August 2024

Dear Colleague,

IMPORTANT: CHANGE TO TEST ELIGIBILITY CRITERIA
for Patients with Intellectual Disability and Developmental Delay (R377 and R29)

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/. A new version of the Directory was published on 31st July 2024, clarifying which paediatric patients are eligible for array testing and/or whole genome sequencing. The tests affected are:

R377 Intellectual disability - microarray only
Testing Criteria: Unexplained moderate/severe/profound global developmental delay or unexplained moderate/severe/profound intellectual disability.

R29 Intellectual disability – whole genome sequencing
Testing Criteria: Unexplained moderate/severe/profound global developmental delay or unexplained moderate/severe/profound intellectual disability, where clinical features are suggestive of an underlying monogenic disorder requiring sequencing and targeted genetic testing is not possible.

1) Patients with autism in the absence of intellectual disability or developmental delay **are no longer eligible for testing**.
2) Patients with isolated behavioural issues **are ineligible for testing**.
3) R377 microarray testing **is not a prerequisite** for R29 testing in patients with moderate/severe/profound intellectual disability in whom a monogenic cause is suspected.

These changes will be implemented by the North East and Yorkshire GLH from September 2024.

The referral information given **must** state how the patient meets the testing criteria:
https://www.england.nhs.uk/wp-content/uploads/2024/07/national-genomic-test-directory-rare-and-inherited-disease-eligibility-criteria-v7.pdf

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

All samples must be accompanied by a laboratory request form, which can be downloaded here:
https://ney-genomics.org.uk/wp-content/uploads/2023/02/411.027-Rare-Disease-Referral-Form-v3.1.pdf

R29 involves whole genome sequencing (WGS). Analysis is most effective as part of a trio (i.e. using samples from the affected individual and both parents). All WGS tests require completion of two forms in addition to the standard laboratory request 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

Turnaround times

Standard reporting targets set by NHSE for testing are 42 days for microarray and 84 days for whole genome sequencing. However, patients should be aware that results may take significantly longer, due to capacity issues within the laboratories. Backlogs in the analysis of WGS tests mean that routine cases are currently taking over a year to report.

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

Newcastle Genetics Laboratory: Shaun Haigh shaun.haigh@nhs.net
Leeds Genetics Laboratory: James Steer james.steer@nhs.net
Sheffield Genetic Diagnostic Service: Emma Shearing emma.shearing@nhs.net

Yours faithfully,

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