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LAB 60

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UKAS policy on accreditation of medical laboratories performing genomic tests

Contents

Changes since last edition

This is the first edition of this publication.

Definitions

- FFPE tissue: Formalin-fixed paraffin-embedded tissue
- Variant: incorporates single nucleotide variants (SNV), copy number variants (CNV), insertions and deletions (indels), Structural rearrangement. Each variant type will be assessed as a separate entity by UKAS. For example, accreditation for SNVs will not automatically imply accreditation for other variant types.

1. Introduction

1.1 Genomic testing services in the UK are provided via numerous different service delivery models, ranging from traditional pathology disciplines (e.g. histopathology/microbiology) offering a small scope of molecular testing on all-in-one/ cartridge-based platforms, through to Genomic Laboratory Hubs offering large, complex, ever-changing testing scopes.

1.2 Other laboratory services (for example Histocompatibility and Immunogenetics (H&I) services and pathogen genomics services) use a lot of the same technical methods as human genomic services; the accreditation principles described in this policy can be applied to any genomics discipline wanting a flexible scope of accreditation.

1.3 This policy details UKAS's risk-based approach to accreditation of genomic services, to ensure accreditation remains appropriate, timely and responsive to user needs. A number of genomic testing methods are listed as examples in this policy; not all current and future methods can be listed, but the principles outlined in this policy are able to be applied to all methods and scenarios.

1.4 UKAS accreditation is based on the demonstration of competence and conformity to the relevant accreditation standard. For medical laboratory services, this includes both the technical competence to perform tests, and the competence to interpret/report those results and offer appropriate clinical advice to users and/or patients.

1.5 Due to the frequency of changes, and the use and application of the same equipment, analytical techniques/measurement principles and technical competence to identify different variants or disorders, many of which are unidentified at the start of the diagnostic process, there may be a benefit for genomics services to be accredited using a flexible scope approach. Flexible scope accreditation is a routine UKAS process, detailed in UKAS publication GEN 4 *UKAS policy and general guidance for the implementation and management of flexible scopes of accreditation*; this policy has been written to supplement GEN 4 and provide support and guidance to genomic services considering applying for flexible scope of accreditation.

The level of flexibility and the boundaries associated with that flexibility shall be defined by each service, in conjunction with UKAS. In accordance with GEN 4, each laboratory shall keep an up-to-date list of accredited activities, including newly modified, introduced or developed activities. The level of detail required in the definitive list of accredited processes/activities will vary depending on the flexible scope boundaries. For example, for Next Generation Sequencing (NGS), the list will need to include, at a minimum, panel name and version number, and application (disorder).

Note that the scope of accreditation for UKAS customers can, and often does, include a combination of fixed scope and flexible scope.

1.6 In line with UKAS publication GEN 4, the implementation and effectiveness of a medical laboratory's management system in controlling its flexible scope of accreditation will be assessed as part of the normal accreditation assessment cycle. Sufficient time will be allowed at assessments to assess the continuing competence of the medical laboratory service to operate its flexible scope. The assessment effort

required for these assessment activities will depend upon the approach taken, the activities involved, risks associated with activities, and the number and complexity of examination methods included.

1.7 Accreditation will be awarded for a flexible scope where the medical laboratory service demonstrates to UKAS that it is competent to manage a flexible scope approach to accreditation. Where a medical laboratory service is unable to maintain its competence or demonstrate compliance with the requirements (for example, if records of changes made and evidence of their associated validation/verification processes are not available, incomplete, or do not demonstrate compliance with the relevant accreditation standard), the flexible scope will be rescinded. Depending on the circumstances the medical laboratory service may be able to revert to a fixed scope of accreditation (see GEN 4 for more information). In this case the UKAS schedule will be updated to list the individual gene variants/disorders/disorder types which the medical laboratory service has demonstrated that it is competent to identify and, where applicable, interpret. Any additions to the list of individual variants/disorders will require an extension to scope (ETS) application to be submitted. Accreditation will remain fixed (i.e. not flexible) until confidence in the competence of the medical laboratory service to manage the flexible scope process is demonstrated. This will require an extension to scope application from the medical laboratory service.

