

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. Testing may be performed at Synnovis, any other NHSE GLH or by other international laboratories where necessary. 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		SAMPLE TYPE:	
First name:		Blood EDTA <input type="checkbox"/> for DNA or gene tests	
Last name:		Lithium Heparin <input type="checkbox"/> for karyotype only	
DOB:	Sex assigned at birth: Male <input type="checkbox"/> Female <input type="checkbox"/> Other <input type="checkbox"/>	CVS <input type="checkbox"/> Amnio <input type="checkbox"/> Fetal blood <input type="checkbox"/> POC <input type="checkbox"/> RNA <input type="checkbox"/>	
NHS number:		Histology sample <input type="checkbox"/> specimen number	
Hospital no:	Family ref no:	Tissue type:	
Postcode:	Life status: Alive <input type="checkbox"/> Deceased <input type="checkbox"/>	Other (please state):	
Non-NHSE funded (please attach invoicing details): <input type="checkbox"/>		Date of collection:	Time of collection:
Ethnicity:		For Departmental Use Only:	

CLINICAL INFORMATION AND FAMILY HISTORY	
<p>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 (<a href="https://hpo.iax.org/app/">https://hpo.iax.org/app/</a>) when possible, see reverse.</p> <p>Is patient pregnant? Yes <input type="checkbox"/> No <input type="checkbox"/> If yes how many weeks gestation? _____</p> <p>Have other members of this family had gene testing? Yes <input type="checkbox"/> No <input type="checkbox"/></p> <p>Please provide details:</p> <p>Has this patient had a bone marrow transplant or a blood transfusion? Yes <input type="checkbox"/> No <input type="checkbox"/></p>	<p>Affected <input type="checkbox"/> Unaffected <input type="checkbox"/></p> <p>Age of onset: _____</p> <p>For familial cases, please include a pedigree with the patient clearly marked:</p> <p>Consanguineous? Yes <input type="checkbox"/> No <input type="checkbox"/></p>

TEST REQUEST:	
URGENT <input type="checkbox"/>	Routine <input type="checkbox"/>
If you are a member of a Clinical Genetics team, is this test: For a consultant <input type="checkbox"/> For a proband <input type="checkbox"/> Type of testing? A diagnostic / predictive / carrier test	If you are <u>not</u> a member of a Clinical Genetics team, is this test diagnostic? Yes <input type="checkbox"/> No <input type="checkbox"/>
Test Directory ID number: _____ (R or M code) <i>This is a mandatory field.</i>	SNP Array <input type="checkbox"/> QF-PCR (rapid aneuploidy) <input type="checkbox"/> Karyotype analysis <input type="checkbox"/> Storage only (no test activation) <input type="checkbox"/> Other (please specify)
<i>Please ensure that this referral meets the testing criteria:</i> <a href="https://www.england.nhs.uk/publication/national-genomic-test-directories/">https://www.england.nhs.uk/publication/national-genomic-test-directories/</a>	

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

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>

**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 - mild		
Global developmental delay - moderate		
Global developmental delay - profound		
Global developmental delay - severe		
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		
Cleft Palate		

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		

CARDIAC	Present	Absent
Tetralogy of Fallot		
Interrupted aortic arch		
Truncus arteriosus		
Other congenital heart disease		
Calcium homeostasis disorder		

Neurology	Present	Absent
Muscular dystrophy		
Myopathy		
Myotonia		
Fatigable weakness		
Peripheral neuropathy		
Distal arthrogryposis		
Schizencephaly		
Holoprosencephaly		
Hydrocephalus		
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		

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

Renal & Urinary	Present	Absent
Multiple renal cysts		
Nephronophthisis		
Hepatic cysts		
Enlarged kidney		
Congenital anomalies of the kidney & urinary tract (CAKUT)		

Other (please specify)	Present	Absent
Hyper/hypo pigmentation following Blaschkos lines (Hypomelanosis of Ito)		
Asymmetry		
Dysmorphism (please specify)		

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