

SKIN BIOPSY FOR INBORN ERRORS OF METABOLISM

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INDICATIONS

- Diagnosis of inherited metabolic disorders
- **Wherever possible, discuss biopsy and arrangements with Department of Newborn Screening and Biochemical Genetics, Birmingham Children's Hospital 0121 333 9942**
- this should include discussion about which specimen bottles and transport medium to use
- confirm instructions for storage and transport with your local laboratory

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

EQUIPMENT

- Forceps: fine dissecting forceps
- Cotton wool balls and gallipots
- Dressing towel
- Plastic apron
- Size 15 scalpel blade and no. 3 handle
- 25 G needle (orange top)
- 23 G needle (blue top)
- 21 G needle (green top) for drawing up lidocaine
- 2 mL syringe
- **Cleaning solution as per unit policy**
- Lidocaine 1%
- Bottles of culture medium
- Sterile gloves
- Steri-Strip™
- Dressings:
 - 1 small transparent dressing (e.g. Tegaderm™/Opsite)
 - gauze swabs
 - elasticated cotton or other bandage

SAMPLE REQUIREMENTS

- $\geq 1 \text{ mm} \times 1 \text{ mm}$ of skin (ideally $2 \text{ mm} \times 2 \text{ mm}$) 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**)

PROCEDURE

Consent

- Inform parents of reason for biopsy, explain procedure and risks including:
 - healing and scarring
 - possibility of contamination
 - poor growth
- Obtain and record **written** consent (**see below for post-mortem sampling**)

Technique

Maintain strict asepsis using 'non-touch' technique

- Wash hands and put on apron and sterile gloves
- Cleanse site
 - ensure cleaning fluid does not pool beneath baby
- Sedation if appropriate
- Inject lidocaine 1%, a little intradermally and remainder subcutaneously to anaesthetise an area $1.5 \times 1 \text{ cm}$
- Wait 5 min to ensure site anaesthetised
- Cleanse again, wipe off and dry using sterile cotton wool or gauze swabs

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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 1 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 G needle and lift to produce 'tenting'
- cut off tip of tent to produce a round 'O' shaped piece of skin approximately 2 mm
- 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 Steri-Strip™ 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 aseptic technique

- Full written consent required for post-mortem biopsy samples
- Take 2 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 diseases 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