

GENOMICS AND MOLECULAR PATHOLOGY DIGITAL SPECIALIST PORTFOLIO MODULES



Institute of Biomedical Science
12 Coldbath Square
London
EC1R 5HL

Tel 020 7713 0214
Web www.ibms.org
Email Elearning@ibms.org

Genomics and Molecular Pathology Digital Specialist Portfolio Modules

Required Modules

- Essentials of Genomics
- Sample Handling
- Nucleic Acid Extraction and Quantification
- Sanger Sequencing
- Polymerase Chain Reaction (PCR)
- Next Generation Sequencing (NGS)

Select Modules- minimum of one

- Haematological Malignancy
- Solid Tumours
- Rare and Inherited Diseases (R&ID)

Optional Modules (at least one will be required)

- Karyotyping
- In-situ Hybridisation
- Microarray
- Pyrosequencing
- Fragment Analysis
- High Resolution Melt Analysis

Selection of Haematological Malignancies or Solid Tumour includes the Fundamentals of Cancer module

- Fundamentals of Cancer

Please note

All learning outcomes (LOs) are met through two pieces of evidence, Q&A as agreed with a training officer and an additional piece of work as selected by the candidate.

A statement of work and reflective statement on each module will be required which will include sign off by the trainer stating that the candidate works in accordance with laboratory procedures, the competence for which should be evidenced in-house and is not part of the portfolio submission.

Indicative Content outlines background knowledge that may be required to meet the LOs and/or knowledge and competences expected to be demonstrated across multiple modules. Knowledge of areas highlighted in the indicative content may be examined during the viva.

Selection options- If one select module is chosen then a minimum of 2 optional modules are required. If one or more select modules are chosen a minimum of one optional module is required.

Module Title	Essentials of genomics
Module code	7216
Rationale/ Aims	This module will enable the candidate to have a sound understanding of the genome and the range of abnormalities that can occur to give rise to disease. Candidates will understand the ethical and legal implications of genomic testing and impacts of the 100K genome project.
Learning outcomes	<ol style="list-style-type: none"> 1. Discuss how common systems for variant nomenclature (International System for Human Cytogenetics Nomenclature, Human Genome Variation Society) are used in practice. 2. Discuss the multiple theories attempting to explain the origin and development of cancer, consider: Somatic Mutation Theory (SMT) and Knudson (two hit) theory. 3. Discuss common conditions caused by aneuploidy, how they may arise, and how they may be detected. 4. Explain deletion, duplication, insertion, inversion, and translocation, how they may arise and how they may be detected. Include examples of how these lesions translate, at the gene, level, into disease. 5. Discuss the importance of the single nucleotide polymorphism in generating variation, both in physiology and pathology. 6. Evaluate the roles of the inheritance of specific genetic lesions, and the de novo development of gene abnormalities, provide examples of dominant, recessive, X-linked (dominant and recessive) and imprinting. 7. Discuss and give examples of ethical, legal and social implications / considerations of genomic testing. 8. Evaluate the influence of the 100k Genome Project and national genomic programmes, and provide examples of how this has or could impact medicine, equity of access, reducing costs, and new discoveries, etc.
Indicative Content	<p>Candidates should have knowledge of the primary, secondary and tertiary structure of nucleic acid strands and their organisation within the cell. Knowledge of the structure and function of genetic apparatus, and its role in pathology. Candidate should understand the processes of translation and transcription. Explain the process of cellular replication, with reference to the cell cycle, DNA duplication, mitosis and meiosis.</p> <p>Explain how genes are expressed, with reference to transcription, gene regulation/silencing, epigenetics (e.g., methylation), introns, exons and splice variants.</p>

