HANDBOOK

RCPA
The Royal College of Pathologists of Australasia

Extension of Scope of Practice in Molecular Genetics

(NPAAC Supervision Certification Modules)

Chemical Pathology

Table of Contents

Glossary

CPDP	RCPA Continuing Professional Development Program
(F)RCPA	(Fellow of the) Royal College of Pathologists of Australasia
IANZ	International Accreditation New Zealand
MDT	Multi-disciplinary team
NATA	National Association of Testing Authorities
NPAAC	National Pathology Accreditation Advisory Council
PPD	Personal Professional Development
RCPAQAP	RCPA Quality Assurance Programs Pty Ltd
SOP	Standard Operating Procedure
WHS	Workplace Health and Safety

SECTION 1

Introduction

1.1 Introduction

The Royal College of Pathologists of Australasia (the College) offers four certification modules in genetic/genomic pathology for those who have attained FRCPA.

These modules have been designed to satisfy the recent NPAAC Requirements, particularly as applicable to supervision of testing involving molecular diagnostics and genomics.

To competently supervise a molecular/genomic service, chemical pathologists need to have adequate and detailed knowledge of the wet and dry laboratory aspects of the technology and bioinformatics analysis. It is recognised that this knowledge may not be the same as the hands-on experience of the scientist in the genomics laboratory; however, supervising pathologists should be familiar with the limitations and strengths of the methodology, the ethical considerations of data use and reporting and the clinical relevance for assessing appropriate requesting and reporting.

It is anticipated that in the next few years, genomic analysis will be integral to the work of pathology laboratories and thus the general Fellowship training program. These modules are designed to provide all chemical pathologists with the opportunity to participate in accordance with NPAAC requirements.

1.2 General aims

The current Chemical Pathology trainee curriculum is regarded to be the current standard and determines the Scope of Practice for Chemical Pathology and includes genotyping for *AAT*, *APOB*, *APOE*, *BCHE*, *HFE*, *TPMT*, *UGT1A1*, etc. The genetic/ genomic certification modules build on discipline-specific Fellowship training. They are designed to build base knowledge, analytical and interpretive skills, quality assurance and communication capabilities required for safe clinical service provision of genetic/ genomic testing in Chemical Pathology.

The purpose of the modules outlined in this Handbook is to offer Fellows the opportunity to gain certification of expertise for a graduated range of genetic/ genomic testing categories specifically for clinical applications within their specific pathology discipline.

While candidates will gain understanding of the breadth of the field, they must remain aware of the limits of their own knowledge and appreciate when it is in the best interests of patients to refer to, or formally consult with, genetic pathologists or other appropriately credentialed colleagues.

Completion of one or more modules will result in the extension of scope of practice within Chemical Pathology to the limits defined for each module. The scope of practice would not extend into other discipline areas – for example, investigation of intellectual disability or prenatal diagnostic testing. Please note: non-invasive prenatal screening and testing is within the scope of Chemical Pathologists who have completed the relevant module, while all other prenatal testing is not.

1.3 Genetic and genomic testing

Genetic and genomic testing includes targeted analysis for presence/ absence of predefined mosaic or clonal genomic variation; screening for undefined variants in a single gene; screening for undefined variants in a limited number of specified genes, and screening for undefined variants in a large number of specified genes.

Testing can also include untargeted screening of all chromosomes (karyotyping), higher resolution screening of all chromosomes (chromosomal microarray), whole exome screening, and whole genome screening.

Specialised genetic, genomic, epigenetic and gene expression tests are also emerging. They include targeted testing for uniparental disomy, chimerism, methylation anomalies and other epimutations, and gene expression profiling.

The goal of all genetic/ genomic investigations is to identify variants that are of direct relevance to the clinical phenotype in question. A growing number of tests may also simultaneously detect additional disease-causing variants that are not directly relevant to the clinical indication for the specified test. Instead, these unanticipated findings may indicate a heightened risk of either having, or developing in the future, another unrelated illness. Additionally, tests that involve screening specified genomic sequences for undefined variants will inevitably identify variants that are of uncertain clinical significance.

The three modules in this handbook are a practical response to the gradient of technical and clinical complexity within genetic and genomic testing, as well the differing practical skill sets required across the spectrum of methods now available.

Given the close relationship between Biochemical Genetic Pathology and Chemical Pathology, with possible boundary overlap, these modules are designed to ensure that the respective scopes of practice are maintained. Chemical Pathologists who complete these modules may subsequently present this evidence if they later apply for additional scope of practice in Biochemical Genetics. Further information about this process is available from the College.

