Genomic Medicine Service

Whole Genome Sequencing (WGS) Test Request
PLEASE DO NOT USE FOR NON-WGS TESTS

RARE AND INHERITED DISEASES PROBAND

NHS

Requesting organisation:	
GLH laboratory to receive sample:	

Patient first name		Life status ☐ Alive ☐ Deceased	Ethnicity *(Please tick on Page 2)*
Patient last name		Family test ☐ Singleton ☐ Trio ☐ Other (provide number):	
Date of birth *(dd/mm/yyyy)*	Hospital number	Additional clinical information (if relevant) *Please include any previous molecular testing with date(s) and any other pertinent clinical information*	
Gender ☐ Male ☐ Female ☐ Other			
NHS number (or postcode if not known)			
Reason NHS Number not available: ☐ Patient not eligible for NHS number (e.g. foreign national) ☐ Other (please provide reason):			

Family members to be tested

First name	Last name	Date of birth *(dd/mm/yyyy)*	Relationship to proband

Test request

Test required **Whole Genome Sequencing**	Test Directory Clinical Indication & code (reason for testing)	Age of onset *State in years and months*
Clinically urgent ☐		

Additional panel(s) (if relevant; mandatory for R89) *(use panels with panel type 'GMS Rare Disease Virtual' - http://panelapp.genomicsengland.co.uk)*	Disease penetrance ☐ Complete ☐ Incomplete ☐ Not known	Specific rare or inherited diseases that are suspected or have been confirmed (OMIM/Orphanet) (if relevant)

HPO terms (https://hpo.jax.org/app/) phenotypes & presence in this individual *(Tick on page 2 or state below if not listed)*

Samples *(being sent to GLH DNA extraction lab)*

☐ Blood (EDTA) ☐ Amniotic fluid ☐ Fetal blood ☐ Chorionic Villus ☐ Fresh Tissue (not tumour)
☐ Stored DNA *(please specify primary source type/refer to sample handling guidance)*

Sample ID	Collection date / time	Sample volume if applicable	Comments

Clinician details

Responsible clinician / consultant	Main contact (if different from responsible clinician/consultant)
Name: Department address: Phone: Email:	Name: Department address: Phone: Email:

☐ **I have attached a copy of the Record of Discussion form**
☐ Patient conversation taken place; Record of Discussion form to follow

First name	Last name	Date of birth *(dd/mm/yyyy)*	NHS number (or postcode if not known)

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

Ethnicity – Please tick the relevant Self Defined Ethnicity code below

White	Mixed	Asian or Asian British	Black or Black British	Other Ethnic Groups
A British ☐	D White and Black Caribbean ☐	H Indian ☐	M Caribbean ☐	R Chinese ☐
B Irish ☐	E White and Black African ☐	J Pakistani ☐	N African ☐	S Any other ethnic group ☐
C Any other White background ☐	F White and Asian ☐	K Bangladeshi ☐	P Any other Black background ☐	Z Not stated ☐
	G Any other mixed background ☐	L Any other Asian background ☐		99 Not known ☐

Additional local identifiers – please use the table below, these will also be displayed in the interpretation portal

Type – O (Test order/Case ID), P (local patient ID), S (local sample identifier)		
Organisation	**Type**	**Identifier**