NW Genomic Laboratory Hub (Manchester Site)
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9th April 2026

USER COMMUNICATION: CHANGES TO THE NHS ENGLAND COMMISSIONED RARE AND INHERITED DISEASE NATIONAL GENOMIC TESTING DIRECTORY

Dear colleagues,

We would like to inform you that a new version of the Rare and Inherited Disease National Genomic Test Directory (version 9) was published on 8th April 2026.

We hope that you received our previous communication, which outlined the proposed changes. A final summary of the changes by specialty, provided by NHS England, is included in the appendix overleaf. Please note that this includes inherited cancer and a small number of minor updates to the proposals we previously shared.

The test directory details the NHS funded tests available and details the eligibility criteria to access testing. We recommend that you refer to the full Test Directory (link above) for comprehensive details of eligibility criteria.

Testing will not be initiated for samples received on or after 1st May 2026 if they no longer meet the v9 eligibility criteria or if the relevant tests are no longer available. In such cases, DNA will be stored and a DNA storage report will be issued to the referring clinician, where appropriate.

To minimise delays in testing, please ensure that test request forms include clear patient details, the appropriate Test Directory indication code(s), relevant clinical information compliant with the NGTD eligibility criteria and the expected clinical utility of the result. We also ask that you provide a contact telephone number or email address so that we may contact you should any queries arise.

All whole genome sequencing (WGS) requests must be accompanied by a WGS specific Test Order Form (TOF) and Record of Discussion (RoD) form (one RoD form per individual). The completed TOF and RoD forms should be submitted to mft.nwglhdnalab@nhs.net.

Please note that an updated version of the WGS Rare Disease TOF is due to be published on the **NHS England website** on 6th May 2026.

For clinicians using the Manchester HIVE test ordering system, it has been updated to reflect the new Test Directory. Please report any issues encountered to your HIVE leads.

If you have any questions regarding these Test Directory changes, please contact the laboratory at mft.genomics@nhs.net, including 'NGTD April update' in the subject line. Additional information about ordering genomic testing and links to genomic testing forms are available on our website.

Yours faithfully,

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APPENDIX

Summary of changes to eligibility criteria in the National Genomics Test Directory – Rare & Inherited Disease version v9 published 8th April 2026

R code	Clinical Indication name	Summary of Changes
Cardiology		
R137	Congenital heart disease – microarray	Added to eligibility criteria: Complex congenital heart disease in a neonate/infant undergoing corrective surgery, in whom other syndromic features may not yet be apparent AND a syndromic diagnosis may impact surgical decision making. Test requests must be approved by clinical genetics to proceed. Amendments to the criteria already present by stating congenital heart disease refers to complex CHD and cleft palate and / or disorder of calcium homeostasis should be present. Provided an example of non-syndromic CHD in the exclusion criteria: isolated VSD/ASD
R125	Thoracic aortic aneurysm or dissection	Removed criterion points 3. 4. and 7. Changed threshold for z score in the definition of thoracic aortic aneurysm in children, from >2 to >3.
R132	Dilated and arrhythmogenic cardiomyopathy	Corrected an error by removing criterion 1c.
R135	Paediatric or syndromic cardiomyopathy	Corrected criterion 2 for clarity.
R140	Elastin-related phenotypes	Added to the overlapping clinical indications that patients must meet the criteria for the overlapping clinical indications if these are felt a better test for their patients.
Multi specialty		
R441	Unexplained death in infancy and sudden unexplained death in childhood	Provided more information in the testing criteria and "where in the pathway" to link with the Joint Agency Response. Clarification that DNA from both biological parents is preferential and to contact the laboratory if this is not possible. Exclusion criteria have been added: Testing should **not** be performed where the likelihood of a monogenic disorder is low for example where the available evidence supports a non-genetic cause of death (e.g. severe infection, suspicious death/homicide investigation). Requesting specialties: Added that these should be in liaison with the designated doctor for child deaths.
Developmental disorders		
R26	Likely common aneuploidy	Removed R297 from the overlapping Clinical Indications as R297 has been retired – see separate entry for R297 for details.