2. General information

2.1 Some genomics equipment can be used for multiple different analysis techniques or measurement principles. For example, a Rotor-Gene can perform PCR and High-Resolution Melt analysis, and an ABI genetic analyser can be used for fragment analysis, Sanger Sequencing or qualitative and quantitative Realtime PCR. In order for a medical laboratory service to be able to claim accreditation for the different analytical techniques/measurement principles available on a single platform, each technique and principle requires specific accreditation application, successful assessment and inclusion on the UKAS schedule of accreditation.

2.2 It is important to note that the same molecular variant can be detected using different measurement principles/technology. For example, the *JAK2 V617F* variant can be detected using Sanger Sequencing, NGS, Pyrosequencing and ddPCR. In order for a medical laboratory service to be able to claim accreditation for the different measurement principles, each principle requires specific accreditation application, successful assessment and inclusion on the UKAS schedule of accreditation. For example, a change in detection of *JAK2 V617F* from pyrosequencing to ddPCR would require an extension to scope (ETS) application to be submitted and successfully assessed before accreditation could be claimed, unless the ddPCR methodology was already accredited under a flexible scope, with boundaries that would include *JAK2 V617F*.

2.3 When an accredited medical laboratory service introduces new equipment (see 2.4), new measurement principles, and/or new sample types, these will require an extension to scope application to be submitted, unless already included in the boundaries of flexible scope accreditation.

2.4 Like for like changes in instrumentation can be made without submission of an ETS application, as long as the processes for which the medical laboratory service was originally accredited remains unchanged. “Like for like” changes include replacement of equipment of the same make and model, or implementation of additional equipment identical to already accredited equipment (i.e. to increase capacity). This principle may also apply when medical laboratory services install new equipment from the same manufacturer, which has undergone a minor upgrade, depending on risk. The medical laboratory shall consult with their UKAS assessment manager as early as possible during the change control process, to confirm whether or not an ETS application is required. Medical laboratory services can claim accreditation for tests run on new instrumentation which does not require an ETS application as soon as the verification process is successfully completed in accordance with their internal procedures. Services shall inform UKAS of the change, which shall be assessed at the next annual assessment, and may require additional assessment effort.

2.5 Regarding the level of flexibility in the scope of accreditation, and the associated boundaries, it is the responsibility of each accredited medical laboratory service to define their own boundaries of competence. UKAS assesses both the technical competence and the clinical/interpretive competence of each medical laboratory service, to confirm competence within the defined boundaries

For example, a medical laboratory service might be accredited, under a flexible scope, for detection and identification of nucleic acid sequence variants using NGS in haematological disorders. Where common equipment, commercial kits/panels and methods are used, accreditation can be claimed for detection and identification of any nucleic acid sequence variant, using NGS, in any haematological disorder. If the medical laboratory service wants to extend its accreditation to use the same equipment, commercial kits/panels and methods to detect nucleic acid sequence variants in solid tumour disorders, this would require an ETS application to be submitted, as UKAS has no assurance of the medical laboratory service's competence to interpret and provide clinical advice on solid tumour disorders. The ETS assessment would likely focus on the interpretation and clinical advice requirements of the standard, whilst not completely omitting assessment of the technical service.

2.6 Section 3 of this document gives examples of examination methods/measurement principles, and the flexible scope accreditation boundaries which could be considered by medical laboratory services. Where named examples of equipment or analysers are included in this policy, they have been added to support reader clarity and understanding. Lists of equipment or analysers are not exhaustive, nor do they imply UKAS endorsement of the named examples.

3. Guidance - Test method examples

3.1 Cartridge-based (sample to result)

Examples include:

- Biocartis Idylla
- Cepheid GeneXpert

These tests and platforms are straightforward to use, requiring direct addition of patient samples to a cartridge for analysis. The test kits are specific for point variants, fusions, or microsatellite instability (for example). Technical competence to perform the testing, and clinical competence with regard to understanding the clinical utility of the test, interpretation of test results and provision of advisory services are assessed by UKAS. These platforms are unlikely to benefit from a flexible scope unless regular changes (e.g. regular addition of cartridges or targets) to the test repertoire are expected.

