

A Matter of Heart: a case of sternal cleft and ectopia cordis

Dr Nikhil Krishnan^{1,2}, Dr Maria Elisabeth Smet^{1,2,3}, Dr Alex Gooi⁴, A/Prof Thushari Indika Alahakoon^{1,2,5,6}



¹Department of Women's and Newborn Health, Westmead Hospital, ²Westmead Institute for Maternal Fetal Medicine (WIMFM), Westmead Hospital, ³Sydney Ultrasound for Women (SUFW), Sydney, Australia, ⁴Heart Centre for Children, Children's Hospital at Westmead, ⁵Centre for Women's Ultrasound, Westmead, NSW, Sydney, Australia, ⁶University of Sydney, Sydney, Australia

Background

Ectopia cordis (EC)

- A rare congenital anomaly with the heart positioned either partially or completely outside the thoracic cavity
- Diagnostic modalities: ultrasound with Doppler, MRI
- Broad spectrum of ectopia: cervical, thoracic, abdominal
- **Cervical and thoracic are fatal, abdominal has better prognosis due to less instance of intracardiac defects (Pius et al., 2017)**
- Management: surgical
 - Mean survival: 4 – 5 days

Sternal Cleft (SC)

- Malformations caused by failure of fusion of the mesenchymal cells at the six weeks gestation
- **2 forms: complete and partial, further characterised into superior or inferior**
 - **Inferior partial sternal cleft associated with ectopia cordis**
- Diagnosis: antenatal ultrasound at 18-26 weeks gestation (Kamal, 2018)
- The presence of other associated syndromic defects must be assessed prior to surgical repair
- Management: surgical repair with primary approximation during neonatal period (due to more compliant thoracic wall)

The Case

33 years old, G2P0

- G1: missed miscarriage, 8/40, surgical management, G2 (index pregnancy): IVF (ovulation induction)

Past Medical History: 1. Endometriosis, 2. LLETZ (2012, 2015) for CIN 3

Antenatal History: O positive blood group, nil antibodies. NIPT low risk

- Morphology Ultrasound at 20+6: No structural anomalies
 - **Cardiac: Increased left ventricular trabeculation. Heart apex lies in the midline. Aorta arises from the anterior thorax.**
- Fetal Echocardiogram at 25+6:
 - Mesocardia with pectus excavatum deformity and suspected defect in lower sternum
 - Apex pulsating very close to the cleft, **Suspected mild ectopia cordis**
- Amniocentesis: 46 XY, no abnormalities detected
- Fetus delivered vaginally at 39 weeks gestation, APGAR scores 9 and 9
- No resuscitation required in acute post-partum period

