Diagnostic trans-iliac bone biopsy: is there a clinical need?
Review of consecutive bone biopsies carried out over five years in a UK specialist centre.

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Background
• Diagnostic bone biopsy has been an important component of metabolic bone services delivery for 50 years.
  • Specialist laboratory facilities are not available in regular hospital or histopathology labs in the UK.
  • Trans-iliac trephine of the pelvis is used (figure 1a).
• The Metabolic Bone Unit at Addenbrooke’s Hospital, Cambridge provides services for the Eastern Rare Bone Disease Network (ERBoN).
  • Comprehensive governance structure; samples of patients with rare skeletal diseases can be stored for up to 30 years.
  • Annual sample utility audits and regular inspections.
  • Inter-laboratory quality control including blind assessment of slide pathologies.
  • Licensed by the Human Tissue Authority.
• Recent Kidney Disease Improving Global Outcomes (KDIGO) guidelines suggest clinical utility of biopsy for two indications:
  • In diagnosis.
  • In guiding complex treatment decisions.

Methods
Case selection
• Eligible rare bone disease cases are presented at our monthly ERBoN multidisciplinary team meeting.
  • Agreement is reached by a minimum of two experienced clinicians before proceeding to bone biopsy.
  • Biopsy is recommended only when there are unanswered clinical/pathological questions despite imaging and biochemical analysis (Bone Assay Service).
  • Patients are consented and given spaced fluorochrome labels.

Procedure
• Orthopaedic surgeon-led procedure involving light, brief (15 minute) general anaesthetic usually in day case theatre.
  • Modified Border trephine trans iliac biopsy taken, transported in 70% ethanol.
  • 1 x 7.5mm trans iliac core taken (+ 1 x 2mm Jamshidi marrow biopsy into formalin for decalcification sent to NHS histopathology for malignancy staining/marrow analysis).
  • Undecalcified bone core embedded in LR white resin, sectioned stained, and specific features interpreted in the calcified tissues (figure 1 b - d).

Results
• During the study period, 430 patients with rare bone diseases were documented in Cambridge Outpatients Bone Registry (COBR)
  • 46 bone biopsies were carried out (20 male, 28 female, mean age 54.4 yrs ± 15.1 SD).
  • 40 patients received fluorochrome labelling prior to biopsy.
  • Underlying renal disease was the primary indication in 21 cases (45.7%), with 16 other specialist bone indications.
  • In 43 samples (93.5%) specific pathological features (figure 2a) were described and 26 of those reports (60%) contained specific new treatment advice.

Conclusions
• Diagnostic bone biopsy was performed in 10.7% of 430 patients with rare bone disease over 5 years.
  • Most biopsies exhibited pathology.
  • Treatment recommendations were made in 60% of these cases based on the full clinical picture and histology.
  • A prospective multi-centre study is needed to fully address the clinical utility of diagnostic bone biopsy.
• Diagnostic bone biopsy, histological interpretation and reporting to clinicians should be an essential component of complex metabolic bone disease services.
  • Where laboratory facilities do not exist locally bone samples should be sent to laboratories that do provide these services.

More details on the Cambridge University Metabolic bone Unit can be found at tinyurl.com/y354swmz