3.2 Non-cartridge-based equipment - Initial examination processes

Initial examination processes can include (note this list is not exhaustive):

i. Sample preparation steps e.g. centrifugation to remove plasma, extraction of specific lineages (e.g. CD3+, CB19+), cell harvest and processing, FFPE block cutting, micro/macro dissection

ii. DNA and RNA extraction and reverse transcription of RNA to cDNA for use in down-stream processes

iii. Quantification of DNA/RNA/cDNA (this is generally not reported and used solely for the purposes of QC prior to downstream processes)

iv. Cell culture for Karyotyping

Although these processes do not produce a reportable result, they are critical to the testing process and shall be captured in the accreditation assessment and records. These steps shall appear on the schedule of accreditation for the medical laboratory service performing them, including whether they are manual, automated, or both.

Commercial kits used for DNA/RNA extraction or preparatory steps are validated by the manufacturers to be used on specific sample types and with specific equipment, therefore a fixed scope of accreditation is usually appropriate.

If a medical laboratory service is accredited for DNA/RNA extraction from a specific sample type using a specific kit and specific equipment, then wishes to extend their accreditation to include DNA/RNA extraction from a different sample type, using the accredited kit and equipment, and the sample type has been validated by the manufacturer (i.e. is listed in the Instructions for Use), this will not require an Extension to Scope application, but the service shall notify their UKAS assessment manager of the change as early as possible. The verification of DNA/RNA extraction from the new sample type will be assessed by UKAS, either as extra assessment effort added to the annual surveillance/reassessment visit, or as a stand-alone chargeable extra assessment. The accredited service can only claim accreditation for this change when the assessment is completed, and the published UKAS schedule of accreditation has been updated.

Where a new DNA/RNA extraction kit is introduced, with no change to the chemistry or sample type, but with a minor change to the methodology (e.g. volume change/timings differences between Maxi and Mini kits), this will not require an Extension to Scope application, but the service shall notify their UKAS assessment manager of the change. Medical laboratory services can claim accreditation for the new kit as soon as the verification process is complete and signed off; the medical laboratory shall inform their UKAS assessment manager when this is completed, so that the UKAS Schedule of Accreditation can be

updated. The new kit will be assessed at the next annual assessment and may require additional assessment effort.

For equipment and methods which are multifaceted (i.e. can be used for DNA and RNA extraction), services which are accredited for DNA extraction on a specific platform shall apply for an extension to scope if they want to include RNA extraction in their accredited scope (and vice versa).

If a service wishes to extend its accreditation to include sample types not validated by the manufacturer, new kits and/or new equipment, these will all require submission of an Extension to Scope application.

If a service regularly changes its sample types, kits and/or equipment, a flexible scope of accreditation for these initial examination processes may be suitable.

3.3 Non-cartridge-based equipment - Analysis & interpretation

3.3.1 Sanger sequencing

Sanger sequencing methodology involves detection and identification of nucleic acid sequence variants (e.g. SNVs, small indels and breakpoints) across a range of disorders, with common equipment, methods and technical competence.

UKAS will assess the competence of a medical laboratory service to perform Sanger sequencing, including primer design and optimisation, and result interpretation/analysis, using the same equipment, kits/ in-house methodology (with optimisation of extension) and chemistry (same Mg, Taq, dNTPs), and analysis software across a range of variant types and disorders, as applicable to their testing and clinical advisory service. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited for detection of any variant type in specific disorder types (e.g. haematological malignancy, somatic solid tumour or germline testing), including confirmatory testing whereby primer optimisation (as above) is required, using the accredited equipment, kits/in-house methodology and chemistry, and versioned software. If a medical laboratory service cannot demonstrate competence and compliance across this range, individual variants/disorders will be assessed and listed on the UKAS schedule as a fixed scope.

