
Guidelines for sending samples to the EB laboratory/Skin service for analysis

The National Diagnostic EB Laboratory undertakes examination of skin biopsy samples for diagnosis of all forms of epidermolysis bullosa and for other genodermatoses. Techniques used include light microscopy (LM) and immunohistochemistry by immunofluorescence microscopy (IF).

We also undertake molecular genetics analysis of junctional EB, dystrophic EB and other genodermatoses.

We carry out prenatal diagnosis (PND) by molecular genetic analysis of chorionic villus samples (CVS) and cells from amniocentesis fluid.

In order to be sure that we can carry out the necessary tests and to help us to provide you with an efficient service, please follow these guidelines. We may not be able to commence work on your samples until we have all the required information.

Should these guidelines below not be adhered to, samples may be rejected.

A. Background information

Please:

1. Complete the supplied request form fully and clearly; one request form per patient.
2. Indicate that consent was obtained from the patient, and whether this is for diagnosis and/or for research. The Human Tissue Act, which came into force on 1st September 2006, requires that we have patient consent for all samples stored by the EB Lab.
3. Give all relevant clinical details, including any family history.
4. Supply any available details of consanguinity.
5. For PND by molecular genetic analysis, give the mutations for the proband and parents, and indicate which lab carried out this diagnosis.
6. Supply your telephone number(s), secure fax number, as well as email address, so that we can contact you if necessary.

B. Sample treatment

1. Blood samples must be collected into EDTA tubes, with volumes between 0.5ml and 10mls.
2. Saliva samples can be collected from patients unable to give blood. These should be collected Oragene®•DNA sample collection kit, but closed and sealed tightly to avoid spillage.

3. Cleaned CVS must be placed in 50mM Tris/HCl, pH 7.0 (**no heparin**).
4. Tubes must be clearly and unambiguously labelled with the patient's name and date of birth (*except for anonymous research samples which should be coded*).
5. Each patient's sample must be placed in a separate plastic bag. This avoids the possibility of cross-contamination if leakage occurs.
6. Skin biopsy sample tubes should be placed in their original labelled bags – sample for IF in 'Michel's Medium' bag. Any tools (petri dishes, scalpels) used to collect/excise the biopsies should be disposed of according to your local procedure for sharps waste.
7. Sample tubes should be safely packed into the BioBottle (if supplied) for transit or otherwise packed in accordance with appropriate transport regulations.

C. Dispatch information

Please:

1. Contact us before you send the sample.
We need to make sure that someone will be available to receive the package.
Opening hours are from 9.00 -17.00.
2. Tell us how the sample will be sent, e.g. courier, Special Delivery mail or regular mail.
This helps us to know where to look for the package when it arrives.
3. Address the package to:

DNA Lab
Specimen Reception
5th Floor Tower Wing
Guy's Hospital
Great Maze Pond Road
London
SE1 9RT
UK

Please put our telephone contact details on the outside of the package

Tel: +44 (0)20 7188 7229

Samples should be sent at ambient temperature. There is no need to refrigerate during transit.

DO NOT SEND ON DRY ICE - DO NOT FREEZE.

4. Contact us when you have sent the sample to let us know that it is on its way. This will tell us when to expect it.

5. If you are sending the sample by courier, please supply the following information as soon as you dispatch it:
 - a) Courier company name
 - b) Consignment reference or Airway Bill number
 - c) Expected arrival date and time.
 This will allow us to track the package.
6. If you are sending the sample by Special Delivery mail, please supply the following information as soon as you dispatch it:
 - a) Tracking reference number
7. Even if the sample is being sent by regular mail, please let us know when you dispatch it.
8. Do not dispatch the package on a Friday. If the sample is ready on a Friday, store it until Monday as follows:
 - a) Skin biopsy sample for IMF (in Michel's Medium) – store at room temperature
 - b) Blood sample/saliva sample – store in a fridge (4°C)
 - c) DNA sample - store in a fridge (4°C)
 - e) d)Cleansed CVS – store in a freezer (-20°C)
9. Please provide us with contact telephone numbers, and an email address if possible, so that we can confirm receipt of the sample.
10. Please inform us if the procedure is cancelled or postponed. This will prevent us wasting time searching for samples that have not been dispatched.
11. Indicate that consent was obtained from the patient, and whether this is for diagnosis and/or for research. The Human Tissue Act, which came into force on 1st September 2006, requires that we have patient consent for all samples stored by the EB Lab.
12. **Please note for biopsy kits that are not to be used immediately, please store the reagents as follows:**
 - a) Michel's medium for IMF, store at room temperature
 - b) RNALater store at room temperature
13. **The stipulated time-frame for a sample to reach the lab, from time of taking is one week. Please ensure patient samples reach us in good time to ensure optimal results are obtained.**