



FACULTY OF SCIENCE

TRAINEE HANDBOOK 2019

GENETIC PATHOLOGY

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

APCI	Acid Cysteine Proteinase Inhibitor
AS ISO	Australian and International Standard
CbD	Case-based Discussion
CGH	Comparative Genomic Hybridisation
CNV	Copy Number Variation
CPDP	Continuing Professional Development Program
DNA	DeoxyriboNucleic Acid
DOPS	Direct Observation of Practical Skills
EBLP	Evidence Based Laboratory Practice
ELISA	Enzyme linked Immunosorbent Assay
FIA	Fluorescent Immunoassay
FISH	Fluorescence in situ Hybridisation
FSc	Faculty of Science
HGVS	Human Genome Variation Society
IEM	Inborn Errors of Metabolism
ISCN	International System for Chromosome Nomenclature
LCSH	Long Contiguous Stretches of Homozygosity
MALDI	Matrix Assisted LASER Desorption/Ionisation
MSc	Master of Science
NAHR	Non-allelic Homologous Recombination
NATA	National Association of Testing Authorities
NBS	Newborn Screening
NHEJ	Non-homologous End Joining
NPAAC	National Pathology Accreditation Advisory Council
PCR	Polymerase Chain Reaction
PhD	Doctorate of Philosophy
QA	Quality Assurance
QC	Quality Control
RCPA	Royal College of Pathologists of Australasia
RNA	RiboNucleic Acid
SNP	Single Nucleotide Polymorphisms
UPD	Uniparental Disomy

SECTION I

Introduction

The Faculty of Science provides a structured Fellowship program to enable scientists to demonstrate competence in the following areas to a standard specified by the RCPA.

1. Use professional judgement in advising clinicians on the requirements for investigations and in carrying out these investigations for patients as a member of the team providing clinical care.
2. Maintenance of safe and effective service through the use of relevant quality assurance and audit tools, to appropriate national standards.
3. Undertake scientific research, including the evaluation of scientific literature, to introduce new scientific procedures or solve diagnostic or therapeutic problems within their field.
4. Apply the principles of evidence-based laboratory practice to inform health care decisions.
5. Provide innovative and strategic direction to the operation of the laboratory.

The scientist will complete the training requirements specified in the curriculum, and will demonstrate competence and attainment of learning outcomes by satisfying all assessment requirements to the standards set by the Faculty of Science, as defined in the curriculum.

General aims & structure of the training program

The general aims of the training program are to provide a structured pathway for scientists working in a pathology context to meet the standards defined by the RCPA of a leading scientist in their field.

These general aims of the training program relate to three areas of professional activity of a leading scientist, i.e.,

- Discipline specific clinical laboratory functions
- Research
- Innovation, Development and Leadership

The Faculty of Science curriculum in Genetic Pathology comprises standards in these three areas as follows:

1. Research standards

- Demonstrate highly developed skills in research, management of time and resources and communication of outcomes and data, whilst independently developing theoretical concepts, acquiring new knowledge and testing hypotheses in the field of Genetic Pathology.

2. Clinical laboratory standards

- Demonstrate competence in applying the techniques, technology and reporting associated with a Genetic Pathology laboratory with a broad case-mix of patients.
- Apply the theoretical and technical expertise in laboratory techniques required to lead the activities of a Medical Genomics or Biochemical Genetics laboratory, including one specialised area of Medical Genomics or Biochemical Genetics.

3. Innovation, development and leadership standards

- Apply, implement and evaluate strategies that guarantee quality assurance, compliance, safety and efficient use of resources fundamental to the operation of a Genetic Pathology laboratory.
- Demonstrate a commitment to the continual improvement and advancement of Genetic Pathology.
- Apply the principles of Evidence Based Laboratory Practice (EBLP) to inform health care decisions.

These standards are elaborated as content areas and specific training outcomes in Section 2 of this handbook. In the Clinical Laboratory Standards section there are specific content areas and training outcomes for Part I and II. Competence in outcomes achieved by Part I of training should be maintained throughout. It is expected that trainees should achieve the outcomes in the Research Standards and Innovation, Development & Leadership Standards gradually throughout their training.

Trainees, with the assistance of their supervisor, should ensure that they engage in appropriate learning activities to achieve each of the outcomes, and therefore the standard. The indicators are statements which guide the assessment process, and describe how the trainee will demonstrate they have met the standard. Specific assessment requirements are detailed in Section 3 of this handbook.

The total time to complete the training program is normally a minimum of 5 years, except when time credits have been granted by the Chief Examiner on the advice of the Principal Examiner for previous experience through a Training Determination. Part I assessment criteria can normally be met and assessed during the third year of training, Part II requirements following another 2 years training.

Administrative Requirements

This handbook should be read in conjunction with the *RCPA Trainee Handbook Administrative Requirements* document on the College website.

Entry requirements

Trainees should be graduates of a university in Australia or New Zealand with a degree at Australian Qualifications Framework level 7 (minimum) with subjects relevant to the field of pathology. If such a degree is awarded by an overseas tertiary education institution the qualifications should be approved by the College. To enter the program, trainees are ordinarily required to have five (5) years post graduate experience working as scientists in a Pathology related field.