Module Title	Sample Handling
Module code	7212
Rationale/Aims	This module enables the candidate to understand the preanalytical variables of samples referred for molecular tests and gain an understanding of the core principles underpinning molecular analysis and samples referred for genomic testing. Candidates will gain knowledge of managing insufficient or inadequate samples as well as understanding the importance of external quality assessment to molecular testing. Candidates will gain knowledge of genomic test directories and how these apply to practice.
Learning outcomes	<ol style="list-style-type: none"> 1. Explain different types of samples and quantity required, for a variety of genomic tests and discuss the appropriate transportation of these samples to the laboratory. 2. Demonstrate accepting the sample arriving in the laboratory with the correct and complete information regarding the patient's identity and the pathological nature of the sample, including different referral forms and consent requirements, and explain the importance of accurate logging of sample details. 3. Discuss factors affecting sample integrity and suitability for downstream testing. 4. Explain, with examples, why some samples require fixation, the need to optimise protocols dependent on which fixative has been used, and the impact and importance of fixation. 5. Discuss competing demands for samples which may be limited and/or valuable. 6. Evaluate the importance of assessment of cellularity in specimens. 7. Recognise and categorise different tissue types which require extra consideration for adequate neoplastic cellularity during tumour assessment and dissection. 8. Demonstrate the management process for failed or insufficient sample extraction and incorrect sample type. 9. Discuss the importance of external quality assessment schemes and their application and limitations in genomics and molecular pathology. 10. Explain the importance of the National Genomic Test Directory, and discuss how it relates to the candidate's practice.
Indicative Content	<p>Candidates require knowledge and understanding of:</p> <p>The importance of the initial phase of good laboratory practice in ensuring incoming material is assessed and processed correctly.</p> <p>The importance of the genomic test directory used in your home nation of practice</p>

Module Title	Nucleic acid extraction and quantitation
Module code	6993
Rationale/ Aims	The candidate will gain knowledge of theoretical methods and demonstrate practical skills in extraction and quantitation of nucleic acids as well as troubleshoot established procedures. In addition, the candidate will be able to identify both the need for, and methods of extraction of, nucleic acids, and subsequent techniques for assessing the quality and concentration of extracted nucleic acids.
Learning outcomes	<ol style="list-style-type: none"> 1. Discuss and give examples from your lab on the impact of pre-analytical specimen collection and handling upon nucleic acid extraction, quality/concentration from a range of sources including blood, fresh/frozen vs formalin-fixed paraffin embedded tissue, aspirates, direct-preparation cytology specimens, swabs etc. 2. Explain the principles of nucleic acid extraction techniques including crude lysis/precipitation/purification, spin-column methods, magnetic bead isolation, and sonication and discuss the rationale for selection of the method used in your laboratory. 3. Summarise the differences in processing of specimens for extraction of DNA vs RNA, and subsequent sample handling and storage. 4. Evaluate the advantages and disadvantages of automated vs. manual methods of nucleic acid extraction. 5. Assess techniques for enrichment and/or increasing extracted nucleic acid yield. 6. Compare methods for assessment of quality/integrity and quantity of nucleic acids in the isolate including spectrophotometry, electrophoresis (including automated methods), fluorometric methods, and quantitative (q)PCR. Discuss a method from your lab explaining why this method is appropriate. 7. Describe and justify techniques used in your lab to reduce cross-contamination between specimens and reagents including use of negative controls and/or extraction "blanks".
Indicative Content	<p>Candidates require knowledge and understanding of:</p> <p>Sample types used in nucleic acid extraction.</p> <p>Methods by which nucleic acids may be extracted and prepared for genetic analysis.</p> <p>The advantages and disadvantages of extraction methods and the quality management of the processes.</p>

Module Title	Sanger sequencing
Module code	7218
Rationale/ Aims	This module will enable the candidate to gain knowledge and understanding of the key principles of Sanger sequencing and understand the advantages and disadvantages of the technique and its clinical applications.
Learning outcomes	<ol style="list-style-type: none"> 1. Explain the principles of Sanger sequencing and discuss its current relevance. 2. Describe the stages in the practical set-up of a typical Sanger assay, including, but not limited to, reagents and analysers. 3. Demonstrate principles of Sanger sequencing analysis using appropriate software. 4. Interpret the sequence of an electropherogram and describe the variants using Human Genome Variation Society (HGVS) nomenclature. 4. Discuss advantages and disadvantages of Sanger sequencing compared to alternative methods for sequencing such as next generation sequencing and pyrosequencing. 5. Discuss relevant quality parameters for Sanger sequencing and factors that may influence outcomes. 6. Discuss examples of the clinical applications of Sanger sequencing. 7. Discuss troubleshooting techniques to obtain optimum sequencing.
Indicative Content	<p>Candidates require knowledge and understanding of: DNA replication processes.</p> <p>The process of the amplification and analysis of a specific sequence of DNA in order to detect abnormalities.</p>

