Fetal Ventriculomegaly

Recommendations of Practice

Background

- Fetal Ventriculomegaly (VM) is usually diagnosed at the routine mid-trimester screening ultrasound scan but can be identified earlier or later in pregnancy as an incidental finding
- There are a wide range of possible underlying conditions and the spectrum of outcome is also wide, ranging from normal to significant neurodevelopmental disability. The aetiology of VM can include normal variation, aneuploidy, genetic conditions and syndromes, central nervous system abnormalities, congenital infection, and intracranial haemorrhage
- Appropriate counselling and investigation should be offered and tailored to the likely diagnosis. If no underlying diagnosis or associated anomalies are found, then aspects of the VM (regression or progression) can be used to offer some indication of prognosis.

Objective

- To guide accurate diagnosis, investigation, and management of women presenting with fetal VM
- To provide a consistent approach to the care of women with fetal VM taking into consideration the wide range of underlying causes and individual women's wishes.

Definition

- Lateral cerebral ventricular measurement increased above the normal cut off.
  - Measurement values:
    Normal: <10mm
    Mild: >10 - <12mm
    Moderate: >12 - <15mm
    Severe: ≥15mm
- May be unilateral or bilateral.

Differential Diagnosis

- Chromosomal and genetic differences
- Infection
- Syndrome
- Abnormal neuroanatomy
- Cerebral haemorrhage
- Ischaemic injury.

Important History

- Maternal history of viral/bacterial infection
- Past obstetric history (ie. fetal/neonatal anomalies, neonatal thrombocytopenia)
- Family history (ie. aneuploidy, syndromes, hydrocephalus)
- Consanguinity.

Ultrasound

- Full anomaly screen
- Appropriate measurement of the lateral ventricle (LV)
    - The plane for measurement is the transventricular (axial) plane. at the level of the frontal horns and cavum septi pellucidi, with the cerebral hemispheres symmetric in appearance
    - The atrium of the LV is measured at the at the level of the parieto-occipital sulcus of the posterior horn perpendicular to the long axis of the ventricle with calipers placed on the inner aspect of the ventricular walls
    - There is no standard measurement for the first trimester and early second trimester. This relies on choroid plexus to ventricle ratio.
- Assessment of the shape of the LV
    - Colpocephaly (teardrop shape) is associated with agenesis of the corpus callosum.
- Shape of the head and cerebellum
    - 'Lemon' shaped head (inward scalloping of the frontal bones) and 'banana' shaped cerebellum (Chiari 2 malformation) are associated with open neural tube defect.
- Identification of the cavum septum pellucidum and corpus callosum
- Posterior fossa assessment including the vermis cerebelli and cisterna magna
- Is there any evidence of intracranial haemorrhage
    - Echogenic lining and irregular hyperechoic mass. With maturation of haemorrhage may be calcifications and porencephalic cyst formation.
- Fetal gender
    - Can aid in prognostication (ie. X-linked hydrocephalus).
- Markers of infection
    - Brain: periventricular calcifications, subependymal cysts
    - Other: liver calcifications, ascites, hepatosplenomegaly, placentomegaly, growth restriction.
- Markers of aneuploidy

Investigation

- Maternal blood
    - CMV, toxoplasmosis, Parvovirus serology (IgM, IgG).
        - Infection can only be ruled out if IgM and IgG negative. In cases of IgM and IgG positive an avidity test maybe helpful in determining timing of infection. It may be required to access previous serology, such as booking bloods, to determine whether infection/seroconversion occurred in pregnancy.
    - Maternal allo-immune anti-platelet antibodies if evidence of severe VM or cerebral haemorrhage

- Amniocentesis
    - Chromosomal microarray (molecular karyotype)
        - Trisomy 21 is the most common genetic difference. LR 3.57.
        - Microarray abnormalities in 3.6% if isolated VM and 11.4% with associated anomalies.
    - PCR for CMV, toxoplasmosis, parvovirus
        - Note: some labs will only do if maternal serology IgM positive. Discussion with microbiology maybe required if recurrence or reactivation is suspected.
    - As NIPT is not a diagnostic test it is not sufficient for aneuploidy testing in this context
- Neurosonogram
    - Performed at MFM Hubs
    - Includes additional views of the fetal brain and spine
- MRI
    - May be advised at the tertiary level depending on ultrasound findings, test results and availability to better aid in prognosis. Especially useful in identifying cortical malformations, haemorrhage, and parenchymal disorders not as easily seen on ultrasound
    - Studies indicate that additional findings on MRI not visible on ultrasound that have implications for prognosis are found in 3-5% of cases when an MRI is offered for mild VM.

Prognosis

- Prognosis depends upon the degree and/or progression of VM and any additional anomalies or genetic conditions identified
- In the context of isolated VM, outcomes are generally very favourable
    - Mild VM: >90% normal outcome
    - Moderate VM: 75-93% normal outcome
- Neurodevelopmental delay has been identified in a small proportion of children who were identified as having isolated mild/moderate VM (6-8%), which is similar to the background rate in the general population
- Those with severe and/or progressive VM have a worse prognosis, in general relating to underlying cause.
- Unilateral VM:
    - Approximately 50 to 60% of cases of VM are unilateral; the remainder are bilateral. The aetiology and outcome of unilateral and bilateral VM are similar; therefore, counselling and management are generally the same.
    - It should be noted that some degree of asymmetry of the lateral ventricles exists in the human fetal brain, and occasionally, this can be appreciated prenatally. By itself, asymmetry (≥2 mm difference between the two lateral ventricles) does not appear to be associated with an adverse neurodevelopmental outcome when both ventricles are <10 mm.

Management

- On-going care will depend upon the underlying aetiology. Other disciplines that may be involved in patient care both antenatally and postnatally including geneticists, neonatologists, neurosurgeons, developmental paediatricians
- In the case of isolated mild or moderate VM, serial scans should continue throughout the pregnancy (~monthly) to identify any VM progression or any additional anomalies not previously seen.
- Postnatal followup:
    - Isolated mild/moderate VM
        - Postnatal paediatric review
        - Head USS +/-MRI as directed by HUS and Paediatrician.

This guideline 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/

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