Training requirements

Training must take place in an RCPA accredited laboratory and is limited to the time period for which that laboratory is accredited in each discipline. Details of RCPA accredited laboratories are available through the College website.

Please note that ordinarily, a maximum of 4 years is to be spent in any one laboratory over the course of the 5-year training program. Individuals should contact the College Registrar if a deviation from this requirement is sought.

Trainees are responsible to ensure that all forms are submitted by the due dates indicated in the handbook and the College website.

Supervision

References

- Policy: Supervision of Training and Accreditation of Supervisors
- Resource Manual for Supervisors

All training must be supervised. More than one supervisor can be nominated if Trainees divide the year between two or more unrelated laboratories. The College recommends that any one supervisor be responsible for no more than two Trainees.

Who can be a supervisor?

The supervisor will normally be a Fellow of the RCPA; however non-Fellows may be approved by the Principal Examiner in the discipline if no Fellow is available. 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.

Day-to-day supervision should primarily be the responsibility of a Fellow of the Faculty of Science, however it is appropriate for senior pathology staff with relevant experience to sign off on some workplace-based assessments.

The role of the supervisor

Supervisors will 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 also ensure that the Trainee has sufficient time and opportunities to carry out the required training activities.

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. In addition to the formal meetings with the Trainee that should occur every three months, they should meet regularly with the Trainee; observe their laboratory performance and interaction with pathologists, peers and clinicians; and review result reporting. This may be delegated to other trainers where appropriate, e.g. 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 described in the RCPA Resource Manual for Supervisors and the RCPA policy on Supervision of training and the Accreditation of Supervisors (hyperlinked above).

Supervisors and Trainees should contact the **RCPA Education Advisor** for assistance with supervision and training issues.

Resources

The resources listed below are useful but do not necessarily cover all the genetic pathology that a trainee should know. None of these is compulsory. Information for examination may come from books and journals outside this list.

Resources for all Genetic Pathology trainees

Books

Medical Genomics

Strachan T, Read A. Human Molecular Genetics. 4TH Edition. 2010. ISBN 9780815341499

[Garland Science Online](#)

Gardner RJM, Sutherland GR Shaffer LG. Chromosome Abnormalities and Genetic

Counselling. 4th Edition. 2011. Print ISBN-13: 9780195375336. [Oxford Medicine Online](#)

Clinical Genetics/Laboratory Interface

Read A, Donnai D. New Clinical Genetics. 2nd Edition. 2011. Scion Publishing Ltd. ISBN

9781904842804. [Scion Publishing web resource](#)

NPAAC Guidelines. All of the Tier 2 and Tier 3 publications plus "Requirements for Medical Testing of Human Nucleic Acids 2012" from Tier 4. ISBN:1741861640. (See website below).

Journals and Websites

Websites

RCPA Education Online <http://www.rcpa.edu.au/Education> (specifically the Ethics, Quality Management and Laboratory Safety eLearning modules)

Online Mendelian Inheritance in Man: <http://www.ncbi.nlm.nih.gov/omim/> (Accessed October 2017)

Biostatistics Collaboration of Australia (BCA) website listing the statistics courses available in Australia: <http://www.bca.edu.au/> (Accessed October 2017)

Information about rare genetic tests

NIH Genetic Testing Registry – <http://www.ncbi.nlm.nih.gov/gtr/> (Accessed October 2017)

NCBI Gene Reviews – <http://www.ncbi.nlm.nih.gov/books/NBK11116/> (Accessed February 2016)

EuroGentest – <http://www.eurogentest.org/> (Accessed February 2016)

RCPA Catalogue of Genetic Tests and Laboratories <http://genetictesting.rcpa.edu.au/> (Accessed February 2016)

Quality assurance/ Best practice guidelines

American College of Medical Genetics Standards and Guidelines for Clinical Genetics

Laboratories https://www.acmg.net/ACMG/Medical-Genetics-Practice-Resources/Technical_Standards_and_Guidelines.aspx (Accessed February 2016)

RCPA Guidelines for Implementation of Massively Parallel Sequencing:

<https://www.rcpa.edu.au/getattachment/7d264a73-938f-45b5-912f-272872661aaa/Massively-Parallel-Sequencing-Implementation.aspx> (Accessed February 2016)

RCPA Standards for Clinical Databases of Genetic Variants V.1.0 2014:

<https://www.rcpa.edu.au/Library/College-Policies/Guidelines/RCPA-Standards-for-Clinical-Databases-of-Genetic-V.aspx> (Accessed February 2016)

European Molecular Genetics Quality Network <http://www.emqn.org/emqn/Best+Practice> (Accessed February 2016)

Clinical Molecular Genetics Society (part of the federated British Society for Genetic Medicine)

<http://www.acgs.uk.com/quality/best-practice-guidelines/> (Accessed February 2016)

Human Genetics Society of Australasia Policies, Guidelines and Position Statements

<https://www.hgsa.org.au/> (Accessed February 2016)

NPAAC Guidelines: <http://www.health.gov.au/internet/main/publishing.nsf/Content/health-npaac-publication.htm> (Accessed February 2016)

European Research Network for evaluation and improvement of screening, Diagnosis and treatment of Inherited Disorders of Metabolism (ERNDIM) <http://www.erndim.org/home/start.asp> (Accessed February 2016)