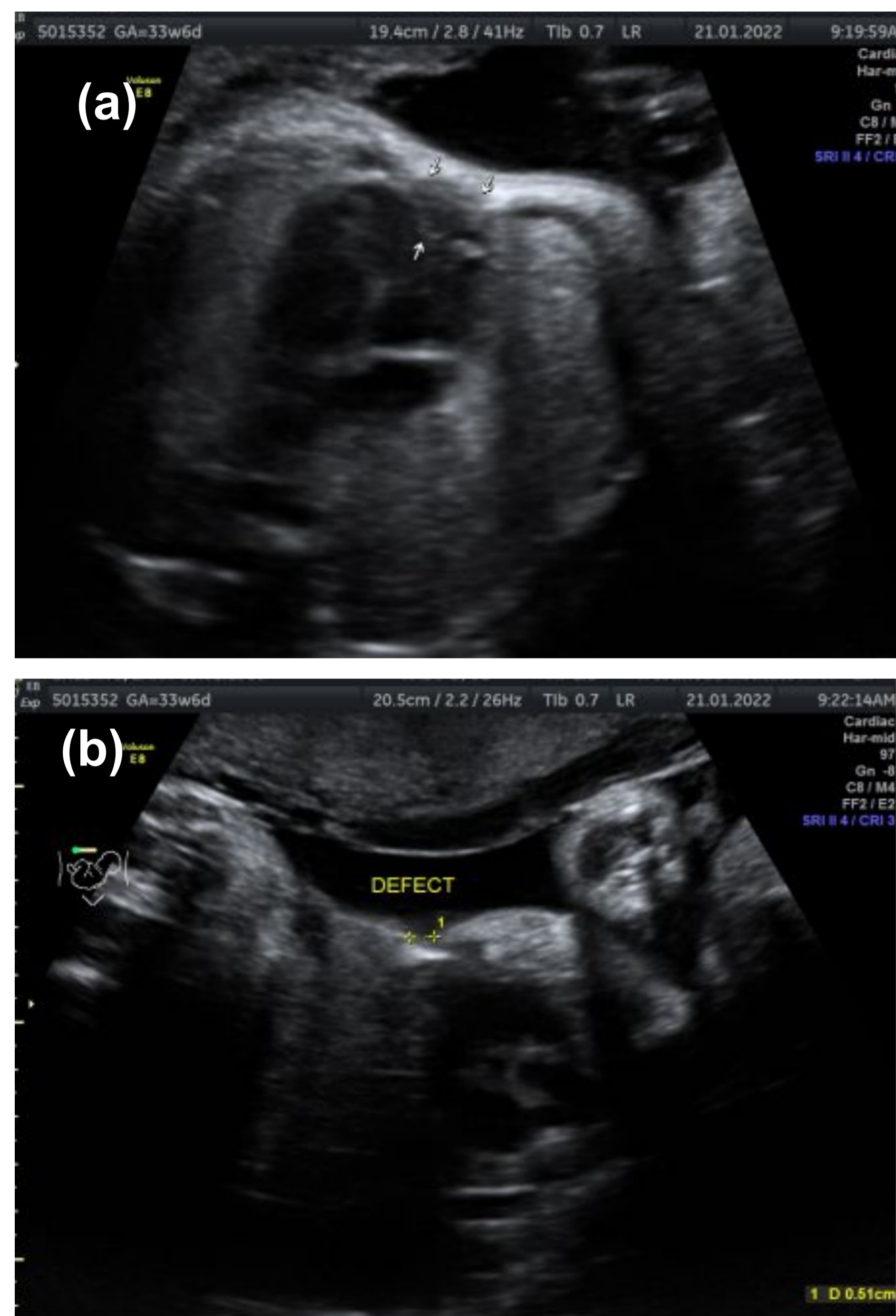


Figure 1. Ultrasound images of fetal heart at 33 weeks and 6 days of gestation.

(a) Sternal cleft indicated by 2 arrows (b) Defect in sternum through which fetal heart protrudes through in every cardiac cycle

Antenatal Investigations



Figure 2. QR code providing access to a collection of videos, of Westmead Hospital Fetal Welfare ultrasound footage of fetal cardiac activity, demonstrating the pulsing of fetal cardiac apex through the sternal cleft, also known as ectopia cordis.

