

Suspected skeletal dysplasia

Inform mother of likely diagnosis
Assess for associated anomalies
Review first trimester screening result (if performed) and risk factors for IUGR
Send TORCH screen
Offer referral to Leeds FMU <3 working days

Re-scan by local Obs US specialist < 3 working days if expertise available

Leeds FMU within 5 days of initial diagnosis

- Full anatomical review including thoracic assessment and all long bones – length, ossification and architecture
- Differential diagnosis is IUGR – assess including uterine artery resistance +/- fetal dopplers
- Offer invasive testing for qf-PCR & array-CGH– ask lab to store DNA
- Consider NIPD testing if appropriate
- Offer recruitment to research studies – RAPID, PAGE, BOOSTB4, 100KG
- Multidisciplinary input from genetics, neonatology and review in the perinatal skeletal dysplasia forum

TOP – advise full PM, skeletal survey and send tissue for molecular genetic studies and DNA storage
Postnatal review by genetics

Continuing pregnancy
Leeds FMU 2-4 weekly
Assess growth, liquor, dopplers
Refine diagnosis

Uncertain outcome or good prognosis likely

Poor outcome expected

Birth in Leeds General Infirmary, usually around term
Multidisciplinary team input
Offer meeting with neonatology to discuss postnatal management
Ensure all relevant specialties aware

Offer hospice team input and support from Bereavement team
Local birth may be preferred
Neonatal review to plan postnatal care