Due to the non-targeted nature of confirmatory testing and the need for primer design and optimisation with method verification/validation, this can only be accredited as a flexible scope. If a medical laboratory service cannot demonstrate competence to manage a flexible scope, confirmatory testing will not be accredited.

If a medical laboratory service wishes to extend its accreditation to include different in-house methodology/chemistry, equipment, kits, or targets out with its accredited scope of competence for clinical advisory services, an extension to scope application will be required.

3.3.2 Pyrosequencing

Most medical laboratory services utilise pyrosequencing for two purposes – sequencing or determining methylation status (e.g. in MGMT).

For use in sequencing the accreditation approach as discussed above for Sanger sequencing applies.

Methylation is usually either kit-based or in-house method based. The same assessment and accreditation principles apply as for sequencing assessment and accreditation. Flexible scope accreditation may be beneficial if the same methodology, kit and/or equipment is used to detect methylation status of different genes, in line with manufacturer's validation, and there are frequent changes to the targets being assessed for methylation status by the accredited medical laboratory service.

3.3.3 Fragment length analysis

The same assessment and accreditation principles apply as for sequencing assessment and accreditation i.e. UKAS will assess a medical laboratory service's competence to perform fragment length analysis, including result interpretation/analysis, using the same equipment, kits/in-house methodology and chemistry, and analysis software across a range of variant types and disorders, as applicable to their testing and clinical advisory service. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited for detection of any variant type (including CNV or methylation status by MLPA kit) in specific rare diseases, cancers or acquired disorders using the accredited equipment, kits/in-house methodology and chemistry, and versioned software. If a medical laboratory service cannot demonstrate competence and compliance across this range, individual variants/disorders will be assessed and listed on the UKAS schedule.

Fragment length analysis involves a number of different methodologies, kits and chemistries. Several examples are listed below. Where a specific example is not listed, the principles of this policy still apply.

3.3.3.1 PCR with resolution by Gel Electrophoresis

The assessment and accreditation principles apply as listed above. If methodology and chemistry are identical for analysis for a regularly changing variant type (e.g. SNV, indel, fusion) then flexible scope accreditation might be appropriate. However, if new methodology (chemistry, PCR conditions, etc) is introduced, then an ETS is required to add targets to scope.

3.3.3.2 Multiplex Ligation-dependent Probe Amplification (MLPA) for detection of CNVs and large rearrangements

(MLPA) methodology involves determination of gene copy number across a range of variants/disorders, using commercial kits with common equipment (normally genetic analysers with fragment analysis capability). The assessment and accreditation principles listed in 3.3.3 apply.

3.3.3.3 Methylation Specific Multiplex Ligation-dependent Probe Amplification (MS-MLPA)

The same principles apply for accreditation of MS-MLPA as accreditation of MLPA for the detection of CNVs and large rearrangements.

If a medical laboratory service is accredited for MLPA for the detection of CNVs and large rearrangements, this does provide assurance that that the service is competent to perform, and be accredited for, MS-MLPA. An extension to scope application will be needed if the service is only accredited for the detection of CNVs and large rearrangements but wants to also be accredited for MS-MLPA, and vice versa.

3.3.3.4 Quantitative Fluorescence PCR (QF-PCR)

This method is used to determine change in gene copy numbers (including mosaicism) for a range of disorders in prenatal or postnatal settings (e.g. Uniparental Disomies (UPD), trisomy, triploidy, mosaicism, maternal cell contamination) using commercial equipment and software.

The assessment and accreditation principles listed in 3.3.3 apply. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited for

determination of copy number changes in named types of disorders (e.g. prenatal, post-natal). If a medical laboratory service cannot demonstrate competence and compliance across this range, individual disorders will be listed on the UKAS schedule.

3.3.3.5 Capillary Electrophoresis - fragment size

Commercial kits

There is a vast range of kits being used by medical laboratory services for the purposes of PCR and resolution by genetic analyser.

