

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 AND FAMILY HISTORY	
Please give as much clinical and genetic information as possible. Interpretation of results depends on the quality of clinical information provided. Please use HPO terms (https://hpo.jax.org/app/) when possible, see reverse.	
<p>Have other members of this family had gene testing? Y/N Please provide details:</p> <p>For familial cases, please include a pedigree with the patient clearly marked:</p>	
Is patient pregnant? Y/N If yes how many weeks gestation? _____	
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	Gene test: _____
Lithium Heparin <input type="checkbox"/> for karyotype only	Test directory clinical indication and ID (R number): _____ https://www.england.nhs.uk/publication/national-genomic-test-directories/ <i>If the clinical indication and code are not provided, a panel will be applied based on the clinical information provided.</i>
CVS <input type="checkbox"/> Amnio <input type="checkbox"/> Fetal blood <input type="checkbox"/> POC <input type="checkbox"/>	Array CGH <input type="checkbox"/>
Other (please state)	QF-PCR (rapid aneuploidy) <input type="checkbox"/>
Date of collection:	Karyotype analysis <input type="checkbox"/>
Time of collection:	DNA storage only <input type="checkbox"/>
For Departmental Use Only:	Other (please specify) _____

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

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Patient first name:	Patient last name:	DOB:	NHS no:																				
			<table border="1"> <tr> <td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td><td></td> </tr> </table>																				

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

Intellectual disability, developmental and metabolic	Present	Absent
Intellectual disability - mild		
Intellectual disability - moderate		
Intellectual disability - profound		
Intellectual disability - severe		
Autistic behaviour		
Global developmental delay		
Delayed fine motor development		
Delayed gross motor development		
Delayed speech and language development		
Generalized hypotonia		
Feeding difficulties		
Failure to thrive		
Abnormal facial shape		
Abnormality of metabolism/homeostasis		
Microcephaly		
Macrocephaly		
Tall stature		

Craniosynostosis	Present	Absent
Bicoronal synostosis		
Unicoronal synostosis		
Metopic synostosis		
Sagittal craniosynostosis		
Lambdoidal craniosynostosis		
Multiple suture craniosynostosis		

Skeletal dysplasia	Present	Absent
Disproportionate short stature		
Proportionate short stature		
Short stature		
Skeletal dysplasia		

Epilepsy	Present	Absent
Seizures		
Generalized-onset seizure		
Focal-onset seizure		
Epileptic spasms		
Infantile encephalopathy		
Atonic seizures		
Generalized myoclonic seizures		
Generalized tonic seizures		
Generalized tonic-clonic seizures		
EEG with focal epileptiform discharges		
EEG with generalized epileptiform discharges		
Multifocal epileptiform discharges		

Neurology	Present	Absent
Muscular dystrophy		
Myopathy		
Myotonia		
Fatigable weakness		
Peripheral neuropathy		
Distal arthrogryposis		
Arthrogryposis multiplex congenita		
Cognitive impairment		
Parkinsonism		
Spasticity		
Chorea		
Dystonia		
Ataxia		
Cerebellar atrophy		
Cerebellar hypoplasia		
Dandy-Walker malformation		
Olivopontocerebellar hypoplasia		
Diffuse white matter abnormalities		
Focal White matter lesions		
Leukoencephalopathy		
Cortical dysplasia		
Heterotopia		
Lissencephaly		
Pachygyria		
Polymicrogyria		
Schizencephaly		
Holoprosencephaly		
Hydrocephalus		

Diabetes	Present	Absent
Neonatal insulin-dependent diabetes mellitus		
Transient neonatal diabetes mellitus		

Renal	Present	Absent
Multiple renal cysts		
Nephronophthisis		
Hepatic cysts		
Enlarged kidney		

Other (please specify)	Present	Absent

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