1.4 Registration

Applicants must complete a registration form, obtainable from the RCPA website and submit this with the application fee. The form must clearly state the modules(s) being applied for. Applications for partial exemption from assessments (recognition of prior learning) must be submitted with supporting documentation at the time of registration.

1.5 Training structure and requirements

The modules are outcomes-based, with no fixed time requirements for completion. Candidates must participate in a range of laboratory-based experiences to achieve the listed outcomes. All specified assessment tasks must be completed satisfactorily to achieve certification.

1.6 Supervision

All training must be supervised by an approved Fellow of the RCPA with the required expertise. Supervisors are expected to monitor and provide regular feedback on the candidate's developing competence. Formal meetings with the candidate are expected to occur at least every three months.

The supervisor will complete a Supervisor Report at the end of the training period, along with a completed and signed portfolio summary sheet. If the training period exceeds twelve months, an

additional annual Supervisor report will be required.

The supervisor will carry out workplace-based assessments or may delegate this responsibility to another suitably qualified pathologist or senior scientist.

1.7 Assessment

Assessment consists of a range of activities and workplace-based assessments to be documented in a portfolio, and formal examinations as prescribed for each module.

Portfolios may be maintained in paper-based and/or electronic format with back-up. Portfolios do not need to be submitted to the College. The supervisor will verify that all entries and assessment tasks have been completed on the portfolio summary sheet to be submitted with the final Supervisor Report for the module.

1.8 Continuing professional development

Activities carried out and documentation associated with the modules may be included, along with general chemical pathology activities, in the relevant categories of the RCPA Continuing Professional Development Program (Professional Performance Framework). Following certification, specialists are expected to undertake ongoing professional development relevant to the modules for which they are certified.

1.9 Note

Module 1 (Targeted testing for presence/ absence of predefined genomic variation by molecular methods) has been rescinded since it has been deemed that the content is covered in the standard Fellowship program for Chemical Pathology. This module is not required or available but numbering of subsequent modules has been retained for consistency with previously circulated documents.

SECTION 2

Learning outcomes and recommended training activities

Module 2: Targeted screening for undefined variants in genes associated with specified clinical phenotypes

The unifying concept of this module is the detection of unknown genomic variation within a pre-defined list of genes for analysis (focused on a specific biochemical phenotype/clinical context) which allows for the practice and development of discipline-specific expertise.

Typically, these assays are 20-30 gene panels, which are run for disease diagnosis or stratification. They may also include single gene or single exon sequencing for specific diseases.

This is complex and specialised training, which builds upon the sound genomic basics within the Chemical Pathology Trainee Curriculum.

Examples of typical clinical applications for this module would include targeted gene panels for diagnosis/predictive testing for pre-defined inherited conditions (e.g. cystic fibrosis, familial hypercholesterolaemia, hereditary haemochromatosis, alpha-1 antitrypsin deficiency, butyrylcholinesterase deficiency, maturity onset diabetes of the young etc.)

Types of genomic variation covered
Targeted panels and associated bioinformatic pipelines may be designed to detect sequence variants and copy number changes involving pre-defined genes/regions on the targeted panel, depending on the nature of the genomic variations contributing to the defined clinical phenotype.

2.1 Learning outcomes

2.1.1 Theoretical and Technical Knowledge

- Methods covered
  - Sanger sequencing
  - Multiplex-ligation primer amplification (MLPA)
  - Massively parallel sequencing (MPS) with a range of library preparation/ bio-informatic filtering
    - Targeted amplicon enrichment
    - Hybridisation-based enrichment
- Knowledge of theory and processes relating to Wet Lab:
  - General practical skills understanding of nucleic acid preparation method(s), quantification/purity/intactness, storage/archiving
  - Knowledge of technical performance, limitations and quality issues associated with different library preparation methodologies (amplicon, hybridisation based, use of unique molecular identifiers (UMI), etc.)
  - Knowledge of technical performance, limitations and quality issues associated with different sequencing technologies.

