

MUTATION ANALYSIS & SKIN MICROSCOPY TEST REQUEST FORM

The National Diagnostic Epidermolysis Bullosa (EB) Laboratory

Guy's Hospital 3rd Floor Bermondsey Wing Great Maze Pond London SE1 9RT

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

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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		PATIENT ETHNICITY	
First name:			British 🗆 Irish 🗆 Any Other White Background 🗆
Last name: DOB:	Gender: Male	Mixed:	White And Black Caribbean □ White And Black African □ White And Asian □ Any Other Mixed Background □
NHS number:		Asian or Asian British:	Indian □ Pakistani □ Bangladeshi □ Any Other Asian Background □
Hospital no:	Family ref no:	Black or Black British:	Caribbean African Any Other Black Background
Postcode:	Life status: Alive Deceased	Other Ethnic Groups:	Chinese Any Other Ethnic Group (please specify:)
Non-NHSE funded (please attach invoicing details):		Not stated 🗆	Not Known 🗆

CLINICAL INDICATIONS			
Segmental overgrowth disorders R110 (only germline mutations)		Cutaneous photosensitivity with a likely genetic cause R23	37 🗌
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			

Please include relevant family history, details of any consanguinity and provisional diagnosis. PLEASE NOTE TESTING WILL NOT COMMENCE UNTIL FULL CLINICAL INFORMATION IS PROVIDED.	For familial cases, please include a pedigree with the patient clearly marked:			
Affected Unaffected CLINICALLY URGENT? Reason for urgency:				
Sample(s) For lab reference use only Skin biopsy details:				

		Pedigree number:	
ELISA			
Saliva		Date & time of sample(s):	
DNA		Rubbed skin: Yes 🗌 No 🗌	
Blood (in EDTA tube)			
Skin: RNA Later		Lesional 🗌 Peri-lesional 🗌 Non-lesional 🗆	
Skin: EM Fixative		Shave 🗆 Punch 🗆 Ellipse 🗆	
Skin: Michel's Medium		Biopsy site:	

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