NHS
South East
Genomic Laboratory Hub

MUTATION ANALYSIS & SKIN MICROSCOPY TEST REQUEST FORM

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

The National Diagnostic Epidermolysis Bullosa (EB) Laboratory & Skin Service, 3rd Floor, Bermondsey Wing, Guy's Hospital.
All enquiries to EBLab@synnovis.co.uk
NOT FOR PATIENT USE: Tel: +44 (0)20 7188 7229

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:	**Sex assigned at birth:** Male ☐ Female ☐ Other ☐
NHS Number:	
Hospital Number:	**Family Ref. Number:**
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:	Indian ☐ Pakistani ☐ Bangladeshi ☐ Any Other Asian Background ☐
Black:	Caribbean ☐ African ☐ Any Other Black Background ☐
Other Ethnic Groups:	Chinese ☐ Any Other Ethnic Group ☐ (please specify: _____)
Not stated ☐	**Not known** ☐

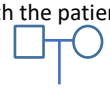
CLINICAL INDICATIONS

- **Ectodermal dysplasia R163** ☐
- **Epidermolysis bullosa and congenital skin fragility R164** ☐
- **Ichthyosis and erythrokeratoderma R165** ☐
- **Palmoplantar keratodermas R166** ☐
- **Autosomal recessive primary hypertrophic osteoarthropathy R167** ☐
- **Xeroderma pigmentosum, trichothiodystrophy or Cockayne syndromeR227** ☐
- **Multiple monogenic benign skin tumours R230** ☐
- **Pigmentary skin disorders R236** ☐
- **Cutaneous photosensitivity with a likely genetic cause R237** ☐
- **Incontinentia pigmenti R239** ☐
- **Epidermodysplasia verruciformis R255** ☐
- **Vascular skin disorders R326 (only germline variants)** ☐
- **Rare genetic inflammatory skin disorders R332** ☐
- **HAVCR2 R424** ☐
- **Other** ☐

CLINICAL INFORMATION, FAMILY HISTORY AND CONFIRMATION OF ELIGIBILITY

Please demonstrate how your patient meets the eligibility criteria for this test. Interpretation of results depends on the quality of clinical information provided (including details of any consanguinity and clinical presentation). Find the eligibility criteria here: https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-inherited-disease-eligibility-criteria-v8.0.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: RNA Later ☐
- Skin: Cell culture Medium ☐
- Blood (in EDTA tube) ☐
- DNA ☐
- Saliva ☐
- ELISA ☐

Date & time sample(s) received:

For lab reference use only

PLEASE ENSURE ALL SAMPLES ARE SENT DIRECTLY TO:

SEGLH Specimen Reception
5th Floor, Tower Wing
Guy's Hospital
Great Maze Pond
London SE1 9RT

Skin biopsy details:

Biopsy site:_____

Shave ☐ Punch ☐ Ellipse ☐

Lesional ☐ Peri-lesional ☐ Non-lesional ☐

Rubbed skin: Yes ☐ No ☐

Date & time of sample(s) taken: _____

Pedigree Number:

CLINICAL UTILITY- Please indicate category and provide details

Patient management (determining therapeutic decisions and/or clinical investigations and/or surveillance programme) ☐
Patient, parents, or adult relative reproductive decision making ☐
Unaffected relatives are seeking predictive testing ☐

CLINICAL DETAILS

Requesting clinician/consultant name: Hospital and department:
NHS.net email:
Phone:

Responsible clinician/consultant name (if different):
Hospital and department:
NHS.net email:
Phone:

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