- Knowledge of theory and processes relating to Dry Lab:
  - Knowledge of primary, secondary and tertiary analysis, variables and limitations
  - Knowledge of relevant bioinformatics issues including performance and limitations of demultiplexing/alignment tools/variant callers, variant annotation strategies, bioinformatic methods of structural variant detection, reference generation strategies for copy number assessment, performance and validation of copy number calling algorithms
  - Knowledge of data architecture, computing/processing/capacity issues and data security/privacy
  - Knowledge of cloud based secondary and tertiary analysis systems

2.1.2 Analytical and interpretive skills

- Detailed working knowledge of HGVS/ISCN nomenclature and its practical application
- Knowledge and application of pathogenicity classification systems for germline variants Management of incidental genomic findings more generally, and variant associated with significant clinical outcomes unrelated to the purpose of testing
- Integration of the genomic variation detected with accompanying biochemical analyte data and the clinical context of the patient in order to provide a clinically appropriate genomic report
- Synthesis and clinical interpretation of laboratory data taking account of the clinical presentation and other relevant non-genomic and genomic investigations

2.1.3 Quality assurance

- Knowledge of specific quality issues, validation and requirements of supervision of MPS (i.e. *Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies, National Pathology Accreditation Advisory Council, First Edition 2017*)
- Knowledge of potential sources of error arising from MPS assays
- Knowledge of ethical, clinical and regulatory structures/framework around germline testing for inherited disease/predictive testing
- Knowledge of technical performance, limitations and quality issues
- Recognition and troubleshooting of challenges
- Competence in monitoring data quality and result verification

2.1.4 Communication and Consultation

- Ability to provide clinically appropriate advice regarding contents of genomic reports including diagnostic implications, prognostic implications and therapeutic implications of detected genomic variations
- Advice on appropriate follow-up genomic testing/other modalities as required (including testing of family members)
- Involvement and ability to communicate in multidisciplinary meetings with pathologists, physicians, scientists and other referring specialists

2.2 Specific considerations/challenges to be addressed in assessment

- Investigations/actions when DNA is of inadequate quantity for testing
- Sanger sequencing primer design
- Sequence with poor Phred quality score
- Trouble-shooting poor-quality sequence
- Challenges associated with homopolymer runs
- Challenges with assay controls – positive and negative
- Trouble-shooting an MPS run with poor quality control metrics – wet lab and/or dry lab MLPA probe design
- Issues with sample depurination
- Trouble-shooting MLPA results – quantitation, standard deviation, abnormal marker patterns, low probe signals, apparent single exon deletion, etc.
- Investigations/actions when control samples perform adequately but one or more samples does not pass QC metrics
- Investigations/actions when testing performed at another laboratory does not match with the result in your laboratory
- Validation process for Sanger sequencing/ MLPA
- Validation process for MPS, including validation of bioinformatic pipeline
- Maintenance of bioinformatic pipeline, including version control, verification of new versions
- Assessment of variants of uncertain clinical significance (must be included)
- Consultation with colleagues regarding an incidental/secondary finding
- Investigations/actions when an external QA program does not award full marks for genotyping
- Investigations/actions when an external QA program does not award full marks for result interpretation/reporting
- Consultation with expert colleagues regarding an incidental/secondary finding, or other complex finding

2.3. Assessment

2.3.1 Portfolio requirements

Item	Requirements (number/type)	Documentation
Case Log	Summaries of all laboratory-based experiences, including supervising the analysis, data interpretation and reporting of at least 150 cases and 100 disease-associated variants.	Log book to include test name; assay type; number of assays/ runs; test failures requiring review. (Appendix 2)
Clinical Consultations	Fifteen (15) technically challenging or unusual cases/consultations involving lab data. Should address challenges as listed in paragraph 2.2. NOTE: evaluation of variants of uncertain significance and assessment of variant pathogenicity MUST be included	Clinical Consultations Sign-Off Form (Appendix 2)
Primary Requestor one-to-ones	Five (5) primary requestor one-on-ones (telephone)	Primary Requestor one-to-one Sign-Off Form

		(Appendix 2)
Process-based Discussions (PdD)	• Detailed discussion (at least 1-page description) of case in addition to PbD cover sheet) on five (5) "challenging" cases that required use of multiple skills, including consideration of lab data. • At least one (1) PbD should be on further workup/management of unusual such as an "additional" or "off target" finding.	PbD Assessment Form (Appendix 2)
Quality Assurance (QA) Activities Log	Eight (8) quality assurance activities (including two (2) compulsory). (**note**: You may include a significant/critical laboratory incident relevant to the module as a QA Audit)	Quality Assurance Activities Log (Appendix 2)
QA Reflection	Reflection on each of above	QA Reflection Form (Appendix 2)
Supervisor Report	One final report, plus additional annual report if module not completed in <12 months.	With sign-off indicating: • the principles of the method are understood; • working knowledge of instrument processes and maintenance requirements; • successful generation of results from each method, at a quality level sufficient for reporting • strong understanding of QC procedures for the method, including internal and external QA; • working knowledge of method anomalies and associated troubleshooting requirements. (Supervisor Report, Appendix 2)
Portfolio Summary sheet	To accompany final Supervisor Report	Portfolio Summary Form

2.3.2 Formal examinations

See Appendix 1

2.3.3 Assessment matrix

See Appendix 3

Module 3 – Sequence-based screening for known and unknown variants in multiple genes, including genes potentially linked to clinical phenotypes that have not been previously diagnosed in the patient

This is complex and specialised training and builds upon Module 2. It requires sound working fluency in dealing with the range of expected challenging outcomes and additional, “off-target” findings.