Clinical and Laboratory Standards Institute: <http://clsi.org/> (Accessed February 2016)

Westgard QC Rules: <https://www.westgard.com/> (Accessed February 2016)

Journals

New England Journal of Medicine

Lancet

Journal of the American Medical Association

Nature Genetics

Genetics in Medicine

American Journal of Human Genetics

American Journal of Medical Genetics

Human Molecular Genetics

Journal of Medical Genetics

Journal of Molecular Diagnostics

European Journal of Human Genetics

Prenatal Diagnosis

Clinical Chemistry

Clinica Chimica Acta

Genetic Testing

Trends in Genetics

Human Mutation

Journal of Inherited Metabolic Disease

Molecular Genetics and Metabolism

Orphanet Journal of Rare Diseases

Genes Chromosomes and Cancer

Clinical Genetics

Additional resources for Medical Genomics trainees

Books

Human Molecular Genetics and Cytogenetics

Gersen, SL, Keagle MB. (Eds.) The Principles of Clinical Cytogenetics.. Springer. ISBN 978-1-4419-1687-7

Shaffer LG, McGowan-Jordan J, Schmid M (eds). An International System for Human Cytogenetic Nomenclature (2013) ISBN: 978-3-318-02253-7

Swerdlow SH, Campo E, Harris NL, Jaffe ES, Pileri SA, Stein H, Thiele J, Vardiman JW (Eds).

WHO Classification of Tumours of Haematopoietic and Lymphoid Tissues. 4th Edition, 2008.

[WHO Press](#). ISBN-13 9789283224310, ISBN-10 9283224310

Genome analysis protocols

Current Protocols in Human Genetics (Wiley) Online ISBN: 9780471142904

Rooney D. Human Cytogenetics: Constitutional Analysis. A Practical Approach. 3rd Edition, 2001, Oxford University Press. ISBN 978-0-19-963839-0

Useful background reading

Brown TA Genomes 3. Garland Science. ISBN 0 8153 4138 5

Hartl DL, Clark AG. Principles of Population Genetics, 4th edition, 2007. Sinauer, ISBN: 0878933085, 9780878933082

Bradley-Smith G, Hope S, Firth HV Hurst JA. Oxford Handbook of Genetics. 2010. Oxford University Press, ISBN 978-0-19-954536-0

Rimoin DL, Connor JM, Pyeritz RE, Korf BR. Emery and Rimoin's Principles and Practice of Medical Genetics. 6th Edition. 2013. Academic Press. ISBN 0123838355, 9780123838353

Scriver, C.R., Beaudet, A.L., Valle, D., Sly, W.S., Vogelstein, B., Childs, B., Kinzler, K.W.
Scriver's [OMMBID The Online Metabolic & Molecular Bases of Inherited Disease](#) (accessed January 2015)

Milunsky A, Milunsky JM. Genetic Disorders and the Fetus. 6th Edition. Wiley-Blackwell. ISBN: 978-1-4051-9087-9

Heim S, Mitelman F. Cancer Cytogenetics: Chromosomal and Molecular Genetic Abberations of Tumor Cells. 3rd Edition. 2009. Wiley. ISBN: 978-0-470-18179-9

Websites

DECIPHER (Mapping the clinical genome) <https://decipher.sanger.ac.uk/> (Accessed February 2016)

ClinGen - Clinical Genome Resource <https://www.clinicalgenome.org/> (Accessed February 2016)

ClinVar <http://www.ncbi.nlm.nih.gov/clinvar/> (Accessed February 2016)

ExAC Exome Aggregation Consortium <http://exac.broadinstitute.org/> (Accessed February 2016)

Additional resources for Biochemical Genetics trainees

Books

Scriver, C.R., Beaudet, A.L., Valle, D., Sly, W.S., Vogelstein, B., Childs, B., Kinzler, K.W.
Scriver's [OMMBID The Online Metabolic & Molecular Bases of Inherited Disease](#) (accessed January 2015)

Saudubray J-M, Berghe G vd, Walter JH (Eds). Inborn Metabolic Diseases. Diagnosis and Treatment. 5th Edition 2012 [Springer Online](#) (accessed January 2015). ISBN 9783642157202

Blau N, Duran M, Gibson KM (Eds). Laboratory Guide to the Methods in Biochemical Genetics. 2008. [Springer Online](#) (accessed January 2015). ISBN 9783540766988

Blau N, Duran M, Gibson KM and Dionisi-Vici C (Eds) Physician's Guide to the Diagnosis, Treatment, and Follow-Up of Inherited Metabolic Diseases. 2014 [Springer Online](#) (Accessed December 2015) ISBN: 978-3-642-40336-1 (Print), 978-3-642-40337-8 (Online)

Zschocke J, Hoffmann G. Vademecum Metabolicum. 3rd edition 2012. Schattauer. ISBN 978-3-7945-2816-5

Websites

Metabolite/Enzyme resources and databases:

UK National Metabolic Biochemistry Network teaching resource

<http://www.metbio.net/metbioHome.asp> (accessed February 2016)

The Urine Metabolome database. <http://www.urinemetabolome.ca/> (accessed February 2016)

Metabolomics Toolbox (Human Metabolome Project) <http://www.hmdb.ca/> (accessed January 2015)

Metabolic and Genetic Information Center <http://metagene.de/appl/index.html> (Accessed February 2016)

NeurometPlus <http://www.neurometplus.com/> (Accessed February 2016)

Reactome - a curated knowledgebase of biological pathways <http://www.reactome.org/> (Accessed February 2016)

Kyoto Encyclopedia of Genes and Genomes: <http://www.genome.jp/kegg/> (Accessed February 2016)

Newborn Screening:

Region 4 Genetics Collaborative <http://www.region4genetics.org/> (Accessed February 2016)

If you have ideas about additional resources, please inform RCPA:

(email rcpa@rcpa.edu.au) so these can be added to future editions of this handbook.

