Shortened Humerus or Femur

Recommendation of Practice

Background

The shortened humerus or femur has been an ultrasound finding associated with a number of conditions, including aneuploidy. In isolation it is more likely to be a variation of normal. In conjunction with other ultrasound features a short femur or humerus could be an indication of an underlying pathology, aneuploidy or syndrome.

Objective

To provide a consistent approach for the accurate diagnosis and management of a fetus found to have a shortened humerus or femur at the 18-20 week scan.

Definition

Short femur or humerus

- Measurement below 5th centile for gestational age
- Measurement less than 0.91 of that predicted by measured biparietal diameter (BPD)

Isolated or associated with other anomalies

Differential Diagnosis

Short femur associations:

- Isolated (2/3)
  - Normal variation or constitutional
  - Early onset fetal growth restriction (FGR)
    - Odds Ratio (OR) 9.7
- Non-Isolated (1/3)
  - Skeletal dysplasia
  - Aneuploidy
    - T21, T18, T13, triploidy
    - Likelihood ratio (LR) T21: 3.32
  - Genetic Syndromes
    - Eg Fraser and Costello Syndrome
  - Multiple Structural Anomalies

Short Humerus associations:

- Normal variation or constitutional
- Aneuploidy
    - LR T21: 4.81
- Skeletal dysplasia
- Early onset fetal growth restriction
    - OR 13

Important History

- History of previous FGR or pre-eclampsia (PET)
- PMedHx associated with FGR
- Assess risk for chromosomal abnormality
    - Review results of prenatal screening and risk assessment
    - Correlation with apriori risk with consideration of adjustment

Ultrasound

Femur and humerus length

- Measured with bone perpendicular to ultrasound beam
- Epiphyseal cartilages visible but not included in measurement
- Measure other long bones

Assessment for other structural abnormalities, evidence of skeletal dysplasia or FGR

- Shortened long bones are an indication for detailed careful ultrasound assessment rather than invasive testing
    - See Skeletal Dysplasia recommendation of practice: https://www.healthpoint.co.nz/public/wahi-rua-new-zealand-maternal-fetal-medicine/?solo=otherList&index=3
- Uterine artery Dopplers
    - Bilateral notching +/- increase resistance is demonstrated in up-to 90% of cases with early onset FGR
    - Associated with an increased risk of abruption, PET and intrauterine fetal demise
- Suboptimal imaging may necessitate follow up repeat scans or referral to exclude other abnormality
    - It is important that the patient is aware that repeat or tertiary scans are being requested because of suboptimal images, and not because of a soft marker identified on scan.

Investigation

Consider referral to a Fetal Medicine Centre for tertiary assessment for:

1. **Evaluation for other causes**
    - Detailed survey for other structural abnormality
    - Markers for skeletal dysplasia and manage accordingly
    - Markers (including uterine artery Dopplers) for early onset FGR and manage accordingly
    - Assess risk for aneuploidy, and establish apriori risk

2. **Calculation of aneuploidy risk for T21**
   - Individual likelihood ratios (LR) to apply to the apriori risk can be accurately calculated using the negative (ie absence of) and positive (presence of) LR for each marker.
   - This can be automatically calculated using the online tool:.
     - Follow the link to download the spreadsheet from the supporting information section: http://onlinelibrary.wiley.com/doi/10.1002/uog.12364/suppinfo

3. **Offer counselling with consideration of advanced screening (ie NIPT) or amniocentesis if:**
   - Adjusted risk > 1:300
   - Other structural abnormality
     - Note: NIPT may not be appropriate here (favour invasive testing)
   - Other indicators of aneuploidy
   - Parents wish definitive testing for aneuploidy rather than screening (favour invasive testing)

On-going Management

Needs ongoing surveillance and follow up (minimum four-weekly) ultrasound for:

- Skeletal dysplasia (late presentation)
  - Consider antenatal genetics referral is this is suspected
- FGR
  - Follow Small for Gestational Age Guideline if surveillance scan detects FGR or drop in growth trajectory
- PET via clinical assessment

This Recommendation of Practice was updated in August 2023 by Dr Jaynaya Marlow 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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