Module Title	Polymerase chain reaction (PCR)
Module code	7214
Rationale/ Aims	<p>The candidate will understand the use of both PCR and real-time PCR within a molecular pathology laboratory.</p> <p>The candidate will be able to demonstrate both practical and theoretical understanding of PCR including developing, optimising, validating and troubleshooting.</p>
Learning outcomes	<ol style="list-style-type: none"> 1. Define the underlying principles of PCR, and consider modifications and their applications, e.g., real-time PCR, multiplex PCR and nested PCR, as it pertains to the local repertoire. 2. Identify each component of the assay and discuss its function. 3. Define performance characteristics (including, but not limited to, accuracy, precision, reproducibility, sensitivity, specificity, etc.) for a range of PCR and/or real-time PCR assays. 4. Discuss what remedial/repeat actions may be required where an individual assay is suboptimal and demonstrate with examples from practice. 5. Explain how data is interpreted, including the use of parameters such as (but not limited to) Ct, threshold limits, and allele discrimination. 6. Discuss the factors to be considered when setting up and optimising a new PCR assay. Consider reagents, primer design, equipment, target, set up and workflow, and end point analysis. 7. Demonstrate application of QC criteria to the acceptance/rejection of a sample result and explain the remedial actions taken.
Indicative Content	<p>Candidates require knowledge and understanding of: DNA replication processes, principles, set-up, and applications of the PCR on a wide range of analytical platforms. Primer design, e.g., amplifying regions of high %GC content, minimising primer dimers.</p>

Module Title	Next Generation Sequencing (NGS)
Module code	7219
Rationale/ Aims	Candidate will gain a detailed understanding of the principles and applications of NGS in the laboratory. The candidate will be able to develop/ optimise/ troubleshoot/ validate this technique within the laboratory.
Learning outcomes	<ol style="list-style-type: none"> 1. Discuss the relevance of NGS in the laboratory and its importance in the wider setting of clinical pathology. 2. Define the underlying principles and chemistry of a range of NGS technologies, for example amplicon-based, hybrid capture and long read sequencing methods. 3. Explain the importance of each major stage of an NGS assay (e.g., the preparation of a library, templating, informatics, interpretation, etc.). 4. Compare and contrast the features of DNA and RNA sequencing applications. Consider the detection of copy number variation, single nucleotide variation, loss of heterozygosity, and structural variants. 5. Compare and evaluate the strengths and limitations of each NGS method against each other and traditional sequencing techniques. 6. Explain processes in the bioinformatic pipelines, and steps included relevant to your practice (e.g. variant calling, alignment, etc.). 7. Discuss possible errors which can occur during NGS and propose possible troubleshooting. 8. Discuss the scope of NGS application with respect to the use of hotspot targeted, large targeted panels, whole genome sequencing and whole exome sequencing, include the advantages and disadvantages, and clinical applications. 9. Demonstrate competence in practical set up and operation of an assay including application of quality control criteria to accept / reject reagents, samples, runs, etc.
Indicative Content	<p>Candidate should understand DNA replication processes, the theory and practice of leading NGS platforms and have knowledge of the basis for Sanger sequencing and or Pyrosequencing.</p> <p>Candidate should have knowledge of simple bioinformatics including use of relevant reference genomes and gene transcripts, base/variant calling and construction, read depth/coverage, use of software to interrogate/visualise variant calls and familiarity with online repositories for variant annotation.</p>