SECTION 2 – CURRICULUM

Research Standards

Standard
<p>Fellows of the Faculty of Science will:</p> <p>Demonstrate highly developed skills in research, management of time and resources and communication of outcomes and data, whilst independently developing theoretical concepts, acquiring new knowledge and testing hypotheses in the field of Genetic Pathology.</p>

Content	Outcomes	Indicator
<p>R 1 – Research</p>	<p>R 1 – Demonstrated ability in carrying out effective research</p> <p>1.1 Comment on recent advances and relevant literature in their field of study</p> <p>1.2 Employ analytical and critical thinking to develop, refine or critique theoretical concepts, and to recognise problems</p> <p>1.3 Develop research proposals and protocols towards testing current hypotheses/ investigating or validating contemporary problems/ acquiring new knowledge in the discipline</p> <p>1.4 Apply statistical and epidemiological concepts and interpret epidemiological/ laboratory data</p> <p>1.5 Critically evaluate own findings and findings of others</p> <p>1.6 Demonstrate an understanding of ethical/ professionalism issues relating to research including but not limited to consent, ethical treatment of humans and animals, confidentiality and privacy, attribution of credit, intellectual property, malpractice and misconduct</p> <p>1.7 Participate in effective and ethical peer review processes as researchers and peer reviewers</p>	<p>R 1 will be evidenced through:</p> <p>At least 2 first author publications, published in the last ten years together with a written discussion that explains the background, interrelatedness and significance of the research. Candidates must detail their own contribution to the research. Individuals with a PhD, or a Masters (by Research) related to the area of expertise in Pathology conferred by a university recognised by the College, may be exempted from this component at the discretion of the Principal Examiner.</p> <p>AND</p> <p>Answers questions in a viva voce examination to the standard approved by the Principal Examiner</p>
<p>R 2 – Management</p>	<p>R 2 – Demonstrated ability in the management of research and research administration</p> <p>2.1 Prioritise outcomes, meet goals and work productively with key stakeholders using effective project management skills</p> <p>2.2 Participate in processes for obtaining funding including applying for grants and other external funding</p> <p>2.3 Use information systems and appropriate resources or technologies to record and communicate research findings</p> <p>2.4 Determine the most cost effective methods to achieve a research goal</p> <p>2.5 Demonstrate flexibility, adaptability, and</p>	<p>All R 2 outcomes could be assessed through:</p> <p>A report, to be submitted in the candidate’s portfolio as detailed in Part II assessment policy</p> <p>AND</p> <p>Answering questions in a viva voce examination to the standard approved by the principal examiners</p>

Content	Outcomes	Indicator
	innovation in management of research	
<p>R 3 – Communication</p>	<p>R 3 – Demonstrated ability in research communication:</p> <p>3.1 Clearly articulate ideas, construct cohesive arguments, and translate and convey technical concepts and information to a variety of stakeholders in a style appropriate to the context</p> <p>3.2 Prepare reports and papers for peer review/ publication that comply with the conventions and guidelines for reporting biomedical research</p> <p>3.3 Defend research methods and findings in peer review and/or viva voce examination</p> <p>3.4 Achieve a significant number of articles in peer-reviewed publications</p> <p>3.5 Support the development of research, capacity of others in teaching, mentoring or demonstrating</p>	<p>Document material presented at weekly laboratory meetings.</p> <p>Document the planning and progress of research towards a higher degree through Annual or 6 monthly report.</p> <p>Publications, presentations and poster abstracts.</p> <p>Document the contribution to research training programs or assisting other scientists/ registrars in conducting research</p> <p>AND</p> <p>Answer questions in a viva voce examination to the standard approved by the principal examiner</p>

Clinical Laboratory Standards – Common – Part I

Genetics (Common core material for both Medical Genomics and Biochemical Genetics)

Standard
<p>Fellows of the Faculty of Science will:</p> <p>Demonstrate competence in applying the techniques, technology and reporting associated with a Genetics laboratory with a broad case-mix of patients</p>

