Genomic Medicine Service

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

RARE AND INHERITED DISEASES

NHS

Requesting organisation:

GLH laboratory:

Proband's first name	Life status ☐ Alive ☐ Deceased; Ethnicity
Proband's last name	Family test ☐ Singleton ☐ Trio ☐ Other (provide number):
Date of birth *(dd/mm/yyyy)*; Hospital number	Relevant clinical information *Please include any previous molecular testing with date(s) and any other pertinent clinical information*
Gender ☐ Male ☐ Female ☐ Other *Please state in clinical information box if karyotypic and/or phenotypic sex differ from given gender*	
Postcode	
NHS number	
Reason NHS Number not available: ☐ Patient not eligible for NHS number (e.g. foreign national) ☐ Other (please provide reason):	

Test request

Clinical Priority
There is currently no urgent WGS pathway, however it may be possible to prioritise cases in exceptional circumstances. Please provide details of why this referral is considered a priority.

Test Directory Clinical Indication & code (reason for testing)

Proband's age of onset *years* *months*

Additional panel(s) (if relevant; **mandatory for R89**)
(use panels with panel type 'GMS Rare Disease Virtual' - https://nhsgms-panelapp.genomicsengland.co.uk/)

Disease penetrance
☐ Complete
☐ Incomplete

Specific rare or inherited diseases that are suspected or have been confirmed

Family members to be tested *(not required for proband only referrals)*

First name	Last name	Date of birth	NHS Number *(or postcode if not known)*	Gender	Deceased	Status	Ethnicity	Relationship to proband

Samples *being sent to GLH DNA extraction lab (only required if also using this form for sample collection)*

First name	Last name	Date of birth	Sample ID	Collection date / time	Sample type	Sample volume	Comments

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 for all individuals**
☐ Patient conversation taken place; Record of Discussion form to follow

Proband first name	Proband last name	Date of birth *(dd/mm/yyyy)*	NHS number

HPO terms are important for the analysis and interpretation of WGS data.
Please enter valid HPO terms present in the proband/family members being tested.
HPO terms can be copied from the lists below, however please note these are not comprehensive lists of available terms.

HPO Terms - Please ensure those given match those available at [(https://hpo.jax.org/app/)](https://hpo.jax.org/app/)	**Present**	**Absent**	**Present**	**Absent**	**Present**	**Absent**
	☐	☐	☐	☐	☐	☐
	☐	☐	☐	☐	☐	☐
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	☐	☐	☐	☐	☐	☐

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
Nephronophthisis
Hepatic cysts
Enlarged kidney
Renal insufficiency

Neurology
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
Neurodegeneration
Dementia

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

Cardiology
Hypertrophic cardiomyopathy
Dilated cardiomyopathy
Cardiomyopathy

Eye Disorders
Cataract
Retinal dystrophy
Macular dystrophy
Microphthalmia
Anophthalmia
Coloboma
Developmental glaucoma
Aniridia
Abnormal anterior eye segment morphology
Nystagmus

Immune Disorders
Immunodeficiency
Abnormal lymphocyte morphology
Abnormal lymphocyte physiology
Abnormal lymphocyte count
Abnormality of neutrophils
Abnormality of humoral immunity
Abnormal inflammatory response
Abnormality of complement system