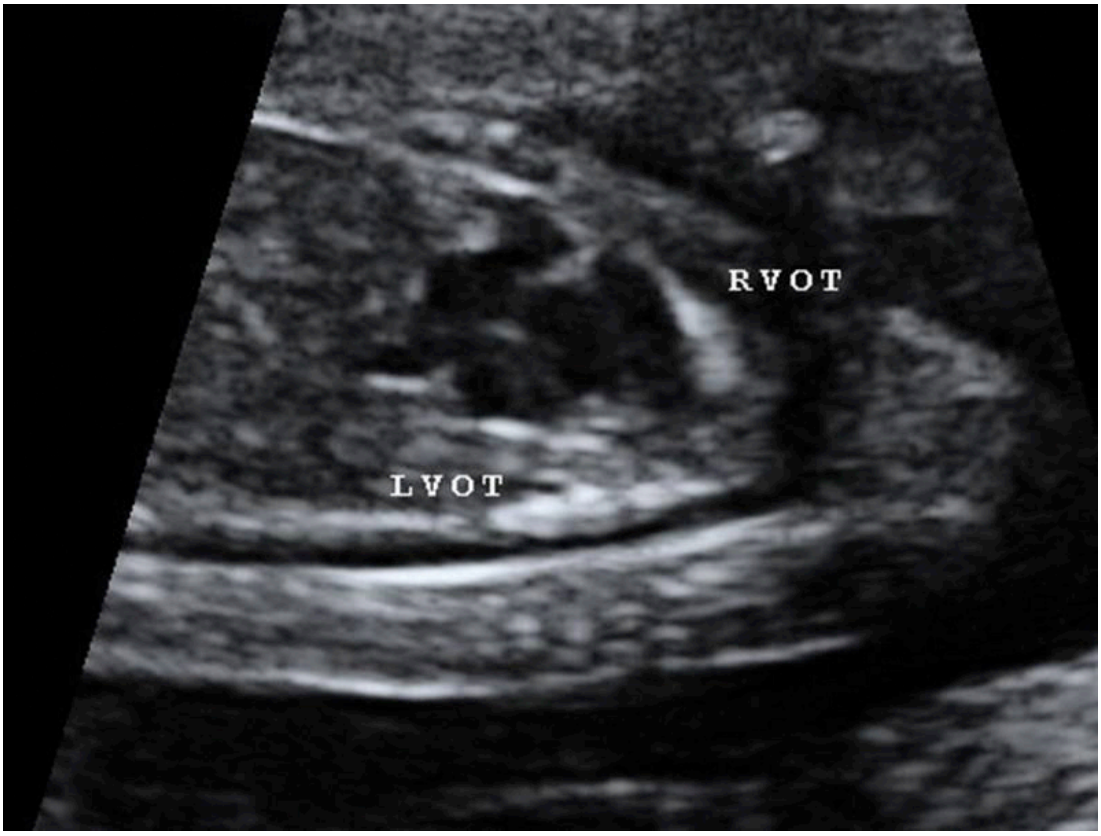

Figure 6: Double Outlet Right Ventricle.