Content	Outcomes	Indicator
<p>G1 – Laboratory practice (Common to Medical Genomics and Biochemical Genetics strands)</p>	<p>G1.1 – Apply these basic principles of scientific laboratory practice</p> <ul style="list-style-type: none"> • The theoretical chemistry and physics relevant to the practice of laboratory genetics • The full range of analytical techniques currently used in laboratory genetics • Statistical analysis • General assay metrics: analytical sensitivity, specificity, accuracy and imprecision; test positive and negative predictive values, diagnostic sensitivity and specificity • Principles of sampling and assay validation 	<p>Answer examination questions and/or complete workplace-based assessments that demonstrate the application of these principles</p>
<p>G2 – Foundations of Genetics (Common to Medical Genomics and Biochemical Genetics strands)</p>	<p>G2.1 – Describe the structure and functions of genetic material, including:</p> <ul style="list-style-type: none"> • Eukaryotic and prokaryotic cell structure and organelle function • Key differences between prokaryotic, eukaryotic and viral genetics • Eukaryotic chromosomes; relationship to chromosome banding • Nucleic acid structure and biology • Genome organisation (including repetitive DNA, transposons, genes and their regulatory regions, pseudogenes, etc.) • Mitochondrial genome • Mitotic cell cycle, including genome replication and repair • Meiosis, recombination and chromosomal segregation • Chromatin; higher-order chromatin folding and fractal packaging <p>G2.2 – Describe how functional gene products are generated</p> <ul style="list-style-type: none"> • Gene expression and regulation • RNA transcription and post-transcriptional processing • RNA-mediated regulation of cell function • Protein translation and post-translational processing and targeting • Protein structure and protein turnover • Enzymes, pathways and signalling 	<p>All of G2 could be evidenced by answering examination and viva voce questions and/or completing workplace-based assessments that describe the foundations of Genetics knowledge</p>

Content	Outcomes	Indicator
	<ul style="list-style-type: none"> • Structures of lipids and sugars • Epigenetic regulation (DNA methylation, histone modifications/ histone code, RNA-mediated regulation of gene expression) • Epigenome organization <p>G2.3 – Describe developmental genetics, including:</p> <ul style="list-style-type: none"> • Gametogenesis, fertilisation and infertility • Pre-implantation morula; placentation; early embryogenesis • Twins and twinning • Chimerism • Mosaicism • Cell differentiation/ migration/developmental fields • Epigenetic features of gametogenesis; early embryogenesis, and stem cells <p>G2.4 – Describe DNA sequence and copy number mutational mechanisms, including:</p> <ul style="list-style-type: none"> • Mechanisms of DNA damage • 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) <ul style="list-style-type: none"> • Oligonucleotide repeat (e.g. triplet repeat) expansion mutagenesis <p>G2.5 – Describe genomic and epigenomic variation, including:</p> <ul style="list-style-type: none"> • 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 assembly, mutation databases and genome browser tools • Genome variant nomenclature – DNA (including cytogenetic and array-detected genomic lesions), RNA and protein level nomenclature • Epigenetic dysregulation and disease <p>G2.6 – Describe inheritance patterns, including:</p> <ul style="list-style-type: none"> • 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 	

Content	Outcomes	Indicator
	<ul style="list-style-type: none"> • Other non-classical forms of inheritance – mitochondrial; pseudoautosomal, uniparental disomy (UPD) and oligonucleotide repeat disorders • Incidence of de novo and inherited chromosomal and large-scale genomic variants • Maternal and paternal age effects <p>2.7 – Describe population genetics, including:</p> <ul style="list-style-type: none"> • Factors determining genetic population structure – mutation rates; selection; migration; random drift • Population description - Hardy-Weinberg equilibrium and the estimation of allele frequencies • Estimation of factors affecting the genetic structure of populations • Deviations from random mating – assortative mating, consanguinity, founder effects • Overview of genetic similarities and differences across human populations • Frequency of genetic diseases in populations 	
<p>G3 Preparation of samples (Common to Medical Genomics and Biochemical Genetics strands)</p>	<p>G3 – Describe and apply the handling, storage and retrieval of laboratory samples and reagents</p> <ul style="list-style-type: none"> • Prepare, label, store and handle reagents correctly • Handle and label patient samples correctly • Comply with the specimen storage and indexation conventions of the laboratory • Use the laboratory information system to retrieve specimens for examination and review • Prepare calibrants and use certified reference materials appropriately • Prepare, isolate, concentrate and purify samples, including tissues, cells, organelles and body fluids • Ensure the stability and correct storage for long-term preservation of patient tissues and samples 	<p>Complete workplace-based assessments and answer examination or viva voce questions to demonstrate correct procedures for the preparation, handling and storage of samples and reagents</p>

Clinical Laboratory Standards – Part I – Medical Genomics

Standard

Fellows of the Faculty of Science will:

Demonstrate competence in applying the techniques, technology and reporting associated with a Genetics laboratory with a broad case-mix of patients

Content	Outcomes	Indicator
<p>MG1 – Laboratory techniques</p> <p>Apply and evaluate the techniques and technology routinely used in the laboratory</p>	<p>MG1.1 – Explain the principles, performance and limitations of genomic methods, including:</p> <ul style="list-style-type: none"> • Microscopy (bright-field and fluorescence) • Banding and karyotype analysis • FISH analysis • CGH and SNP array technologies and analysis • PCR-based assays (end point and real-time), qualitative and quantitative and analysis • Gel-based hybridisation and analysis • Nucleic acid fragment separation and quantitation by both gel and capillary electrophoresis • DNA sequencing (Sanger) and analysis • DNA sequencing (massively parallel) and analysis (library preparation, sequencing methodologies, informatics and data interpretation) • Whole genome amplification • Copy number and CNV determination using sequencing and other molecular methods <p>MG1.2 – Evaluate genomic test data, including:</p> <ul style="list-style-type: none"> • Calculating and interpreting statistical data related to the reproducibility and validity of diagnostic and screening tests, including sensitivity, specificity, positive/ negative predictive values, receiver operating characteristics analysis, spectrum & bias, predictive values, likelihood ratio, and Bayes' theorem • Apply non-parametric statistics • Apply methods of bivariate statistical analysis • Apply methods of multivariate statistical analysis (as applicable to the analysis of large data sets obtained from a single sample) • Apply methods of measuring and improving data quality • Evaluating and expressing uncertainty in measurement 	<p>MG1 will be evidenced by answering examination and viva voce questions and providing a portfolio of work that demonstrates competence in applying and evaluating medical genomics laboratory techniques</p>