Module Title	Haematological Malignancies
Module code	7211
Rationale/Aims	<p>This module aims to provide candidates with an understanding of the molecular pathology of lymphoid and myeloid neoplasia that can be applied to their professional practice.</p> <p>Candidates will gain a solid understanding of the molecular pathology of lymphoid and myeloid neoplasms. Candidates will gain knowledge of sample requirements, related tests analysed in different pathology disciplines, the pathway of analysis leading to molecular testing and the importance of molecular testing in diagnosis, prognosis and treatment.</p>
Learning outcomes (What the students will be able to demonstrate upon completion of the module)	<ol style="list-style-type: none"> 1. Explain the principles of haemopoiesis from the multipotent stem cell to the mature cell and discuss why this knowledge is important in the investigation of haematological malignancies. 2. Describe presenting clinical symptoms (constitutional features, fatigue/night sweats/weight loss) and discuss initial routine screening tests (FBC, CRP, LDH) which may raise suspicion of a haematological malignancy. Consider possible differential diagnoses. 3. Define the impact on testing of sample type: consider peripheral blood, bone marrow trephine and bone marrow aspirate. 4. Define common terminology used in FBC, blood morphology and bone marrow reports. 5. Explain the importance of "pre-malignant haematological conditions" for example, but not limited to: Monoclonal B-cell lymphocytosis (MBL), monoclonal gammopathy of undetermined significance (MGUS), and clonal haematopoiesis of indeterminate potential (CHIP). 6. Define the key features of myeloid neoplasms as a continuum of disease: MPN <MPN/MDS overlap> MDS <AML/MDS overlap> AML. Consider the current guidelines (WHO, ICC and ELN) and how these are applied within the laboratory. 7. Define the key features of lymphoid neoplasms as B cell and T cell malignancies. Consider the current guidelines (WHO, ICC and ELN) and how these are supported within the laboratory 8. Discuss, with examples from practice, how genetic testing is influenced by results from other pathology disciplines, e.g., blood sciences, immunophenotyping, cellular pathology. 9. Describe the importance of genomic information in both the context of diagnosis, prognosis and treatment decision making, and monitoring of

	<p>minimal residual disease. Highlight the importance of a molecular service across the lifespan of the disease/ patient experience.</p> <p>10. Describe the function of at least 2 of named targeted therapies and explain how their efficacy is predicted by applied molecular testing.</p>
<p>Indicative Content</p>	<p>Candidates should have knowledge and understanding of:</p> <p>The principles of lymphopoiesis and myelopoiesis, to include normal processes with reference to relevant cytokines and receptor pathways, and awareness of the variety of laboratory tests which are included in the WHO Classification of Haematolymphoid Tumours (e.g. blast count, LDH, EPO levels).</p> <p>The principles of pharmacogenetics and targeted therapies.</p> <p>That some genetic variants are associated with germline predisposition and how this is determined.</p> <p>Candidates should be able to:</p> <p>Navigate and broadly understand blood science reports (haematology, chemistry and immunophenotyping), including common terminology used, which have indicated molecular pathology testing and understand how this supports diagnosis (including differential diagnosis), therapy and prognosis.</p>

Module Title	Solid Tumours
Module code	7213
Rationale/ Aims	This module will enable candidates to gain an in-depth understanding of the molecular pathology of solid tumour cancers, including features of malignancy, methods of analysis, an understanding of therapies including precision/personalised and the organisation and delivery of a clinical tumour genomic profiling -service.
Learning outcomes	<ol style="list-style-type: none"> 1. Compare and contrast the clinical features of common solid tumour malignancies. 2. Explain the classification and cell biology of carcinomas, sarcomas, neurological cancers, and melanomas. 3. Recognise laboratory techniques which identify features of malignancy such as standard tinctorial methods, immunocytochemistry, and <i>in situ</i> hybridisation techniques, and discuss their use in diagnosis. 4. Summarise the appropriate genomic testing strategy dependent on disease, (to include examples from carcinoma, sarcoma, neurological and melanomas) stage, and other test results as defined by the most up to date version of the National Genomic Test Directory as applicable to your practice. 5. Describe the function of a range of named targeted therapies (e.g., monoclonal antibodies, small molecular inhibitors, immunotherapy etc.) and discuss how their efficacy is predicted by molecular pathology testing. 6. Discuss the role of pharmacogenetics in guiding the use of cancer chemotherapy. 7. Discuss the importance of metastasis, tumour heterogeneity, relapse, and acquired therapeutic resistance mechanisms in common solid tumours. 8. Discuss the necessity of salvage pathways in suboptimal specimens.
Indicative Content	<p>Candidates require knowledge and understanding of:</p> <p>The pathophysiology of solid organ cancer, including the lung, gastrointestinal tract, breast, prostate and neuroblastoma, and how these are diagnosed.</p> <p>The principles of pharmacogenetics and targeted therapies.</p> <p>Genetic variants associated with germline predisposition and how this is determined.</p> <p>Candidates should be able to:</p> <p>Navigate and broadly understand histopathology reports, including common terminology used, which have indicated molecular pathology testing and understand how this supports diagnosis (including differential diagnosis, therapy and prognosis).</p>

