Royal United Hospitals Bath
NHS Foundation Trust

Four Unusual Case Studies of the Fetal Thorax

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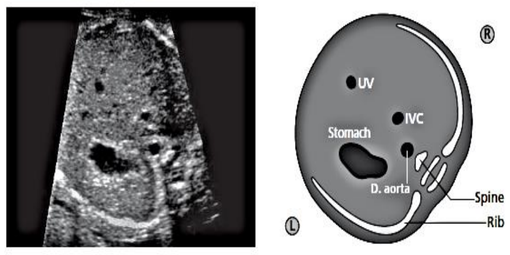
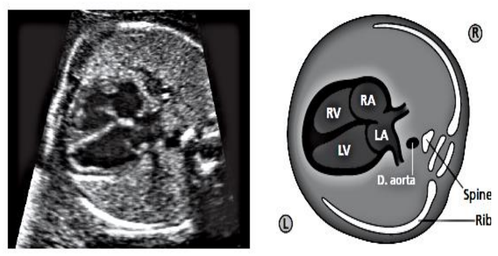
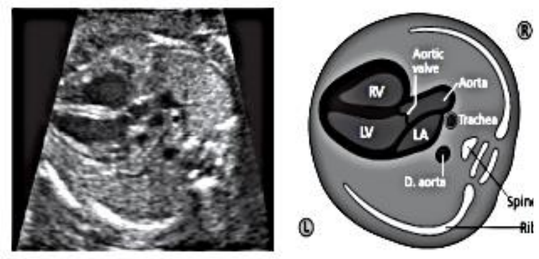
Visceral situs/laterality



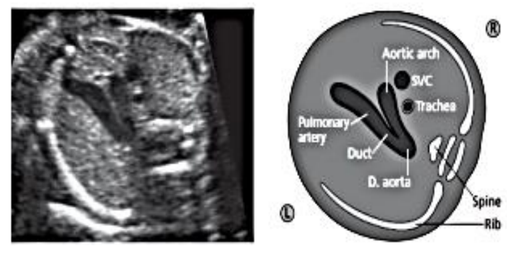
4 chamber view (4CH)



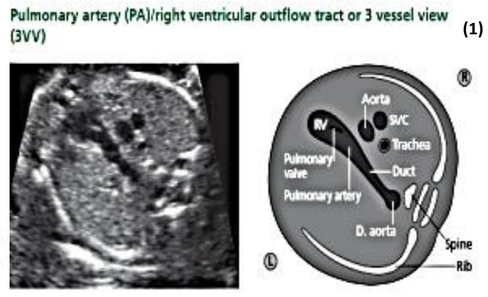
Aorta (AO)/left ventricular outflow tract



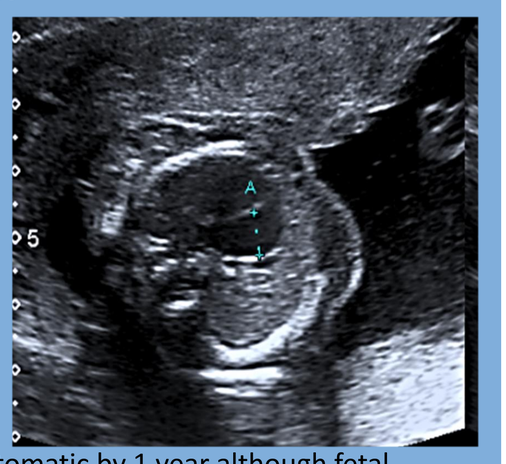
3 vessel and trachea view (3VT)



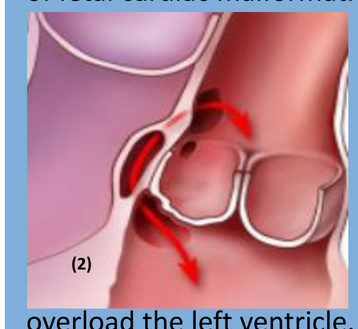
Pulmonary artery (PA)/right ventricular outflow tract or 3 vessel view (3VV) (1)



