

Genomic Medicine Service	RARE AND INHERITED DISEASES FAMILY MEMBER	
Whole Genome Sequencing (WGS) Test Request		
PLEASE DO NOT USE FOR NON-WGS TESTS		

Requesting organisation:
GLH laboratory:

First name	Life status Alive Deceased	Ethnicity
Last name	HPO terms (https://hpo.jax.org/app/) phenotypes & presence in this individual (if relevant)	
Date of birth (dd/mm/yyyy)	Hospital number	
Gender Male Female Other	Specific rare or inherited diseases that are suspected or have been confirmed	
Postcode		
NHS Number		
Reason NHS Number not available: Patient not eligible for NHS number (e.g. foreign national) Other (please provide reason):		

Relevant clinical information <i>Please include any previous molecular testing with date(s) and any other pertinent clinical information</i>
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Relationship to proband	For the condition being tested, please describe the individual's disease status Affected Unaffected	Age of onset <i>State in years and months</i>
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Proband details

Proband first name	Proband NHS number (or postcode if not known)
Proband last name	Proband date of birth (dd/mm/yyyy)

Test request

Test required Whole Genome Sequencing	Test Directory Clinical Indication & code (reason for testing)
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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
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Responsible clinician / consultant

Main contact (if different from responsible clinician/consultant)	Main contact (if different from responsible clinician/consultant)
Name:	Name:
Department address:	Department address:
Phone:	Phone:
Email:	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)										
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HPO terms phenotypes and presence in this individual
Please confirm the HPO terms that have been assessed, and select whether they are present or absent

Suggested useful terms below

HPO Terms (https://hpo.jax.org/app/)		
	Present	Absent

Intellectual disability, developmental and metabolic
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
Bicoronal synostosis
Unicoronal synostosis
Metopic synostosis
Sagittal craniosynostosis
Lambdoidal craniosynostosis
Multiple suture craniosynostosis

Skeletal dysplasia
Disproportionate short stature
Proportionate short stature
Short stature
Skeletal dysplasia

Diabetes
Neonatal insulin-dependent diabetes mellitus
Transient neonatal diabetes mellitus

Renal
Multiple renal cysts
Nephronopththisis
Hepatic cysts
Enlarged kidney

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

Epilepsy
Seizures
Generalized seizures
Focal seizures
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