The unifying concept of this module is the application of massively parallel methodologies to screen for disease-causing and disease-associated genomic variants in multiple genes, including genes which can be linked to biochemical phenotypes that have not been previously diagnosed in the patient. The purpose of genome-wide testing would be to bioinformatically extract panels of genes relevant to biochemical phenotypes, or for genome-wide analysis of biochemical disorders.

Noting the limitation of the Chemical Pathologists from unfocussed (genome wide) DNA analysis of samples from patients with these conditions – i.e. the purpose of this module is the diagnosis of phenotypes related to chemical pathology.

Examples of typical clinical applications include: –

- Diagnosis of biochemical disorders with diverse genetic aetiologies
- Whole genome/ exome sequencing for inherited biochemical disorders

The approach used to detect disease-linked variants will also yield the following outcomes:

- False positive results
- False negative results
- Detection of variants of uncertain clinical significance
- Detection of variants with partial penetrance or expressivity
- Findings that may allow reproductive choices

Wide scope sequencing may also yield other additional or “off-target” findings:

- Medically treatable disorders
- Serious, incurable conditions
- Variants conferring susceptibility to disease
- Variants with pharmacogenetic implications
- Mis-attributed paternity
- Close consanguinity

Types of genomic variation covered
Bioinformatic “pipelines” can be targeted to screen MPS output for nucleotide variants, copy number changes and structural variants/fusions across a large number of specified genes up to the level of all exons of all known genes, or even the whole genome.

3.1 Learning outcomes

3.1.1 Theoretical and Technical Knowledge

- Methods covered
  - MPS using amplicon-based and hybridisation capture-based assays
- Knowledge of theory and processes relating to Wet Lab:
  - General practical skills and understanding of nucleic acid preparation method(s), quantification/purity/intactness, storage/archiving
  - Knowledge of technical performance, limitations and quality issues associated with different library preparation methodologies (amplicon, hybridisation based, use of unique

molecular identifiers (UMI), etc.)
    - Knowledge of technical performance, limitations and quality issues associated with different sequencing technologies
- Knowledge of theory and processes relating to Dry Lab:
    - Knowledge of primary, secondary and tertiary analysis, variables and limitations
    - Knowledge of relevant bioinformatics issues including performance and limitations of demultiplexing/alignment tools/variant callers, variant annotation strategies, bioinformatic methods of structural variant detection, reference generation strategies for copy number assessment, performance and validation of copy number calling algorithms
    - Knowledge of data architecture, computing/processing/capacity issues and data security/privacy
    - Knowledge of cloud based secondary and tertiary analysis systems

3.1.2 Analytical and interpretive skills

- Detailed working knowledge of HGVS/ISCN nomenclature and its practical application
- Knowledge and application of pathogenicity classification systems for germline variants
- Management of incidental genomic findings more generally, and variant associated with significant clinical outcomes unrelated to the purpose of testing
- Integration of the genomic variations detected with accompanying biochemical analyte data and the clinical context of the patient in order to provide a clinically appropriate genomic report
- Synthesis and clinical interpretation of laboratory data taking account of the clinical presentation and other relevant non-genomic and genomic investigations

3.1.3 Quality assurance

- Knowledge of specific quality issues, validation and requirements of supervision of MPS (i.e. *Requirements for Human Medical Genome Testing Utilising Massively Parallel Sequencing Technologies, National Pathology Accreditation Advisory Council, First Edition 2017*)
- Knowledge of potential sources of error arising from MPS assays
- Knowledge of ethical, clinical and regulatory structures/framework around germline testing for inherited disease/predictive testing
- Knowledge of technical performance, limitations and quality issues
- Recognition and troubleshooting of challenges
- Competence in monitoring data quality and result verification

