15 May 2025

Dear Colleague,

IMPORTANT: CHANGE TO NATIONAL TEST DIRECTORY

Removal of Array Testing for patients eligible for Whole Genome Sequencing

NHSE genetic laboratories are required to deliver testing according to the National Genomic Test Directory https://www.england.nhs.uk/publication/national-genomic-test-directories/.

In the Test Directory version published on 2nd January 2025 the following microarray tests were removed:

R59.2 Epilepsy

R69.3 Hypotonic infant

R83.2 Arthrogryposis

R84.2 Cerebellar anomalies

R86.2 Hydrocephalus

R87.2 Cerebral malformation

R88.2 Severe microcephaly

R89.2 Ultra-rare disorders

R100.2 Craniosynostosis

Array testing will therefore be unavailable for these clinical indications. For patients meeting the eligibility criteria **whole genome sequencing (WGS)** testing is available. WGS can detect small sequence variants, small copy number variants, as well as large copy number variants and structural variants previously tested by microarray. This change is therefore predicted to increase the diagnostic yield and de-duplicate work.

WGS analysis is most effective as part of a trio (i.e. using samples from the affected individual and both parents) and we request that trios be submitted for testing wherever possible. All WGS tests require completion of the following forms:

1) WGS GMS test order form for rare disease (one form per trio)
https://www.england.nhs.uk/wp-content/uploads/2024/07/gms-test-order-form-rare-disease-v1.5.pdf

2) Record of Discussion Regarding Genomic Testing (one form per individual)
https://www.england.nhs.uk/wp-content/uploads/2021/09/nhs-genomic-medicine-service-record-of-discussion-form.pdf

The referral information given **must** state how the patient meets the testing criteria:
https://www.england.nhs.uk/wp-content/uploads/2025/01/rare-and-inherited-disease-eligibility-criteria-V7.1-OFFICIAL-2.pdf

If the patient does not meet the criteria for testing or insufficient clinical information is provided the test request will be rejected.

These changes will be implemented by the North East and Yorkshire GLH from 1st June 2025.

Turnaround times

Standard reporting targets set by NHSE for testing are 84 days for whole genome sequencing. However, patients should be aware that results may take longer, due to capacity issues within the laboratories.

If you have any questions regarding this change in service, please do not hesitate to contact your local genetics laboratory:

Newcastle Genetics Laboratory: Ciaron McAnulty ciaron.mcanulty@nhs.net
Leeds Genetics Laboratory: Andrea Coates andrea.coates1@nhs.net
Sheffield Diagnostic Genetics Service: Nick Beauchamp nick.beauchamp@nhs.net

Yours faithfully,

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