Content	Outcomes	Indicator
	<p>MG1.3 – Describe the analysis and validation of large-scale data sets (such as array and massively parallel sequencing data), including:</p> <ul style="list-style-type: none"> • Test validation and verification of results • The sources of bias and artefacts in genome analysis • Assess and interpret mosaicism (somatic and germline) • Principles of design and scripting of algorithms required to analyse large data files • Threshold call parameters and their limitations • Risks associated with analysing high volume data • Limits associated with analysing large data files • The principles, requirements and functioning of laboratory information systems used for instrument interfacing, flagging of results and generating interpretive comments • The components and limitations of large-scale genome data analysis pipelines (quality assessment; data flow; data validation; mapping; dynamic querying; assembly visualization) • How to implement trouble-shooting procedures • Statistical approaches to analysing large-scale data sets 	
<p>MG2 – Somatic genetics</p>	<p>MG2 - Describe somatic genetics, including:</p> <ul style="list-style-type: none"> • Cancer cell biology • Chromosomal breakage syndromes • Genomic and epigenomic architecture of tumourigenesis • Genomic and epigenomic features of aging 	<p>MG2 will be evidenced by answering examination and viva voce questions on somatic genetics</p>
<p>MG3 – Investigation of genomic disorders</p>	<p>MG3.1 – Explain the strategies for use of appropriately selected analyses for detection of constitutional genomic variants, including:</p> <ul style="list-style-type: none"> • Chromosomal gains and losses • Specified and unspecified balanced chromosomal rearrangements • Specified and unspecified copy number variants/long contiguous stretches of homozygosity/uniparental disomy • Specified and unspecified nucleotide-level mutations at the genome level • Specified and unspecified nucleotide-level mutations at the single gene level • Use of specific non-disease associated polymorphisms (i.e., for linkage analysis in pre-implantation genetic diagnosis; detection of maternal cell contamination; sample identification, uniparental disomy, etc.) • Specified methylation anomalies and other epimutations • Gene expression analysis 	<p>MG3 will be evidenced by a portfolio of work including case reports, answering viva voce and written examination questions interpreting the output of data to assist in the diagnosis and management of conditions</p>

Content	Outcomes	Indicator
	<p>MG3.2 Explain the strategies for use of appropriately selected analyses for detection of somatic genomic variants including (e.g., screening for genome variants in DNA derived from mosaic/chimaeric tissues, such as fetal cells and cell-free DNA in maternal blood; tumour material; constitutional chimaerism; etc.):</p> <ul style="list-style-type: none"> • Whole chromosomal gains and losses in DNA/cells from mosaic tissues • Specified and unspecified balanced chromosomal rearrangements in DNA/cells from mosaic tissues • Specified and unspecified copy number variants/ loss of heterozygosity/ uniparental disomy in DNA from mosaic tissues • Specified and unspecified nucleotide-level mutations in DNA from mosaic tissues • Quantitative analysis for specific mutations in DNA from mosaic tissues • <i>in situ</i> detection in cells of specified mosaic mutations • Specified uniparental disomies, methylation anomalies and other epimutations in DNA from mosaic tissues • Gene expression analysis <p>MG3.3 – Analyse genomic data, including:</p> <ul style="list-style-type: none"> • Genetic mapping of Mendelian characters (linkage analysis) • The use of clinical data in the interpretation of constitutional genomic anomalies detected by karyotyping, array, massively parallel sequencing or other molecular methods (including autosomal and sex chromosome aneuploidy; polysomies; structural anomalies; translocations; microdeletion syndromes; loss of heterozygosity; uniparental disomy; identity-by-descent) • Use of clinical data in the interpretation of somatic genomic and epigenetic anomalies detected in constitutional mosaicism and malignancy by karyotyping, array, DNA sequencing or other molecular methods (including chromosomal aneuploidy; polysomies; structural anomalies; translocations; microdeletion syndromes; loss of heterozygosity and uniparental disomy; identity-by-descent) • Annotation, navigation and data mining from major genome browsers • Genome and reference sequence annotation tools • Sequence assembly and alignment 	