R code	Clinical Indication name	Summary of Changes
R27	Paediatric disorders	Added criteria for unexplained epilepsy as the separate clinical indication for Epilepsy (R59) has been retired. Added to additional text in the overlapping clinical indications for R14. Added psychiatry as a requesting specialty as separate Clinical Indication for Intellectual Disability has been retired.
R29	Intellectual disability (WGS)	Clinical indication retired. Patients to be tested under R27 Paediatric disorders where patients meet the criteria for R27.
R377	Intellectual disability – microarray only	Clinical indication retired. Patients to be tested under R27 Paediatric disorders where patients meet the criteria for R27.
R48	Prader-Willi syndrome	Additional criteria added to promote appropriate referrals.
R69	Hypotonic infant	Addition of R452 Silver Russel Syndrome / Temple syndrome to overlapping CIs. Removal of confirmatory STR CITT R69.6 as the generic code for confirmation test should be used instead.
Endocrinology		
R314	Ambiguous genitalia	Changed name of Clinical Indication by removing "presenting neonatally". Amended the criteria that includes non neonatal presentations.
R142	Glucokinase-related fasting hyperglycaemia	Added specialist midwifery to the requesting specialties.
R146	Differences in sex development	Removed R297 as an overlapping clinical indication as R297 has been retired (see separate entry) and added R468 Possible sex chromosome aneuploidy or structural rearrangement – Targeted Chromosome Analysis, as new overlapping CI.
R452	Silver Russell Syndrome and Temple Syndrome	Added to the testing criteria, clinical features suggestive or Temple Syndrome for different age groups.
R453	Monogenic short stature	Added table of primary investigations in children referred to secondary and tertiary care with short stature from the British Society for Paediatric Endocrinology and Diabetes recommendations. Added clarifying statement in the overlapping clinical indications for R52 Short stature – SHOX deficiency. To state that this test should be ordered if you suspect SHOX deficiency as SHOX is not include on the panel test for R453 due to technical limitations.
R267	Temple syndrome – maternal uniparental disomy 14	Retired Clinical Indication. Patients to be tested under R452 Silver Russell Syndrome and Temple Syndrome, where they meet the testing criteria.
R180	Congenital adrenal hyperplasia diagnostic test	Added an additional criterion in the testing criteria: Female adult with raised 17-OHP and at least one of the following: hirsutism, frontal baldness, delayed menarche or infertility.

R code	Clinical Indication name	Summary of Changes
R388	Linkage testing for congenital adrenal hyperplasia	Retired Clinical Indication as the generic code for linkage testing should be used instead, R409 Linkage testing for recognisable Mendelian disorders.
R293	Albright hereditary osteodystrophy, pseudohypoparathyroidism pseudopseudohypoparathyroidism, acrodysostosis and osteoma cutis	Added Paediatrics as a requesting specialty
R154	Hypophosphataemia or rickets	Criteria added to the testing criteria for patients with low ALP that requires one of the following: • early dental loss OR • rickets-like changes on X-ray OR • chronic musculoskeletal pain OR • atypical femoral fractures OR • poor healing fractures
R223	Inherited phaeochromocytoma and paraganglioma excluding NF1	Removed from the 6th testing criteria "renal cell cancer (any age)".
R158	Lipodystrophy	Amended the Clinical Indication name FROM Lipodystrophy – childhood onset TO: Severe insulin resistance and lipodystrophy syndromes. Amended testing criteria to remove need for childhood onset and added in severe insulin resistance. Additional genes added to the panel to align with these changes.
Fetal		
R445	Common aneuploidy testing - NIPT	Amended Clinical Indication name FROM: Common aneuploidy testing – NIPT TO: T21, T18, T13 aneuploidy testing – NIPT (previous history). Amended testing criteria to specify the three aneuploidy syndromes that are tested for. Made amendments to criteria so that R445 and R470 (new CI) are aligned.
R470	T21, T18, T13 aneuploidy testing – NIPT NHS Fetal Anomaly Screening Programme (FASP)	New Clinical Indication.
R318	Recurrent miscarriage with products of conception available for testing	Removed R297 as an overlapping clinical indication as R297 has been retired (see separate entry) and added R464 Recurrent miscarriage where products of conception are not available for testing - parental karyotype, as a new overlapping Clinical Indication.
R22	Fetus with a likely chromosomal abnormality	Added to the criterion for death or stillbirth from 24 weeks to clarify this is referring to intrauterine death.
R304	NIPD for cystic fibrosis - haplotype testing	Error correction, removal of "where parents are consanguineous" from criterion 1.
R306	NIPD for Apert syndrome - variant testing	Removed from criteria testing where there has been a previous pregnancy with confirmed Apert syndrome.