3.1.4 Communication and Consultation

- Close link with a clinical genomics service, particularly for whole genome/ exome services
- Ability to provide clinically appropriate advice regarding contents of genomic reports including diagnostic implications, prognostic implications and therapeutic implications of detected genomic variations
- Advice on appropriate follow-up genomic testing/other modalities as required (including testing of family members)
- Involvement and ability to communicate in multidisciplinary meetings with pathologists, physicians, scientists and other referring specialists

3.2 Specific considerations/challenges to be addressed in assessment

- Evaluating primary specimen for suitability for molecular testing
- Investigations/actions when signal is detected in a no-template control
- Investigations/actions when DNA is of inadequate quantity for testing
- Investigations/actions when DNA is of inadequate quality/amplifiability for testing
- Investigations/ actions when sex discrepancy, discordant result, sample mix up identified
- Investigations/actions when results may be confounded by repetitive DNA sequences/ pseudogenes
- Investigations/actions when testing performed at another laboratory does not match with the result in your laboratory
- Trouble-shooting a whole exome/genome sample/run with poor quality control metrics – Wet Lab and/or Dry Lab metrics
- Validation process for a targeted assay based on bioinformatic filtering of whole genome/exome sequencing data
- Panel selection from WES/WGS data, including technical and clinical considerations
- Approach to targeted genes which are not adequately covered for clinical reporting
- Investigations/actions when an external QA program does not award full marks for genotyping
- Investigations/actions when an external QA program does not award full marks for result interpretation/reporting
- Consultation with expert colleagues regarding an incidental/secondary finding, or other complex finding

3.3. Assessment

3.3.1 Portfolio requirements

Item	Requirements (number/type)	Documentation
Case Log	Summaries of all laboratory-based experiences, including supervising the analysis, data interpretation and reporting of at least 150 variants (including 100 disease-associated variants). (**note**: this may include multiple variants from a single case)	Log book to include test name; assay type; number of assays/ runs; test failures requiring review. (Appendix 2)
Clinical Consultations	Fifteen (15) technically challenging or unusual cases/consultations involving lab data. Should address challenges as listed in paragraph 3.2.	Clinical Consultations Sign-Off Form (Appendix 2)
Process-based Discussions (PdD)	• Detailed discussion (at least one (1) page description of case in addition to PbD cover sheet) on five (5) “challenging” cases that required use of multiple skills, including consideration of laboratory data. (note: discipline-specific discussion topics to be defined) • At least one (1) PbD should be on further workup/management of	PbD Assessment Form (Appendix 2)

	unusual such as an “additional” or “off target” finding.	
MDT Participation	Five (5) MDT attendances.	Multidisciplinary Meeting Sign Off Form (Appendix 2)
Quality Assurance (QA) Activities Log	Eight (8) quality assurance activities (including two (2) compulsory). (**note**: You may include a significant/critical laboratory incident relevant to the module as a QA Audit)	Quality Assurance Activities Log (Appendix 2)
QA Reflection	Reflection on each of above	QA Reflection Form (Appendix 2)
Supervisor Report	One final report, plus additional annual report if module not completed in <12 months.	With sign-off indicating: • the principles of the method are understood; • working knowledge of instrument processes and maintenance requirements; • successful generation of results from each method, at a quality level sufficient for reporting; • strong understanding of QC procedures for the method, including internal and external QA; • working knowledge of method anomalies and associated trouble-shooting requirements. Supervisor Report (Appendix 2)
Portfolio Summary sheet	To accompany final Supervisor Report	Portfolio Summary Form

3.3.2 Formal examinations

See Appendix 1

3.3.3 Assessment matrix

See Appendix 3

Module 4 - Cell Free DNA (cfDNA) and Single Nucleotide Polymorphisms (SNPs) for the purpose of Non-Invasive Prenatal Screening (NIPS)

Under development

The Curriculum Development Working Group is currently developing this module for the Recognition of Prior Learning Pathway.

SECTION 3

Appendices

Assessment

Assessment is by

- Formal examinations (see Appendix 1)
- A portfolio of evidence of having participated in a sufficient number and type of work activities (see Appendix 2)
- Satisfactory progress (supervisor reports) (see Appendix 2)

An assessment matrix is provided in Appendix 3.

Portfolio requirements

Portfolio activities are carried out in the workplace provides evidence that fellows have engaged in the appropriate number and type of work-based activities to build base knowledge, analytical and interpretive skills, quality assurance and communication capabilities relevant to genetics/genomic testing in Chemical Pathology. Requirements or each module are summarised in a table at the end of the module in Section 2.