Module Title	Rare and Inherited Disease (R&ID)
Module code	7217
Rationale/Aims	This module will enable the candidate to understand genetic inheritance patterns and the molecular basis of the pathogenicity of R&ID. The candidate will be able to perform analytical testing for rare and inherited diseases and have knowledge of different testing methods available. Candidates will understand consenting processes for genetic testing and the impact on the patient and the laboratory.
Learning outcomes	<ol style="list-style-type: none"> 1. Explain, with examples from candidate's practice, the genetic basis and pathogenicity of inherited diseases: include autosomal dominant, autosomal recessive, X-linked, imprinted mitochondrial disorders and cancer related to tumour suppressor genes. 2. Recognise the importance of genomic test request management with reference to the relevance National Genomics test Directory including prenatal testing and inherited cancer, give examples of their clinical application. 3. Discuss, with reference to specific diseases, the benefits and disadvantages of targeted pathogenic variant detection. 4. Demonstrate the application of variant interpretation guidelines with reference to at least 2 clinical indications relevant to your laboratory. 5. Explain maternal cell contamination in prenatal testing and discuss steps undertaken to detect this. 6. Discuss with examples the testing for late onset disorders. 7. Demonstrate the process behind the management of presymptomatic/familial testing and discuss the importance of controls and primer design. 8. Demonstrate calculation of prior risk based on family history of genetic disease e.g. cystic fibrosis and subsequent residual risk post testing. 9. Describe the process for consenting of genetic testing, discuss the implication to the patient, the laboratory and the laboratory's role in reporting incidental findings.
Indicative Content	<p>Candidates should have knowledge and understanding of:</p> <p>Genetic inheritance and its relationship to pathological conditions/disease.</p> <p>The pathophysiology and genetic lesions in rare and inherited diseases.</p> <p>Disease-specific testing strategies relevant to their laboratory such as targeted mutation testing for haemochromatosis, cystic fibrosis and triplet repeat disorders such as Fragile X, Myotonic Dystrophy and Huntington's Disease, multiplex ligation-dependent probe amplification for deletion and duplication disorders and microsatellite analysis for aneuploidy, and NGS/WGS for learning disability.</p>

Module Title	Karyotyping
Module code	7215
Rationale/ Aims	Candidate will be able to demonstrate both practical and theoretical understanding of karyotyping in the laboratory. Candidate will be able to develop/ optimise/ troubleshoot/ validate this technique within the laboratory.
Learning outcomes	<ol style="list-style-type: none"> 1. Identify the key phases of the cell cycle required for karyotyping and discuss their place in leukocyte tissue culture. 2. Define the underlying principles of karyotyping. 3. Describe how your lab uses tissue culture to obtain banded metaphase chromosome preparations, identifying the key stages and reagents used. 4. Explain, giving examples of specific disorders, the clinical use for karyotyping. 5. Recognise common numerical and structural abnormalities from a karyogram. 6. Describe the advantages and limitations of karyotyping techniques, compared to other cytogenetic methods. 7. Describe the most common chromosome banding techniques used in karyotyping, including G banding, and their chemical basis. 8. Demonstrate analysis of G-banding on a range of samples and report the results using the International System for Human Cytogenomic Nomenclature. 9. Discuss troubleshooting techniques to obtain optimum chromosome preparations for analysis and give an example from your practice.
Indicative Content	Candidates require knowledge and understanding of the cell cycle, mononuclear leukocyte tissue culture and microscopy. Candidates should have the ability to follow standard operating procedures to run, troubleshoot and quality manage assays.