R code	Clinical Indication name	Summary of Changes
R307	NIPD for Crouzon syndrome with acanthosis nigricans - variant testing	Removed from criteria testing where there has been a previous pregnancy with confirmed Crouzon syndrome.
R308	NIPD for FGFR2-related craniosynostosis syndromes - variant testing	Removed from criteria testing where there has been a previous pregnancy with confirmed FGFR2 related craniosynostosis.
R309	NIPD for FGFR3-related skeletal dysplasias - variant testing	Removed from criteria testing where there has been a previous pregnancy with confirmed FGFR3 related skeletal disorder.
R433	NIPD for monogenic diabetes, subtype glucokinase	Amended Clinical Indication name FROM: NIPD for monogenic diabetes, subtype glucokinase TO: Monogenic diabetes, subtype glucokinase - NIPT
Gastrohepatology		
R177	Hirschsprung disease	Retired Clinical Indication. Patients to be tested under R438 Paediatric pseudo-obstruction syndrome, where they meet the testing criteria.
R438	Paediatric pseudo-obstruction syndrome	Added to "where in pathway": "This test should be used where testing for Hirschsprung disease is required".
Inherited cancer		
R208	Inherited breast cancer and ovarian cancer	Error correction: removal of need to reach criteria 2 in the criterion 4b.
R210	Inherited MMR deficiency (Lynch syndrome)	Amended the Lynch related cancers for sebaceous adenomas and carcinoma so that it now reads: at least two sebaceous adenomas, one or more sebaceous carcinoma. A single sebaceous adenoma does not constitute a Lynch-related cancer. Amendment to the sentence above the table of associated tests to now read: Please note that not all the associated tests will be undertaken in every case. The clinical presentation will indicate the tests that are necessary.
R211	Inherited polyposis and early onset colorectal cancer - germline test	Gynaecology added to requesting specialties.
R414	APC Associated Polyposis	Additional 8th criteria added to the testing criteria and change to criteria 7. Removal of R359 Childhood solid tumour panel, from overlapping clinical indications as R359 has been retired, see separate entry for the details. Added dermatology to requesting specialties.
R215	Hereditary diffuse gastric cancer	Amendments to criteria 1e, 1g and 1h.
R359	Childhood solid tumours	Retired Clinical Indication. Two new Clinical Indications replace R359. These are R456 and R457. Testing should only be ordered under codes R456 and R457 where patients meet the testing criteria for these new Clinical Indications.
R220	Wilms tumour with features suggestive of predisposition	Retired Clinical Indication. Patients should be tested under the new Clinical Indication R456.
R358	Familial rhabdoid tumours	Retired Clinical Indication. Patients should be tested under the new Clinical Indication R456.

