


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

CLINICAL INFORMATION, FAMILY HISTORY AND CONFIRMATION OF ELIGIBILITY	
<p>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: <a href="https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-inherited-disease-eligibility-criteria-v8.0.pdf">https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-inherited-disease-eligibility-criteria-v8.0.pdf</a></p> <p>Is patient pregnant? <b>Y/N</b>      If yes how many weeks gestation? _____</p>	<p>Have other members of this family had gene testing? <b>Y/ N</b> Please provide details:</p> <p>For familial cases, please include a pedigree with the patient clearly marked:</p> 
Affected <input type="checkbox"/> Unaffected <input type="checkbox"/>	Age of onset: _____ Patients to be tested: Patient only <input type="checkbox"/> Patient and both parents <input type="checkbox"/> Other <input type="checkbox"/>
CLINICALLY URGENT? <input type="checkbox"/>	

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

SAMPLE TYPE:	TEST REQUEST:
Blood EDTA <input type="checkbox"/> for DNA or gene tests (if not EDTA please state) Extracted DNA <input type="checkbox"/> CVS <input type="checkbox"/> Amnio <input type="checkbox"/> Fetal blood <input type="checkbox"/> POC <input type="checkbox"/> Other (please state e.g. buccal swab)	Gene test: _____ Test directory clinical indication and ID (R number): _____ <a href="https://www.england.nhs.uk/publication/national-genomic-test-directories/">https://www.england.nhs.uk/publication/national-genomic-test-directories/</a> <i>If the clinical indication and code are not provided, a test or panel will be applied based on the clinical information provided.</i> DNA storage only <input type="checkbox"/> Other (please specify) _____
Date of collection: Time of collection: For Departmental Use Only:	

Note: Please ensure the latest version of this request form is used, found on our website: [www.southeastgenomics.nhs.uk](http://www.southeastgenomics.nhs.uk)

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Patient first name:	Patient last name:	DOB:	NHS no:
			<div style="display: flex; justify-content: space-between;"> <div style="width: 20px; height: 20px; background-color: black;"></div> <div style="width: 20px; height: 20px; background-color: black;"></div> <div style="width: 20px; height: 20px; background-color: black;"></div> <div style="width: 20px; height: 20px; background-color: black;"></div> <div style="width: 20px; height: 20px; background-color: black;"></div> <div style="width: 20px; height: 20px; background-color: black;"></div> <div style="width: 20px; height: 20px; background-color: black;"></div> <div style="width: 20px; height: 20px; background-color: black;"></div> </div>

**HPO terms phenotypes and presence in this individual – please tick**  
Please confirm the HPO terms that have been assessed, and select whether they are present or absent

Bleeding disorder	Present	Absent
Bruising susceptibility <a href="#">HP:0000978</a>		
Epistaxis <a href="#">HP:0000421</a>		
Prolonged bleeding after surgery <a href="#">HP:0004846</a>		
Prolonged bleeding after dental extraction <a href="#">HP:0006298</a>		
Gingival bleeding <a href="#">HP:0000225</a>		
Petechiae <a href="#">HP:0000967</a>		
Menorrhagia <a href="#">HP:0000132</a>		
Post partum haemorrhage <a href="#">HP:0011891</a>		
Intracranial haemorrhage <a href="#">HP:0002170</a>		
Cerebral haemorrhage <a href="#">HP:0001342</a>		
Gastrointestinal haemorrhage <a href="#">HP:0002239</a>		

Thrombocytopenia / Platelet disorder	Present	Absent
Thrombocytopenia <a href="#">HP:0001873</a>		
Congenital thrombocytopenia <a href="#">HP:0001905</a>		
Macrothrombocytopenia <a href="#">HP:0040185</a>		
Abnormal platelet function <a href="#">HP:0011869</a>		
Abnormal platelet granules <a href="#">HP:0011883</a>		
Glanzmann Thrombasthenia <a href="#">ORPHA:849</a>		
Bernard Soulier syndrome <a href="#">ORPHA:274</a>		
Gray Platelet syndrome <a href="#">ORPHA:721</a>		
Thrombotic Thrombocytopenic Purpura <a href="#">ORPHA:54057</a>		

Thrombosis / Thrombophilia	Present	Absent
Venous thrombosis <a href="#">HP:0004936</a>		
Deep venous thrombosis <a href="#">HP:0002625</a>		
Pulmonary embolism <a href="#">HP:0002204</a>		
Arterial thrombosis <a href="#">HP:0004420</a>		
Stroke <a href="#">HP:0001297</a>		
Cerebral venous thrombosis <a href="#">HP:0005305</a>		

Other (please specify)	Present	Absent
Sensorineural hearing impairment <a href="#">HP:0000407</a>		
Presenile cataracts <a href="#">HP:0007819</a>		
Renal insufficiency <a href="#">HP:0000083</a>		
Ocular albinism <a href="#">HP:0001107</a>		
Neutrophil inclusion bodies <a href="#">HP:0008264</a>		

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