

## TRAINEE HANDBOOK 2019



## Genetic Pathology (Medical Genomics)

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It is essential to read this Handbook in conjunction with the ***Trainee Handbook – Administrative requirements*** which is relevant to all trainees. This has information about the College's structure and policies, together with details of requirements for registration, training and examination applications.

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## GLOSSARY

AMC	Australian Medical Council
AS ISO	Australian and International Standard
BEA	Board of Education and Assessment
CbD	Case-based discussion
CISH	Chromogenic in situ hybridisation
CPD	Continuing professional development
DOCS	Directly observed communication skills
DOPS	Directly observed practical skills
DP	Dry practical examination
FISH	Fluorescence in situ hybridisation
IANZ	International Accreditation New Zealand
MDT	Multi-disciplinary team
MLPA	Multiplex ligation-dependent probe amplification
NATA	National Association of Testing Authorities
PCR	Polymerase chain reaction
QA	Quality assurance
QC	Quality control
RACP	Royal Australasian College of Physicians
RCPA	Royal College of Pathologists of Australasia
RT-PCR	Reverse transcriptase-polymerase chain reaction
SAQ	Short answer question
SNP	Single nucleotide polymorphism
SO	Structured oral examination
UCSC	University of California, Santa Cruz
UPD	Uniparental disomy
WHS	Work health and safety
WP	Wet practical examination

## SECTION 1

### INTRODUCTION

The deciphering of the human genome and associated epigenome is creating many new opportunities to improve the diagnosis and management of many human diseases arising from inherited, sporadic or somatic genomic variants.

Genetic pathologists contribute to the multidisciplinary range of skills required within pathology services to aid in the diagnosis, management and treatment of patients with disorders arising from genomic mutations. They are expected to have sufficient clinical knowledge and judgement, as well as scientific knowledge and skills, to:

- deal with unexpected, atypical and complex work;
- respond to challenging enquiries from clinical colleagues and concerned patients;
- recognise and respond quickly to the clinical relevance of new genetic advances; and
- provide professional leadership and guidance, particularly in the arenas of clinical effectiveness and governance.

#### ***Scope and setting of practice of a genetic pathologist***

The objective of RCPA training in genetic pathology is to ensure the supply of medically trained laboratory practitioners required to contribute to the provision of comprehensive genetic diagnostic services.

Genetic diagnostic laboratories serve the needs of a diverse array of patient groups and rely on input from staff who collectively ensure the production of relevant accurate genomic data and the clinical interpretation of results in the context of the clinical question that prompted the test request.

From an organisational viewpoint, genetic pathologists enhance the clinical focus of laboratory service provision; strengthen the clinical focus of quality assurance activities; enhance clinically-directed service development and innovation, and also guide clinical input into undergraduate and postgraduate education and training.

The key factor that distinguishes a genetic pathologist from a senior genetic laboratory scientist is the clinical focus of their basic medical and subsequent specialist training. The clinical aspects of genetic pathology training extend the scope of practice into areas of clinical service provision that are frequently beyond the comfort zones of all but the most experienced genetic laboratory scientists. These aspects include:

- delivery of clinical interpretive and consultative services, with activities focused on delivering laboratory testing services that optimally address the clinical questions raised by the referring clinician;
- use of clinical judgement and knowledge to deal with unexpected, atypical and complex work;
- responding to challenging enquiries from clinical colleagues;
- rapid recognition of clinical relevance of new genetic advances; and
- provision of professional leadership, particularly in the arenas of clinical effectiveness and governance.

In addition to sound background knowledge of cell biology and human genetics, genetic pathologists require a growing range of computing, informatics and statistical skills to analyse the high volumes of genomic data, which are generated by technology platforms such as massively parallel sequencing.

Until recently RCPA genetic pathology training encompassed three major sub-disciplines: *molecular genetics*, which involves genetic and functional analysis of human genomic and epigenetic variants; *cytogenetics*, involving evaluation of human chromosomal variants; and *biochemical genetics*, which addresses the specialised challenges associated with inborn errors of metabolism.

The historic boundaries between these three sub-disciplines have become blurred with the evolution of new technologies and there is now a seamless progression from microscopy-based visualisation of the human genome compacted into chromosomes through to molecular assessment for the presence or absence of specific single nucleotide changes.

In response to these developments, the RCPA now offers two five-year training programs in genetic pathology:

- Medical genomics (outlined in this Handbook)
- Biochemical genetics (outlined in the Biochemical Genetics Handbook)

## **Medical Genomics**

The scope of practice for a genetic pathologist working in medical genomics is as follows:

- diagnostic detection and interpretation of genomic/ epigenomic variants in symptomatic patients (children, adults, fetuses)
- pedigree analysis
- diagnostic assessment of segregation in kindreds of disease-causing mutations or genomic regions
- diagnostic detection and interpretation of mosaic genomic variants (e.g. tumour material; constitutional mosaicism; constitutional chimerism; fetal cells in maternal blood, etc.)
- quantitative assessment of mosaic genomic variants
- predictive/pre-symptomatic assays in unaffected relatives (or a fetus) to determine the risk of inheritance of a familial disorder
- population-based screening for genomic abnormalities (as a component of antenatal and newborn screening programs)
- application of probability, statistics, bioinformatic databases, and other aspects of computer science relevant to the practice of genetic pathology.
- providing advice to laboratory and clinical colleagues regarding selection and interpretation of results of medical genomic assays.

## **PERSONAL CHARACTERISTICS NEEDED**

In brief, the distinguishing traits of a laboratory medical geneticist are:

- specialised knowledge and understanding of the relevance of human genetics to medical practice, particularly for the clinical interpretation of individual patient test results;
- practical appreciation of the areas of clinical practice to which genetic diagnostic laboratories contribute;
- ability to use clinical judgement and knowledge to deal with unexpected, atypical and complex work;
- ability to work predominantly at the interface between the genetic diagnostic laboratory, computer science informatics, patients and clients, and clinical colleagues responsible for their care;
- familiarity with the information systems and data analysis tools required to analyse genomic data;
- ability to work in and lead, where required, multidisciplinary teams in pathology services;
- ability to offer clinically-oriented leadership to multi-professional teams in genetic diagnostic laboratory services, particularly in areas of clinical effectiveness and governance;

- ability to improve laboratory quality assurance by conducting clinically meaningful audit of genetic diagnostic laboratory activities;
- ability to manage ongoing continuing professional development requirements;
- ability to critically evaluate research findings and to contribute effectively to medical research in the discipline;
- ability to contribute to the education and training of clinical and scientific colleagues.

## GENERAL AIMS OF THE TRAINING PROGRAM

The medical genomics training program positions the genetic pathologist to practise as a specialist in laboratory medical genomics, and also to maintain professional skills by ongoing active engagement in the life-long task of continuing professional development. Training is also targeted to equip the genetic pathologist to contribute effectively to translational research and development, undergraduate and post-graduate teaching, as well as other associated professional activities.

Furthermore, the RCPA policy on patient expectations of pathologists specifies that pathologists will:

- Demonstrate and maintain competence
- Be respectful of patients
- Treat specimens respectfully
- Foster constructive collegiality and teamwork within the laboratory
- Be part of the medical team looking after patients
- Provide accurate and timely results
- Be professional in their approach
- Be involved in appropriate accreditation and quality activities
- Provide value for public and private expenditure.

The general aims of the training program relate to four general functions of genetic pathologists who specialise in medical genomics:

- discipline-specific functions as a medical specialist in the laboratory
- role as a manager in the laboratory
- research and scholarship
- professional qualities

These functions are elaborated as specific training outcomes and activities in **Section 2**.

It should be understood that the training outcomes are intended as guides. Whilst training should be comprehensive, it is recognised that not all subjects can be covered in the same detail. Adaptations may also be required to meet local needs.

## TRAINING REQUIREMENTS

To gain the Fellowship in genetic pathology (medical genomics) requires five (5) years of accredited training and satisfactory completion of the assessment program detailed in this Handbook. No more than four (4) years training in one laboratory will be allowed.

Evidence of participation in research activities includes completion of a substantial research project on a topic relevant to medical genomics in the advanced phase of training. Recognition is also provided for research training aspects associated with peer-reviewed activities such as quality assurance, presentations at scientific meetings, publications and/or progress towards, or successful completion of a PhD or MD thesis.

### Exemptions from training time

Trainees who have trained in areas of relevance to genetic pathology may be given recognition of this prior learning towards their five (5) years of approved training. Trainees who have trained with the RCPA in another discipline (ie, haematology, anatomical pathology, immunology, chemical pathology or microbiology), or who have trained in genetic pathology in a similar organisation to the RCPA, may be granted retrospective accreditation of this training.

Trainees who have gained a fellowship from the RACP in clinical genetics (general or cancer) may also have retrospective accreditation of training time.

Please refer to the *RCPA Trainee Handbook – Administrative Requirements* for essential information regarding training limitation, retrospective accreditation of training and temporary suspension of training. Training time exemption tables on the RCPA website provide a guide to retrospective training time credits. However, all applications for retrospective accreditation of training require individual assessment.

## RESEARCH

Research is a component of RCPA advanced training. A research project is component of the part II assessment. Additional research activities include portfolio activities such as peer reviewed quality assurance activities, presentations at scientific meetings, publications and/or progress towards, or successful completion of a PhD or MD thesis.

Planning for the research project component of the part II assessment may commence at any point in training but must not be completed until after having passed the part I assessment. The project demonstrates the trainee's ability to plan, perform and present the results of a scientific investigation in medical genomics.

With prior approval from the College, up to eight months of full-time research training may be accredited provided that the goals of the research project are met.

Trainees who wish to commence a PhD or MD in this research training must have completed the part I assessment prior to enrolment in the PhD or MD. During research training, trainees must complete the required workplace-based assessment tasks of the laboratory training program. Training beyond 5 years is usually necessary to obtain RCPA fellowship and the award of a PhD or MD degree.

See **Appendix 5** for detailed research project requirements.

## SUPERVISION

All training must be supervised. More than one supervisor can be appointed if trainees divide the year between two or more unrelated laboratories. The College recommends that a supervisor should have responsibility for no more than two trainees at a time.

The supervisor will normally be a Fellow of the College, who has a recognised scope of practice in medical genomics. Fellows of the Faculty of Science, who have expertise in relevant aspects of medical genomics, may also provide supervision for trainees. The Board of Education and Assessment may approve a non-Fellow if no Fellow is available locally.

In some circumstances, the Board of Education and Assessment may also require supplementary mentoring supervision provided in-house by a Fellow of the College with recognised scope of practice in another pathology discipline.

If the trainee spends significant periods working in an area where the supervisor has no personal involvement, the supervisor must certify that suitable supervision is being provided. The supervisor must also ensure that adequate supervision is arranged in their absence.

In some circumstances shared supervision may be necessary, but there must be a nominated primary supervisor with overall responsibility. Trainees working towards higher academic degrees (e.g. PhD), who find that their research supervisor is not suitable to be the RCPA training supervisor, should nominate an RCPA Fellow as co-supervisor.

Workplace-based assessment forms are to be signed off by supervisors or laboratory staff members recognised by supervisors to be proficient in the practical aspect that is being assessed, including non-Fellow laboratory scientists.

Supervisors should devise a prospective training (or research) program on initial registration and annually. This should be devised in collaboration with the trainee and submitted to the RCPA. Supervisors should verify beforehand that the trainee has sufficient time and opportunities to carry

out the required training activities. The program should be reviewed at regular, documented meetings between supervisor and trainee.

Supervisors, and others to whom aspects of training have been delegated, are expected to monitor and provide regular feedback on the development of the trainee's competence. A formal meeting with the trainee should occur every three months. Supervisors should meet regularly with the trainee; observe their laboratory performance and interaction with scientists, peers and clinicians; and review result reporting. This may be delegated to other trainers where appropriate, eg, when the trainee is on secondment to another laboratory for a segment of training.

The formal duties of supervisors, such as requirements to report the trainee's progress to the Board of Education and Assessment, are outlined in **Appendix 6** and more fully described in the RCPA Induction Manual for Supervisors and the RCPA policy on the Role of the Supervisor. Please refer to these documents for detailed information.

## ASSESSMENT

Assessment is by formal examination and by submission of a portfolio, which is a record of workplace-based assessment and other achievements during training. The periodic and annual supervisor's reports are also kept in the portfolio. The requirements are summarised below. Please refer to the Appendices for details.

### *Examinations*

- 1. Basic Pathological Sciences examination:** usually taken before or during the first year of training. All trainees are required to undertake or apply for exemption from the Basic Pathological Sciences examination. See **Appendix 2** for detailed requirements.
- 2. Genetic Pathology (Medical Genomics) part I examination:** RCPA trainees may not take this examination until the third (3<sup>rd</sup>) year of training. The goal is to ensure that trainees can appropriately mix the laboratory/scientific and clinical elements of genetic pathology. Laboratory management does not feature in the part I examination, except where there is major overlap with scientific/technical areas, such as quality assurance/quality control. See **Appendix 3** for detailed requirements.
- 3. Genetic Pathology (Medical Genomics) part II examination:** trainees who pass part I are eligible to sit the part II examination, ordinarily in the final year of training. The focus is on integration of technical/scientific knowledge with clinical and management elements. Clinical elements are emphasised more than management. The goal of this examination is to determine whether the candidate is competent to function as a consultant. See **Appendix 4** for detailed requirements.

All durations refer to full-time training (or part-time equivalent) in an accredited laboratory. If it is found that after the submission of an examination application form, a trainee has not met the eligibility criteria for sitting an examination/s, e.g. insufficient accredited training time, the trainee will be withdrawn from the examination/s. The trainee will be eligible for a full refund of the examination fee.

### *Research project*

The research project should be a substantial amount of laboratory-based work, undertaken after having passed the part I assessment, See **Appendix 5** for detailed requirements.

### *Portfolio*

The **portfolio** is a physical collection of workplace-based assessment forms and other documents that provide evidence that trainees have successfully completed a range of activities that form part of their daily work in the laboratory. The portfolio records the trainee's progress in developing technical skills and professional values, attitudes and behaviours that are not readily assessed by formal examinations. Details of the portfolio requirements to be completed before the part I and part II examinations are in **Appendix 7**.

Trainees are responsible for initiating their workplace-based assessments and ensuring that they have completed the required number by the required dates. They should identify opportunities to have their competencies assessed, negotiate an appropriate time for assessments by suitably qualified assessors and ensure that assessors have the appropriate form. Assessments should be able to be done regularly without significant disruption to workplace productivity.

## RESOURCES

Texts, journals and weblinks are in the [Genetics](#) section of the RCPA website. Other peer-reviewed resources should be consulted as necessary for comprehensive coverage, especially contemporary reviews and key papers in the general medical genomics literature.

