

# Isolated Choroid Plexus Cyst



This guideline was updated in April 2015 by Dr Joana De Sousa, with input from members of the New Zealand Maternal Fetal Medicine Network.



## Background

Midtrimester soft markers have been in use for many years, and controversy continues as to the significance of some.

Choroid plexus cysts are one of several recognised soft markers. Approximately 1-3% of the normal population will have choroid plexus cysts (CPC) identified within the fetal head at midtrimester ultrasound.

Choroid plexus cysts have an association with Trisomy 18 with a likelihood ratio (LR) of 9. **However most fetuses with choroid plexus cysts do not have chromosomal abnormalities, especially if this is an isolated finding.**

Detection and reporting of a feature suggestive of chromosomal anomaly has consequences. These include significant patient anxiety, the need for further counselling, significant investment in time and expense, the decision to proceed with further invasive testing, and the risk of miscarriage from invasive testing.

There are effective prenatal screening tests available for chromosomal abnormality with detection rates up to 90%. The second trimester ultrasound has a much lower detection rate and is therefore not intended as a screening test for chromosomal abnormality.

## Objective

To provide guidance and a consistent approach for the accurate diagnosis and management of fetuses presenting with choroid plexus cysts at the 18-20 week scan.

## Definition

- Well circumscribed echolucent areas within the choroid plexus in the lateral ventricle
- CPC should not be reported unless > 5mm in maximum dimension



## Differential Diagnosis

1. Normal variant
2. Associated with Trisomy 18.
  - 30-50% of Trisomy 18 have CPC
  - 80% of Trisomy 18 will also have other structural abnormalities
  - Likelihood Ratio for Trisomy 18 is 9 (95% CI 4.2-19.1)
  - Number of cysts and size of cysts does not change LR
3. Not associated with increased risk of Trisomy 21. LR is 1.0 (95% CI 0.1-9.4)

## Important History

1. Assess risk for chromosomal abnormality
2. Review results of prenatal screening and risk assessment

## Ultrasound

**Choroid plexus cysts are an indication for detailed careful ultrasound assessment rather than invasive testing**

Suboptimal imaging may necessitate follow up repeat scans to exclude other abnormality. Referral to tertiary anatomy scan is triggered if there are additional features identified.

### Choroid plexus cysts

- Sonographic well circumscribed echolucent discrete small cysts found within the choroid plexus within the lateral ventricle
- Imaging should be in the transverse plane of the fetal head at the same level for evaluation of lateral ventricles

- Inspect bilaterally for CPC
- CPC should not be reported unless > 5mm in maximum dimension
- Cystic spaces within the choroid less than this size should be considered “mottled” choroids, and unlikely to be significant

### **Assessment for other structural abnormalities ( particularly those associated with T18)**

- Ventriculomegaly
- Cystic hygromas
- Neural tube defects
- Cardiac abnormalities
- Diaphragmatic hernias
- Omphalocele
- Careful assessment of hands: Clenched hands, overlapping digits
- Growth restriction
- Single umbilical artery

### **Investigation**

Offer counselling and amniocentesis if:

- Other structural abnormalities suggestive of chromosomal anomaly

### **Prognosis**

Isolated CPC unilateral or bilateral (particularly with normal hands)

- Are a normal variant
- Does not increase risk of aneuploidy
- Are transient

No known adverse outcome if karyotype is normal





## On-going Management

Isolated CPC unilateral or bilateral (particularly with normal hands) in low risk

- Does not require further discussion or investigation
- Does not require follow up ultrasound

## References

- Bethune M. Management options for echogenic intracardiac focus and choroids plexus cysts: A review including Australian Association of Obstetrical and Gynaecological Ultrasonologists consensus statement. *Australas Radio*. 2007 Aug;51(4): 324-9
- SOGC Clinical Practice Guidelines. Fetal soft markers in obstetric ultrasound. *J Obstet Gynecol Can* 2005; 27(6): 592-612
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