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| Genomic Medicine Service Whole Genome Sequencing (WGS) Test Request PLEASE DO NOT USE FOR NON-WGS TESTS | RARE AND INHERITED DISEASES FAMILY MEMBER |  |
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|---------------------------------|
| 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> | | |
| 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) <div style="border: 1px solid black; width: 100px; height: 20px; margin-top: 5px;"></div> |
| Proband last name | Proband date of birth (dd/mm/yyyy) <div style="border: 1px solid black; width: 100px; height: 20px; margin-top: 5px;"></div> |

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| 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) | |

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|-----------|------------------------|-----------------------------|----------|
| 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) |
| 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) |
| | | | <div style="display: flex; justify-content: space-around;"> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> <div style="border: 1px solid black; width: 15px; height: 15px;"></div> </div> |

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 |
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| Intellectual disability, developmental and metabolic |
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| 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 |
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| Neonatal insulin-dependent diabetes mellitus |
| Transient neonatal diabetes mellitus |

| Renal |
|----------------------|
| Multiple renal cysts |
| Nephronophthisis |
| 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 |