## SECTION 2

# LEARNING OUTCOMES AND RECOMMENDED TRAINING ACTIVITIES

In Section 2 of the Handbook, the four broad functions of the genetic pathologist who specialises in medical genomics are elaborated as sets of training outcomes and suggested training activities.

The learning outcomes are denoted as to be achieved early in training [E] or at a more advanced level [A]. Competence in outcomes achieved early in training should be maintained throughout. Trainees are not expected to do every activity in the lists but should select those that are likely to meet their needs, being mindful of the range of learning opportunities offered by their laboratory.

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# 1 DISCIPLINE-SPECIFIC KNOWLEDGE, SKILLS AND LABORATORY PROCESSES

Pathologists with expertise in medical genomics contribute to the diagnosis and management of disease by analysing genomic and epigenetic variants. Clinical interpretation of results is central to the role of providing an effective diagnostic service and offering clinically useful advice to clinicians. Medical genomic specialists have expertise in the genomic and epigenetic basis of human diseases, with a particular focus on constitutional disorders rather than on cancer or infectious diseases.

Genetic pathologists serve to strengthen the clinical focus of laboratory services, enhance the clinical focus of quality assurance activities and enrich clinically-directed service development and innovation. Together with specialist medical scientists, genetic pathologists are responsible for running laboratories with both manual and automated components and ensuring the quality of the results. Genetic pathologists must understand a wide variety of genomic and epigenetic analytical techniques, be able to solve problems that arise in the laboratory and make informed decisions with regard to the selection of laboratory methods most applicable to specific clinical tasks. Genetic pathologists must also have expertise in the selection and functioning of computing and laboratory information systems upon which the efficient operation of the laboratory depends.

By the end of training, trainees are expected to be fully knowledgeable and technically competent in routine genomic and epigenomic investigations and be competent to provide advice to clinicians. They should also have observed and reflected on the ways that senior medical genetic specialists fulfil the role of medical specialist in the laboratory and have participated in the more demanding aspects of the role, as appropriate for their stage of training, assuming increasing levels of responsibility as they progress. They should also know how to access expertise in all these areas and to consider where their own specific interests lie and how to develop them further.

The following lists of learning outcomes and activities serve to guide what trainees should achieve by the end of training.

## 1.1 Cell biology, genetics and epigenetics (refer to Appendix 1A for details)

### Outcomes

- [E] \* Explain basic eukaryotic and prokaryotic cell biology;
- [E] Explain mutational mechanisms;
- [E] Explain the nature and range of consequences arising from genomic and epigenomic variation;
- [E] Explain the nature and cellular basis of human inheritance patterns;
- [E] Explain the nature of and basis for genome variation in populations.

## 1.2 Whole organism and tissue biology (refer to Appendix 1B for details)

### Outcomes

- [E] Explain human life cycle-associated changes in the biology of tissues and organs;
- [E] Explain genetic/epigenetic disease-associated perturbances to normal tissues and organ function;

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\* [E] Early stage learning outcome.

## Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Maintain current knowledge of basic sciences underpinning human medical genetics/genomics by reading relevant journals, text books, on-line resources and other relevant material;
- Attendance at clinical meetings;
- Case-based discussions;
- Research and scholarly activities.

### 1.3 Genomic and epigenetic testing in clinical practice

#### Outcomes

- [E] Explain categories of genomic/epigenomic testing (*refer to Appendix 1C for details*);
- [E] Explain the range of clinical scenarios for which genomic/epigenomic testing has clinical utility (*refer to Appendix 1C for details*);
- [E] Explain the range of therapeutic and management options, as well as life choices influenced by genomic/epigenetic test results (*refer to Appendix 1C for details*).

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Maintain current knowledge of medical genomics by reading relevant journals, text books, on-line sources and other relevant material;
- Maintain current knowledge of therapeutic and investigative aspects of other disciplines that are relevant to medical genomics;
- Attendance at clinical genetic clinics (general, dysmorphology, cancer, cardiovascular, neurogenetics, fetal medicine meetings, prenatal clinics);
- Clinical meetings;
- Attendance at multidisciplinary pathology meetings;
- Consultations with medical colleagues about genetic investigation of patients;
- Case-based discussions.

### 1.4 Investigative pathways

#### Outcomes

- [E] Explain and follow guidelines for laboratory assessment of possible genetic/epigenetic aetiologies of disorders including, but not limited to clinical indications listed in *Appendix 1C, Clinical indications for genomic/epigenetic investigations*.

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Direct observation of communications skills (DOCS);
- Case-based discussions;
- Attendance/presentations at clinical meetings;
- Consultations with medical colleagues about genetic investigation of patients.

## 1.5 Genomic/epigenomic variant detection strategies

### Outcomes

- [E] Conduct laboratory diagnoses of genomic/epigenetic disorders by appropriately targeted analysis of nucleic acids and associated functional tests (refer to *Appendix 1D, Genomic/epigenomic variant detection strategies*).

### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Direct observation of practical skills (DOPS);
- DOCS.

## 1.6 Pre-analytical processes

### Outcomes

- [E] Explain strategies for minimising risk of pre-analytical sample error;
- [E] Demonstrate competence in evaluating clinical requests for genetic testing;
- [E] Demonstrate proficiency in liaising with clinical referrers;
- [E] Demonstrate competence in selecting cost-effective testing pathways for clinical requests.

### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- DOCS.

## 1.7 Laboratory procedures

### 1.7.1 Handling, storage and retrieval of laboratory samples, reagents and data

#### Outcomes

- [E] Prepare, label, store and handle reagents correctly;
- [E] Handle and label patient samples correctly;
- [E] Use laboratory information systems in recording patient and request information, including a storage and retrieval system for specimens, results, comments and final reporting;
- [E] Comply with the specimen storage and indexation conventions of the laboratory;
- [E] Use the laboratory information system to retrieve reports/specimens for examination and review to satisfy clinical audit and/or research purposes;
- [E] Prepare, isolate, concentrate and purify samples for cell-based analysis (*refer to Appendix 1E for details*);
- [E] Prepare, isolate, concentrate and purify samples for nucleic acid analysis (*refer to Appendix 1E for details*);
- [E] Ensure the stability and correct storage for long-term preservation of patient tissues and samples;
- [E] Knowledge of the principles of database organization and maintenance of data integrity;
- [E] Understanding of IT security issues, privacy principles and ISO standards, codes of practice;
- [E] Understanding of laboratory and health information management, including standards for interoperability and data sharing.

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Read laboratory manuals and NATA/RCPA, IANZ and other relevant guidelines;

- During assay evaluation retrieve selected specimens;
- Use LIMS to retrieve specimens with particular abnormalities for clinical review.

### 1.7.2 Practical skills

#### Outcomes

- [E] Proficient in microscopy (bright-field and fluorescence);
- [E] Proficient in banding and karyotype analysis;
- [E] Proficient in FISH analysis;
- [E] Proficient in array technologies and analysis;
- [E] Proficient in PCR-based assays (end point, quantitative and real-time) and analysis;
- [E] Proficient in gel-based hybridisation and analysis;
- [E] Proficient in fragment separation, electrophoresis and analysis;
- [E] Proficient in DNA sequencing (classical) and analysis;
- [E/A] Proficient in DNA sequencing (massively parallel) and analysis (*library preparation, sequencing methodologies, informatics and data analysis*).

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- DOPS – refer to Appendix 8 for details.

### 1.7.3 Analysis and validation of laboratory data

#### Outcomes

- [E] Understand methods used to assess the validity of tests specified in *Section 1.7.3.1* below (*Knowledge required to evaluate genomic tests*);
- [E] Demonstrate competence in monitoring data quality and verifying results in accordance with laboratory procedures;
- [E] Understand the sources of bias and artefacts in genome analysis (*e.g. age of DNA samples; DNA quality, chimerism, etc.*);
- [E] Demonstrate competence in the assessment and interpretation of mosaicism (somatic and germline);
- [E] Explain principles of design and scripting of algorithms required to analyse large data files
- [E] Knowledge of threshold call parameters and their limitations;
- [E] Knowledge of risks associated with analysing high-volume data (*e.g. curse of dimensionality; bias in cross validation*);
- [E] Understand limits associated with analysing large data files (*e.g. algorithmic complexity; intractability*);
- [E] Understand the principles, requirements and functioning of laboratory information systems used for instrument interfacing, flagging of results and generating interpretive comments;
- [E] Understand the components and limitations of high-volume genome data analysis pipelines (quality assessment [*checking signal-noise ratio; etc.*]; data flow; data validation; mapping; dynamic querying; assembly visualization);
- [E] Understand how to implement trouble-shooting procedures;
- [A] Knowledge of statistical approaches to analysing high volume data.

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Maintain current knowledge by reading relevant journals and text books to supplement laboratory manuals;
- Follow up problem cases;

- Perform calibration procedures on as many platforms as possible (activity reflection);
- DOPS.

### 1.7.3.1 Knowledge and skills required to evaluate genomic test data

#### Outcomes

- [E] Calculate and interpret statistical data related to the reproducibility and validity of diagnostic tests, including sensitivity, specificity, positive predictive value, receiver operating characteristics analysis, spectrum & bias, predictive values, likelihood ratio, and Bayes' theorem;
- [E] Understand and apply non-parametric statistics;
- [E] Understand, apply and interpret methods of bivariate statistical analysis;
- [E] Understand, apply and interpret methods of multivariate statistical analysis (*as applicable to the analysis of large data sets obtained from a single sample*);
- [E] Demonstrate competence in validating large data sets;
- [E] Understand, apply and interpret methods of measuring data quality;
- [E] Demonstrate competence in evaluating and expressing uncertainty in measurement.

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Refresh knowledge of general statistical concepts and procedures relevant to medical genomics;
- Review the literature and other documentation on defining the reproducibility and validity of diagnostic tests;
- Research and scholarly activities;
- Quality activities.

## 1.8 Post-analytical processes

### 1.8.1 Interpreting genomic data

#### Outcomes

- [E] Proficient in the use of ISCN and HGVS nomenclature for genomic variants;
- [E] Proficient in genetic mapping of Mendelian characters (linkage analysis), particularly as a tool for estimating genetic risks facing an individual in families where the disease-causing mutation remains unknown;
- [E] Proficient in the clinical evaluation of constitutional genomic anomalies detected by karyotyping, array or other molecular methods (*including autosomal and sex chromosome aneuploidy; polysomies; structural anomalies; translocations; microdeletion syndromes; loss of heterozygosity; uniparental disomy; identity-by-descent*);
- [E] Proficient in the clinical evaluation of somatic genomic and epigenetic anomalies detected in constitutional mosaicism and malignancy by karyotyping, array or other molecular methods (*including chromosomal aneuploidy; polysomies; structural anomalies; translocations; microdeletion syndromes; loss of heterozygosity and uniparental disomy; identity-by-descent*);
- [E] Advanced skills in understanding annotation, navigating and data mining from major genome browsers (Ensembl, UCSC);
- [E] Knowledge and understanding of genome and reference sequence annotation tools;
- [E] Proficient in sequence assembly and alignment;
- [E] Proficient in contextualising DNA sequence data using informatics tools (both proprietary and open-source);
- [E] Proficient in evaluating the potential pathogenicity of an unknown genomic variant (coding and non-coding intragenic regions, as well as extragenic regions);

- [E] Working knowledge of the bioinformatic resources required to analyse and visualise massively parallel sequencing data (e.g. commercial/proprietary suites such as Cartagenia, Alamut/AlamutHT, HGMD professional, Ingenuity, GenGo, Omicia, and academic/freeware mapping/alignment algorithms and tools available on github such as Novoalign, BWA, tmap, IGV, Galaxy and GATK);
- [E] Working knowledge of the bioinformatic tools/suites required to CGH and SNP array data (e.g. proprietary tools such as Cartagenia; and web-based databases such as DECIPHER and Database of Genomic Variants);
- [E] Understanding of network knowledge base analysis of high-throughput genomic data networks.

### 1.8.2 Developing and reporting a professional opinion

#### Outcomes

- [E] Proficient in the synthesis and clinical interpretation of laboratory data taking account of relevant clinical information and aided, where required, by validated bioinformatic and database resources, and peer-reviewed published data;
- [E] Proficient in assessing familial recurrence risks arising from small- and large-scale genomic/chromosomal anomalies;
- [E] Proficient in reporting summary clinical insights that may be relevant to the test outcome, as well as any relevant familial recurrence risks;
- [E] Proficient in taking account of the influence of ethnogeographic origins, particularly when reporting negative genotype findings;
- [E] Add concise guidance about follow-up testing, when appropriate, for test verification, functional assessment of findings and of other family members;
- [E] Use the laboratory information system to design algorithms for investigating and reporting different clinical scenarios;
- [E] Use these algorithms, alert limits, etc, to draw attention to results requiring additional attention and to develop protocols for reflex testing;
- [E] Report in accordance with the relevant regulatory framework;
- [E] Recommend and use standardised information structures, terminology and units for requesting and reporting, e.g. use of formal terminologies;
- [A] Explain evidence-based advice, guideline development, prediction and research, and describe the knowledge and information tools that can be used to help with this.

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Perform in teaching, grand rounds, clinical rounds and contribute effectively to these;
- Participate in signing out;
- Participate in developing/adapting expert system;
- Review the departmental list of medical genomic investigations and define appropriate reporting;
- DOPS.

## 2 FUNCTIONS OF THE MEDICAL GENOMICS SPECIALIST AS A MANAGER

The genetic pathologist will usually be working with a team of people. As a manager, the medical genomics specialist must be fully conversant with topics as disparate as budgeting, safety, privacy, certification and quality, as well as having to represent the department to higher authorities.

By the end of training, trainees will not be expected to take on the responsibilities of a senior manager; however, they are expected to have become familiar with managerial tasks by observing and reflecting on managerial duties and by participating in activities that are appropriate to their stage of training, assuming increased levels of responsibility as they progress. Knowledge of management principles and associated skills are best developed by active participation in regular department management meetings, observing laboratory preparation for NATA inspections, and so on.

The following lists of learning outcomes and activities serve to guide what trainees should achieve by the end of training.

### 2.1 Quality Management

#### Outcomes

- [E] Implement findings from internal and external quality control activities to laboratory procedures;
- [E] Document, notify and apply corrective actions, employing laboratory information systems where appropriate, in the event of incidents, errors and adverse events;
- [E] Promote timely and appropriate use of pathology investigations;
- [A]<sup>†</sup> Contribute to continuous quality improvement by participation in audit;
- [A] Contribute to the development and implementation of quality policy within the laboratory;
- [A] Apply, review and plan for internal quality assessment and accreditation according to accepted norms such as the AS ISO 15 189:2009 standard;
- [A] Apply internal quality assurance processes – standardization, calibration and evaluation of analytical techniques;
- [A] Understand external quality assurance, technical performance and proficiency testing schemes;
- [A] Practice in accordance with the national regulatory guidelines for genetic diagnostic laboratories;
- [A] Explain the relative benefits and disadvantages of the design and operating characteristics of a particular instrumentation or platform.