Module Title	In-situ hybridisation (ISH)
Module code	7220
Rationale/ Aims	Candidates will be able to demonstrate specialist expertise in theoretical and practical aspects of ISH. Candidates will be able to perform ISH staining, assess quality of stained preparations and understand the analysis of different probe sets in clinical context.
Learning outcomes	<ol style="list-style-type: none"> 1. Describe the range of genetic abnormalities that may occur at the chromosomal level and the extent to which these can be detected by ISH. 2. Recognise different visualisation techniques, such as fluorescence and chromogenic and describe advantage and disadvantage of each. Suggest appropriate sizes for a probe. 3. Explain the difference between break-apart, fusion, enumeration probe sets, and list common clinical applications of each. 4. Identify and prepare suitable samples for ISH techniques 5. Demonstrate ISH and discuss measures of internal quality assessment 6. Demonstrate the use of appropriate microscopy techniques to visualise and capture images of stained material. 7. Assess quality of stained material and resolve problems by identifying appropriate remedial action. 8. Distinguish between positive, negative, and equivocal results of break apart/fusion probe sets and, using named examples, explain the clinical implications. 9. Distinguish between positive, negative, and equivocal results of enumeration/deletion probe sets and, using named examples, explain the clinical implications.
Indicative Content	Candidates should have knowledge and understanding of DNA, RNA, mRNA and nucleic acids. Candidates should understand and be able to use microscopes. Candidates will need to be able to determine the presence of defined chromosome and gene lesions within a cell sample.

Module Title	Microarray
Module code	7221
Rationale/ Aims	Candidates will gain knowledge and understanding of the principles of microarray and how this technology compares to comparative molecular techniques. Candidates will be able to set up, run and troubleshoot assays as well as use the appropriate software. Candidates will be able to interpret results in line with relevant guidance.
Learning outcomes	<ol style="list-style-type: none"> 1. Discuss the benefits and limitations of microarray analysis compared to other techniques. 2. Describe principles of microarray and discuss the application of comparative genomic hybridisation and SNP arrays. 3. Demonstrate practical assay set up (relating to probes, solid phase, a reporter system), and the operation of assay. 4. Discuss troubleshooting techniques applicable to microarray. 5. Explain basic bioinformatic processes and steps as they apply to microarrays. 6. Demonstrate application of QC criteria to; accept / reject reagents, samples, and runs. 7. Demonstrate microarray analysis using appropriate software, including interpretation of results with awareness of relevant guidelines. 8. Discuss clinical applications of microarray with examples from the candidate's practice.
Indicative Content	Candidates require knowledge and understanding of DNA replication, hybridisation of genetic material in a sample with complimentary sequences fixed to a solid phase, and their identification. Candidates should have knowledge of quality management processes relating to running and accepting results from analysis.

Module Title	Pyrosequencing
Module code	7222
Rationale/ Aims	This module will enable candidates to apply knowledge of the pyrosequencing technique in the laboratory they will be able to demonstrate both practical and theoretical understanding of pyrosequencing. Candidates will be able to develop/ optimise/ troubleshoot/ validate this technique within the laboratory.
Learning outcomes	<ol style="list-style-type: none"> 1. Discuss, with examples, clinical applications of pyrosequencing as appropriate to your laboratory. 2. Explain the principles of pyrosequencing and discuss why this technique is used. 3. Describe the key stages in the practical set-up of a pyrosequencing assay and discuss how these may be optimised. 4. Demonstrate competence in practical set up and operation of the assay. 5. Identify and correctly interpret appropriate control samples for pyrosequencing. 6. Analyse sequencing results using appropriate software, including interpretation of single nucleotide variants and short indels. 7. Describe common issues with pyrosequencing and explain how these can be identified and resolved. 8. Discuss the advantages and disadvantages of pyrosequencing compared to other common methods such as Sanger sequencing and NGS.
Indicative Content	<p>Candidates should have knowledge of the basics of formation of a nucleotide sequence.</p> <p>Candidates should be able to follow standard operating procedures to set up and run analysis including the application of quality management processes.</p>

Module Title	Fragment Analysis
Module code	7223
Rationale/Aims	This module will enable candidates to apply knowledge of fragment analysis to the laboratory and to demonstrate both practical and theoretical understanding of fragment analysis, including developing/ optimising/ troubleshooting/ validating this technique within the laboratory.
Learning outcomes	<ol style="list-style-type: none"> 1. Explain the principles of fragment analysis and discuss the range of clinical applications of two types of fragment analysis e.g., SNP genotyping, indel identification, and microsatellite marker analysis. 2. Compare methods of visualising fragment analysis, e.g. gel electrophoresis and capillary electrophoresis. Discuss the benefits and clinical applications of each. 3. Discuss the factors to be considered when setting up and optimising a new fragment analysis assay, including reagents, equipment, target and end point analysis. 4. Define success criteria for fragment analysis assays performed within your laboratory, including how the data is interpreted and parameters such as background noise, sensitivity, peak intensity. 5. Demonstrate ability to perform standard fragment analysis and interpretation relevant to your laboratory. 6. Apply QC criteria to the acceptance/ rejection of a sample result and justify the acceptance/rejection decision. 7. Demonstrate what remedial / repeat actions may be required where an individual assay is suboptimal.
Indicative Content	<p>Candidates should have knowledge and understanding of DNA and DNA replication, DNA extraction and PCR amplification.</p> <p>Candidate should be able to run assays, understand and apply quality management process including internal and external quality assessment.</p>

