SKIN BIOPSY FOR INBORN ERRORS OF METABOLISM

INDICATIONS
- Diagnosis of inherited metabolic disorders
- Check with your laboratory for local arrangements

Skin biopsy is often collected for histological analysis. Contact your local histopathology department for advice on sample handling

EQUIPMENT
- Forceps: fine non-bend watchmaker’s or dissecting
- Cotton wool balls and gallipots
- Dressing towel
- Size 15 scalpel blade and no. 3 handle
- 25 gauge needle (orange top)
- 23 gauge needle (blue top)
- 21 gauge needle (green top) for drawing up lidocaine
- 2 mL syringe
- Chlorhexidine gluconate 0.05%
- Lidocaine 1%
- Bottles of culture medium
- Sterile gloves
- Steristrips
- Dressings:
  - 1 small transparent dressing (e.g. Tegaderm/Opsite)
  - Gauze swabs
  - Elasticated cotton or other bandage

SAMPLE REQUIREMENTS
- At least 1 mm × 1 mm of skin (ideally 2 mm × 2 mm) from preferred site (e.g. inner side of forearm or posterior aspect just above elbow)
- Choose site carefully as even a small scar on coloured skin will be very obvious
- If post-mortem, take skin from over scapula as this leaves less obvious damage (see Post-mortem specimens below)

PROCEDURE

Consent
- Explain procedure and reassure parents
- Obtain and record consent

Technique

Maintain strict asepsis using ‘no touch’ technique
- Sterilise site with chlorhexidine gluconate 0.05% and dry off with a gauze swab
- Inject lidocaine 1%, a little intradermally and remainder subcutaneously to anaesthetise an area 1.5 × 1 cm
- Wait 5 min to ensure site anaesthetised
- Cleanse again using chlorhexidine gluconate 0.05%, wipe off and dry using sterile cotton wool or gauze swabs

Method A
- Using fine forceps, grip a fold of skin between blades so that a length of skin 3 mm × 2 mm protrudes
- Slice off in one stroke by running scalpel blade along upper edge of forceps blades
- If skin too thick or oedematous to grip, proceed to method B
Method B
- Pierce skin with 23 or 21 gauge needle and lift to produce ‘tenting’
- Cut off tip of tent to produce piece of skin approximately 2 mm with a round ‘O’ shape
- Place into culture medium bottle immediately (lid of bottle removed by assistant for shortest possible time)
- Complete request form with:
  - clinical details
  - date and time of sampling

Dressing wound
- Although it may bleed freely, wound is usually partial thickness and should not require stitching
- Apply pressure to stanch bleeding
- apply Steristrips and sterile dressing, bandage if necessary
- Remove bandage after a few hours, but leave dressing for several days
- Reassure parents that scar, when visible, will be seen as a fine line

Transport
- Once sample taken, send to Inherited Metabolic Diseases Laboratory as soon as possible
- if unable to arrange transport immediately, store sample at +4°C for maximum of 12 hr before despatch
- do not freeze sample

POST-MORTEM SPECIMENS
- In accordance with Human Tissue Act, post-mortem samples must be taken only on licensed premises (or satellites thereof). Check with your pathology laboratory manager

Specimens taken after death present a high risk of infection and possible failure of culture. Follow strict asepsis technique

- Take two biopsies from over scapula (as this leaves less obvious damage), as soon as possible after death, ideally before 48 hr have elapsed
- Send sample to Inherited Metabolic Disease Laboratory immediately, or store at +4°C before dispatch for maximum of 12 hr, do not freeze
- Include clinical details, date and time of sampling, and date and time of death on request form