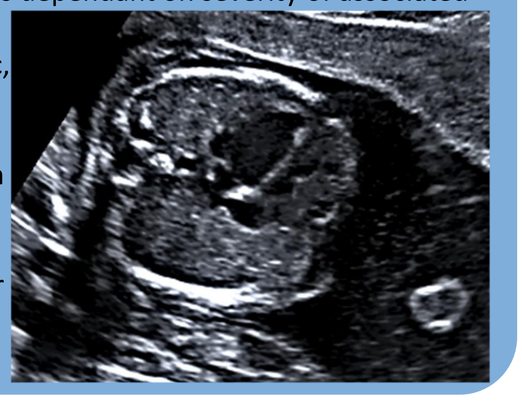
Aortic Tunnel



Ultrasound Appearances: Normal Situs. Enlarged Left ventricle. Disproportionate dilatation of the aortic arch.
Further Investigation & Diagnosis: Further ultrasound assessments in tertiary unit. Normal karyotyping. Diagnosed with aortic ventricular tunnel (AVT).
Literature: AVT is a congenital, extra-cardiac channel which connects the ascending aorta above the mitral valve to the cavity of the left ventricle. Incidence is unknown but certainly rare accounting for approx. 0.1% of fetal cardiac malformations.(3) Many cases are diagnosed postnatally when patients often become symptomatic by 1 year although fetal echocardiography is considered reliable after 18 weeks gestation(2).


(2)

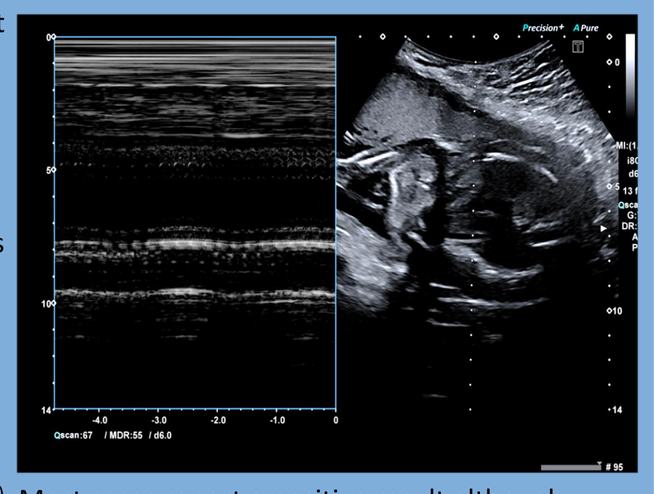
The scale of this condition is dependant on severity of associated anomalies and whilst mild cases may be asymptomatic, if left unmanaged this condition can fatally overload the left ventricle. Surgical management is often successful and achieved through catheter closure of the 'tunnel' under cardiopulmonary bypass(4).



Outcome: Unfortunately due to the complexity and poor prognosis of this particular case, this pregnancy was sadly terminated at 23^{+6}.

Supraventricular Fibrillation

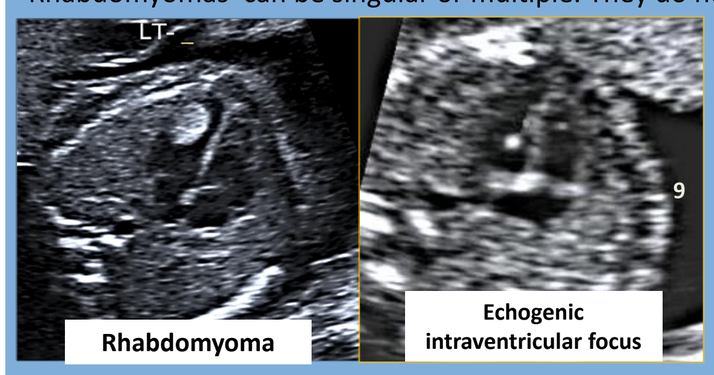
Ultrasound Appearances: Fetal cardiac fibrillation at around 254Bpm . Hydrops
Further investigation & Diagnosis: Fetal echocardiogram. Diagnosed with Supraventricular tachycardia (SVT). Postnatal Electrocardiogram.



