East GLH Rare Disease Test Directory – Semi-Rapid Clinical Indications

The Rare and Inherited Disease Genomic Test Directory includes four clinical indications delivered using trio whole genome sequencing (WGS) that in specific circumstances can be delivered as a Semi-rapid singleton whole exome virtual gene panel test. These four indications are listed in the table below.

This option is available to provide a semi-rapid turnaround for diagnostic testing in patients where a rapid diagnosis will direct immediate treatment or medical care. The expected turnaround time for issuing results is 6 weeks.

The eligibility criteria for semi-rapid tests is provided for each clinical indication in the Test Directory Eligibility Criteria document:
https://www.england.nhs.uk/publication/national-genomic-test-directories/

Authorisation must be sought from the specialist provider prior to ordering a semi-rapid test (as per the R14 rapid pathway). Please contact the specialist provider team using the email addresses below prior to submitting a patient for this test.

Please use your local rare disease non-WGS process for ordering the semi-rapid test (see section below). A record of the consent discussion with the patient and /or family should be retained within the medical record.

Consider arranging for parental blood samples to be sent for DNA storage to enable future WGS trio testing if required.

Please contact the Cambridge Genomics Laboratory for further guidance: geneticslaboratories@nhs.net

Clinical Indication Code	Test code	Clinical Indication	Provider	Provider contact email address
R15	R15.5	Primary immunodeficiency or monogenic inflammatory bowel disease	North Thames GLH, Great Ormond Street FT	gosh.geneticslab@nhs.net
R98	R98.3	Likely inborn error of metabolism	Central and South GLH, Birmingham Women's and Children's FT	bwc.metabolicgenomics@nhs.net
R135	R135.3	Paediatric or syndromic cardiomyopathy	South West GLH, North Bristol Trust	nbn-tr.cardiacservice@nhs.net
R257	R257.3	Unexplained young onset end-stage renal disease	South West GLH, North Bristol Trust	nbn-tr.swglhrenalservice@nhs.net

Process for Ordering Semi-Rapid Rare Disease Testing

1. Contact appropriate specialist testing provider using the details above.
   Copy the Cambridge Laboratory email address in your communication:
   geneticslaboratories@nhs.net

2. Once authorised by the specialist testing provider, use your local standard rare disease non-WGS process to order the semi-rapid genetic test. Indicate on the test order:
   - that the request is for a "Semi Rapid Singleton WES"
   - the clinical indication required and the clinical information for the patient
   - that approval has been given by the specialist testing provider (or send copy of the email with the test order form)

3. Arrange for the test order and EDTA blood sample to be sent to the Cambridge, Leicester or Nottingham Genomics Laboratories

Process for Ordering Semi-Rapid Rare Disease Testing in CUH EPIC

In EPIC, select one of the following test codes to order genomic testing:

LAB9956	Rare Disease Genomic Testing
LAB7298	Genomic Test Request on Stored Sample

- Select the required Clinical Indication.
- Select "not for WGS".
- Provide clinical information for the patient.
- In free text box indicate "Semi Rapid Singleton WES" and indicate that approval has been given by the specialist testing provider (forward email to geneticslaboratories@nhs.net or add copy of email to patient notes in EPIC).