NHS
South East
Genomic Laboratory Hub

TEST REQUEST FORM FOR NON-WGS GENETIC TESTS

Genetics Laboratories, 5th Floor, Tower Wing, Guy's Hospital, Great Maze Pond, London, SE1 9RT
020 7188 1696/1709
Clinical enquiries to gst-tr.viapathgeneticsadmin@nhs.net
General enquiries to gst-tr.southeastglh@nhs.net

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	
First name:	
Last name:	
DOB:	**Sex assigned at birth:** Male ☐ Female ☐ Other ☐
NHS number:	
Hospital no:	**Family ref no:**
Postcode:	**Life status:** Alive ☐ Deceased ☐
Non-NHSE funded (please attach invoicing details): ☐	
Ethnicity: _____	

SAMPLE TYPE:	
Blood EDTA ☐ for DNA or gene tests	
Lithium Heparin ☐ for karyotype only	
CVS ☐ **Amnio** ☐ **Fetal blood** ☐ **POC** ☐ **RNA** ☐	
Histology sample ☐ specimen number	
Tissue type:	
Other (please state):	
Date of collection:	**Time of collection:**
For Departmental Use Only:	

CLINICAL INFORMATION, FAMILY HISTORY AND CONFIRMATION OF ELIGIBILTY

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: https://www.england.nhs.uk/wp-content/uploads/2018/08/rare-inherited-disease-eligibility-criteria-V7.1-OFFICIAL.pdf

Is patient pregnant? Yes ☐ No ☐ If yes how many weeks gestation? _____

Have other members of this family had gene testing? Yes ☐ No ☐

Please provide details:

Has this patient had a bone marrow transplant or a blood transfusion? Yes ☐ No ☐

Affected ☐ Unaffected ☐

Age of onset: _____

For familial cases, please include a pedigree with the patient clearly marked:

Consanguineous?
Yes ☐ No ☐

TEST REQUEST:

URGENT ☐ **Routine** ☐

If you are a member of a Clinical Genetics team, is this test:

For a consultand ☐ For a proband ☐

Type of testing? A diagnostic / predictive / carrier test

If you are not a member of a Clinical Genetics team, is this test diagnostic?
Yes ☐ No ☐

Test Directory ID number: _____
(R or M code)
This is a mandatory field.

Please ensure that this referral meets the testing criteria:
https://www.england.nhs.uk/publication/national-genomic-test-directories/

- **SNP Array** ☐
- **QF-PCR (rapid aneuploidy)** ☐
- **Karyotype analysis** ☐
- **Storage only (no test activation)** ☐
- **Other (please specify)**

CLINICIAN DETAILS:

Requesting clinician / consultant Name:	Reporting clinician/ consultant *(if different)*
Hospital & department:	Name:
	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

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Patient first name:	Patient last name:	DOB:	NHS no:

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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