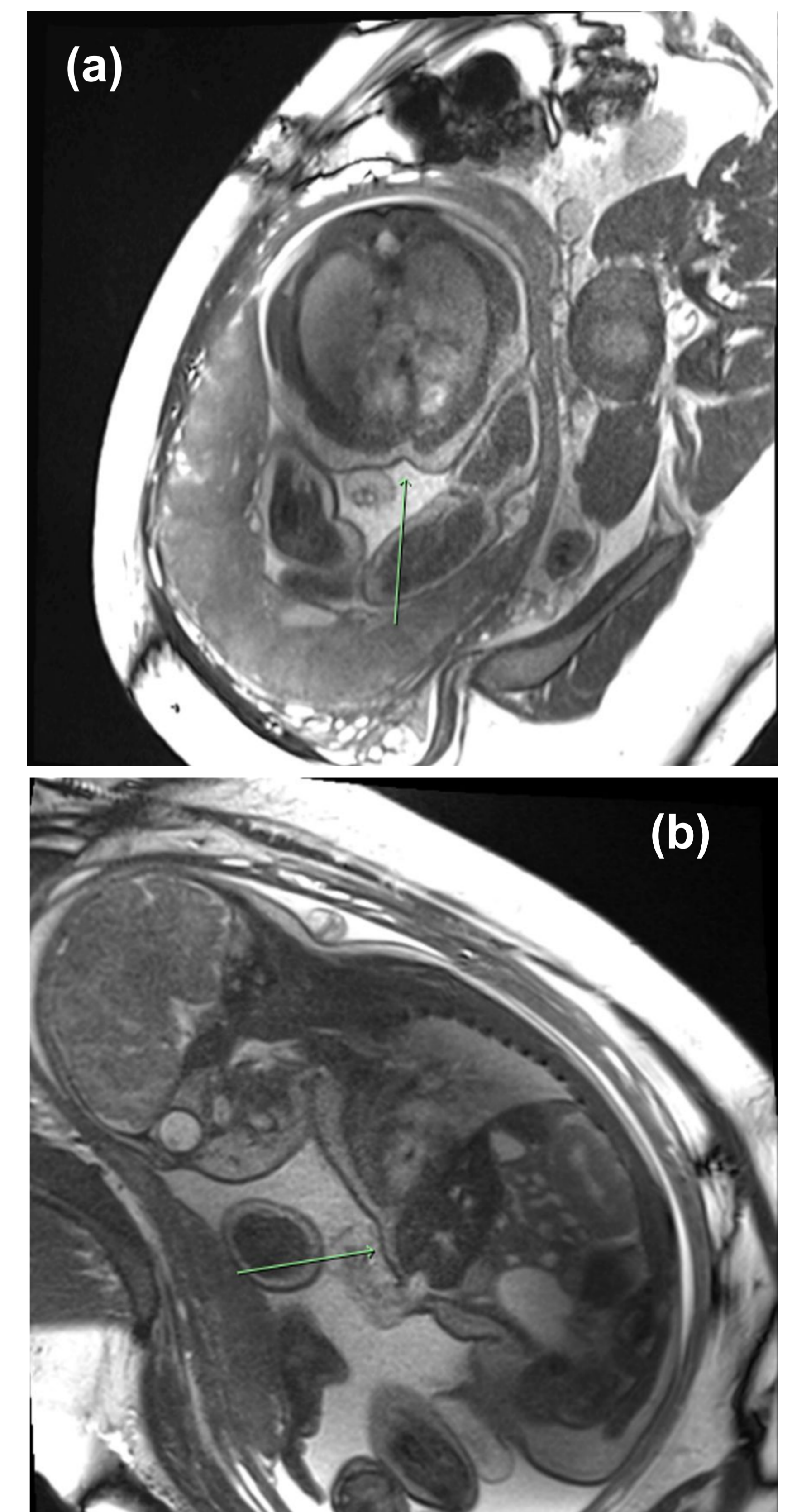


Figure 3. Multiphase fetal MRI images conducted at 37 weeks 5 days gestation. (a) Axial image, with arrowing showing central indentation of anterior inferior chest wall, corresponding to site of sternal defect on ultrasound (b) Sagittal image, with anterior bulge in the abdominal wall immediately inferior to the region described in (a).

Discussion

Imaging Modalities

- Diagnosis of EC can be made as early as 10-12 weeks gestation via ultrasonography
- Diagnosis of SC however is multimodal
 - MRI often thought to be complementary prenatally (Pasoglou et al., 2012)
 - **However, MRI conducted at 37 weeks 5 days gestation was UNABLE to directly visualise the sternum as a discrete structure. The lower chest of the fetus had an indeterminate appearances with slight central indentation and abdominal wall bulging inferior to this indentation**
 - **Post birth: chest X ray, computed tomography (CT) is GOLD STANDARD**

Mode of Delivery (MOD) and Survival

Ectopia Cordis

- Isolated ectopia cordis: delivery > 37 weeks and birth weight > 2500 g → survival exceeds 50% (Smith et al., 2014)

No clear literature on safest MOD in sternal cleft

Surgical Repair of Defect

- For neonates undergoing reparative cardiac surgery for congenital defects, birth at 37 – 38 weeks is associated with higher post-operative complications and longer hospital stay, with mortality in-hospital morbidity and mortality significantly higher after birth at 37 weeks vs 39.5 weeks (Costello et al., 2014)
- SC repair prognosis: **best outcomes with early primary closure in isolated SC**

Conclusion

EC diagnosed with fetal echocardiogram and ultrasound with Doppler
SC diagnosed with antenatal ultrasound, or post-birth X Ray + CT

No clear guidelines on safest mode of delivery for EC or SC
• Presence of associated congenital would guide MOD

Timing of delivery: should be aimed for ≥ 39 weeks gestation

EC repair: timing is multifactorial (assess severity of cardiac defect(s) and whether EC is isolated), presence of other syndromic defects, access to tertiary level facilities)

SC repair: early primary closure before 3 months

References

- Costello, J., Pasquali, S., Jacobs, J., He, X., Hill, K., Cooper, D., Backer, C. and Jacobs, M., 2014. Gestational Age at Birth and Outcomes After Neonatal Cardiac Surgery. *Circulation*, 129(24), pp.2511-2517.
- Engum, S., 2008. Embryology, sternal clefts, ectopia cordis, and Cantrell's pentalogy. *Seminars in Pediatric Surgery*, 17(3), pp.154-160.
- Gabriel, A., Donnelly, J., Kuc, A., Good, D., Doros, G., Matusz, P. and Ioukas, M., 2014. Ectopia cordis: A rare congenital anomaly. *Clinical Anatomy*, 27(8), pp.1193-1199.
- Kamal, Y., 2018. Sternal Cleft: Appropriate Approach to Diagnosis and Treatment. *Surgery and Case Studies: Open Access Journal*, 1(3).
- Pasoglou, V., Tebache, M., Rausin, L., Demarche, M. and Collignon, L., 2012. Sternal cleft: Prenatal multimodality imaging. *Pediatric Radiology*, 42(8), pp.1014-1016.
- Pius, S., Abubakar Ibrahim, H., Bello, M. and Bashir Tahir, M., 2017. Complete Ectopia Cordis: A Case Report and Literature Review. *Case Reports in Pediatrics*, 2017, pp.1-6.
- Smith, B., Flyer, J., Edwards, E., Soll, R., Horbar, J. and Yeager, S., 2020. Outcomes for Ectopia Cordis. *The Journal of Pediatrics*, 216, pp.67-72.
- Torre, M., Rapuzzi, G., Carlucci, M., Pio, L. and Jasonni, V., 2011. Phenotypic spectrum and management of sternal cleft: literature review and presentation of a new series. *European Journal of Cardio-Thoracic Surgery*, 41(1), pp.4-9.