For full functionality of this form, use Adobe Reader or Acrobat. Web browser or Outlook views may be limited.

Genomic Medicine Service

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

RARE AND INHERITED DISEASES

NHS

Requesting organisation:

GMS 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*
Sex assigned at birth ☐ Male ☐ Female	
Postcode	
NHS number	
Reason NHS Number not available: ☐ Patient not eligible for NHS number (e.g. foreign national) ☐ Other (please provide reason):	
Proband's age at onset of clinical features *years* *months*	

Test request

Test Directory Clinical Indication & code (reason for testing)

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

State if specific rare disease is suspected or confirmed

Clinical Priority There is no urgent pathway, however prioritisation may be possible in exceptional circumstances. Please provide reason for urgency.

Reason for diagnostic test (Please provide additional information with other relevant clinical information above)

☐ Patient management: determining therapeutic decisions and/or clinical investigations and/or surveillance programme
☐ Patient, parents, or adult relative reproductive decision making
☐ Unaffected relatives are seeking predictive testing

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)*	Sex assigned at birth	Deceased	Status	Ethnicity	Relationship to proband

Samples *being sent to GMS DNA extraction lab* ***(only required if also using this form for sample collection, please provide blood samples in EDTA)***

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

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

Also required - provide HPO terms on page 2

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

HPO terms are NECESSARY for the analysis and interpretation of WGS data and cannot commence until provided. Please ensure HPO terms match those available at https://hpo.jax.org/app/

HPO Terms At **least** one HPO term is required									
	Present	**Absent**	Unknown	**Present**	**Absent**	Unknown	**Present**	**Absent**	**Unknown**
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The following list of HPO terms is provided as a guide but is not an exhaustive list. More terms are available at **https://hpo.jax.org**

Cardiology	**Developmental**	**Neurology**
Hypertrophic cardiomyopathy	Intellectual disability, moderate	Muscular dystrophy
Dilated cardiomyopathy	Intellectual disability, profound	Myopathy
Cardiomyopathy	Intellectual disability, severe	Myotonia
Immunology	Global developmental delay	Peripheral neuropathy
Immunodeficiency	Generalized hypotonia	Cognitive impairment
Abnormal lymphocyte count	Failure to thrive	Spasticity
Abnormality of humoral immunity	Abnormal facial shape	Chorea
Abnormal inflammatory response	Abnormality of metabolism/homeostasis	Dystonia
Opthalmology	Microcephaly	Ataxia
Cataract	Macrocephaly	Cerebellar atrophy
Retinal dystrophy	Tall stature	Cerebellar hypoplasia
Macular dystrophy	Short stature	Seizure
Renal	Skeletal dysplasia	
Multiple renal cysts	Hearing impairment	
Hepatic cysts		

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