The assessment and accreditation principles listed in 3.3.3 apply. Each kit, and variant type it detects, shall be assessed by UKAS and documented on the schedule of accreditation. Examples include detection of fragment length size, deletions, known variants, repeat expansions, or linkage markers, CF analysis, triplet repeats, microsatellite instability, Short Tandem Repeats (STRs, including chimerism analysis).

For kit-based tests resolved using capillary electrophoresis, UKAS will assess the competence of the medical laboratory service to use these kits and analyse outcomes. If a medical laboratory service demonstrates competence and compliance, it may be accredited for detection of any variant types or specific variants offered by the kit in any disorder using the accredited equipment, kits, methodology and chemistry, and versioned software (flexible scope). If a medical laboratory service cannot demonstrate competence and compliance across this range, competence to detect specific variants (e.g. JAK2 V617F, STR loci for chimerism analysis) will be assessed and listed on the UKAS schedule.

If a medical laboratory service changes a kit (and therefore chemistry) or adds a new kit with new target type, and wishes to claim accreditation for the new kit, an ETS application and assessment will be required.

In-house methodology (single/known target)

Most in-house methods are likely to be validated for a single target type, which will be listed on the UKAS schedule of accreditation.

As most in-house methods are likely to be validated for a single target type, it is likely an ETS application will be needed if a medical laboratory service changes their in-house methodology, and that there will be little benefit in including this method in a flexible scope of accreditation.

In-house methodology (confirmatory testing)

Due to the non-targeted nature of confirmatory testing and the need for primer design and optimisation with method verification/validation, this can only be accredited as a flexible scope. If a medical laboratory service cannot demonstrate competence to manage a flexible scope, confirmatory testing will not be accredited.

3.3.3.6 Southern Blotting for detection of large gene rearrangements

UKAS will assess the competence of a medical laboratory service to perform southern blotting, including primer design and interpretation/analysis, using the same equipment, kits/ in-house methodology and chemistry and analysis software, across a range of large gene rearrangements, as applicable to their service and clinical advisory services. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited

for detection of any rearrangement, using the accredited equipment, kits/in-house methodology and chemistry and versioned software. If a medical laboratory service cannot demonstrate competence and compliance across this range, individual variants/disorders will be assessed and listed on the UKAS schedule.

If a medical laboratory service wishes to extend its accreditation to include different in-house methodology/chemistry, equipment, kits, or targets out with its accredited scope of competence for clinical advisory services, an extension to scope application will be required.

3.3.3.7 Qualitative Real Time PCR and Qualitative Reverse Transcriptase PCR (RTPCR)

For example, *JAK2 V617F* SNP detection, *BCR-ABL1* fusion detection.

Commercial kits

For kit-based tests resolved using real-time analysers:

UKAS will assess the competence of the medical laboratory service to use these kits and analyse outcomes. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited for detection of any pathological variant relevant to the service's medical competency/pathological discipline, using the accredited equipment, kits/methodology and chemistry and versioned software. If a medical laboratory service cannot demonstrate competence and compliance across this range, individual variants/disorders will be assessed and listed on the UKAS schedule.

In-house methodology

Most in-house methods are likely to be validated for a single known target or target type which will be listed on the UKAS Schedule of Accreditation. UKAS will assess the competence of a medical laboratory service to perform the in-house method, including primer design, interpretation/analysis, and clinical advisory services.

As most in-house methods are likely to be validated for a single target type, it is likely that any changes will require different primer conditions and different methodology, meaning an ETS application will be needed if a medical laboratory service wants to include the new method in their accredited scope, and that there will be little benefit in including this method in a flexible scope of accreditation.

3.3.3.8 Droplet Digital PCR

The assessment and accreditation approach as detailed for Qualitative Real Time PCR and Qualitative Reverse Transcriptase PCR (RTPCR) applies where known targets are being identified.

Due to the non-targeted nature of confirmatory testing and the need for primer design and optimisation with method verification/validation, it can only be accredited as a flexible scope. If a medical laboratory service cannot demonstrate competence to manage a flexible scope, confirmatory testing will not be accredited.