#### Activities

Select Activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Complete the [Quality Management](#) eLearning module in RCPA Education Online and print a copy of the certificate of completion for the portfolio;
- Review summaries of relevant requirements for laboratory accreditation and performance, for example the NATA Checklist for Laboratory Accreditation;
- Review recent audit assessment reports of your laboratory and identify any contentious issues; Opportunity for entry into written communication, management and ethics log
- Participate in a quality audit (activity reflection);

<sup>†</sup> [A] Advanced level learning outcome

- Review laboratory internal QC procedures and update if required;
- Review external QA program results and any remedial actions;
- Participate in the establishment of a new in vitro diagnostic test;
- Participate in case/slide/laboratory/clinical rounds, peer review meetings, external quality assurance (e.g. HGSA/RCPA QAP, EMQN and ASoC) and continuing professional development activities;
- Read current literature on QA strategies, risk management, informatics and evidence based medicine in chemical pathology laboratories;
- Participate in workflow checks to ensure effective and efficient laboratory function;
- Recognise, report and analyse quality problems when they arise in the laboratory;
- Participate in implementing plans for testing and evaluating measures to improve the quality of laboratory practice and patient care;
- Identify the source of water supply in your laboratory. Review the grading of water purification in your laboratory and its quality control;
- Explain laboratory sterilization procedures and the associated measures of quality control;
- Explain the consequences of inappropriate QC limits in terms of assay out-of-control.

## 2.2 Laboratory Safety

### Outcomes

- [E] Explain and act in accordance with relevant WHS legislation, especially relating to biohazard, chemical, radiation and physical safety;
- [E] Explain and act in accordance with relevant waste disposal legislative requirements;
- [E] Explain and act in accordance with in-house laboratory waste disposal procedures;
- [A] Plan, review and apply laboratory safety procedures, which ensure effective protection of self and staff against infection, radiation, toxic, gas, chemical, electrical and fire hazards;
- [A] Evaluate risk assessment processes, in accordance with legal aspects of investigation and disclosure;
- [A] Proficiently evaluate incident reports and near misses to identify opportunities for improvements in practice;
- [A] Contribute to staff protection in the event of an adverse event in the laboratory.

### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Complete the [Laboratory Safety](#) eLearning module in RCPA Education Online and print a copy of the certificate of completion for your portfolio;
- Complete workplace health and safety training in the laboratory (see laboratory safety checklist);
- Review the Australian WHS Act 2011/ WHS standards in your own jurisdiction;
- Participate in WHS drills and meetings, especially fire safety;
- Schedule meeting with workplace WHS Officer;
- Review incident reports and explore relevant improvements.

## 2.3 Compliance with Legislation

### Outcomes

- [A] Demonstrate basic knowledge of requirements of Approved Pathology Provider (Australia) or other relevant undertakings;
- [A] Demonstrate basic knowledge of regulatory requirements of NATA, IANZ or other relevant accrediting authorities;

- [A] Operate with awareness of the potential for medical litigation and the role of pathologists as defendants or consultants, and apply appropriate risk management strategies;
- [A] Ensure laboratory compliance with current requirements for notifiable diseases;
- [A] Identify acceptable standards of billing practice appropriate to the work setting.

### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Review summaries and seek advice from appropriate senior staff;
- Locate sources of pathology financing information, e.g. Medicare Benefits Schedule, Health Insurance Act or other documentation relevant to your jurisdiction;
- Document incidents and discussions with medico-legal implications and discuss with supervisor or a senior colleague;
- Review laboratory manuals and State/Territory/country legislation regarding notifiable diseases;
- Maintain currency with the relevant requirements for notifiable diseases;
- Reflective activities (aspects of laboratory practice).

## 2.4 Managing People

### Outcomes

- [E] Review and use orientation and training protocols for new staff;
- [E] Describe procedures for staff selection, training and performance appraisal;
- [E] Display skills in conflict resolution in the workplace;
- [E] Recognise and cope with stress in self and others;
- [E] Behave in accordance with equal opportunity and antidiscrimination practices in the workplace;
- [E] Be familiar with the RCPA policy on bullying and harassment. Refer to Appendix 1 of the RCPA Trainee Handbook - Administrative Requirements;
- [A] Provide supervision and constructive feedback to staff;
- [A] Contribute to the preparation of duty rosters.

### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Become involved in the overall organization of departmental activities;
- Participate in staff and business meetings in the department;
- Observe administrative procedures in relation to selection and appointment of staff;
- Reflect on observations of interactions in the workplace;
- Observe and develop insight into the knowledge levels and skills of laboratory colleagues;
- Participate in training on giving and receiving feedback and/or read articles on the subject;
- Participate in a conflict resolution course and/or read articles on the subject;
- Assist in the orientation and mentoring of junior colleagues;
- Participate as trainee representative on College committees;
- Participate in procedure for developing a business case for extra staff;

## 2.5 Managing resources

### Outcomes

- [A] Demonstrate understanding of management skills required for development and use of resources in the laboratory;
- [A] Contribute to strategic planning in the department;
- [A] Contribute to the preparation of business plans;
- [A] Describe budgetary considerations in an established genetics pathology laboratory;
- [A] Describe issues concerned with the assessment, procurement, installation, maintenance and use of laboratory equipment and electronic information systems in the laboratory environment and evaluate cost effectiveness;
- [A] Be familiar with procedures used to ensure regular and preventative maintenance of laboratory equipment;
- [A] Review and benchmark the performance of laboratory equipment in terms of breakdowns, and repair frequency, as distinct from planned preventative maintenance;
- [A] Identify sources of funding for laboratory testing;
- [A] Respond effectively to laboratory-related complaints;
- [A] Understand the structure and function of local, state and public sector service providers.

### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Review and discuss with senior staff laboratory budget reports including income, expenditure, salary, overtime, annual leave and sick leave costs, maintenance and consumables costs;
- Participate as an observer in committees concerned with resource management;
- Participate in evaluating the cost-effectiveness of current and proposed laboratory procedures and equipment in the context of limited resources;
- Teach colleagues to use new laboratory equipment and IT software and hardware;
- Attend training sessions concerned with implementing new technology, noting costs and benefits of the technology;
- Access Medicare Benefits Schedule and other documents relevant to your jurisdiction;
- Take part in drawing up an annual department budget and identifying the fixed, variable and discretionary costs;
- Participate in drawing up a tender for new laboratory equipment (opportunity for activity reflection);
- Perform time and motion studies in your own lab and visit other labs of similar size and view their procedures;

## 2.6 Information fundamentals

### Outcomes

- [E] Understand statistical concepts, methods and tools used to assess the accuracy, uncertainty, variation and reproducibility of test results, including data for both individual patients and populations, and to be able to determine confidence levels, reference or expected values and the clinical significance of testing;
- [E] Understand the role and scope of informatics in laboratory medicine, including concepts of information architecture, quality and analysis, systems design, and specialised sub-domains such as bioinformatics, imaging and statistics;
- [E] Explain the basics of laboratory systems architecture and the movement of data for communication of requests, reports and instrument interfacing;

- [E] Identify the information technology environment in which the laboratory information system operates, including integrated systems (i.e. hospital information systems, back-ups, reporting and network structure);
- [E] Describe meaningful and secure use of electronic health records in pathology practice.

### **Activities**

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Access and read documents and view video presentations relating to informatics to be found in RCPA Education Online
- Participate in departmental and clinical meetings;
- Network and share information with colleagues;
- Plan, organise and review teaching activities, together with supervisor, peers and laboratory staff;
- Participate in College activities and meetings.

### 3 RESEARCH AND SCHOLARLY ACTIVITIES

Genetic pathologists require a sound understanding of research methodology and an ability to critically evaluate research findings. These skills are essential for critical appraisal of the benefits and deficiencies associated with new medical and scientific tests and procedures. Research skills allow the medical genomics specialist to contribute to the body of knowledge and ongoing practice improvements in the discipline, as well as to maintain professional competence throughout their career, and also to contribute to the education of colleagues, trainees and the wider public.

By the end of training, trainees should be sufficiently skilled in the methods of scientific inquiry to be able to critically appraise scientific literature and to conduct a small-scale laboratory investigation or participate in larger-scale studies. An appreciation of the challenges associated with formulating and answering even apparently simple questions comes only from active involvement in research projects. Trainees should have developed the self-discipline to support the habit of lifelong self-education. Through personal experience and observation, they should have sufficient understanding of teaching and learning to be able to mentor and supervise junior staff and also to conduct educational sessions for students, colleagues and for the general community.

The following lists of learning outcomes and activities serve to guide what trainees should achieve by the end of training.

#### 3.1 Critical appraisal and research skills

##### Outcomes

- [E] Critically evaluate scientific and laboratory results from a literature review and audit;
- [E] Be able to distinguish a medical audit from medical research;
- [E] Understand and apply the methods used to establish evidence-based practice;
- [E] Understand and apply processes for validating new genetic tests;
- [E] Critically evaluate sources of medical information, discriminating between them in terms of their currency, format, authority and relevance;
- [E] Apply and interpret basic statistical and epidemiological concepts and data;
- [E] Demonstrate skill in developing a research proposal, conducting appropriate research activities and writing up for peer review/publication;
- [E] Comply with the requirements of relevant bodies concerned with ethics in human and animal research;
- [A] Prepare reports and papers for publication that comply with the conventions and guidelines for reporting biomedical research;
- [A] Knowledge and understanding of disease gene discovery strategies (including pedigree segregation analysis, linkage analysis, study of patients with novel genomic variants, position-independent routes to finding genes, twin studies, genetic studies of complex diseases [use of IBD, QTL, LD, association and sib-pair analysis, etc.]);
- [A] Proficient in the critical evaluation of bioinformatic resources;
- [A] Contribute to data analysis and publication in the department.

##### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Organise and present data;
- Prepare scientific articles in the field of medical genomics for publication in a peer-reviewed journal;
- Give an oral presentation or poster presentation at scientific/professional congress or symposium;
- Read and apply NPAAC Guideline: Requirements for the Development and Use of In-House In Vitro diagnostic Devices 2007;

- Participate in and present cases, reviews, original work, to peers at grand rounds, specialist meetings, journal club, etc.;
- Participate in the establishment of a new in vitro diagnostic test;
- Contribute to preparing grant applications, research proposals and ethics submissions;
- Search clinical and laboratory databases to collect, organise and analyse data;
- Use a standard bibliographic application (e.g. EndNote) to download citations and organise them into a personal database;
- Read reference materials on basic statistical and epidemiological concepts;
- Access appropriate resources for relevant information, e.g. text books, journals, databases and other electronic media, including the [research and scholarship](#) section of RCPA Education Online;
- Part II research project.

### 3.2 Self-Education and Continuing Professional Development

#### Outcomes

- [E] As part of a personal continuing education strategy, practise the habit of identifying and documenting own learning needs, planning educational strategies to meet them, monitoring achievements through self-assessment and reflecting on the outcomes;
- [E] Identify personal learning preferences and reflect on how effective they are in developing competence;
- [E] Demonstrate up to date knowledge of and ability to appraise medical and pathological literature and innovations in areas relevant to medical genomics.

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Formulate a personal learning plan;
- Complete an online learning style inventory and explore a variety of ways to learn;
- Apply various computer-based instructional tools, such as electronic tutorials for confirming or updating knowledge and skills;
- Review the [RCPA Continuing Professional Development Program](#) documentation to identify and apply activities and recording strategies that may be applicable. Participate in the RCPA Continuing Professional Development Program;
- Select relevant mentors to guide professional activities;
- Regularly review journals relevant to genetic pathology and participate in or lead discussions on contemporary issues;
- Participate in and present personal work at relevant educational meetings and journal clubs;
- Integrate national and international developments into personal and laboratory practice (opportunity for activity reflection).

### 3.3 Educating colleagues and others

#### Outcomes

- [E] Develop and evaluate educational materials to enable regular participation in teaching programs;
- [E] Prepare and deliver formal and informal teaching sessions at undergraduate and postgraduate level and reflect on their effectiveness;
- [E] Contribute to the growth of scientific knowledge among laboratory personnel, peers, medical students and other health professionals;
- [E] Translate and convey technical concepts and information in an understandable manner to people without a background in genetic pathology;

[E] Organise teaching programs and postgraduate meetings.

### **Activities**

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Participate in and contribute to formal and informal departmental teaching sessions, clinicopathological meetings, conference presentations;
- Organise a scientific staff continuing education program and provide a list of learning objectives associated with each presentation (opportunity for entry into Teaching sessions log);
- Prepare posters or educational articles of scientific investigations in genetic pathology and present to peers and other health professionals;
- Review or develop educational materials for non-pathologists, eg, LabTests Online;
- Identify and record examples in which training deficiencies lead to lab problems and then implement staff training to remedy identified deficiencies (opportunity for activity reflection);
- Mentor students and other trainees and advise on effective preparation for examinations;
- Read relevant journals, including articles on effective teaching strategies;
- Participate in training on the effective teaching and supervision of adult learners in laboratory and clinical settings, e.g, "Teaching on the Run" if offered in your hospital;
- Seek evidence of your own teaching effectiveness;
- Case-based discussions.

## **3.4 Providing Data for Planning and Evaluation**

### **Outcomes**

[A] Identify requirements for reporting and costing of clinical and laboratory information and requirements in the provision of new services;

[A] Become familiar with the Benchmarking in Pathology Program.

### **Activities**

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Assemble information regarding costs, test selection and possible test numbers to assist in health service planning. e.g. the introduction of a new genetic test, or a business case for a new analytical instrument or software (opportunity for activity reflection);
- Part II research project.

## 4 PROFESSIONAL QUALITIES

Genetic pathologists work effectively with the laboratory and clinical team to ensure timely, appropriate and accurate patient diagnoses. They also perform tests and procedures that require them to ensure patient safety, comfort, confidentiality and privacy. They respect patient confidentiality and rights and conduct themselves in a professional manner at all times, being responsible and accountable to colleagues and the community.

During training, trainees should reflect on and strive to adopt the attitudes and values that underpin professional practice and take advantage of opportunities to extend themselves in these areas, so that by the end of training they are fully able to assume their professional responsibilities.

The following lists of learning outcomes and activities serve to guide what trainees should achieve by the end of training.

