NHS
South East
Genomic Laboratory Hub

MUTATION ANALYSIS & SKIN MICROSCOPY TEST REQUEST FORM

The National Diagnostic Epidermolysis Bullosa (EB) Laboratory

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All fields are mandatory. Illegible, unclear or incomplete forms will result in delays or rejection.

CONSENT STATEMENT: It is the referring clinician's responsibility to ensure that the patient/carer knows the purpose of the test and that the sample may be stored for future diagnostic testing. In signing this form the clinician has obtained consent for testing, storage and for the use of this sample and the information gathered from it to be shared with members of the donor's family through their health professionals (if appropriate). The patient should be advised that the sample may be used anonymously for quality assurance and training purposes. If the patient does not wish information to be shared please write this clearly in the clinical summary box.

PATIENT DEMOGRAPHICS	
First name:	
Last name:	
DOB:	**Gender:** Male ☐ Female ☐ Other ☐
NHS number:	
Hospital no:	**Family ref no:**
Postcode:	**Life status:** Alive ☐ Deceased ☐
Non-NHSE funded (please attach invoicing details): ☐	

PATIENT ETHNICITY	
White:	British ☐ Irish ☐ Any Other White Background ☐
Mixed:	White And Black Caribbean ☐ White And Black African ☐ White And Asian ☐ Any Other Mixed Background ☐
Asian or Asian British:	Indian ☐ Pakistani ☐ Bangladeshi ☐ Any Other Asian Background ☐
Black or Black British:	Caribbean ☐ African ☐ Any Other Black Background ☐
Other Ethnic Groups:	Chinese ☐ Any Other Ethnic Group ☐ (please specify: _____)
Not stated ☐	**Not Known** ☐

CLINICAL INDICATIONS

Segmental overgrowth disorders R110 (only germline mutations)	☐	**Cutaneous photosensitivity with a likely genetic cause R237**	☐
Ectodermal dysplasia R163	☐	**Incontinentia pigmenti R239**	☐
Epidermolysis bullosa and congenital skin fragility R164	☐	**Epidermodysplasia verruciformis R255**	☐
Ichthyosis and erythrokeratoderma R165	☐	**Vascular skin disorders R326 (only germline mutations)**	☐
Palmoplantar keratodermas R166	☐	**Rare genetic inflammatory skin disorders R332**	☐
Autosomal recessive primary hypertrophic osteoarthropathy R167	☐	**Other**	☐
Xeroderma pigmentosum, Trichothiodystrophy or Cockayne syndrome R227	☐		
Multiple monogenic benign skin tumours R230	☐		
Pigmentary skin disorders R236	☐		

CLINICAL INFORMATION, FAMILY HISTORY AND CONFIRMATION OF ELIGIBILTY

Please demonstrate how your patient meets the eligibility criteria for this test. Interpretation of results depends on the quality of clinical information provided. Find the eligibility criteria here: https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-inherited-disease-eligibility-criteria-V7.1-OFFICIAL.pdf

For familial cases, please include a pedigree with the patient clearly marked:

Affected ☐ Unaffected ☐

CLINICALLY URGENT? ☐ **Reason for urgency:** _____

Sample(s)

Skin: Michel's Medium	☐
Skin: EM Fixative	☐
Skin: RNA Later	☐
Blood (in EDTA tube)	☐
DNA	☐
Saliva	☐
ELISA	☐

For lab reference use only

Skin biopsy details:

Biopsy site: _____

Shave ☐ Punch ☐ Ellipse ☐

Lesional ☐ Peri-lesional ☐ Non-lesional ☐

Rubbed skin: Yes ☐ No ☐

Date & time of sample(s): _____

Pedigree number:

CLINICIAN DETAILS

Requesting clinician / consultant	Responsible clinician / consultant *(if different)*
Name:	Name:
Hospital & department:	Hospital & department:
NHS email:	NHS email:
Phone:	Phone:

Note: Please ensure the latest version of this request form is used, found on our website: www.southeastgenomics.nhs.uk