Literature: Antenatal SVT is a rare finding but also the most common form of fetal tachycardia(5). The rapid contraction of the cardiac muscles during SVT prevents blood filling the ventricles to be efficiently pumped around the body. Decreased cardiac output will eventually lead to cardiac failure, hydrops and eventually death(6,7 &8). Whilst there are no clear indicators for fetuses who will develop postnatal STV, prognosis is significantly improved with antenatal treatment. Fetal SVT can be controlled with trans -placental anti-arrhythmic medication without affecting maternal cardiac output(6&8). Most cases report a positive result although there is a higher incidence of post natal SVT for those detected in later gestations where preterm delivery also poses a potential risk(7).
Outcome: Condition stabilised antenatally with anti-arrhythmic medication. Live delivery of a baby girl at term. Single SVT episode 2 days post delivery. Condition remaining stable with medicinal support. No subsequent events. She is showing otherwise normal development and her Consultants hope she has simply 'grown out' of her condition. Plan to reduce dosage with a view to cease medication in the near future. She is currently 15 months old.

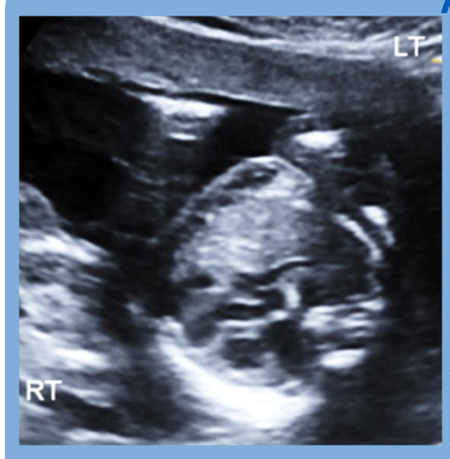
Rhabdomyoma

Ultrasound Appearances: An 8 x 5 x 10mm echogenic mass noted in the left ventricle.
Differential Diagnoses: depending on the location of the anomaly, appearances may mimic an echogenic intraventricular focus (these are typically <3mm)(9)
Further investigation & Diagnosis: Postnatal MRI & genetic analysis.
Literature: A rhabdomyoma is the most common primary cardiac tumour found in infants and children(10) and are strongly associated to Tubers Sclerosis (TS) with a prevalence ranging from 50-90%(13,17). TS is an autosomal multisystem disorder which can also target the central nervous system, skin, retina and kidneys; early detection is therefore considered critical(13).
Rhabdomyomas' can be singular or multiple. They do not grow in size, but actually remain consistent or resolve entirely during infancy(12, 13). Occasionally, the space occupying lesion(s) has/have an impact on cardiac output which typically manifests as an arrhythmia and often responds well to postnatal medication(12). Due to their low risk their presence alone is not a concern however, antenatal detection of a rhabdomyoma not only anticipates the likely need for neonatal cardiovascular support; but also the potential diagnosis of TS sanctions appropriate parental prenatal counselling coupled with access to postnatal multidisciplinary care(11,12 &13).

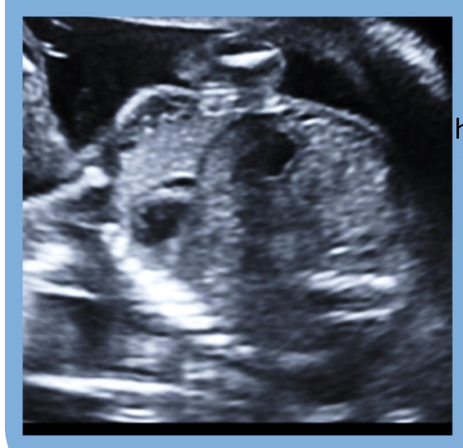


Outcome: Live delivery of a baby boy at term. Medicated with propranolol to maintain healthy cardiac output. Postnatal MRI confirmed no intracranial masses. Cardiac rhabdomyoma remains unchanged in size. Normal infant kidneys. No skin lesions. No seizers. He is currently 13 months old and showing normal milestone development. Still awaiting final results of genetic analysis however the presence of TS is considered unlikely at this stage.