Appendix 2 contains the forms and logbook pages for recording the portfolio activities. Please file the hard copy forms in a **portfolio folder** with separate sections as in the table provided.

A soft copy **portfolio summary** (Excel spreadsheet) should also be compiled so that the fellow can keep track of what has been completed. The spreadsheet can be downloaded from the RCPA website. It is the fellow's responsibility to keep both hard and soft copy records up-to-date.

The portfolio summary spreadsheet should be appended to the annual supervisor reports and will be reviewed by the Registrar, Board of Education and Assessment and the Chief Examiner. Signatories and Fellows may be contacted to confirm evidence of satisfactory completion.

Appendix 1: Examinations

There will be a multi-station examination consisting of three stations, each of twenty minutes, with two examiners for each. These will include stations focussing on practical cases; for example: practical cases for variant curation in Module 3.

Candidates will be allowed 30 minutes pre-reading time, during which they may review data and reports, making notes where applicable, prior to discussion with the examiners.

The examination will focus on ability to:

- Analyse and interpret findings
- Explain principles of the test methods used, identifying any limitations
- Apply quality management principles and troubleshooting methods to explain and prevent possible sources of error
- Communicate clinically relevant conclusions and advice to referring doctors, including discussion of uncertainties
- Discuss broader health implications and/or ethical considerations relevant to the testing performed

Appendix 2: Forms and logbook pages for portfolio

RCPA The Royal College of Pathologists of Australasia	**Medical Genomics** **Chemical Pathology** **Routine Case Log**

How to use this form

From the beginning of training, Fellows should log all laboratory-based experiences, including analysis and reporting. Only runs that the Fellow has been directly involved with should be logged.

A minimum of 150 cases with 100 disease-associated variants should be recorded during the Module.

The log book should include:

- Test name
- Assay type
- Number of assays/ runs
- Test failures requiring review

At the end of each Module, the log should be sighted and signed off **on the Supervisor Report**.

Fellow Name	**Fellow ID**	**Module of Training** (please tick) ☐ Module 2 ☐ Module 3

	Date	***Test name***	***Assay type***	***Number of assays/ runs***	***Test failures requiring review***
1					
2					
3					
4					
5					
6					

7					
8					
9					
10					
11					
12					
13					
14					
15					
16					
17					
18					
19					
20					

Final Outcome (please tick) ☐ Competent ☐ Not Competent

Signature of Assessor	**Signature of Fellow**

Name of Laboratory

Medical Genomics
Chemical Pathology
Clinical Consultations Sign-Off Form

How to use this form
From the beginning of training, Fellows should log consultations with clinical colleagues that involve significant, difficult or unusual cases.
A minimum of (fifteen) 15 consultations should be recorded during the Module.

Consultation type* should be noted on the form as:** ***ORAL:*Telephone Outpatient* ***(TOP) OR*** *Telephone Inpatient* ***(TIP)***
WRITTEN: *Outpatient* ***(OP) OR*** *Inpatient (IP)*

At the end of each Module, this form and appended case lists should be sighted by the supervisor and signed off.

Fellow Name	**Fellow ID**	**Module of Training** (please tick) ☐ Module 2 ☐ Module 3

	Date	***Headline summary of case***	***Issue(s) raised by the case***	***Consult type***	***Fellow's role in the case***
	example	***e.g. Hereditary haemochromatosis***	***Clinician seeking guidance on diagnostic possibilities and investigations***	***TOP / TIP OP / IP***	***Advice offered; review of results; follow-up discussion with referring clinician***
1					
2					
3					
4					
5					
6					
7					

8					
9					
10					
11					
12					
13					
14					
15					
Final Outcome (please tick) ☐ Competent ☐ Not Competent					
Signature of Assessor			**Signature of Fellow**		
Name of Laboratory					

Medical Genomics
Chemical Pathology:
Primary Requestor one-on-one Sign Off Form

How to use this form
This form is for Modules 2 only; five (5) consultations should be recorded.
Consultation type may be: *Telephone Outpatient* ***(TOP) OR*** *Telephone Inpatient* ***(TIP)***

At the end of each Module, this form and appended case lists should be sighted by the supervisor and signed off.