3.3.3.9 High resolution melt analysis/melt analysis

The assessment and accreditation approach as detailed for Qualitative Real Time PCR and Qualitative Reverse Transcriptase PCR (RTPCR) applies.

3.3.3.10 Quantitative Real Time PCR for purposes of Minimal Residual Disease detection (MRD)

Commercial kits

For kit-based quantitative tests using real-time analysers:

UKAS will assess the competence of the medical laboratory service to use these kits and analyse outcomes. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited for detection of any pathological variant relevant to the service's medical competency/pathological discipline, using the accredited equipment, kits/methodology and chemistry and versioned software. If a medical laboratory service cannot demonstrate competence and compliance across this range, individual gene targets will be assessed and listed on the UKAS schedule. This will require additional assessment effort and associated charges.

In-house methods

- Single known targets e.g. *BCR::ABL1 (e1a2, e13a2, e14a2)*, *PML::RARA (bcr1, bcr 3)* gene fusions

  Most in-house methods are likely to be validated for a single known target or target type. UKAS will assess the competence of a medical laboratory service to perform the in-house method, including primer design, interpretation/analysis, and clinical advisory services.

  Flexible scope accreditation in this area may only be beneficial if changes are being made frequently.

- Patient Specific targets (e.g. rare *BCR::ABL1* transcripts / TCR gene rearrangements MRD)

  Due to the patient-specific nature of this MRD testing and the need for individual primer design and optimisation with method verification/validation, this can only be accredited as a flexible scope. If a medical laboratory service cannot demonstrate competence to manage a flexible scope, patient-specific MRD testing will not be accredited.

3.3.4 Panel based Next Generation Sequencing (NGS)

Next Generation Sequencing (NGS) methodology involves detection and identification of nucleic acid sequence variants (e.g. SNVs, small indels and breakpoints) across a range of types of acquired disorders and rare diseases, with common equipment, kits/panels, methods and technical competence.

UKAS will assess the competence of a medical laboratory service to perform NGS, including interpretation/analysis, using the same equipment, library preparation and enrichment methodology, and software/bioinformatics pipeline across a range of variants and disorders, as applicable to their service. If a medical laboratory service demonstrates competence and compliance (including panel design and verification), it will be flexibly accredited for detection of any variant type in named types of disorders (for example, rare disease, endocrine disease, cardiac disease), using the accredited equipment, kits/panels and software/bioinformatics pipeline.

If a medical laboratory service cannot demonstrate competence and compliance across this range, individual variants/disorders will be assessed and listed on the UKAS schedule. Library preparation, enrichment method and sequencer shall all be documented on the Schedule of Accreditation for fixed scopes of accreditation.

If a medical laboratory service wishes to extend its accreditation to include different in-house methodology/chemistry, equipment, kits, or targets out with its accredited scope of competence for clinical advisory services, an extension to scope application will be required.

3.3.5 Arrays

Array methodologies involve detection and identification of SNPs, changes to gene copy number alterations or methylation status across a range of types of disorders (e.g. prenatal, haematological malignancy, somatic solid tumour or germline testing) with common equipment, kits, methods and technical competence.

UKAS will assess the competence of a medical laboratory service to perform array testing, including interpretation/analysis, using the same kits/CHIPS and equipment across a range of types of disorder, as applicable to their service. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited for determination of copy number changes/methylation status in named types of disorders. If a medical laboratory service cannot demonstrate competence and compliance across this range, individual disorders will be listed on the UKAS schedule.

If a medical laboratory service wishes to extend its accreditation to include different in-house methodology/chemistry, equipment, kits, or targets out with its accredited scope of competence for clinical advisory services, an extension to scope application will be required.

3.3.6 G-band chromosome analysis/Karyotyping

G-band chromosome analysis/Karyotyping methodologies involve detection and identification of chromosomal aberrations or rearrangements across a range of test specialties (as per, for example the NHSE test directory) and sample types (with common equipment, kits, methods and technical competence.