Content	Outcomes	Indicator
	<ul style="list-style-type: none"> • Contextualising DNA sequence data using informatics tools • Evaluating the potential pathogenicity of an unknown genomic variant • The bioinformatic resources required to analyse and visualise massively parallel sequencing data • Bioinformatic tools/suites required to analyse and visualise CGH and SNP array data • Network knowledge base analysis of high-throughput genomic data networks <p>MG3.4 – Developing and reporting a professional opinion</p> <ul style="list-style-type: none"> • Outline the use of ISCN and HGVS nomenclature for genomic variants • Interpret and synthesise laboratory data in the context of relevant clinical information, validated bioinformatic and database resources and peer-reviewed published data • Assess familial recurrence risks arising from small- and large-scale genomic/chromosomal anomalies • Explain the influence of ethnogeographic origins, particularly when reporting negative genotype findings • Suggest concise guidance about follow-up testing, when appropriate, for test verification, functional assessment of findings and of other family members • Design algorithms for investigating and reporting different clinical scenarios • Apply these algorithms, alert limits, etc, to draw attention to results requiring additional attention and to develop protocols for reflex testing • Report in accordance with the relevant regulatory framework 	
<p>MG4 – Clinical molecular genetics</p>	<p>MG4 – Explain the use of genomic testing in the following clinical contexts:</p> <ul style="list-style-type: none"> • Diagnostic testing • Predictive/pre-symptomatic genetic testing • Genotyping (constitutional and somatic) to predict drug responsiveness/ toxicity/ side-effects (targeted therapeutics, pharmacogenetics/pharmacogenomics) • Genotyping plasma DNA or circulating tumour cells for prognostic and therapeutic prediction • 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 	<p>MG4 will be evidenced by answering viva voce and written examination questions to explain genomic testing</p>

Content	Outcomes	Indicator
	<ul style="list-style-type: none"> • 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 Laboratory Standards – Part II – Medical Genomics

Standard

Fellows of the Faculty of Science will:

Apply the theoretical and technical expertise in laboratory techniques required to lead the activities of a Medical Genomics laboratory, including one specialised area of Medical Genomics

Content	Outcomes	Indicator
MG5 Advanced laboratory techniques in Constitutional Genetic Testing	<p>MG5.1 – Detail your experience and contribution with an advanced laboratory technique used in Constitutional Genetic Testing.</p> <p>MG5.2 – Describe the development of an advanced technique used in your field of expertise and its application to the analysis of a pathological disorder. Evaluate the science &/or technology underpinning the technique and describe the contributions of some key authors involved in its development</p>	All Part II outcomes in MG5 to MG8 (Advanced laboratory techniques) will be evidenced by answering viva voce examination questions & submitting Faculty of Science Reports to the satisfaction of the Principal Examiner.
OR MG6 Advanced laboratory techniques in Cancer Genetics	<p>MG6.1 – Detail your experience and contribution with an advanced laboratory technique used in Cancer Genetics (germline or somatic mutations)</p> <p>MG6.2 – Describe the development of an advanced technique used in your field of expertise and its application to the analysis of a pathological disorder. Evaluate the science &/or technology underpinning the technique and describe the contributions of some key authors involved in its development</p>	
OR MG7 Advanced laboratory techniques in Reproductive Genetics	<p>MG7.1 – Detail your experience and contribution with an advanced laboratory technique used in Reproductive Genetics</p> <p>MG7.2 – Describe the development of an advanced technique used in your field of expertise and its application to the analysis or prevention of a pathological disorder. Evaluate the science &/or technology underpinning the technique and describe the contributions of some key authors involved in its development</p>	
OR MG8 Advanced laboratory techniques in Population Genetics	<p>MG8.2 – Describe the development of an advanced technique used in your field of expertise and its application to the analysis or prevention of a pathological disorder. Evaluate the science &/or technology underpinning the technique and describe the contributions of some key authors involved in its development</p>	

Content	Outcomes	Indicator
<p>PLUS</p> <p>MG9 Instrumentation</p>	<p>MG9.1 – Describe the principles of operation of an advanced system or apparatus in your field of expertise</p> <p>MG9.2 – Explain the significance of this instrument to a specialised area of Medical Genomics</p>	<p>Answer viva voce examination questions to the satisfaction of the Principal Examiner, describing scientific principles supported by appropriate formulae and statistics, limitations, error detection and troubleshooting, along with how the apparatus or system has advanced Medical Genomics</p>
<p>And</p> <p>MG10 Advanced pathology science</p>	<p>MG10.1 – Describe the genetic basis of a group of disorders that are linked by multiple causative genes, multiple modes of inheritance or by a common pathogenic pathway</p> <p>MG10.2 – Describe the pathogenic mechanisms linking the mutations to dysfunction at the cellular and clinical levels and the implications for the investigation and diagnosis of this group of disorders</p>	<p>Answer viva voce examination questions & submit Faculty of Science Reports to the satisfaction of the Principal Examiner</p>

Clinical Laboratory Standards – Part I – Biochemical Genetics

Standard
<p>Fellows of the Faculty of Science will:</p> <p>Demonstrate competence in applying the techniques, technology and reporting associated with a Genetics laboratory with a broad case-mix of patients</p>

