Shortened Humerus or Femur

New Zealand Maternal Fetal Medicine Network

This guideline was updated in August 2015 by Dr Jay Marlow, with input from members of the New Zealand Maternal Fetal Medicine Network.

Background

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

Objective

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

Definition

Short femur or humerus

- measurement below 2.5 centile for gestational age
- measurement less than 0.91 of that predicted by measured BPD

Isolated or associated with other anomalies

Differential Diagnosis

Short femur associations:

- **Isolated (2/3)**
  1. Normal variation or constitutional
  2. Early onset fetal growth restriction (FGR)

- **Non-Isolated (1/3)**
  3. Skeletal dysplasia
  4. Aneuploidy
     - T21, T18, T13, triploidy
     - Likelihood ratio T21 3.32
  5. Genetic Syndromes
     - Eg Fraser and Costello Syndrome
  6. Multiple Structural Anomalies
- **Short Humerus associations**
  1. Normal variation or constitutional
  2. Aneuploidy
     - Likelihood ratio T21 4.81
  3. Skeletal dysplasia
  4. Early onset fetal growth restriction

Important History

1. History of previous FGR or PET
2. PMedHx associated with FGR
3. Assess risk for chromosomal abnormality
   a. Review results of prenatal screening and risk assessment
   b. Correlation with *apriori* risk with consideration of adjustment

Ultrasound

Femur and humerus length

- measured with bone perpendicular to ultrasound beam
- with 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
- 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, pre-eclampsia 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 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: http://onlinelibrary.wiley.com/doi/10.1002/uog.12364/suppinfo
3. **Offer counselling with consideration of advanced screening (ie NIPS) or amniocentesis if:**
   - Adjusted risk > 1:300
   - Other structural abnormality
     - Note: NIPS may not be appropriate here (favour invasive testing)
   - Other indicators of aneuploidy
   - Parents wish definitive testing for aneuploidy rather than screening (favour invasive testing)

- NB: NIPS is currently only available to the patient with an out-of-pocket cost in NZ

On-going Management

Needs ongoing surveillance and follow up ultrasound for:

- Skeletal dysplasia (late presentation)
  - Consider antenatal genetics referral is this is suspected
- FGR
- PET

References

- Papageorghiou AT, Fratelli N, Leslie K, Bhide A, Thilaganathan. Outcome of fetuses with antenatally diagnosed short femur. Ultrasound Obst Gynecol 2008; 31:507-511
- Weisz B, David AL, Chitty L, Peebles D, Pandya P, Patel P, Rodek CH. Association of isolated short femur in mid-trimester fetus with perinatal outcome. Ultrasound Obst Gynecol 2008; 31:512-516
- Agathokleous M, Chaveeva P, Poon LCY, Kosinski P, Nicolaides KH. Meta-analysis of second trimester markers for trisomy 21. Ultrasound Obst Gynecol 2013; 41:247-261