Absent Right Lung



Ultrasound Appearances: Extreme dextrocardia with the apex of heart pointing towards right scapula. Otherwise normal fetal echocardiography
Differential diagnoses: Dextrocardia or right sided diaphragmatic hernia (apparently intact diaphragm).
Further investigation & Diagnosis: Fetal echocardiogram & antenatal MRI in tertiary unit.
Literature: Lung agenesis is a rare anomaly accounting for approx. 1 in 50,000 births(17). The exact aetiology is uncertain, however it is believed to occur during weeks 4-5 of embryology(14). There is an approximate mortality rate of 50% within the first 5 years of life, however there are some reported cases where patients' have not presented until middle aged(14) . There are several factors which can influence prognosis including: classification (Agenesis, Aplasia or Hypoplasia), cardiac positioning, the presence of genetic anomalies (if any) and access to healthcare(15). The malformed pulmonary trunk can present potentially fatal associated anomalies (as exampled in this case study) however antenatal diagnosis significantly improves prognosis. This not only gives parents access to appropriate counselling to facilitate informed decisions but also access to tailored appropriate multidisciplinary postnatal care(16).

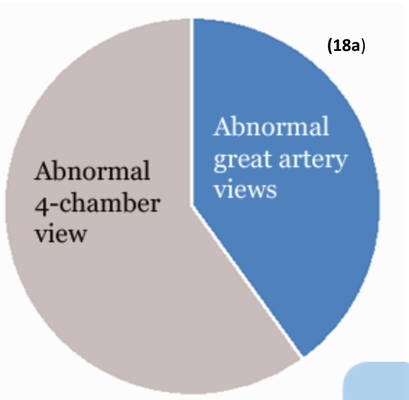


Outcome: Preterm live delivery of a baby girl in specialist unit at 38/40. Initial assessment in NICU showed she had oesophageal atresia, a trachea-oeophageal fistula and a gastric perforation which were repaired on day 1. She was discharged home at 3 ½ months. She is currently 3 years old and showing otherwise normal healthy development.

Optimisation and Pitfalls

Ultrasound optimisation in fetal echocardiography is key to effective assessment. It is important Sonographers methodically and systematically assess the fetal heart, evaluating the size, position and structure of each segment(16); of the 50% of cardiac anomalies detected during the anomaly scan, 60% are identified on the four chamber view alone(18a). Whilst the ability to identify anomalies is important, knowledge of normal variants and differential diagnoses will also improve accuracy in detecting anomalies. In order to comply with HCPC registration, sonographers are required to continue professional development (CPD) and keep up-to-date with current professional practice(17). CPD can be achieved by reflecting on an event which may have simply occurred during day-to-day practice but also accessing reputable online resources or attending a training days can enhance knowledge(1&16).
Not only is operator competency an important factor in thorough assessment of the fetal heart but also access to equipment which meets FASP standards(1&16). Whilst selecting a 'fetal heart' pre-set will generally optimise the image, skilful and effective adjustments of pre and post processing parameters will further improve image quality(16&18). The angle of isonation can be revised through subtle sliding, rocking and pressing movements of the transducer(16) and, assuming that room lighting is appropriate the operator may also consider other external limitations such as fetal lie and patient habitus(18).