Fellow Name	**Fellow ID**	**Module 2 only**

	Date	***Headline summary of case***	***Issue(s) raised by the case***	***Consult type***	***Fellow's role in the case***
	example	***e.g. Hereditary haemochromatosis***	***Clinician seeking guidance on diagnostic possibilities and investigations***	***TOP / TIP***	***Advice offered; review of results; follow-up discussion with referring clinician***
1					
2					
3					
4					
5					

Final Outcome (please tick) ☐ Competent ☐ Not Competent

Signature of Assessor	**Signature of Fellow**
Name of Laboratory	

Medical Genomics
Chemical Pathology
PbD Assessment Form
Process based Discussion

Fellow Name	**Fellow ID**	**Module of Training** (please tick) ☐ Module 2 ☐ Module 3
Assessor Name	**Assessor position** ☐ Pathologist ☐ Other (please specify)	

Techniques/methods (tick the box that applies).

1. ☐ Sanger sequencing
2. ☐ Multiplex-ligation primer amplification (MLPA)
3. ☐ Massively parallel sequencing (MPS) with a range of library preparation/ bio-informatic filtering
4. ☐ Targeted amplicon enrichment
5. ☐ Hybridisation-based enrichment
6. ☐ MPS using amplicon-based and hybridisation capture-based assays
7. ☐ Other (insert)_____

Please comment on whether these aspects of the Fellow's performance	**Yes**	**No**	**N/A**
Understands the principles of the method			
Has a working knowledge of instrument processes and maintenance requirements			
Has observed all phases of an assay successfully and been involved in the production of a valid result that can be reported			
Able to explain the Quality Controls procedures for this method, including internal and external Quality Assurance.			
Able to discuss anomalies and resolve uncertainties for the method			
Able to explain maintenance and trouble-shooting requirements for the method			

Please comment on other relevant aspects, especially on aspects for improvement (use the reverse side if insufficient room)

Final Outcome (please tick)
☐ Competent ☐ Not Competent

Signature of Assessor	**Signature of Fellow**

Name of Laboratory

Medical Genomics
Chemical Pathology
Clinical/Multidisciplinary Meeting Sign Off Form

How to use this form

This form is to be used to record that the Fellow has fulfilled the following

Module 3 only: present cases at a minimum of five (5) clinical or laboratory meetings throughout the Module

The supervisor is asked to sign after each meeting to verify off the Fellow's participation. Fellows should retain a list of the cases/entities presented at each meeting in the portfolio. At the end of the module, this form and appended case lists should be sighted by the supervisor and signed off.

Fellow Name:	**Fellow ID:**	**Module 3 only**

	Meeting date	**Brief description of meeting; subject(s) of discussion**	**Did Fellow present cases? Y/N**
1			
2			
3			
4			
5			
6			
7			
8			
9			
10			

Final Outcome (please tick) ☐ Competent ☐ Not Competent

Signature of Assessor	**Signature of Fellow**

Name of Laboratory

Medical Genomics
Chemical Pathology
Quality Assurance Activities Log

How to use this form
Eight (8) activities should be selected from the list. Items 9 and 10 are compulsory.
Use the Reflection Form in this handbook to write a brief reflection on what you learned from doing each activity (photocopy as many copies of the form as you need).
Keep the forms in your portfolio along with other specified documents if required

At the end of each rotation, the log should be sighted and signed off by the supervisor.

Fellow Name	**Fellow ID**	**Module of Training** (please tick) ☐ Module 2 ☐ Module 3

	Quality activity	***Summary of Fellow's role in the activity or comment (where applicable)***	***Date***
1	Analyse the design and operating characteristics of a particular instrument or platform		
2	Work through the development of a new in vitro diagnostic test and the associated IQA processes		
3	Review relevant AS ISO standards (list documents reviewed)		
4	Review relevant NPAAC standards and guidelines (list documents reviewed)		
5	Review the laboratory's quality policy, including policy guiding response to unsatisfactory QAP results		
6	External QAP (particularly involvement with the HGSA/QAP, EMQN, ASoC programs)		
7	Active involvement in preparations for laboratory accreditation		

Medical Genomics
Chemical Pathology
Quality Assurance Activities Log

How to use this form
Eight (8) activities should be selected from the list. Items 9 and 10 are compulsory.
Use the Reflection Form in this handbook to write a brief reflection on what you learned from doing each activity (photocopy as many copies of the form as you need).
Keep the forms in your portfolio along with other specified documents if required

At the end of each rotation, the log should be sighted and signed off by the supervisor.