### 4.1 Ethics and Confidentiality

#### Outcomes

- [E] Practise ethically, which includes:
- promptness of reporting;
  - interacting appropriately with others;
  - respect for patient autonomy;
  - knowing when to seek opinion from others;
  - financial probity;
  - recognising and handling conflict of interest;
  - recognising, acknowledging and responding adequately to personal mistakes, responding effectively to complaints;
- [E] Comply with legal, ethical and medical requirements relating to patient records and documentation, including confidentiality, informed consent and data security;
- [E] Differentiate between ethically appropriate and ethically inappropriate procedures;
- [E] Identify appropriate courses of action in regard to unprofessional conduct by or ill health in a colleague;
- [E] Recognise, acknowledge and respond adequately to mistakes that impact on patient care.
- [E] Comply with copyright and intellectual property rules;
- [E] Describe strategies to ensure equity of access to pathology testing for patients.
- [E] Maintain patient safety, comfort, confidentiality, privacy while performing tests and procedures;
- [E] Respond effectively and ethically to serve the needs of patients and their families;
- [E] Act in accord with principles of beneficence and non-maleficence;
- [E] Advocate for, and protect, patient rights.

#### Activities

Select activities that establish knowledge/ proficiencies (where applicable, retain records for portfolio):

- Review appropriate literature and guidelines including the National Patient Safety Education Framework;
- Read the most recent Australian Medical Association Code of Ethics;
- Read the Australian Medical Council Good Medical Practice Code of Conduct;
- Access and read documents relating to cultural competence, including those concerning indigenous people, such as Aboriginal and Torres Strait Islander and Maori people;
- Reflect on professional behaviour of self and others, identifying potential for ethical dilemmas and strategies to deal with them;
- Complete the 6 [Ethics eLearning modules](#) in RCPA Education Online (mandatory).

- Complete relevant activities from the [Monash University Clinical Ethics Resource](#) (optional);
- Read the NHMRC document - National Statement on Ethical Conduct in Human Research (2007, updated 2009);
- Case-based discussions;
- Opportunity for entry into written communication, management and ethics log.

## 4.2 Communication

### Outcomes

- [E] Appreciate the different levels of understanding of genetic diseases among specialist and non-specialist colleagues as well as among patients and families;
- [E] Use appropriate language in all communications, showing awareness of cultural and linguistic diversity and level of understanding of the requestor;
- [E] Listen well and obtain relevant clinical information;
- [E] Produce concise, grammatically correct written reports directed towards the clinical question being asked;
- [E] Demonstrate respectful interpersonal communication skills such as active listening and accepting and offering appraisal;
- [E] Comply with guidelines for handling sensitive information;
- [E] Consult with clinical specialists and pathologists on issues of patient care and professional practice and in seeking and providing referral opinion on difficult cases;
- [E] Advise clinicians on the appropriate choice and performance of laboratory procedures and the interpretation and relevance of pathological findings, taking into account clinicians' and patients' needs;
- [E] Advise laboratory staff about testing methodologies, quality assurance techniques and delineating protocols for the issuing of results;
- [E] Pay prompt attention to communicating urgent and critical results.

### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Participate in effective discussion of the detail of medical cases with colleagues and clients;
- Participate in effective discussion about medical/diagnostic uncertainty with colleagues, clients;
- Prepare concise and effective written and verbal reports about patient cases;
- Participate in training sessions on communications, cross-cultural communications, presentation skills, etc;
- Document telephone communication of pathological findings, interpretations, clarification of requests and complaints where appropriate, seeking feedback on the quality of your communication from supervisors and colleagues;
- Read documents relating to etiquette and proper use of electronic communications;
- Consult style guides for correct use of grammar and terminology for written communications;
- Communicate urgent results and document appropriately;
- Become familiar with policies and procedures relating to printing of results, including incomplete requests and site of printing (e.g. the ward);
- Part II research project;
- Case-based discussions.

### 4.3 Collaboration and teamwork

#### Outcomes

- [E] Contribute effectively to the activities of laboratory and health care teams, recognizing responsibilities and limitations of own role;
- [E] Consult with laboratory colleagues, other medical practitioners, pathology informaticians and health care professionals;
- [E] Contribute effectively to inter-disciplinary team activities, such as peer review sessions and other education and quality activities, recognizing responsibilities and limitations of own role;
- [E] Promote the role of pathologists as vital contributors to patient care;
- [E] Communicate directly with clinicians about results, when appropriate.

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Identify the elements of an effective team and reflect on your observations of teams in your work place and others with which you interact;
- Participate in departmental and clinical meetings;
- Network and share information with colleagues, using available technologies;
- Plan, organise and review teaching activities, together with supervisor, peers, laboratory staff;
- Participate in mentoring programs;
- Participate in College activities and meetings;
- Work in a collegial and forthright manner with colleagues;
- Case-based discussions.

### 4.4 Cultural competence

#### Outcomes

- [E] Demonstrate an awareness of cultural diversity and the ability to function effectively, and respectfully, when working with and treating people of different cultural backgrounds. Diversity includes but is not limited to ethnicity, gender, spiritual beliefs, sexual orientation, lifestyle, beliefs, age, social status or perceived economic worth;
- [E] Apply knowledge of population health, including issues relating to health inequities and inequalities; diversity of cultural, spiritual and community values; and socio-economic and physical environment factors; to specialist pathology practice
- [E] Apply knowledge of the culture, spirituality and relationship to land of Aboriginal, Torres Strait Islander and/or Māori peoples to specialist pathology practice and advocacy

#### Activities

Select activities that establish knowledge and proficiency (where applicable, retain records for portfolio):

- Access and read documents relating to cultural competence, including those concerning indigenous people, such as Aboriginal and Torres Strait Islander and Maori people
- Participate in departmental and clinical meetings;
- Network and share information with colleagues;
- Plan, organise and review teaching activities, together with supervisor, peers and laboratory staff;
- Participate in mentoring programs;
- Participate in College activities and meetings;
- Case-based discussions;
- Complete the [Cultural Competence eLearning modules](#) in RCPA Education Online and print the email verifying completion for your portfolio: OR provide evidence of completion of cultural competence training provided by your employer, if a registered health services provider

## Section 3

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## Appendix 1

### Essential topics in Medical Genomics

#### *A. Cell biology, genetics and epigenetics*

##### Cell biology

- Eukaryotic and prokaryotic cell structure and organelle function
- Key differences between prokaryotic, eukaryotic and viral genetics
- Eukaryotic chromosome structure and function; relationship to chromosome banding.
- Nucleic acid structure and biology
- Genome organisation (including repetitive DNA, transposons, gene regulatory regions, pseudogenes, etc)
- Mitochondrial genome (structure and function)
- Cell cycle, including genome replication and repair
- Meiosis, recombination and chromosomal segregation
- Chromatin structure and function; higher-order chromatin folding and fractal packaging.
- Gene structure, expression and regulation
- RNA transcription and post-transcriptional processing.
- RNA-mediated regulation of cell function
- Protein translation and post-translational processing and targeting
- Epigenetic regulation (DNA methylation, histone modifications/ histone code, RNA-mediated regulation of gene expression)
- Epigenome organization
- Epigenetic inheritance through mitosis and meiosis

##### Mutational mechanisms

- Mechanisms of DNA damage, including UV light, ionising radiation and chemicals
- Purine and pyrimidine stability and metabolism
- Defects of DNA replication and DNA repair
- Repeat mediated non-allelic homologous recombination (NAHR), gene conversion and non-homologous end joining (NHEJ)
- Oligonucleotide repeat (e.g. triplet repeat) expansion mutagenesis
- Copy number mutations

##### Genomic and epigenomic variation

- Natural copy number, structural and sequence variation in the human genome,
- Effects of mutations on normal cell function (loss-of-function; gain-of-function; dominant-negative)
- Genome variant nomenclature – DNA (including cytogenetic and array-detected genomic lesions): RNA, and protein level nomenclature
- Epigenetic dysregulation and disease
- Epimutation
- Disorders of genomic imprinting (constitutional and somatic)

##### Inheritance patterns

- Standard patterns and modifiers of mendelian inheritance
- Cell biological basis of mendelian inheritance
- Complex, multifactorial and quantitative traits

- Epigenetic inheritance through mitosis and meiosis
- Epigenetic influences on mendelian patterns - imprinting, X chromosome inactivation and other trans-generational phenomena
- Other non-classical forms of inheritance – mitochondrial; pseudoautosomal, uniparental disomy (UPD) and oligonucleotide repeat disorders
- Origin, incidence and inheritance of chromosomal and large-scale genomic variants.
- Maternal and paternal age effects

### **Population Genetics/ Miscellaneous**

- Factors determining genetic population structure – mutation rates; selection; migration, and random drift
- Population description - Hardy-Weinberg equilibrium and the estimation of gene frequencies
- Estimation of factors affecting the genetic structure of populations
- Deviations from random mating – assortative mating; consanguinity
- Overview of genetic similarities and differences across human populations
- Frequency of genetic diseases in populations
- History of genetics in medicine
- Contemporary insights from hominid phylogenetics

### ***B. Whole organism and tissue biology***

- Gametogenesis, fertilisation and infertility
- Pre-implantation morula; placentation; early embryogenesis
- Twins and twinning
- Chimerism
- Mosaicism – confined placental mosaicism, constitutional; malignant change
- Uniparental disomy
- Cell differentiation/ migration/developmental fields
- Cancer cell biology
- Epigenetic features of gametogenesis; early embryogenesis, and stem cells
- Chromosomal breakage syndromes
- Genomic and epigenomic architecture of tumourigenesis
- Genomic and epigenomic features of aging

### ***C. Genomic and epigenetic testing in clinical practice***

#### **Clinical categories of testing**

- Diagnostic testing
- Predictive/pre-symptomatic genetic testing
- Genotyping (constitutional and somatic) to predict drug responsiveness/toxicity/side-effects (targeted therapeutics, pharmacogenetics/pharmacogenomics)
- Genotyping (constitutional) to avoid transfusion reaction/ transplant rejection/ graft v host reaction
- Genotyping (somatic) to determine clinical subcategories
- Therapeutic monitoring (quantitative genomic and transcriptome analysis)
- Carrier testing
- Population screening
- Prenatal testing (fetal tissues – amniocentesis; CVS; fetal blood)

- Prenatal testing (“cell-free” nucleic acids and fetal cells in maternal blood)
- Pre-implantation genetic testing
- Parentage testing

### **Clinical indications for genomic/epigenetic investigations**

- Clinical presentations in all medical specialty areas are listed to indicate the breadth of the examination. Unlisted emerging clinical scenarios, which are of practical utility may also be included.
- Cardiovascular (congenital heart disease; inherited dyslipidaemias; ischaemic heart disease; inherited cardiomyopathy [HOCM and dilated]; inherited arrhythmias; hypertension)
- Connective Tissue – skin, bone, blood vessels (e.g. Marfan syndrome and disorders of fibrillin; Ehlers-Danlos syndrome; pseudoxanthoma elasticum, cutis laxa and disorders of elastin, osteogenesis imperfecta, skeletal dysplasias (more frequently occurring e.g. achondroplasia, hypochondroplasia, Stickler syndrome; Leri-Weill syndrome, camptomelic dysplasia); epidermolytic diseases; ectodermal dysplasia – anhydrotic and others; ichthyosis; DNA repair anomalies)
- Endocrine (diabetes mellitus types 1 and 2; parathyroid anomalies, calcium and bone mineralisation disorders; adrenalopathies; obesity)
- Fertility (Infertility [male and female]; Premature ovarian failure)
- Gastrointestinal (GI congenital anomalies; cystic fibrosis; pancreatic; inflammatory bowel disease; hepatic iron overload; Wilson’s disease)
- Gene- and cell-based therapies (in particular, therapeutic interventions that may be aided by laboratory monitoring programs)
- Haematology – haemoglobinopathies; inherited red cell dyscrasias; bleeding disorders (e.g. haemophilias, vWD, TTP/atypical HUS); thrombophilia; haematologic neoplasms and related disorders; HLA system (for transfusion and stem cell transplantation), and blood groups (e.g. red cell and platelet antigens/antibodies and their consequences for transfusion and pregnancy).
- Immunology (HLA complex and transplantation genetics; primary immunodeficiencies; inherited complement defects)
- Inborn errors of metabolism (the more frequently occurring or clinically significant inborn errors presenting in childhood/ early adulthood, as well as the categories of disorders detected by newborn screening. i.e. phenylketonuria, including the management of pregnancy, homocystinuria, galactosaemia, organic acidaemias, porphyrias, etc.)
- NB – categories of inborn errors of metabolism are comprehensively described in the [Biochemical Genetics Trainee Handbook](#)
- Infectious diseases (genetic and epigenetic contribution to susceptibility to infections – tuberculosis; severe bacterial infections; parasitic infections (e.g. toxoplasmosis, malaria); viral (e.g. chronic active hepatitis, HIV)
- Neonatology (common birth defects; ambiguous genitalia; seizures; floppy infant; arthrogyposes)
- Neurology (weakness/inherited myopathies, including myotonic dystrophy; inherited myasthenias; inherited periodic paralyses; spinal muscular atrophies; inherited motor neurone diseases; inherited neuropathies – motor/ sensory/ autonomic; inherited ataxias; inherited paraplegia; epilepsy syndromes; movement/basal ganglia disorders; demyelinating disorders; dementia)
- Obstetrics / Fetal Medicine (pre-conception genetic testing; first trimester screening; pre-implantation genetic diagnosis; spontaneous miscarriage; recurrent abortion; pre-eclampsia; major congenital malformations presenting to fetal medicine; stillborn infant)
- Ocular (colour blindness; strabismus; defects of the lens and cornea; optic atrophy and congenital blindness; retinal and choroidal degenerations; glaucoma)

- Oncology (breast; ovarian; colorectal; melanoma; prostate; brain; lymphoma; phacomatoses, phaeochromocytoma; thyroid; retinoblastoma; etc)
- Otic (congenital and inherited late-onset deafness)
- Paediatrics and development (developmental delay/ cognitive impairment; pervasive developmental disorders; hypotonia; cerebral palsy; developmental regression; epilepsies; neuronal migration anomalies; microcephaly/ megacephaly; movement disorders; premature aging; phacomatoses; mitochondrial anomalies; common bone dysplasias, e.g. achondroplasia; hypochondroplasia; etc)
- Pharmacology (genotyping to predict responses to, or side effects from, drugs; monogenic disorders predisposing to adverse drug reactions [e.g. malignant hyperthermia, G6PD deficiency, porphyrias])
- Psychiatry (schizophrenia; bipolar disorder; depression; addictive disorders)
- Renal tract (congenital anomalies of the genitourinary system; renal cystic diseases; renal dysplasia; nephrotic disease; salt-wasting nephropathies)
- Respiratory (ciliary defects; cystic fibrosis; asthma; emphysema; bronchiectasis)

**Range of therapeutic interventions/management options/ life choices that may be influenced by genetic test results, eg,**

- Inborn errors – (dietary management, metabolic inhibitors/activators, cofactors, plasma exchange/plasmapheresis, chelation, organ transplantation, enzyme replacement, chaperone therapy, gene therapy, stop-codon read-through)
- Neurodegenerative disorders (e.g. Huntington disease, spinocerebellar ataxia, mendelian dementias) – assist in life choices
- Familial cancer (e.g. Breast/ovarian, colorectal) – cancer surveillance, risk-reducing surgery
- Cardiac disease (e.g. familial hypercholesterolaemia, long QT syndrome) – health maintenance programs, drugs, implants

***D: Genomic/epigenomic variant detection strategies***

**Constitutional genomic variants**

- Screening for chromosomal gains and losses;
- Screening for unspecified balanced chromosomal rearrangements;
- Screening genome for unspecified copy number anomalies/ loss of heterozygosity/ UPD;
- Screening genome for unspecified nucleotide-level mutations;
- Screening gene(s) for unspecified nucleotide-level mutations;
- Targeted screening for specific chromosomal translocations/ inversions/ copy number gains and losses;
- Testing for specific copy number anomalies/ loss of heterozygosity;
- Testing for specific nucleotide-level mutations;
- Testing involving use of specific non-disease associated polymorphisms (required for preimplantation genetic diagnosis; maternal cell contamination; etc.);
- Targeted testing for uniparental disomy, methylation anomalies and other epimutations;
- Gene expression analysis.

