TECHNIQUE OF SKIN BIOPSY FOR THE INVESTIGATION OF INBORN ERRORS OF METABOLISM

Principle

Many of the enzymes known to be deficient in inborn errors of metabolism are expressed in skin fibroblasts. Enzyme activities can be measured in fibroblast cultures after they have been grown in culture for 2-4 weeks. The results of such assays can definitively diagnose many inborn errors of metabolism and are often necessary before proceeding to pre-natal diagnosis.

Laboratory notification

Discuss with laboratory before arranging test.

Materials required

A 3 – 5 mm punch biopsy is a very useful viable alternative to a skin biopsy taken with a scalpel blade which if available is to be preferred.

Pharmaceutical

Ampoule of lignocaine 1% or 2% - MAY NOT ALWAYS BE ESSENTIAL
Hibitane in spirit
Alcohol swabs
1 medium square of Elastoplast about 3cm^2

Other

Skin biopsy consent form
Forceps - fine, non-bend watchmaker's forceps are best, but otherwise dissecting forceps
Scalpel blade and handle
25-gauge (orange) needle
23-gauge (blue) needle
2mL syringe
Gauze swabs
Cling or other bandage
Bottle of cell culture medium (***sterile transport medium***) (***Do not use Agar***) eg Ham's F10, MEM etc. A sterile container containing Ham's F10 can be obtained from main lab Chem Path at the Children's Hospital. This sterile transport medium can also be supplied by first class post on request if a skin biopsy is to be taken at an outlying hospital.

Approximate length of test

Thirty minutes

Patient preparation

(*NB* The most important aspect is ***STRICT ASEPSIS*** using a '*no touch*' technique.)

1 Swab the area to be biopsied. The inner side of the forearm or the posterior aspect just above the elbow are the preferred sites. Sterilise the site with Hibitane in spirit and clean off with an alcohol swab.

2 Inject lignocaine a little intradermally and the remainder subcutaneously to anaesthetise an area 1.5 x 1cm **(often not necessary)**

3 Wait 5 minutes; ensure site is anaesthetised

4 Cleanse again using Hibitane; wipe off and dry

5 **Sampling**

Method A - normal procedure

Grip a fold of skin between blades of forceps so that a length of skin about 8mm by 2 mm protrudes. If the skin cannot be gripped because it is too thick or oedematous proceed to Method B. For method A slice off protruding skin (about 2mm x 2mm) in one stroke by running the scalpel blade along the upper edge of the forceps blades. A full thickness of skin is needed so that the subdermal layer is included. Place in the culture medium (lid of bottle removed for shortest possible time by assistant).

Method B - for thicker skin

Pierce skin with a modest size needle (23 gauge blue) and lift up to produce 'tenting'. Lop off tip of 'tent' to detach a round piece of skin of about 2mm circumference. Put immediately into the culture medium.

6 **Mending the damage**

The biopsy site is usually mostly of partial skin thickness and although it may bleed freely should not need stitching. Staunch with pressure. Apply Elastoplast for several days. Patients can be re-assured that the scar - when visible - is simply seen as a fine line. Such marks on coloured skin are very obvious - therefore choose your site carefully.

7 The sample should be sent with a completed request form to the cell culture laboratory as soon as possible either by first class post or by special transport. If transport cannot be arranged immediately, the sample should be stored at +4°C

(*Do not freeze*) prior to dispatch.

1 Post mortem samples

If a specimen is taken after death there is a high risk of infection and a possible failure of culture. A skin biopsy should be taken at the beginning of the post-mortem to minimise contamination and the biopsy site rigorously decontaminated as described above. In this situation, the above procedure should be adhered to but 2 separate biopsies from different sites should be obtained and these placed in **two** separate pots of transport medium ideally with added Fungizone if this is available. Successful culture is most likely if the biopsy is taken within 48 hrs of death but can be successful up to 8 days after death in exceptional circumstances

Notes

1 Under normal circumstances biopsies should not be taken without prior arrangements with the laboratory undertaking the test.

2 Large biopsies are less likely to culture than small ones - do not take large deep samples.

3 **A signed consent form must accompany each sample.**

Reference

Holton JB ed. UK Directory of Laboratories Diagnosing Inborn Errors of Metabolism, 3rd edition 1988, appendix pp 83-5.