Module Title	High Resolution Melt [HRM] Analysis
Module code	7224
Rationale/ Aims	This module will enable candidates to apply knowledge and competence of HRM analysis in the laboratory. Candidates will be able to develop/ optimise/ troubleshoot/ validate this technique within the laboratory.
Learning outcomes	<ol style="list-style-type: none"> 1. Define the principles of HRM analysis and discuss the range of applications. 2. Discuss the factors to be considered when setting up and optimising a new HRM assay. Consider reagents, equipment, target and end point analysis. 3. Discuss the benefits and limitations of HRM compared with alternative approaches for methylation / mutation screening as applicable to your laboratory. 4. Demonstrate ability to perform a HRM analysis. 5. Demonstrate interpretation of HRM analysis and discuss the clinical implications of the results and any further tests that may be required. 6. Apply QC criteria to the acceptance/ rejection of a sample result and explain the decision to accept/reject. 7. Demonstrate what remedial / repeat actions may be required where an individual assay is suboptimal.
Indicative Content	Candidate should have knowledge and understanding of DNA, DNA replication processes, DNA amplification and understand the use of fluorescent dyes. Candidate should be able to follow standard operating procedures to set up, run and troubleshoot assays undertaken. Candidates should understand quality management processes and how this applies to HRM analysis.

Module Title	Fundamentals of Cancer
Module code	7267
Rationale/ Aims	This module will enable candidates to gain knowledge in the fundamentals of cancer, its development and its clinical management. A background understanding is considered essential for those working in solid tumour molecular pathology and haematological malignancy diagnostics.
Learning outcomes	<ol style="list-style-type: none"> 1. Define “cancer” and discuss its hallmarks. 2. Describe, with examples, the risk factors for cancer, including environmental, genetic, lifestyle and infections. 3. Discuss mechanisms of carcinogenesis: consider activation of oncogenes, inactivation of tumour suppressors, evasion of apoptosis genes, and defective DNA repair genes etc. 4. Define the following as they relate to the management of cancer: diagnosis, prognosis, therapy stratification, pharmacogenomics. Use examples from your laboratory practice. 5. Discuss the role of molecular testing in terms of interdisciplinary working and the patient pathway. 6. Describe the molecular processes involved in cancer development, growth, and metastasis. 7. Discuss the difference between somatic and germline variants in cancer development and how this may influence patient management
Indicative Content	<p>Candidates require knowledge and understanding of:</p> <p>The developing state of knowledge in molecular pathology and how this impacts on result interpretation and patient pathways.</p> <p>The Knudson (two-hit) hypothesis as a theory of cancer causation.</p> <p>National Genomics Test Directory relevant to their practice and testing strategies.</p> <p>Genetic variants associated with germline predisposition and how this is determined.</p> <p>Molecular pathology in the diagnosis and management of cancer.</p>

About this version

Document title: IBMS Specialist Portfolio Genetics and Molecular Pathology Digital Modules

Produced by: Qualifications

Contact: Elearning@ibms.org

T: + 44 (0)20 7713 0214

Version: Version 1

Date active: March 2024

Copyright and disclaimer

This document and its contents including the IBMS logo are the property and trademarks of the Institute of Biomedical Science (IBMS). The copyright on this material is owned by the IBMS (unless otherwise explicitly stated). This document or no part of it may be copied, reproduced, republished, downloaded or transmitted in any way, other than for your own personal, non-commercial use.

Prior written permission must be obtained from the IBMS, using the contact details above, for any other use of this material. All rights are reserved.



copyright © Institute of Biomedical Science 2024

About IBMS publications

The IBMS publishes a wide range of professional and scientific publications and guidance.