R code	Clinical Indication name	Summary of Changes
R456	Embryonal tumour of possible germline origin	New Clinical Indication.
R457	Sarcoma of possible germline origin	New Clinical Indication.
R224	Inherited renal cancer	Amendments to criteria 2. and 6.
R254	Familial melanoma	Various changes to the testing criteria.
R365	Fumarate hydratase-related tumour syndromes	Amendment to criteria b.
R444	NICE approved PARP inhibitor treatment	Amendments to criteria R444.1 for breast cancer to align with updated CDF criteria.
Metabolic		
R450	Diagnostic testing for Isovaleric acidaemia	Confirming in the criteria that genetic testing is only where required as part of the diagnostic testing pathway. Removal of the sentence: In the case of isovaleric acidaemia, this means that testing is almost exclusively used at those in whom biochemical results indicate a likely pseudodeficiency allele is present.
Mitochondrial		
R315	POLG-related disorder	Removal of R59 as overlapping clinical indication as R59 is retired. Addition of R27 as an overlapping Clinical Indication with note that this should be used, or other relevant broader test, where clinical features are not strongly suggestive of POLG-related disorder and a broader differential diagnosis is under consideration.
Mosaic and structural chromosomal disorders		
R297	Possible structural chromosomal rearrangement – karyotype or Targeted Chromosome Analysis	Clinical Indication retired and replaced with separate Clinical Indications for each of the clinical scenarios where R297 would have been requested. The new Clinical Indications are; R463, R464, R465, R466, R467 and R468
R463	Cytogenetic characterisation of a genomic abnormality – Karyotype or Targeted Chromosome Analysis	New Clinical Indication
R464	Recurrent miscarriage, products of conception not available – parental karyotype	New Clinical Indication
R465	Familial cytogenetic rearrangement - Karyotype or Targeted Chromosome Analysis	New Clinical Indication
R466	Unexplained infertility - karyotype	New Clinical Indication
R467	Gamete donors - karyotype	New Clinical Indication
R468	Possible sex chromosome aneuploidy or structural rearrangement – Targeted Chromosome Analysis	New Clinical Indication
R298	Possible structural or mosaic chromosomal abnormality - FISH	Removal of R297 as an overlapping Clinical Indication as this has been retired. Addition of two new Clinical Indications in overlapping Clinical Indications; R463 and R465.

R code	Clinical Indication name	Summary of Changes
Musculoskeletal		
R104	Skeletal dysplasia	Addition to the testing criteria to clarify that isolated short stature, without evidence of an underlying abnormality of the bones is not appropriate to test via this indication. Other overlapping indications may be more appropriate in these cases. Overlapping Clinical Indications added: R453 Monogenic short stature R52 Short stature – SHOX deficiency Requesting specialties added: Paediatrics Endocrinology
R415	Cleidocranial dysplasia (CCD)	Clinical indication retired. Patients to have R104 testing instead, where they meet the testing criteria.
R101	Ehler Danlos syndrome with a likely monogenic cause	Additional testing criteria has been added and states that testing should NOT be used to exclude a diagnosis. Overlapping clinical indication added: R125 Thoracic aortic aneurysm or dissection panel includes the COL1A1, COL3A1, COL5A1, COL5A2, PLOD1 and FKBP14 genes and may be a better option for patients presenting with aortic/arterial dilatation or rupture without additional features of EDS. Clarification may be sought from the highly specialised National Ehlers Danlos services: lnwh-tr.edslondonoffice@nhs.net or eds.sheffield@nhs.net
R102	Osteogenesis imperfecta	Addition to the testing criteria: Referrals where non-accidental injury is suspected should be discussed with the OI Highly Specialised Service before requesting genomic testing
R284	Van der Woude syndrome	Clinical indication retired. Patients to have R27 testing instead, where they meet the testing criteria.
Neurology		
R471	Neurodegenerative Disorders, adult onset – Prenatal Exclusion Testing	New Clinical Indication
R55.5 R56.4 R57.6 R60.4 R61.5 R78.6 R381.4 R84.5	Confirmatory STR testing for various neurology clinical indications	Retired Clinical Indication Test Types. The multi purpose test R443 Confirmation test, should be used instead.
R57	Childhood onset dystonia, chorea or related movement disorder	Removal of R29 Intellectual disability as an overlapping Clinical Indication.
R58	Adult onset neurodegenerative disorder	Retired Clinical Indication. Testing for the four conditions previously all grouped into this single Clinical Indication have now been separated out into four new Clinical Indications: • R458 Young onset or familial dementia,