There is minimal benefit to flexible scope accreditation in cell culture and harvesting techniques and equipment.

UKAS will assess the competence of a medical laboratory service to perform G-band chromosome analysis/Karyotyping, including interpretation/analysis, using the same kits and equipment across a range of sample types and test specialties, as applicable to their service. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited, for example, for determination of copy number changes in named types of test specialties e.g. reproductive disorder, cardiac, immunology, prenatal *etc.* relevant to their technical and advisory competency. If a medical laboratory service cannot demonstrate competence and compliance across this range, individual disorders will be listed on the UKAS schedule.

If a medical laboratory service wishes to extend its accreditation to include different in-house methodology/chemistry, equipment, kits, or targets out with its accredited scope of competence for clinical advisory services, an extension to scope application will be required.

3.3.7 Fluorescence in situ hybridisation (FISH)

FISH testing involves detection and identification of chromosomal aberrations or rearrangements across a range of sample types (e.g. prenatal blood, postnatal blood, products of conception, bone marrow, solid tumour) common equipment, kits, methods and technical competence.

UKAS will assess the competence of a medical laboratory service to perform FISH testing, including knowledge of probe-type application (break apart, fusion) and interpretation/analysis, using the same kits, probe types and equipment across a range of types of disorder, as applicable to their service. If a medical laboratory service demonstrates competence and compliance, it will be flexibly accredited for determination of variant detection using these defined probe types. If a medical laboratory service cannot

demonstrate competence and compliance across this range, individual targets, probes and disorders will be listed on the UKAS Schedule.

FISH is usually performed on either Formalin-Fixed Paraffin-Embedded (FFPE) samples or liquid samples. Medical laboratories accredited for FISH on only one of these sample types will need to submit an ETS application if they want to include the other sample type in their accredited scope.

If a medical laboratory service wishes to extend its accreditation to include different in-house methodology/chemistry, equipment, kits, or targets out with its accredited scope of competence for clinical advisory services, an extension to scope application will be required.

3.3.8 Whole Genome Sequencing (WGS)

Medical laboratory services will need to examine the level of change and flexibility required when considering whether to adopt a fixed or flexible scope for the management of WGS activities. How boundaries are set will depend upon whether the laboratory is performing an end-to-end workflow in-house or whether it is spilt across several entities where the lab only holds accreditation for only one part of the workflow (see UKAS publication TPS 71)

Laboratories performing wet work only (for example) might find this can be managed appropriately via fixed scope. If kit/reagent and analysers are unlikely to change over time or the bioinformatics pipeline is locked down and validated for known outcomes, then these sections of the workflow could be managed as a fixed scope. The infrequent changes to reagents, methods or pipelines could be managed using the existing extension to scope process.

However, if the laboratory is seeking flexibility to detect unknown variants, change wet work methodology and routinely make changes to the analysis pipeline or set panels, a flexible scope is likely to be more appropriate.

3.3.9 Bioinformatics pipelines

Each bioinformatics pipelines can be used to interpret different genetic sequences to identify a range of disorders.

UKAS will assess the competence of a medical laboratory service to manage and use commercial pipelines, as well as developing pipelines in-house, where applicable. The UKAS schedule will list the name of each pipeline and indicate whether it is a commercial or in-house-developed pipeline.

The version number of each pipeline will not usually be listed on the UKAS schedule (although there may be some occasions where this is necessary), and version changes to pipelines will not usually require an extension to scope unless the scope of the pipeline significantly changes, for example addition of different variant types, changes to speciation, resolution changes, application to additional organ systems. However, medical laboratory services shall validate/verify any changes between each version. These records will be assessed by UKAS during the annual assessments.

Other externally controlled software is used by some genomics laboratories (e.g. the Heidelberg Classifier software for classification of brain tumours). Accreditation of these pieces of software will be dealt with on a case-by-case basis, but the general principles of accreditation and ISO 15189 always apply, in that the accredited medical laboratory service shall validate and/or verify the functioning of the software and be able to demonstrate to UKAS that they have control over / awareness of changes made to the externally managed software.