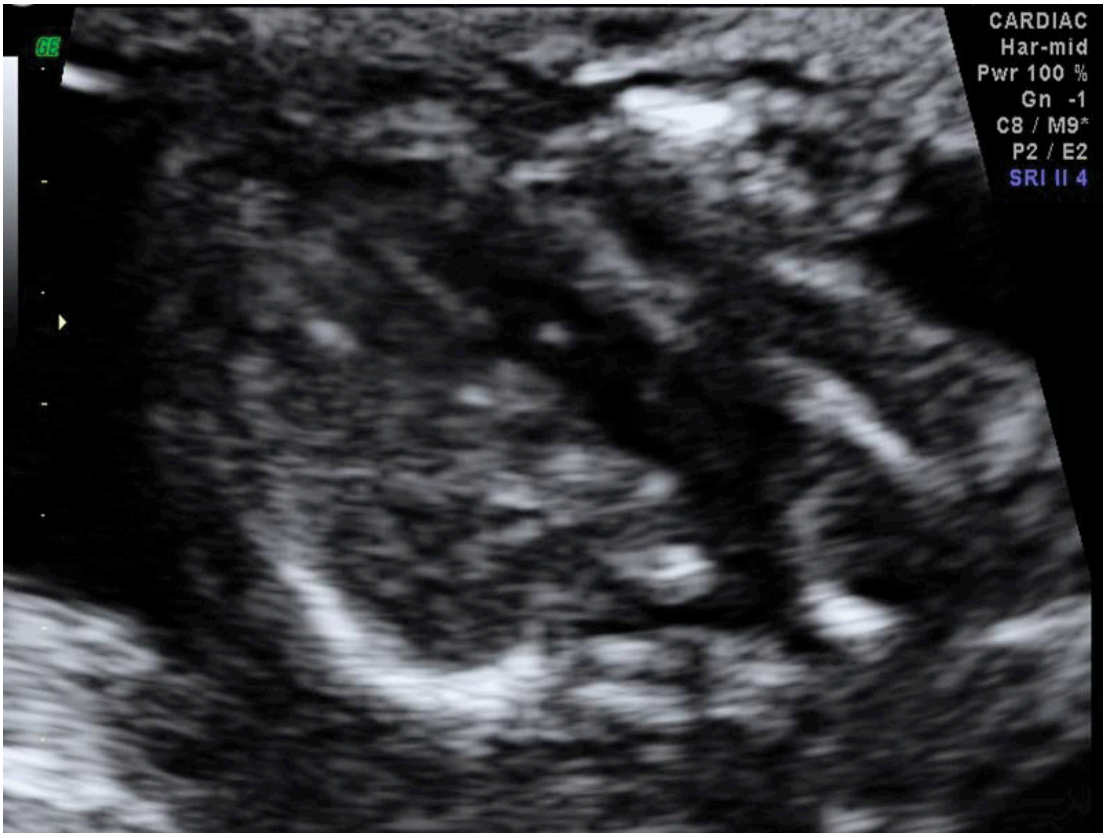

Figure 7: Persistent Truncus Arteriosus.

Examples of these include double outlet right ventricle (DORV) and persistent truncus arteriosus (PTA). These two pathologies each have several variations or sub types which contribute to their complexity.

In DORV (Figure 6) both the aorta and pulmonary artery arise completely or almost completely from the right ventricle and there is always an associated VSD. There are four sub types which are based on the location of the VSD.[4]

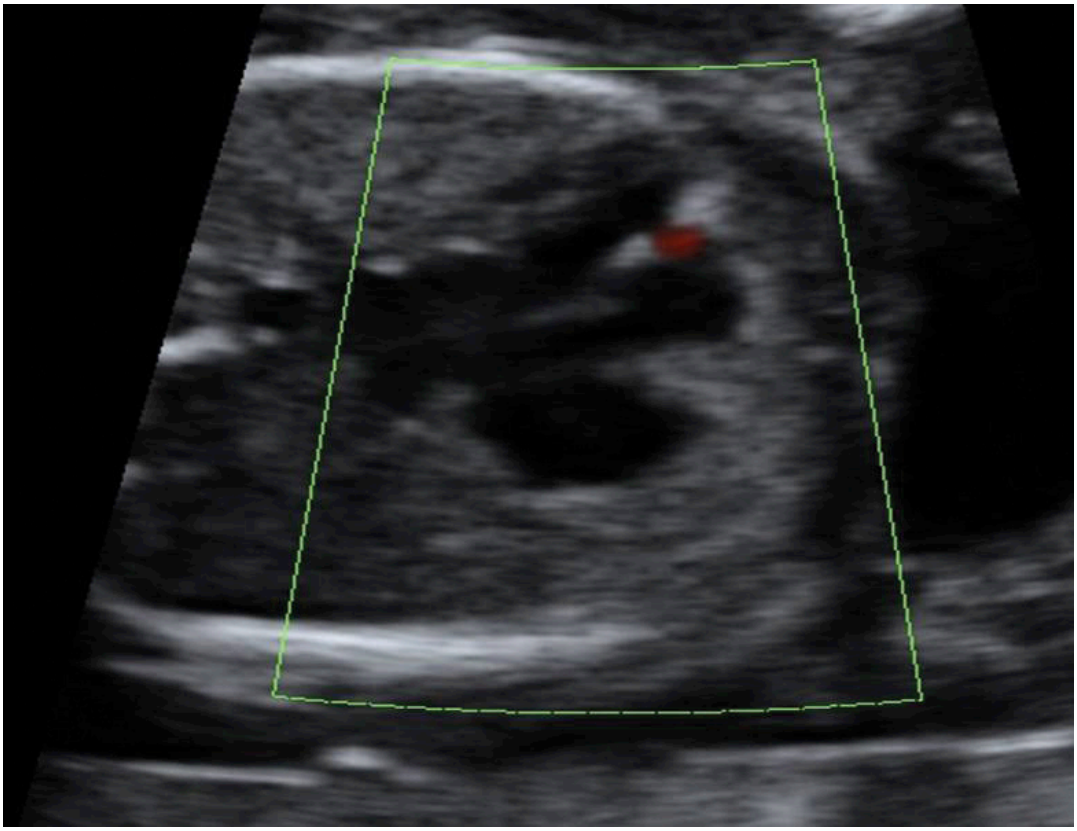

Figure 8: Ventricular septal defects.