**Somatic genomic variants**

(includes screening for genome variants in DNA derived from mosaic/chimeric tissues, e.g. fetal cells in maternal blood; tumour material; constitutional chimerism; etc.)

- Screening for chromosomal gains and losses in DNA/cells from mosaic tissues

- Screening for unspecified balanced chromosomal rearrangements in DNA/cells from mosaic
- Screening genome for unspecified copy number anomalies/ loss of heterozygosity/ uniparental disomy in DNA from mosaic tissues
- Screening genome for unspecified nucleotide-level mutations in DNA from mosaic tissues
- Screening gene(s) for unspecified nucleotide-level mutations in DNA from mosaic tissues
- Targeted screening for specific chromosomal translocations/ inversions/ copy number gains and losses in DNA/cells from mosaic tissues
- Testing for specific copy number anomalies/ loss of heterozygosity in DNA/cells from mosaic tissues
- Testing for specific nucleotide-level mutations in DNA from mosaic tissues
- Quantitative analysis for specific mutations in DNA from mosaic tissues
- Screening cells *in situ* for specified mosaic mutations
- Targeted testing for uniparental disomy, methylation anomalies and other epimutations in DNA from mosaic tissues
- Gene expression analysis

## ***E. Laboratory procedures***

### **Cell culture, selection and processing for whole cell-based genetic analysis**

- Processing of samples referred for cytogenetic analysis (whole blood; buccal cells; amniocentesis; chorionic villus sampling; fetal blood; skin biopsy; bone marrow; solid tumour biopsies, etc.)
- Cell culture and selection
- Sterile techniques
- Long-term storage of cells
- Overcoming challenges arising from pseudomosaicism, mosaicism and chimerism (confined placental mosaicism; constitutional mosaicism; cancer; fetal cells in maternal blood; twin chimerism; etc.).
- Culture, synchronisation, harvest and fixing of metaphase cells for cytogenetic analysis
- Slide making and banding (various band staining techniques)

### **Nucleic acid preparation, storage and labelling**

- Processing of samples referred for molecular analysis (whole blood; buccal cells; amniocentesis; chorionic villus sampling; fetal blood; skin biopsy; bone marrow; solid tumour biopsies, etc.)
- DNA/ RNA extraction techniques
- Nucleic acid quality determinants
- Nucleic acid quality assessment
- Long-term storage/ archiving of nucleic acids
- DNA labelling for FISH, microarray, other hybridisation-based procedures

### **PCR**

- PCR optimisation and troubleshooting
- Contamination issues
- Principles and practice of PCR primer design
- Quantitative PCR analysis
- RT-PCR (including *in situ* PCR)
- Cloning by PCR
- PCR mutagenesis

## **Methylation analysis**

- Restriction enzyme-based methodologies
- Bisulfite modification-based methodologies
- Quantitative methylation analysis
  - CpG sites
  - epialleles
- Bisulfite modification-free methodologies

## **Urgent clinical testing**

- Prenatal – rapid aneuploidy testing (amniocentesis, CVS, fetal blood)
- Postnatal (urgent blood)
- Haemato-oncology (rapid FISH/PCR)

## Appendix 2

### Basic Pathological Sciences Examination

All trainees must pass or be [exempted](#) from the Basic Pathological Sciences examination. The examination may be taken before commencement of training and is open to registered trainees as well as any medical graduate or medical student.

Although a pass in Basic Pathological Sciences is not a prerequisite for attempting Part I examination, a pass or exemption must be achieved before proceeding to sit the Part II examination.

The purpose of the Basic Pathological Sciences Examination is to assess familiarity with the most important pathological processes and biological principles of disease that form essential knowledge for any medical graduate who considers a career in the pathological disciplines.

The examination has become necessary because pathology may no longer be taught as a “core” discipline in some Australasian medical schools, hence an understanding of basic patho-biological processes is no longer guaranteed in many medical graduates. Such knowledge is essential for a successful start and satisfactory progress in the training program.

#### ***Examination Format and Content***

The examination is a single 2.5 hour paper of 100 one-best-answer multiple choice questions, based on the [BPS syllabus](#) on the RCPA website.

The syllabus reflects knowledge that appears in current, authoritative texts as well as newer knowledge that may not yet appear in textbooks.

The topics cover the basic mechanisms of disease that trainees need to understand so they are equipped to train in their chosen discipline and to understand pathology disciplines other than their own chosen field. To cite just a few examples, the microbiology trainee needs to know what a septic infarct looks like; the chemical pathology trainee needs to know about the anatomical pathology changes seen in metabolic syndrome; the anatomical pathology trainee needs to understand why certain antibodies are used in routine diagnosis and the genetic pathology trainee needs to understand how enzyme deficiencies may lead to morphological changes.

The syllabus is primarily based on Chapters 1-11 of the Professional Edition of Robbins and Cotran Pathologic Basis of Disease (9th ed. 2015. Elsevier) by Abul K. Abbas, Vinay Kumar, and Jon C. Aster. References to supplementary materials are also given, which explain details more clearly than the textbook or contain helpful diagrams. As much as possible these references are from Open Access journals, but for copyright reasons the actual articles are not able to be placed on the College website.

## Appendix 3

### Part I assessment

Assessment in part I is by

- Formal examinations;
- A portfolio of evidence indicating completion of a sufficient number and type of work-based activities;
- Satisfactory progress (supervisor) reports

See assessment matrix in **Appendix 9**.

Examinations are prepared in accordance with [RCPA Guideline 3/2015 Quality Framework for RCPA Examinations – Written, Practical and Oral](#).

#### ***Part I formal examinations***

The part I examination comprises written, practical and oral assessments and is considered the barrier examination in medical genomics.

The purpose of the part I assessment is to ensure that trainees have spent time in the laboratory and absorbed the information there, as well as having the competencies required to integrate clinical and laboratory/scientific aspects of medical genomics. Management does not feature in the part I examination, except where there is major overlap with scientific/technical areas, such as quality control.

Trainees are eligible to sit the part I examination after 24 months of accredited laboratory training time.

#### **Phase I:**

**Written paper:** This is a 3 hour and 15-minute paper comprising short answer questions, extended matching questions and worked problems in medical genomics, held at a local centre.

Trainees who fail the written paper will be required to re-sit the following year. Trainees who pass or achieve a borderline grade may proceed to Phase II.

#### **Phase II:**

A “**dry**” **practical examination** of 3 hours and 15 minutes, conducted at a RCPA-nominated venue, which assesses ability to interpret and report laboratory genomic and epigenetic data relevant to a range of clinical scenarios. The examination comprises standard diagnostic material that represents the range of materials handled by typical medical genomic laboratories. In most cases there will be a single correct answer or a clear differential diagnosis determined by the presenting clinical features of the case.

**Oral examination** conducted at an RCPA-nominated venue with two 20-minute stations, each with a standardised set of questions covering a broad range of complex laboratory technical and professional issues, as well as more complex multidimensional problems. A pass is required in both stations.

Candidates who fail the dry practical and/or oral examination will be required to re-sit the following year. Candidates who fail one Phase 2 examination but pass the other do not have to re-sit the passed Phase 1 examination and/or the passed Phase 2 examination if these exams were passed at a sufficiently high standard and if the failed examination is undertaken in the following year.

Candidates who have been invited to the dry practical and oral with a borderline grade for the written examination must achieve a clear pass in both the dry practical and oral examinations to be awarded an overall pass for Part I.

## ***Portfolio***

The portfolio is a record of activities undertaken by the trainee associated with their daily work during the entire period of training. Trainees are advised to commence these activities at the earliest possible time after starting training and to have satisfactorily completed the required number in order to achieve eligibility for the part I examination.

### **The portfolio requirements are set out in Appendix 7.**

Completed portfolio forms should be filed in a folder. The front page of the portfolio should be a print-out of an updated [summary spreadsheet](#). The updated portfolio must be made available to supervisors whenever they are required to prepare a report.

A print copy of the spreadsheet should be sent to the College with the annual and pre-examination supervisor report.

NOTE: The portfolio itself should not be sent to the College unless requested for audit.

## ***Supervisor Reports***

Trainees must submit a completed supervisor report at the following times:

- Annual supervisor report with appended portfolio summary spreadsheet
- Immediately after a laboratory rotation
- Pre-examination supervisor report with appended portfolio summary spreadsheet

It is the trainee's responsibility to ensure that the pre-examination supervisor report is completed and submitted by the due date. Failure to do so may jeopardise training time accreditation or finalisation of examination results. Refer also to **Appendix 6** Guidelines for Completing the Supervisor Report Form.

Any signatory to a supervisor report may be contacted by either the chief examiner or the registrar of the Board of Education and Assessment to confirm evidence of satisfactory completion.

**Summary of assessment requirements for Part I**

Item	Completion	Assessed by	Comments
Supervisor reports: end of rotation, annual and pre-exam reports with portfolio summary spreadsheet as specified	See RCPA web site for submission dates	Reviewed by BEA Registrar and chief examiner or delegate.	See <b>Appendix 6</b>
Portfolio to be signed off by supervisor or delegate	Before part I written exams	Portfolio summary spreadsheet is checked for completeness by BEA Registrar and chief examiner. If not satisfactory, the candidate may be required to undertake further activities.	Portfolio items are to be reviewed by the supervisor when preparing the supervisor's report. The portfolio should not be sent to the College unless requested for audit.  <b>See Appendices 7 and 8</b>
Written examination consisting of short answer and worked problems		Chief examiner. Written responses are independently marked by genetic pathology examiners.	Questions set by the Examinations Subcommittee.
'Dry' practical examination	After passing written examination	Chief examiner. Written responses are independently marked by genetic pathology examiners.	Questions set by the Examinations Subcommittee
Oral examination	Following dry practical examination	Chief examiner and genetic pathology examiner(s)	Questions set by the Examinations Subcommittee

**Assessment calendar**

Please refer to the [RCPA Trainee Handbook – Administrative Requirements](#) (on the RCPA website) for key assessment dates.

## Appendix 4

### Part II assessment

Assessment in part II is by

- Formal examination;
- Research project;
- A portfolio of evidence indicating completion of a sufficient number and type of work-based activities;
- Satisfactory progress (supervisor) reports

See assessment matrix in **Appendix 9**.

Examinations are prepared in accordance with [RCPA Guideline 3/2015 Quality Framework for RCPA Examinations – Written, Practical and Oral](#).

The goal of assessment in part II is to determine whether the candidate has the knowledge, skills and communication ability necessary to function as a consultant. The focus is on integration of technical/scientific knowledge with clinical and managerial elements. The emphasis is clinical with a lesser emphasis on management.

The form of the part II examination, will depend on the type of training undertaken, the laboratory's accreditation status and other considerations which may arise (including performance in specific areas raised during the part I examination).

Appropriate post-graduate or research qualifications e.g. a PhD or MD in a directly relevant field, may be considered suitable for part or all of the part II laboratory training requirements

#### **Part II formal examination**

**Oral examination:** The assessment in the final year of training will involve an oral examination conducted at an RCPA-nominated venue with two 20 minute stations, each with a standardised set of questions. The topics examined may include quality assurance, safety, management, medico-legal issues, communication and teamwork as well as technical aspects of medical genomics. Questions on interpretation of diagnostic material handled by medical genomics laboratories may also be included. The issues are those which a recently qualified Fellow is likely to have to deal with.

A pass is required in both stations. A repeat examination later in the same year may be offered to unsuccessful candidates, at the discretion of the chief examiner.

#### **Research project**

The research project should be a substantial piece of laboratory-based work. A research proposal must be approved by the supervisor before commencing the research. The project must be completed after the part I examination and the report (in triplicate) submitted on the due date, prior to the part II oral examination, for review by two examiners. If, in the opinion of the examiners, the project work is inadequate, the trainee will be asked to revise and resubmit the work. Please see **Appendix 5** for guidelines.

Trainees are advised to keep their own copy as the copies sent to the College will not be returned.

#### **Portfolio**

Eligibility for the part II examination includes having completed the portfolio of evidence of having participated in a sufficient number and type of workplace activities.

The part II portfolio requirements are set out in **Appendix 7**.

The portfolio folder must include all completed portfolio forms. The front page of the portfolio should be a finalised portfolio summary spreadsheet. The portfolio itself should not be sent to the College unless requested for audit. Any signatory within the portfolio may be contacted to confirm evidence of satisfactory completion.

## Supervisor Reports

Trainees must continue to submit annual supervisor reports for each year of training, as well as at the end of each laboratory rotation, plus an additional pre-examination supervisor report. The annual and pre-examination reports must have an updated print copy of the portfolio [summary spreadsheet](#). These will be reviewed by the chief examiner and the registrar of the Board of Education and Assessment.

Trainees are responsible for submitting the pre-examination supervisor report by the due date. Failure to do so may jeopardise the accreditation of training time or finalisation of examination results. See **Appendix 6**.

## Summary of part II assessment requirements

A pass or exemption in the Basic Pathological Sciences examination is required before enrolling for the part II examination.