R code	Clinical Indication name	Summary of Changes
		• R459 Young onset or complex Parkinson disease, • R460 Amyotrophic lateral sclerosis, • R461 Cerebral amyloid angiopathy.
R458	Young onset or familial dementia	New Clinical Indication, replacing R58.
R459	Young onset or complex Parkinson disease,	New Clinical Indication, replacing R58.
R460	Amyotrophic lateral sclerosis	New Clinical Indication, replacing R58.
R461	Cerebral amyloid angiopathy	New Clinical Indication, replacing R58.
R59	Early onset or syndromic epilepsy	Retired Clinical Indication. Patients who meet the testing criteria for R27 should have this testing instead. The genes for the R59 panel are already on R27.
R378	Linkage testing for Duchenne or Becker muscular dystrophy	Retired Clinical Indication. The multi purpose test R409 Linkage testing for recognisable Mendelian disorders, should be used instead.
R82	Limb girdle muscular dystrophies, myofibrillar myopathies and distal myopathies	Addition to where in the pathway that provides information about the Highly Specialised Service.
R85	Holoprosencephaly	Change in Clinical Indication name: FROM: Holoprosencephaly - NOT chromosomal TO: Holoprosencephaly Amendments to the testing criteria and to where in the pathway. Overlapping Clinical Indications added: • R27 Paediatric disorders or R89 Ultra-rare and atypical monogenic disorders tests should be used in individuals with congenital malformations, dysmorphism or other complex or syndromic presentations • R26 Likely common aneuploidy • R28 Congenital malformation and dysmorphism syndromes – microarray
R221	Familial tumours of the nervous system	Amendments to the testing criteria.
R222	Neurofibromatosis 1	Additional testing criteria: • Axillary/inguinal freckling • Malignant Peripheral Nerve Sheath Tumour
R337	CADASIL	Removal of R58 from overlapping Clinical Indications. Addition of R461 Cerebral amyloid angiopathy as an overlapping Clinical Indication where a broader differential diagnosis is under consideration.
Respiratory		
R190	Pneumothorax – familial	Testing criteria amendment: FROM Primary spontaneous pneumothorax with no identifiable cause, AND one of: i) a first degree relative with primary spontaneous pneumothorax, OR ii) Characteristic radiological features of Birt-Hogg-Dubé syndrome on chest imaging TO

R code	Clinical Indication name	Summary of Changes
		1. Primary spontaneous pneumothorax with no identifiable cause AND a first degree relative with primary spontaneous pneumothorax, OR 2. Characteristic radiological features of Birt-Hogg-Dubé syndrome on chest imaging
R192	Surfactant deficiency	Retired Clinical Indication. Patients should be tested for R462 Childhood interstitial lung disease, where they meet the testing criteria.
R462	Childhood interstitial lung disease	New Clinical Indication.
Dermatology		
R239	Incontinentia pigmenti	Addition to the testing criteria to reference IKBKG- related immunodeficiency. Addition to requesting specialties: Immunology (+/- Paediatrics).
Ultra rare and atypical monogenic disorders		
R89	Ultra-rare and atypical monogenic disorders	Addition to the testing criteria: Requests that include any panel for inherited cancers must be taken to the UKCGG/CanGene-CanVar National Multidisciplinary team meeting and receive delegate approval to proceed.
Multi purpose tests		
R370	Validation test	Change of name: FROM: Validation test TO: Validation of unaccredited findings Additional clarification is provided in the testing criteria as to when this test should be used.
R443	Confirmation test	Expansion of testing criteria to clarify when this Clinical Indication should be used. Change in requesting specialties so that only genomics laboratory is the requestor.
R447	Diagnostic discovery – validation/confirmation findings	Change of name: FROM: Diagnostic discovery - validation/confirmation of findings TO: Validation of WGS Diagnostic discovery findings