- The VSD is sub aortic, resembling TOF
- The VSD is sub pulmonic with the pulmonary artery posterior and left of the aorta, resembling transposition of the great arteries (TGA)
- The VSD is located below both great arteries
- The VSD is remote from both great arteries.

Aortic or pulmonary stenosis or atresia may occur with any of these types. DORV is also commonly associated with other cardiac or extra cardiac abnormalities and/or aneuploidy.

Pesistent truncus arteriosus (Figure 7) is a single large truncal vessel which arises from centre of the heart and supplies the systemic, pulmonary and coronary circulations.[4] The valve cusps of this single vessel are multiple and often thickened and dysplastic and the valve may be stenotic or reguritant.[3] PTA is also associated with a VSD which the truncal vessel usually overrides. The aorta and pulmonary artery both arise from this vessel but the sub types are based on the connection of the pulmonary arteries to the common trunk.[5]

- The main pulmonary artery arises from the left lateral aspect of the common trunk and then divides into the right and left pulmonary arteries
- Both pulmonary arteries arise separately from the left posterolateral section of the common trunk, close to each other
- Both pulmonary arteries arise separately from the lateral sections of the common trunk, farther from each other.

PTA is also commonly associated with other cardiac or extra cardiac abnormalities and/or aneuploidy.

A final cardiac anomaly that remains difficult to diagnose is the isolated VSD (Figure 8).

While neither complex nor rare it is difficult to image due to its small size in an otherwise normal heart. The defect can be located in any part of the septum although peri membranous and muscular are the most common. Imaging of the four chamber heart from different angles using properly set colour flow Doppler is necessary to recognize a VSD. Due to these difficulties the rates of false positive and false negative remain high.

Results

The ultrasound findings of the 53 cases scanned in our Department were analysed and compared to the findings in the cardiologist's reports. During the study years the overall diagnostic accuracy of our Department increased from 34% to 60%. (Table 1)

The scans performed in the early years of the study tended to describe the abnormal anatomy but showed a reluctance to name any particular pathology. As training and confidence levels increased the less complex pathologies were correctly identified by our Department and confirmed by the cardiologist.

The more complex pathologies remain difficult to assess. An explanation for this is that they are less commonly seen making recognition more difficult. There are also many variations and sub types and there is often overlap of findings from more than one recognized pathology making it difficult to place a definite diagnosis on some findings. This may leave only a description of the anatomy for the report in even the most experienced hands. However it is this description and differential diagnoses that are now offered in many reports which are then sent to the cardiologist for review.

Through the years our department has increased the number of views imaged in our routine FAS from the 4CH and outflow tracts to include the aortic and ductal arches, IVC/SVC inflow, the bifurcation of the main pulmonary artery and the confluence of the main pulmonary artery with the aorta. We have become increasingly aware of the effect of these views on the developing fetal heart. Although we have not had any formal training program as in some studies[6] we have learned from each other and now train our junior staff and students accordingly.

The eight pathologies most commonly seen during the study years have been presented here.

Due to the relatively low patient numbers some pathologies were only seen once, e.g. AVSD or not at all such as TGA or bicuspid aortic valve. For this reason these have not been included in our audit.

Table 1: Results of appraisal.

Pathology	Number of Cases	% Correct in 1st Half of Study	% Correct in 2nd Half of Study
TOF	8	50	75
HLHS	6	30	90
Coarctation of the Aorta	8	0	75
RAA	3	-	100
PLSVC	2	0	100
DORV	4	0	0
PTA	2	0	0
VSD	8	30	60
Overall	53	34	60

Conclusion

In spite of the low patient numbers involved the skills, training and level of confidence in our Department have improved over the 8 years of the study period, particularly in regards to some of the more commonly seen and less complex cardiac pathologies. However there is still a tendency to describe the more complex pathologies.

While anatomical descriptions and differential diagnoses are offered in our reports further improvement is required. We believe that audits such as these offer an important learning tool and encourage other ultrasound departments to undertake a similar exercise.

Acknowledgement

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