Item	Completion	Assessed by	Comments
All supervisor reports: end of rotation, annual and pre-exam reports with portfolio summary spreadsheet as specified.	See RCPA web site for submission dates	Reviewed by BEA registrar, chief examiner, or delegate.	See <b>Appendix 6</b> .
Research project proposal	Before commencing project	Signed off by supervisor	See <b>Appendix 5</b> .
Research project	Before the part II oral examination	Assessed by chief examiner and two other examiners.	Trainee & supervisor declarations to be completed. 3 copies of the project to be submitted for examination See <b>Appendix 5</b>
All portfolio items to be signed off by supervisor or delegate.	Requirements to be completed before part II oral exam	Portfolio summary spreadsheet is checked for completeness by the BEA registrar and chief examiner If not satisfactory, the candidate may be required to undertake further activities	Portfolio items are to be reviewed by the supervisor when preparing the supervisor's report The portfolio should not be sent to the College unless requested for audit.  See <b>Appendixes 7 and 8</b>
Oral examination	After submission of completed portfolio and achieving a clear pass in the supplementary practical exam if required.	Examiners with at least 5 years post-Fellowship experience.	Questions set by the Examinations Subcommittee

## Assessment calendar

Please refer to the [RCPA Trainee Handbook – Administrative Requirements](#) (on the RCPA website) for key assessment dates.

## Appendix 5

### Research project guidelines

The research project should be a substantial piece of laboratory-based work demonstrating the trainee's ability to plan, perform and present the results of a scientific investigation in medical genomics. The aim is for trainees to gain experience in research, planning and critical analysis of the literature, to gain insight into the limitations and pitfalls of research, to gain skills in critical evaluation of research conducted by others and to improve their skills in written scientific communication. Published case reports and purely clinical research will not suffice for this purpose.

Project planning may begin at any stage of training. A proposal must be completed and signed off by the supervisor before commencing project work. The project must be completed after the part I examination and the report must be submitted in triplicate on the due date, prior to the part II oral exam.

#### **The research and resulting report must be completed before sitting the part II examination.**

The report should be presented in a standard suitable for publication in a peer-reviewed journal, adhering to the norms of scientific writing. Suggested section headings include:

- Abstract
- Introduction
- Materials and methods (including ethical consideration and patient recruitment)
- Results
- Discussion (including problems encountered)
- Conclusions
- Lessons learned, plans for the future

The project will be assessed using the following criteria:

- Are the project aims (scope, purpose, desired outcomes) well formulated?
- Is the background described in sufficient detail to provide a rationale for the project?
- Are relevant concepts and findings from the literature critically reviewed; do they shed light on the subject matter of the project?
- Are the study design, materials and methods appropriate to the project aims; do they reflect an adequate amount of effort?
- Are the findings presented well, interpreted appropriately and discussed adequately?
- Are the conclusions justified and do they relate to the aims?
- Are the lessons derived from project discussed adequately; are the implications related to the candidate's own situation; are the plans for the future realistic?
- Is the report written in sound English, without grammatical and typographical errors?
- Has a standard, consistent method of citing the literature been used?

If the project has resulted in a published first author research article, or if it has been accepted for publication, a copy of the article with a covering letter will suffice instead of a report. Reports must be submitted in triplicate with the completed cover sheet and declaration of originality. If the project is assessed as inadequate, it will need to be revised and resubmitted. The award of Fellowship will be delayed until the project has been graded satisfactory.

Alternatives to the research project are:

- A paper relating to medical genomics of which the trainee is senior or sole author, published in a refereed medical journal;
- A completed thesis in the area of medical genomics, which has been accepted for a PhD or MD degree.



## Medical Genomics Research project proposal

### How to use this form

The purpose of the proposal is to enable your supervisor to ascertain whether your plan is feasible and whether the resulting project is likely to meet the expected standard. It is important to consult your supervisor while developing the proposal. Commence only after receiving your supervisor's approval.

The aim of the research project is for trainees to gain experience in research, planning and critical analysis of the literature, to gain insight into the limitations and pitfalls of research, to gain skills in critical evaluation of research conducted by others and to improve written scientific communication skills.

The proposal should be completed within 6 months (or FTE) of having completed the Part I examinations.

Trainee name	Trainee ID	Stage of training
		Y1 Y2 Y3 Y4 Y5 if > Y5 please specify

**Research project title**

**Preliminary literature review** – *please attach separate pages.*

*This should be brief – just enough to summarise and evaluate current knowledge in the field and show why the topic is important.*

**Methodology** - *please attach separate pages.*

*Outline and justify the method/s you propose to use. Specify the type of data to be collected, list the variables of interest and the method/s of analysis, including statistical methods. Include a detailed list of equipment and any other resources you will need.*

**Ethics approval**

*If ethics approval is needed, state the committee from which it will be obtained. Indicate how long this will take*

**Schedule** - *please attach separate pages.*

*Include a project schedule with target dates and a Gantt chart for each phase.*

**Declaration by laboratory supervisor**

I hereby give approval for trainee .....to undertake the dissertation specified in this proposal. I confirm that this proposal was completed within 6 months (or FTE) of the trainee having completed the Part I examination.

**Supervisor name (print)**

**Supervisor signature and date**

**Declaration by project supervisor (if different to the laboratory supervisor)**

I hereby agree to supervise trainee .....while undertaking the dissertation specified in this proposal. I confirm that this proposal was completed within 6 months (or FTE) of the trainee having completed the Part I examination.

**Supervisor name (print)**

**Supervisor signature and date**

 <b>RCPA</b> The Royal College of Pathologists of Australasia	<b>Medical Genomics</b> <b>Research project cover page</b>
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**Please attach this cover page to the research project when submitting for examination.**

Name of trainee.....

Name of supervisor.....

Laboratory.....Date submitted.....

Title of research project.....

Who conceived of this project? If not you alone, please describe your involvement.

Was ethical approval required for the project?     No                     Yes. Give details.....

Were patient samples used in this project?     No                     Yes

If yes, who collected the samples?

Who stored the samples?

Who tested the samples?

Who entered the data?

Who analysed the data?

How was the project funded? (eg laboratory budget, research grant, commercial grant, other?)

Please state any potential or actual conflict of interest associated with the project

Has the work been, or will it be, submitted for publication?     No                     Yes

Who assisted you with the project? (name, position)

**Trainee declaration:** "I certify that I undertook this project during my accredited training in medical genomics. The project is original and has not been used by any other trainee in this laboratory. I have read and understand RCPA Policy 10/2002 on Plagiarism and Cheating in Examinations.

Trainee signature.....date.....

**Supervisor declaration:** I certify Dr. ....undertook this project during training in medical genomics. The work is original and has not been used by any other trainee in this laboratory. I have reviewed this project report and read the RCPA project requirements and believe it is suitable for submission to the RCPA examiners.

Supervisor signature.....date.....

## Appendix 6

### Guidelines for completing the Supervisor Report Form

Please refer to the following documents:

- [Information about the role and responsibilities of supervisors and resources to support supervision](#)
- [The RCPA policy on the Supervision of Training and Accreditation of Supervisors](#)

The [supervisor report form](#) should be completed by the supervisor in consultation with other pathologists and laboratory staff with a significant role in the trainee's training program and with reference to the trainee's portfolio.

Please refer to the portfolio requirements which are set out in Appendix 7.

Supervisors should be mindful that scoring trainee performance is of critical importance in early notification of underperforming trainees so that remedial action can be initiated early in training, if appropriate. Experience tells us that most trainees score 3, which indicates that they are performing at the expected level of training. A score of 1 or 2 identifies to the College/CJCT an underperforming trainee and flags the need for evaluation for trainee support pathways.

Trainees must make their up-to-date portfolio and logbooks available to the supervisor for the annual, rotation and pre-examination reviews. For the pre-examination review, a print-out of the portfolio summary spread sheet must also be made available.

#### Submitting the supervisor report

It is the trainee's responsibility to ensure that the supervisor reports are completed and submitted by the due dates.

Any signatory in a supervisor's report may be contacted by either the chief examiner or the registrar of the Board of Education and Assessment to confirm evidence of satisfactory completion.

Please post the report form by the due date to

The Royal College of Pathologists of Australasia  
207 Albion Street  
Surry Hills NSW 2010  
AUSTRALIA

**Faxed reports will not be accepted.**

## Appendix 7

### Portfolio requirements

The table below sets out guidelines to assist trainees to compile the portfolio, the logbook and the portfolio summary spreadsheet.

Portfolio activities are carried out in the workplace and provide evidence that the trainee is developing technical skills and professional values, attitudes and behaviours that are not readily assessed by formal examinations.

Trainees should start accumulating evidence for the portfolio as early as possible in training and aim to have half of them underway or complete before the part I examination.

**Appendix 8** contains the forms and logbook pages for recording these workplace activities. Please file the (hard copy) forms in a **portfolio folder** with separate sections numbered as in the table.

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

The supervisor should review and sign off completed portfolio forms and logbook on the annual, rotation and pre-exam supervisor report.

The portfolio summary spreadsheet should be appended to the pre-exam supervisor's report and submitted to the RCPA prior to the oral examination at a time determined by the RCPA. The summary will be reviewed by the Registrar of the Board of Education and Assessment and the Chief Examiner. The signatories and trainees may be contacted to confirm evidence of satisfactory completion.

**Note:** The actual portfolio should not be sent unless requested for audit.

	Item	Part I	Part II	Comments
1.	Laboratory safety checklist	Checklist to be completed within 3 months of starting training.  eLearning module to be completed during training		<b>Checklist:</b> one required. <b>Appendix 8</b>  <b>Certificate of completion</b> of eLearning safety module (see point 11 below)
2.	Direct Observation of Practical Skills (DOPS)	3 per year. All pre-part I activities to be completed before part I practical exam	All activities must be completed before the part II examination	Assess competencies in a range of essential laboratory and bench tasks. See <b>Appendix 8</b> for <i>DOPS</i> guidelines and form. Assessed by supervisor or other appropriately qualified person.
3.	Directly Observed Communication Skills (DOCS)	Phone through results First year – 2 if satisfactory Later years – 1 each year.  Oral presentations: 1 per year		Assess effectiveness of communication. See <b>Appendix 8</b> for <i>DOCS</i> guidelines and forms (phone and oral presentations). Assessed by the supervisor or other appropriately qualified person.
4.	Case-based discussions (CbD)	1 per year before the part I exam (total of 2)	1 per year between part I and part II exams (total of 2)	CbDs offer insights into the trainee's clinical and laboratory judgment and ability to present and discuss a complex clinical case. See <b>Appendix 8</b> for <i>CbD</i> guidelines and form. Assessed by supervisor or other appropriately qualified person.
5.	Clinical consultations (telephone or outpatient/inpatient consultations)	Minimum two (2) per week	Minimum two (2) per week	See <b>Appendix 8</b> for clinical consultations sign off form. Record in logbook. Each logged entry must be sighted and signed off by supervising consultant(s) on the day of the consultation.

	Item	Part I	Part II	Comments
				All logged entries must be reviewed by supervisor every three months to ensure a broad spread of clinical consultations.
6.	Multidisciplinary clinical meetings, clinical or laboratory meetings	Attend minimum one (1) per week and present cases at four (4) per year	Attend minimum one (1) per week and present cases at 4 per year	See <b>Appendix 8</b> for clinical meetings sign off form. Recorded on form and signed by supervisor to verify trainee's involvement.
7.	Quality Assurance activities	Five (5) per year. Fifteen (15) to be completed before part I examination	Five (5) per year to be completed between part I and II exams.	See <b>Appendix 8</b> for quality assurance activities log form, noting the minimum annual requirements for: <ul style="list-style-type: none"> <li>• incident assessment</li> <li>• quality audit</li> <li>• internal QC procedures</li> <li>• involvement in preparations for laboratory accreditation</li> </ul>
8.	Professional qualities	Minimum one (1) item on the list must be completed each year.	All to be completed before part II examination	See <b>Appendix 8</b> for written communication, management and ethics-related activities form.
9.	Teaching sessions for students, laboratory colleagues or other audiences	Minimum 1/year	Minimum 1/year	See <b>Appendix 8</b> for teaching session log form. Record in logbook and signed off by supervisor at the 3 monthly meetings and end-of-year review.
10.	Research project or acceptable research alternative		Completed before part II exam	See <b>Appendix 8</b> for guidelines, proposal form and cover page.
11	eLearning modules Refer to Section 2 Learning outcomes and recommended training activities for weblinks	The following RCPA e-learning modules are required to be completed during training: Quality Management Laboratory Safety Ethics Cultural Competence		A <b>Certificate of completion</b> can be printed when the module has been completed (a workbook is required for the Ethics module).  Note: A cultural competence certificate issued by a recognised health service provider can substitute for the RCPA cultural competence module certificate.
12.	Supervisor reports with brief reflection (max 1 page) by the trainee on the supervisor's comments	End-of-rotation, annual and pre-examination reports.	End-of-rotation, annual and pre-examination reports.	See <b>Appendix 6</b> for Guidelines for completing the Supervisor Report Form.

## Appendix 8

### Forms and Logbook pages

This appendix contains master copies of forms and logbook pages to be used to record activities for the portfolio. Please make as many copies as you need and file the completed forms in the portfolio folder. The forms are:

- Laboratory safety checklist
- Direct observation of practical skills (DOPS)
- Instructions for trainees and supervisors
- Assessment form
- Direct observation of communication skills (DOCS)
- Instructions for trainees and supervisors
- Assessment forms (phoning results, oral presentations)
- Case-based discussions (CbD)
- Instructions for trainees and supervisors
- Assessment forms (phoning results, oral presentations)
- Clinical consultations supervisor sign-off form
- Clinical meetings sign off form
- Quality assurance activities log and forms for significant incident reports/reflections
- Reflection form
- Written communication, management and ethics log
- Teaching sessions log
- Research project guidelines
- Research project proposal form
- Research project cover page

	<b>Laboratory safety checklist</b>
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This form is designed to confirm that trainees have understood and are able to apply laboratory safety instruction provided by the employer as it relates to the RCPA curriculum. It covers the essentials for new trainees and is the basis for subsequent learning that will be assessed and eventually lead to the ability to function in a laboratory management role as a pathologist.