Fellow Name	**Fellow ID**	**Module of Training** (please tick) ☐ Module 2 ☐ Module 3

	Quality activity	***Summary of Fellow's role in the activity or comment (where applicable)***	***Date***
8	Participate in a workflow check of effective/ efficient laboratory function		
9	**MANDATORY ACTIVITY** Review and update laboratory internal QC procedures. Include reports in portfolio.		
10	**MANDATORY ACTIVITY** Significant incident: Involvement in assessment, reporting and review, focussing particularly on the quality issues that were identified and addressed. **Minimum of one**. OR Quality audits: Conduct. Where possible, include comparison with relevant national/international guidelines. **Minimum of one**. Include documentation in portfolio. Use the reporting form in supplied in this Handbook		

Supervisor name…….....

Signature…….…… Date…….....

Medical Genomics Chemical Pathology Quality Assurance Reflection Form

Fellow Name	**Fellow ID**	**Module of Training** (please tick) ☐ Module 2 ☐ Module 3

Nature of activity

Your role in the activity

Your reflection on what you learned from your involvement in this activity

Points to consider: -

- *Type of Activity*
- *Actions undertaken*
- *Findings*
- *Resource considerations*
- *Ethical considerations*

Fellow signature	**Date**
Supervisor name (please print) and signature	**Date**

Name of Laboratory

Medical Genomics Supervisor Report Chemical Pathology

Please review the Fellow's portfolio and logbook before completing this report

Fellow Name (please print)	**Fellow ID**	**Module of Training** (please tick) ☐ Module 2 ☐ Module 3

Name of Organisation

Training period:

_____/_____/_____ to ___/_____/_____

Name of supervisor (please print) Supervisor RCPA ID no.

Please inspect the forms in the Fellow's portfolio and use the Table below to record evidence of completion

Portfolio items for which there is a minimum requirement				
	Previous total (if applicable)	Number in current year/ module	Cumulative Total	Minimum for completion of module
Case Log				150
Clinical Consultations				15
Process-based discussion (PbD)				5
Primary requestor one-on-ones				5 (Module 2 only)
MDT Participation				5 (Module 3 only)
QA Activities				8
QA Reflection				1 per each QA activity
Previous Supervisor's reports				1 report/ 12 months
Portfolio Summary				

Does the print-out of the portfolio summary spreadsheet accurately record the contents of the portfolio?

☐ Yes ☐ No

Please score the Fellow's performance using this scale

1 = Performance currently falls far short of expected standards for level of training.
There is a serious problem that may have implications for accreditation of the current training period. The problem must be stated clearly on the final page.

2 = Performance currently falls short of expected standards for level of training.
There is an area of lower than expected performance. The problem must be stated clearly on the final page.

3 = Performance is consistent with the expected level of training.
About 80% of fellows will merit this grade.

4 = Performance is better than expected for level of training.
About 10% of fellows will merit this grade.

5 = Performance is exceptional.
Very few fellows will merit this grade.

N/A = Not Applicable to this training period

	Score
The principles of all methods are understood	
Working knowledge of method anomalies and associated trouble-shooting requirements	
Working knowledge of instrument processes and maintenance requirements	
Successful generation of results from each method, at a quality level sufficient for reporting	
Strong understanding of QC procedures for the methods, including internal and external QA	
Ability to provide clinically appropriate advice regarding contents of genomic reports	
Ability to provide appropriate follow-up genomic testing/other modalities as required	
Ability to communicate in multidisciplinary meetings with pathologists, physicians, scientists and other referring specialists	

Overall evaluation

Have the outcomes of this module been satisfactorily achieved?
☐ Yes ☐ No

Is specific further professional development required? If yes, please outline process
☐ Yes ☐ No

Signatures

Fellow (please PRINT name and sign)	Date
Supervisor name (please PRINT name and sign)	Date
Other senior staff member/second supervisor (if applicable) (please PRINT name and sign)	Date
Head of Department (please PRINT name and sign)	Date

Comments by Fellow:

Please return this Supervisor's Report to:

The Royal College of Pathologists of Australasia
207 Albion Street
Surry Hills NSW 2010 AUSTRALIA
Faxed reports will not be accepted.

THE ROYAL COLLEGE OF PATHOLOGISTS OF AUSTRALASIA USE ONLY

Signature

Registrar/Deputy Registrar, Board of Education and Assessment (please PRINT name and sign)	Date

Appendix 3: Assessment matrix

	Portfolio of workplace activities						Supervisor report	Oral exam
	Case Log	Consult-ations	Primary requestor	PbD	MDT	QA Activity		
Theoretical & technical knowledge	Y	Y	Y	Y	Y		Y	Y
Analytical and interpretive skills		Y	Y	Y	Y	Y	Y	Y
Quality assurance				Y	Y	Y	Y	Y
Communication & consultation		Y	Y	Y	Y	Y	Y	Y