(18a)



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1.Fetal Anomaly Screening Programme (2010) FASP: 18 $^{+0}$ to 20^{+6} Week Fetal Anomaly Scan National Standards and Guidance of England. Copyright NHS Fetal Anomaly Screening programme 2010. Pages 67-70 **2.** McKay, R (2007) Aorto-ventricular tunnel. *Orphanet Journal of Rare Diseases* 2:41. **3.** Ma, X., Li, J., Zhang, Q., Kong, X., Yuan, G., Wang, Z., Zou, C. (2018) Aorto-left ventricular tunnel with anomalous origin of right coronary artery and bicuspid aortic valve: a case report. *Journal of cardiothorasic surgery*. 13:81. **4.** Kathare, P., Subramanyam, R., Dash, T., Muthuswamy, K., Raghu, K., Konety, N. (2015) Diagnosis and Management of aorto-left ventricular tunnel. *Annals of Paediatric Cardiology*. May-August 8 (2) 103-107. **5.** Nicholaides, K., Huggon, I., Xidre, V. (2019) Fetal Cardiac Scanning [online] *Online training resource accessed through FMF portal* **6.** Hinkle, K., Peyvandi, S., Stiver, C., Killen, A., Weng, H., Etheridge, A., Puchalski, M. (2017) Postnatal Outcomes of Fetal Supraventicular Tachycardia: A Multi Centre Study. *Paediatric Cardiology* 38:1317-1323. **7.** Kang, S., Howe, D., Coleman, M., Granapragasam, J. (2015) Foetal supraventricula tachycardia with hydrops fetalis: a role for direct intraperitoneal aminodrone. *Cardiology in the young* 25, 447-453. **8.**Schmolling, J., Renke, K., Richter, O., Pfeiffer, K., Schlebusch, H., Höller, T. (2000) Digoxin, Flecainide and Aminodarone Transfer across the placenta and the effects of an elevated umbilical venous pressure on the transfer rate. *Therapeutic Drug Monitoring* October 2000 Volume 22 Issue 5 p 582-588. **9.** Goe, A., Weerakkody, Y (2019) Echogenic intracardiac focus. [online] Radiopaedia. Avaliable from https://radiopaedia.org/articles/echogenic-intracardiac-focus?lang=gb [accessed 23/9/19]. **10** Ekmekci, E., Ozkan, B., Yildiz, M., kocakaya, B (2018) Prenatal diagnosis offetal cardiac thabdomyoma associated with tuberscleriosis: a case report. *Case reports in Womens health.* volume 19 **11.** Ide, T., Miyoshi, T., Katsuragi, S., Neki, R., Kurosaki, K-I., Shiraishi, I., Yoshimatsu, J., Ikeda, T (2018) Prediction of postnatal arrhythmia in detuses with cardiac rhabdomyoma. *The Journal of Maternal-Fetal & Neonatal* Medicine. 32:15, 2463-2468. **12**. Bejiqi, R., Retkoceri, R., Bejiqi, H. (2017) Prenatally diagnosis and outcome of fetuses with cardiac rhabdomyoma- single centre experience. *Open Access Maced J Med Sci.* 15; 5(2):193-196. **13.** Mitel, A., Burloiu, C (2017) Cardiac rhabdomyomas- The importance of prenatal diagnosis for tubero sclerosis. *European Journal of paediatric neurology*. Volume 21. **14.**Kisku, K., Panigrahi, M., Sudhakar, R., Nargarajan, A., Ravikumar, R., Daniel, R. (2008) Agenesis of lung – A report of two cases. *Lung India* Jan-Mar 25 28-30. **15.** Zhand, Y., Fan, M., Ren, W., Xie, L., Ding, C., Sun, W., Wang, Y., Guo, Y. Cai, A (2013) Prenatal diagnosis of fetal unilateral lung agenesis complicated with cardiac malposition. *BMC Pregnancy and Childbirth* March 2013 .13 (79) **16.** Public Health England (2017) Fetal Cardiac e-Learning. [Online] Online *training resource accessed through e-Learning For Health.* **17.**HCPC (2019) Contunuing Professional Development. [online] avaliable from https://www.hcpc-uk.org/cpd/ accessed 3/10/19. **18 & 18a** Nicolaides, K., Huggon, I., Zidere, V., (2019) Fetal cardiac scanning e-learning programme. Iaccessed through FMF online training portal.