- I have completed the laboratory's orientation program for new staff
- I have participated in a laboratory safety induction program or educational session
- I have reviewed the laboratory safety manual
- I know how to work safely with radioactivity
- I know where to find the laboratory safety equipment and how to use it
- I have known immunity to hepatitis B (natural or vaccine)
- I have been vaccinated and/or screened for other infectious diseases as required by my laboratory
- I know how and when to wash my hands and carry this out
- I wear enclosed shoes in the laboratory and tie back long hair if applicable
- I wear appropriate protective clothing (gown, gloves, goggles, mask as needed) and always remove it before leaving the laboratory
- I cover any cuts or wounds before working in the laboratory
- I never eat or put anything in my mouth whilst in the laboratory
- I know how to handle blood and other body substances and tissues to avoid transmission of infection to myself and others
- I know how to prevent sharps injury
- I am aware of all relevant bacterial, viral, prion, radiation, chemical, gas, electrical and fire hazards and preventive strategies
- I know what to do in an emergency
- I know the procedure for reporting safety-related breaches and incidents
- I know where to find information about legislative requirements for laboratory safety
- I know where to find detailed information about laboratory hazards such as dangerous chemicals
- I know the laboratory's waste disposal procedures
- I always clean up after myself
- I set up my workspace and ensure correct posture and lifting technique so as to avoid strain and injury
- I have completed my laboratory's training requirements for biological, chemical and fire safety, first aid and resuscitation
- I have completed the laboratory safety eLearning module in RCPA Education Online

Trainee name:

Witness name: *(supervisor or other senior member of staff)*

Please print:

Please print

date

Signature

Signature

## Direct Observation of Practical Skills (DOPS) Assessment

The purpose of the Direct Observation of Practical Skills (DOPS) assessment is to indicate trainees' acquisition of practical laboratory skills; to show that they can work safely in the laboratory; and to provide feedback to trainees about their progress by highlighting strengths and areas for improvement, thereby encouraging their professional development.

Each DOPS assessment is intended to demonstrate competence using a **different type** of instrument or technique\*. Where possible, this will include all aspects of testing including the following: specimen collection and processing; instrument preparation, operation and maintenance; understanding the clinical significance of results; and the preparation of meaningful reports.

Activities to be completed prior to the first practical examination (part I):

1. Sample reception, data entry, test assignment, checking, sample retrieval and sendaway procedures
2. Sample handling, cell separation/purification, culture set up, maintenance and harvesting
3. Reagent preparation/automated liquid handling, avoiding contamination
4. Nucleic acid preparation method(s), quantification/purity/intactness, storage/archiving
5. DNA labelling for FISH, microarray, other hybridisation based procedures
6. Microscopy (bright-field and fluorescence)
7. Banding and karyotype analysis
8. FISH analysis
9. Array technologies and analysis
10. PCR-based assays (end point, quantitative and real-time) and analysis
11. Gel-based hybridisation and analysis
12. Fragment separation, electrophoresis and analysis
13. DNA sequencing (classical) and analysis
14. DNA sequencing (massively parallel) and analysis

Post-part I activities:

1. Laboratory audits
2. Determination of test costs
3. Preparation of business cases

Trainees should initiate the DOPS assessment by requesting an appropriate assessor to observe them when they consider they are proficient. It is important for the assessor to observe the trainee doing the entire activity. The assessor can be the supervisor or a suitably qualified scientist. Assessors who are RCPA Fellows (including Fellows of the Faculty of Science) may record this as a quality activity in their annual CPD submission.

### ***Grading and standards and outcomes of assessment***

Each aspect of the trainee's performance should be graded. The "n/a" option should be used if the assessor has not observed that aspect or is otherwise unable to comment. Assessors should complete the DOPS form in the presence of the trainee and spend 5-10 minutes providing immediate feedback, discussing strengths and areas for improvement. Areas requiring further development should be identified, agreed and recorded on the DOPS form.

The final outcome should be graded according to whether the standard of performance is as expected for the stage of training. The level of competence should be such that the trainee would be able to perform the task safely without supervision, usually at the level of a competent junior scientist. A trainee whose performance does not meet the standard will be able to repeat the assessment without penalty.

### ***Record keeping***

DOPS forms must be fully completed, signed and dated by the trainee and the assessor. The forms must be retained in the trainee's portfolio and be available for review when the annual supervisor's report is being prepared and signed off. Only DOPS where the standard has been met need be recorded in the portfolio.

		<b>Medical Genomics</b> <b>DOPS Assessment form</b> <b>Direct Observation of Practical Skills</b>		
<b>Trainee name</b>		<b>Trainee ID</b>	<b>Stage of training (circle)</b> Y1 Y2 Y3 Y4 Y5 if >Y5, please specify:	
<b>Assessor name</b>		<b>Assessor position</b> <input type="checkbox"/> Pathologist <input type="checkbox"/> Scientist <input type="checkbox"/> Snr trainee <input type="checkbox"/> Other (pls specify)		
<b>Instrument or technique (tick the box that applies).</b> <b>Pre-part I: all activities must be completed prior to part I examinations.</b> 1. <input type="checkbox"/> Sample reception, data entry, test assignment, checking, sample retrieval and send-away procedures 2. <input type="checkbox"/> Sample handling, cell separation/purification, culture set up, maintenance and harvesting 3. <input type="checkbox"/> Reagent preparation/automated liquid handling, avoiding contamination 4. <input type="checkbox"/> Nucleic acid preparation method(s), quantification/purity/intactness, storage/archiving 5. <input type="checkbox"/> DNA labelling for FISH, microarray, other hybridisation based procedures 6. <input type="checkbox"/> Microscopy (bright-field and fluorescence) 7. <input type="checkbox"/> Banding and karyotype analysis 8. <input type="checkbox"/> FISH analysis 9. <input type="checkbox"/> Array technologies and analysis 10. <input type="checkbox"/> PCR-based assays (end point, quantitative and real-time) and analysis 11. <input type="checkbox"/> Gel-based hybridisation and analysis 12. <input type="checkbox"/> Fragment separation, electrophoresis and analysis 13. <input type="checkbox"/> DNA sequencing (classical) and analysis 14. <input type="checkbox"/> DNA sequencing (massively parallel) and analysis <b>Post-part I</b> 1. <input type="checkbox"/> Laboratory audits 2. <input type="checkbox"/> Determination of test costs 3. <input type="checkbox"/> Preparation of business cases				
Number of hours spent performing the method prior to DOPS assessment		Has the trainee completed the laboratory's usual training process for this method? <input type="checkbox"/> yes <input type="checkbox"/> no		
<b>Please comment on whether these aspects of the trainee's performance are as expected for the stage of training</b>			<b>Yes</b>	<b>No</b>
Understands the principles of the method				
Understands and complies with the laboratory documentation, package inserts, manuals, etc.				
Has completed an assay successfully and produced a valid result that can be reported				
Able to explain the QC procedures for this method, including internal and external QA				
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)				
<b>Final outcome</b> (please circle) As expected for the stage of training Below expected for the stage of training		Date of DOPS	Time taken for DOPS	Time taken for feedback
<b>Name (print) and signature of assessor</b>		<b>Signature of trainee</b>		
<b>Name of laboratory</b>				

## **Directly Observed Communication Skills (DOCS) Assessment**

### **Instructions for Trainees and Supervisors**

Trainees should demonstrate that they are able to communicate clearly and distinctly in English, are able to respond to questions pertaining to medical genomics, answer in a clear and logical manner and be confident that the communication has been understood. Two different types of DOCS activities must be completed before the part I examination:

**DOCS form for phone through of results:** The telephone DOCS form is used to assess one-to-one telephone communications. As this is a formative assessment, the minimum requirement is to assess the trainee's first two telephone consultations in the first year of training and then at least one more complex consultation early in each subsequent year of training until the year of sitting the part I examination. Supervisors may decide that further DOCS assessments are needed beyond the minimum requirement

The supervisor should be present and able to hear both parties when the trainee rings through a laboratory result. The purpose of the assessment is to evaluate a trainee's ability to communicate results and provide useful interpretative insights. At the beginning of each call the trainee should disclose to the clinician that the supervisor is also present.

**DOCS form for oral presentation:** The oral presentation DOCS form is used to assess presentation skills at grand rounds or clinicopathological case discussion meetings. The trainee presents the pathology results for particular cases to a clinical audience and is assessed on ability to discuss the pathology and the diagnostic implications. This should be done at least annually and be assessed by the supervisor or appropriate delegate.

Trainees should initiate DOCS assessments by requesting the supervisor or other suitably qualified staff to observe them at suitable opportunities. It is important for assessors to observe trainees doing the entire activity. Assessors who are RCPA Fellows can note this as a quality activity in their annual CPDP submission.

### ***Grading, standards and outcomes of assessment***

Each aspect of the trainee's performance should be graded. The "n/a" option should be used if the assessor has not observed that aspect or is otherwise unable to comment. As DOCS has a key formative role, assessors should complete DOCS forms in the presence of trainees and also reserve 5-10 minutes to offer immediate feedback. The assessor should discuss strengths as well as areas for improvement with the trainee. Feedback should be given sensitively, in a suitable environment. Areas for development should be identified, agreed and recorded on the form.

The final outcome should be graded according to whether the standard of performance is as expected for the stage of training. A trainee whose performance does not meet the standard will be able to repeat the assessment with no penalty.

### ***Record keeping***

DOCS forms must be fully completed, signed and dated by the trainee and the assessor. The forms must be retained in the trainee's portfolio and be available for review when the annual supervisor's report is being prepared and signed off. Only "Satisfactory" DOCS need to be recorded in the portfolio.

		<b>Medical Genomics</b> <b>Phoning through results</b> <b>Directly Observed Communication Skills (DOCS)</b> Form to be completed by the observer	
<b>How to use this form</b> The supervisor should be present when the trainee rings through an important laboratory result (eg on speaker phone) and assess the trainee's ability to communicate the result and provide interpretative discussion. At the beginning of the call the trainee should disclose to the clinician that the supervisor is listening on speaker phone. Completed forms that indicate that the trainee has met the standard are to be retained in the portfolio and should be sighted by the supervisor and signed off on the annual supervisor report.			
<b>Trainee name</b>		<b>Trainee ID</b>	<b>Stage of training (please circle)</b> Y1 Y2 Y3 Y4 Y5 Other (specify):
<b>Observer/Assessor name</b>		<b>Observer/Assessor position</b> <input type="checkbox"/> Genetic Pathologist <input type="checkbox"/> other (specify):	
<b>Case lab. number:</b>		<b>Clinical issue(s):</b>	
<b>Please indicate against each item below whether the trainee's performance is as expected (or better than expected) for the stage of training</b>			
			<b>Yes</b>
			<b>No</b>
			<b>n/a</b>
<b>Introduction</b>			
Trainee states name, laboratory, verify identity of clinician, state reason for calling, allow time for clinician to find patient records			
Trainee has disclosed the observer's presence			
<b>Information sharing - quality of communication</b>			
Speak clearly at an appropriate pace for comprehension			
Invite additional clinical information			
Invite questions			
Invite opinions			
Confirms (directly or indirectly) that clinician has understood the shared information			
<b>Information sharing - quality of information</b>			
Accurate synthesis of clinicopathological findings			
Suggest appropriate further investigations			
Discuss appropriate management plan			
<b>Closing</b>			
Explain what can be done if problem is not resolved			
Provides contact details for follow-up, including offering clinical consultation if needed			
<b>Please comment on both strengths and areas requiring improvement:</b>			
<b>Final outcome</b> (please circle) As expected for the stage of training Below expected for the stage of training		<b>Assessment date</b>	<b>Time taken for assessment</b>
<b>Name (print) and signature of assessor</b>		<b>Signature of trainee</b>	
<b>Name of laboratory</b>			

 <b>RCPA</b> The Royal College of Pathologists of Australasia		<b>Medical Genomics</b> <b>Oral presentation</b> <b>Directly Observed Communication Skills (DOCS)</b> Form to be completed by the observer				
<b>How to use this form</b> One DOCS oral presentation that indicates that the trainee has met the standard is required per year. The trainee should present the pathology results for particular cases to a predominantly clinical audience. The supervisor or delegate should assess the trainee's ability to present and discuss the results and the diagnostic implications. Completed DOCS forms should be held in the portfolio and should be sighted by the supervisor and signed off on the annual supervisor report.						
<b>Trainee name</b>		<b>Trainee ID</b>	<b>Stage of training (please circle)</b> Y1 Y2 Y3 Y4 Y5 Other (specify):			
<b>Observer/Assessor name</b>		<b>Observer/Assessor position</b> <input type="checkbox"/> Genetic Pathologist <input type="checkbox"/> other (specify):				
<b>Case lab. number:</b>		<b>Clinical issue(s):</b>				
<b>Please indicate against each item below whether the trainee's performance is as expected (or better than expected) for the stage of training</b>				<b>Yes</b>	<b>No</b>	<b>n/a</b>
<b>Planning and organisation</b> Ideas organized into clear, concise, logical order Uses transitions and repetition to keep audience on track Content indicates effective prior planning						
<b>Content</b> Clearly defined and explained subject and main messages Stayed focused on main messages throughout Supplied appropriate amount of detail, examples, evidence Effective visual aids – visible to audience						
<b>Delivery</b> Language is clear, appropriate to purpose and audience Enunciates clearly, audibly, at appropriate pace Responsive to audience reaction – adapts delivery to meet their needs Responsive to audience questions, comments Uses the chosen technology competently						
<b>Please comment on both strengths and areas requiring improvement:</b>						
<b>Final outcome (please circle)</b> As expected for the stage of training Below expected for the stage of training		<b>Assessment date</b>	<b>Time taken for assessment</b>	<b>Time taken for feedback</b>		
<b>Name (print) and signature of assessor</b>		<b>Signature of trainee</b>				
<b>Name of laboratory</b>						

## **Case-based Discussion (CbD) Assessment**

Throughout training, trainees should seek opportunities to present and discuss cases with experienced colleagues and receive feedback. The CbD form should be used to formally record at least one (1) of these sessions per year. At least two (2) CbD forms should be signed off as satisfactory before the part I examination, and a total of four (4) before the part II examination.

The early CbDs should be for routine situations and those with frequently occurring, manageable complications. The later cases completed between the part I and part II examinations are expected to have clinical-laboratory complexities or novelty, which may justify their publication as case reports or presentation at a national conference.

Case presentations selected for CbD assessment represent excellent opportunities to prepare for the oral examinations. CbD assessments also provide supervisors and peers with useful insights into a trainee's level of progress in areas such as ability to interpret and relate pathological results to clinical findings; to plan appropriate investigations, and make decisions in relation to patient care, including decisions with ethical and legal dimensions. CbD assessments also create opportunities for supervisors to provide feedback to trainees about their progress. Feedback is important and should highlight both strengths and any areas requiring improvement, thereby further encouraging the trainee's professional development.

Trainees are responsible for initiating the CbD assessments. The trainee should select two (2) recent cases in which s/he has been involved clinically or through laboratory tests. The assessor should select one (1) of these for the trainee to present and discuss. The trainee should select a suitable assessor, who should be an RCPA Fellow but does not need to be the listed supervisor. The trainee may present the case within a suitable clinical-laboratory meeting at which the assessor is present, after which the trainee and assessor should meet for individualised discussion. Alternatively, the trainee should request to meet with the assessor at a mutually convenient time for about 30 minutes. The presentation/discussion should take about 15-20 minutes. A further 5-10 minutes should be allowed for the assessor to give immediate feedback and complete the CbD form. In addition to the formal CbD assessment, supervisors are encouraged to have an informal discussion of the second case prepared by the trainee.

