04 March 2026

Dear Colleague,

IMPORTANT: CHANGES TO NATIONAL TEST DIRECTORY FROM APRIL 2026

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/.

From 1st April 2026 the following significant changes will be implemented:

- **Microarray testing as a first-line test will now only be available for patients with clinical features highly suggestive of a specific chromosomal cause.** For example, individuals with features characteristic of Williams syndrome or 22q11 deletion syndrome. The chromosomal disorder suspected should be specified on the test request form.

- **Patients with non-syndromic mild developmental delay, mild intellectual disability or isolated Neurodiversity are not eligible for genomic testing.**

- Paediatric patients with **unexplained moderate/severe/profound global developmental delay** or **unexplained moderate/severe/profound intellectual disability** or **congenital malformations** and/or **dysmorphism** are eligible for **whole genome sequencing (R27: Paediatric disorders).** 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.

- **R29: Intellectual disability and R59: Epilepsy WGS (virtual) panels have been retired, and all patients will be analysed using the larger and more comprehensive R27: Paediatric disorders panel**

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.

In addition, several changes have been made to eligibility criteria for multiple indications. Please refer to the latest version of the National Test Directory V9 from 1st April here: https://www.england.nhs.uk/publication/national-genomic-test-directories/ to ensure your patient is eligible before sending for testing.

These changes will be implemented by the North East and Yorkshire GLH from 1st April 2026.

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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