Isolated Choroid Plexus Cyst

Recommendations of Practice

Background

A choroid plexus cyst (CPC) is a cystic structure found in the lateral ventricles of the fetal brain during ultrasound assessment. They are grouped in the category of 'soft markers' which are fetal sonographic findings that are not abnormalities themselves, but can be associated with chromosomal or non-chromosomal differences

CPC have an association with Trisomy 18 with a likelihood ratio of 7-9. However, most fetuses with an isolated finding of CPC do not have Trisomy 18.

Objective

To provide guidance and a consistent approach for the accurate diagnosis and management of fetuses identified to have CPCs at the mid-trimester screening ultrasound.

Definition

A well circumscribed echolucent structure within the choroid plexus in the lateral ventricle. A CPC should only be reported if measuring ≥5mm in maximum dimension. CPCs may be single or multiple, unilateral or bilateral and do not affect brain structure or function.

Differential Diagnosis

- Normal variant
- Association with Trisomy 18 (Edward Syndrome)
    - 30-50% of Trisomy 18 fetuses have CPCs
    - 80% of fetuses with Trisomy 18 will have other structural abnormalities seen on ultrasound
    - CPCs as a soft marker have a likelihood ratio (LR) for Trisomy 18 of 7-9 when seen in isolation
    - The number and size of cysts do not change the LR.
- Not associated with increased risk of Trisomy 21. LR is 1.0 (95% CI 0.1-9.4).

History

- Assess risk for chromosomal abnormality
- Review results of antenatal screening and mid-trimester anatomy scan.

Ultrasound

- The presence of a CPC (≥5mm) should prompt a carefully detailed ultrasound assessment to look for structural abnormalities (the fetal hands in particular).
- Features of Trisomy 18 include:
    - Fetal growth restriction
    - Polyhydramnios
    - 'Strawberry' shaped cranium
    - Clenched fists or overlapping fingers
    - Heart defects
    - Omphalocele
    - Single umbilical artery.

Investigation

- If a CPC is seen in isolation, no further testing is recommended
- If structural anomalies are identified on ultrasound, offer counselling and amniocentesis for genetic testing.

Prognosis

An isolated CPC(s) is typically transient and there are no associated adverse outcomes in the absence of aneuploidy.

On-going management

Isolated CPCs do not require further investigation or follow-up.

This Recommendation of Practice was updated in March 2023 by Dr Kristy Wolff with input from members of Wāhi Rua NZMFM Network.

The most up to date version of this Recommendation of Practice can be found on Healthpoint Wāhi Rua: New Zealand Maternal Fetal Medicine Network (NZMFM) webpages: https://www.healthpoint.co.nz/public/wahi-rua-new-zealand-maternal-fetal-medicine/

References

- Audibert, F, et al. No. 348 – Joint SOGC-CCMG Guideline: Update on prenatal screening for fetal aneuploidy, fetal anomalies, and adverse pregnancy outcomes. *JOGC* 2017; 39(9):805-817
- Bethune, M. Management options for echogenic intracardiac focus and choroid plexus cysts: A review including Australian Association of Obstetrical and Gynaecological Ultrasonologists consensus statement. *Australas Radio*, 2007; 51(4):324-329
- Martins, J, et al. Society for Maternal-Fetal Medicine Consult Series #57: Evaluation and management of isolated soft ultrasound markers for aneuploidy in the 2nd trimester. *AJOG* 2021; 225(4):PB2-B17
- Taliento, C, et al. Role of sonographic second trimester soft markers in the era of cell-free DNA screening options: A review. *Reprod Med,* 2022; 3:222-234