Each CbD case discussion should cover one or more of the different aspects of practice indicated on the CbD form.

### ***Grading, standards and outcome of assessment***

Each aspect of the trainee's performance should be graded. The "n/a" option should be used if the assessor has not observed that aspect or is otherwise unable to comment. The assessor should discuss strengths as well as areas for improvement with the trainee. Feedback should be given sensitively, in a suitable environment. Areas for development should be identified, agreed and recorded on the CbD form.

The final outcome should be graded according to whether the standard of performance is as expected for the stage of training. A trainee whose performance does not meet the standard will be able to repeat the assessment with no penalty.

### ***Record keeping***

The CbD forms must be fully completed, signed and dated by the trainee and the assessor. The forms must be retained by the trainee in his/her portfolio. Only the CbD for which the trainee has met the standard need to be recorded in the portfolio.

 <b>RCPA</b> <b>Medical Genomics</b> The Royal College of Pathologists of Australasia <b>Case-based Discussion (CbD)</b> <b>Assessment form</b>						
<b>Trainee name</b>	<b>Trainee ID</b>	<b>Stage of training</b> Y1 Y2 Y3 Y4 Y5 if > Yr5, please specify:				
<b>Assessor name</b>	<b>Assessor position</b> <input type="checkbox"/> Pathologist <input type="checkbox"/> Scientist <input type="checkbox"/> Snr trainee <input type="checkbox"/> Other (please specify)					
<b>Focus of discussion (tick as many as apply)</b>						
<input type="checkbox"/> Diagnostic testing <input type="checkbox"/> Predictive/presymptomatic testing <input type="checkbox"/> Genotyping to predict drug responsiveness/toxicity/ side effects <input type="checkbox"/> Carrier testing <input type="checkbox"/> Prenatal testing <input type="checkbox"/> Population screening, including newborn <input type="checkbox"/> Cancer testing (diagnostic/prognostic)		<input type="checkbox"/> Therapeutic monitoring <input type="checkbox"/> Pre-implantation genetic testing <input type="checkbox"/> Ethical issues <input type="checkbox"/> Pre-analytical issues <input type="checkbox"/> Analytical issues <input type="checkbox"/> Interpretive and other post-analytical issues <input type="checkbox"/> Urgent testing (prenatal, postnatal, haemato-onc.) <input type="checkbox"/> other (please specify)				
<b>Complexity of case</b> (tick box) <input type="checkbox"/> low <input type="checkbox"/> medium <input type="checkbox"/> high						
<b>Brief description of case presented, discussed and assessed (use the reverse side if insufficient room)</b>						
<b>Why was this case selected for discussion?</b> (use the reverse side if insufficient room)						
<b>Does this case broaden the trainee's experience by being different from previous cases that have been discussed?</b> <input type="checkbox"/> yes <input type="checkbox"/> no <input type="checkbox"/> n/a						
<b>Please comment on whether these aspects of the trainee's performance are as expected for the stage of training</b>				<b>Yes</b>	<b>No</b>	<b>n/a</b>
Ability to present case clearly and concisely						
Good understanding of clinical issues relating to the case						
Good understanding of laboratory issues relating to the case						
Depth of understanding and awareness of current literature relevant to this case						
Ability to interpret results in a balanced and rational way						
Ability to provide and clearly communicate well reasoned professional advice						
Ability to clinically correlate laboratory test results with patient features.						
Ability to suggest further relevant or more useful tests towards the management of the patient in relation to diagnosis and monitoring including prognostication.						
Ability to communicate findings to a non-medical person (e.g. patient, lawyer)						
Understanding of management and financial aspects of the case						
Overall laboratory and clinical judgment						
Please comment on both strengths and areas requiring improvement:						
<b>Final outcome</b> (please circle) As expected for the stage of training Below expected for the stage of training			<b>Date of CbD</b>	<b>Time taken for CbD:</b>	<b>Time taken for feedback:</b>	
<b>Name (print) and signature of assessor</b>			<b>Signature of trainee</b>			
<b>Name of laboratory</b>						



The Royal College of Pathologists of Australasia

## Medical Genomics Clinical consultations sign off form

### How to use this form

From the beginning of training, trainees should log consultations with clinical colleagues that involve significant, difficult or unusual cases. **A minimum of one consultation per week should be recorded during training.**

**Consultation type should be noted on the form as Telephone (T) Outpatient (OP) Inpatient (IP)**

Each logged entry must be sighted and signed off by supervising consultant(s) on the day of the consultation. At the end of each year, this form and appended case lists should be sighted by the supervisor and signed off on the annual supervisor report.

Trainee name		Trainee ID No			Stage of training (circle): Y1 Y2 Y3 Y4 Y5 (if > Y5, please specify):	
Date	Headline summary of case	Issue(s) raised by the case	Consult type	Trainee's role in the case	Consultant initials	
<i>example</i>	<i>e.g. hyperammonaemia</i>	<i>Clinician seeking guidance on diagnostic possibilities and investigations</i>	<i>T/ IP</i>	<i>Advice offered; review of results; follow-up discussion with referring clinician</i>		
1						
2						
3						
4						
5						
6						
7						
8						
9						

Supervisor name..... Signature..... Date.....



**Medical Genomics  
Clinical/multidisciplinary meeting sign off form**

**How to use this form**

This form is to be used to record that the trainee has fulfilled the following requirements:  
 attend a minimum of two (2) clinical meetings per week throughout training  
 present cases at a minimum of four (4) clinical or laboratory meetings per year throughout training.  
 The supervisor is asked to sign after each meeting to verify off the trainee's participation. Trainees should retain a list of the cases/entities presented at each meeting in the portfolio. At the end of each year, this form and appended case lists should be sighted by the supervisor and signed off on the annual supervisor report.

<b>Trainee name:</b>	<b>Trainee ID:</b>	<b>Stage of training (circle)</b> Y1 Y2 Y3 Y4 Y5 (if > Y5 please specify):
----------------------	--------------------	---

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

Supervisor name..... Signature..... Date.....

 <p>The Royal College of Pathologists of Australasia</p>		<h2>Medical Genomics</h2> <h3>Quality assurance activities log</h3> <p>Page 1 of 2</p>		
<p><b>How to use this form</b>            Five (5) activities per year should be selected from the list. Fifteen (15) are to be completed before the part I examination. Note activities 13-15 have a minimum requirement.            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 and also signed off on the annual supervisor report.</p>				
<b>Trainee name</b>		<b>Trainee ID</b>	<b>Stage of training (circle)</b> Y1 Y2 Y3 Y4 Y5 (if > Y5 please specify):	
	<b>Quality activity</b>	<b>Summary of trainee's role in the activity or comment (where applicable)</b>		<b>Date</b>
	<b>Supervisor signature</b>			
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	Participate in laboratory review of processes for measuring and improving the quality of laboratory practice and patient care			
4	Review relevant AS ISO standards (list documents reviewed)			
5	Review relevant NPAAC standards and guidelines (list documents reviewed)			
6	<b>MANDATORY ACTIVITY.</b> Complete the <a href="#">Quality Management</a> eLearning module in RCPA Education Online and attach the certificate of competence			
7	Internal QAP			
8	Review the laboratory's quality policy, including policy guiding response to unsatisfactory QAP results			
9	External QAP (particularly involvement with the HGSA/QAP, EMQN, ASoC programs)			
10	Active involvement in preparations for laboratory accreditation (minimum of one prior to part II examination)			

		<b>Medical Genomics</b> <b>Quality assurance activities log</b> Page 2 of 2			
<b>Trainee name</b>		<b>Trainee ID</b>	<b>Stage of training (circle)</b> Y1 Y2 Y3 Y4 Y5 (if > Y5 please specify):		
	<b>Quality activity</b>	<b>Summary of trainee's role in the activity or comment (where applicable)</b>		<b>Date</b>	<b>Supervisor signature</b>
11	Contribute to laboratory's processes for reviewing current literature on its discipline-specific QA strategies, risk management, informatics and evidence-based medicine				
12	Participate in a workflow check of effective/ efficient laboratory function				
13	Review and update laboratory internal QC procedures. <b>Minimum of one (1) prior to part I examination and two (2) prior to part II examination.</b> Include reports in portfolio				
14	Significant incident: Involvement in assessment, reporting and review, focussing particularly on the quality issues that were identified and addressed. One per year. <b>Minimum of three (3) prior to part I, and five (5) in total.</b> Include reports in portfolio. Use the reporting form in supplied in this Handbook				
15	Conduct quality audits. Where possible, include comparison with relevant national/international guidelines. <b>Minimum of one per year.</b> Five in total. Include documentation in portfolio				

Supervisor name..... Signature..... Date.....

		<b>Medical Genomics</b> <b>Significant incident report form</b>	
<b>Trainee name</b>		<b>Trainee ID (RCPA)</b>	<b>Stage of training</b> Y1 Y2 Y3 Y4 Y5 if > Y5, please specify
Nature of incident: what happened and why was it significant?			
What led to the incident?			
Action taken at the time of the incident. Could it have been handled differently?			
Review of similar incidents			
Actions taken (or needed) to prevent future similar incidents			
Reflection by trainee (use reverse if insufficient space)			
Trainee signature			Date
Supervisor name (please print) and signature			Date
Name of laboratory			

 <b>RCPA</b> The Royal College of Pathologists of Australasia		<b>Medical Genomics Reflection form</b>	
<b>Trainee name</b>		<b>Trainee ID (RCPA)</b>	<b>Stage of training</b> Y1 Y2 Y3 Y4 Y5 if > Y5, please specify
Nature of activity			
Your role in the activity			
Your reflection on what you learned from your involvement in this activity			
Trainee signature			Date
Supervisor name (please print) and signature			Date
Name of laboratory			



The Royal College of Pathologists of Australasia

## Medical Genomics Professional qualities log

### How to use this form

This form is to be used to record that the trainee has performed at least **1 activity per year** from the list below. 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 and also signed off on the annual supervisor report.

Trainee name		Trainee ID	Stage of training ( <i>circle</i> ) Y1 Y2 Y3 Y4 Y5 (if > Y5 please specify):		
	Activities	Summary of completed activities (additional details to be included in portfolio)	Date	Supervisor signature	
1	Develop working familiarity with correct use of grammar, terminology and etiquette for written and electronic (email) communications.				
2	Attend departmental management committees, budget meetings, other management-related meetings, ethics review committees				
3	Undertake significant management roles, e.g., chairperson, secretary, treasurer of medical genetics-related committees				
4	Complete relevant activities within the <a href="#">Monash University Clinical Ethics Resource</a> . Review Code of Ethics documents from national organisations – AMC, AMA, NHMRC				
5	<b>MANDATORY ACTIVITY.</b> Complete the 6 <a href="#">Ethics eLearning modules</a> in RCPA Education Online. Get supervisor sign-off.				
6	<b>MANDATORY ACTIVITY.</b> Complete the <a href="#">Cultural Competence eLearning module</a> in RCPA Education Online. Attach email verifying completion OR provide evidence of completion of cultural competence training provided by your employer, if a registered health services provider.				
7	<b>MANDATORY ACTIVITY.</b> Complete the <a href="#">Laboratory Safety eLearning module</a> in RCPA Education Online RCPA Education Online				
8	Other (please specify)				

Supervisor name ..... Signature ..... Date.....



## Medical Genomics Teaching sessions log

**How to use this form**

From the beginning of training, trainees should log each teaching session conducted for students, laboratory colleagues or other audiences. A minimum of one (1) per year is required. At the end of each rotation, the log should be sighted and signed off by the supervisor and also signed off on the annual supervisor report.

**Trainee name:**

**Trainee ID:**

**Stage of training (circle)**

Y1 Y2 Y3 Y4 Y5 (If > Y5 please specify):

	Date	Topic presented	Audience	Duration of session
1				
2				
3				
4				
5				
6				
7				
8				
9				
10				

Supervisor name..... Signature..... Date.....

## Appendix 9 Assessment Matrix

	Outcomes to be assessed <i>(Outcomes are organised according to the RCPA common curriculum framework)</i>	Assessment method										
		Part I				Part II			Portfolio			
		Written exam (SAQ)	Wet practical exam	Dry practical exam	Structured oral exam	Research project: scientific investigation	Research thesis, (eg PhD)	Structured oral exam	DOPS	CbD	DOCS	Suggestions for portfolio evidence of activity*
1.1	Cell biology, genetics and epigenetics	Y										
1.2	Whole organism and tissue biology	Y							Y			2,4
1.3	Genomic and epigenetic testing in clinical practice	Y		Y	Y			Y		Y	Y	2,4
1.4	Investigative pathways	Y		Y	Y			Y		Y	Y	3,2
1.5	Genomic/epigenomic variant detection strategies	Y	Y	Y	Y				Y		Y	2
1.6	Pre-analytical processes	Y									Y	
1.7.1	Handling, storage and retrieval of laboratory samples, reagents and data		Y						Y			
1.7.2	Practical skills (techniques/ instruments)								Y			
1.7.3	Data analysis, validation	Y	Y	Y	Y			Y	Y		Y	5,9
1.8.1	Interpreting genomic data	Y	Y	Y					Y			3
1.8.2	Developing and reporting a professional opinion		Y	Y	Y			Y	Y			3,9
2.1	Quality management							Y			Y	5,9,7
2.2	Laboratory safety							Y			Y	1,5,9
2.3	Compliance with legislation							Y			Y	9
2.4	Managing people										Y	6,9
2.5	Managing resources										Y	6,9
2.6	Information fundamentals		Y		Y				Y	Y		4,5
3.1	Research and critical appraisal					Y	Y				Y	
3.2	Undertake self-education and CPD	Y		Y	Y			Y				9
3.3	Educate colleagues staff, patients/families									Y	Y	9,8
3.4	Provide data for planning and evaluation					Y	Y				Y	9
4.1	Ethics and confidentiality									Y	Y	7
4.2.1	Communication - oral									Y	Y	3
4.2.2	Communication – report writing											7
4.2.3	Communication – academic writing					Y	Y					
4.3	Collaboration and teamwork									Y	Y	3
4.4	Cultural competence										Y	2,3,6,7,8,9
<b>* Portfolio categories</b> 1: Safety checklist, incident reports 2 : Patient consultations 3: Attendance/presentations at clinical / multidisciplinary meetings 4: Research and scholarship activities 5: Quality activities 6: Attendance at management meetings 7: Written communication, management and ethics log 8: Teaching sessions log 9: Activity reflection												