Content	Outcomes	Indicator
<p>BG1 – Metabolism</p>	<p>BG1.1 – Describe normal physiology and biochemistry, including but not limited to:</p> <ul style="list-style-type: none"> • Fluid and electrolyte balance • Acid-base regulation • Intermediary metabolism • Oxidative phosphorylation • Lipids and lipoproteins • Lysosome and peroxisome metabolism • Purines and pyrimidines • Bilirubin and porphyrins • N- and O-glycosylation of proteins • Neurotransmitters and brain metabolism • Creatine metabolism • Calcium metabolism • Trace metal metabolism • Vitamin metabolism 	<p>BG1 will be evidenced by answering viva voce and written examination questions on all aspects of metabolism</p>
<p>BG2 – Laboratory techniques</p> <p>Apply and evaluate the techniques and technology routinely used in the laboratory</p>	<p>BG2.1 – Evaluate biochemical genetic tests and patient results:</p> <ul style="list-style-type: none"> • Calculate and interpret statistical data including sensitivity, limits of detection, specificity, positive predictive value, receiver operating characteristics analysis, bias, predictive values, likelihood ratio and Bayes theorem • Explain how to determine reference ranges • Statistical techniques for method comparisons • Apply parametric and non-parametric statistics and multiple of median analyses • Apply and interpret methods of bivariate statistical analysis • Calculate uncertainty in measurement • Use spread-sheets to analyse data sets: summarising, descriptive statistics, charting • Apply the bioinformatic and biostatistical methodology required to assess and clinically interpret multi-analyte analyses 	<p>BG2 will be evidenced by a portfolio of work, including examples of method development and validation, and answering viva voce and written examination questions that evaluate and explain the principles of diagnostic assays and their output</p>

Content	Outcomes	Indicator
	<p>BG2.2 – Describe the general principles of instrument operation and analysis and validation of laboratory data:</p> <ul style="list-style-type: none"> • Apply laboratory techniques and use equipment • Describe strengths and limitations of different assay techniques • Monitor and verify results in accordance with laboratory procedures including internal QC acceptance/ rejection and using QC data to monitor changes and trends • Outline the purpose of clearly defining the limits of detection for analytes • Perform calibration procedures on platforms and analytes, routine preventative maintenance procedures and checks to determine suitable assay performance • Explain the principles, requirements and functioning of laboratory information systems used for instrument interfacing, flagging of results and generating interpretive comments • Describe how to implement trouble-shooting procedures <p>BG2.3 – Describe the principles, uses and limitations of the following methods in Biochemical Genetics</p> <ul style="list-style-type: none"> • Sample preparation: protein elimination, dialysis and ultrafiltration, solvent extraction, solid phase extraction, derivatisation • Immunoassay methods: ELISA and FIA, antibody specificity and calibration • Enzymatic assays: end-point and kinetic • Spectrophotometry and fluorimetry • Thin layer chromatography • Electrophoresis: cellulose acetate, capillary, PAGE and isoelectric focusing • Liquid Chromatography: modes (reverse and normal phase, ion exchange), influence of column dimensions and particle size on chromatography • Gas chromatography: phases used, isothermal vs temperature programmed • Mass spectrometry: ion sources (electron impact, electrospray, APCI, MALDI) and analysers (quadrupole, time-of-flight, ion trap, tandem MS), isotopic patterns and use of stable isotopes as internal standards and in-vitro and in-vivo tracers, fragmentation of molecules in different MS modes, interpretation of mass spectra, different scan modes • Enzyme assays: tissue preparation, enzyme kinetics, assay conditions, use of artificial substrates, use of ratios 	<p>BG2 will be evidenced by a portfolio of work, including examples of method development and validation, and answering viva voce and written examination questions that evaluate and explain the principles of diagnostic assays and their output</p>

Content	Outcomes	Indicator
<p>BG3 – Investigation of Inborn errors of metabolism</p>	<p>BG3.1 – Newborn screening (NBS):</p> <ul style="list-style-type: none"> • Describe the significance of NBS program coverage, collection time, transit times, turnaround times and refusals • Describe methods used for NBS and the strengths and limitations vs equivalent methods used in routine chemical pathology • Describe NBS protocols: action limits, repeat analysis, repeat sample from baby, clinical referrals • Explain abnormal screening results due to prematurity, contamination, intravenous nutrition, maternal disorders, carrier status or samples collected at the wrong time • Describe the practical limitations of NBS sample collection: paper characteristics, haematocrit effects, factors causing rejected samples • Describe second-tier screening assays • Describe algorithms for confirmatory testing • Evaluate outcomes for diagnosed patients • Explain the significance of incidence/prevalence • Explain the significance of sensitivity, specificity and positive predictive value and their impacts on the screened population and health professionals • Describe secondary uses for stored NBS cards and protocols and legislation relevant to this <p>BG3.2 – Explain the investigation, clinical symptoms, biochemical changes and management of IEMs affecting the following areas of metabolism and organelle biogenesis:</p> <ul style="list-style-type: none"> • Amino acids • Organic acids • Hyperammonaemia and urea cycle • Carbohydrate metabolism • Glucose homeostasis • Fatty acid oxidation • Ketone body metabolism • Lysosomal storage disorders • Peroxisomal disorders • Porphyrias • Metal metabolism • Mitochondrial oxidative phosphorylation • Vitamin/cofactor metabolism • Purines and pyrimidines • Pterins and biogenic amines • Neurotransmitter metabolism • Congenital disorders of glycosylation • Creatine metabolism • Hereditary disorders of electrolyte and trace element metabolism • Cholesterol, sterols and bile acids 	<p>BG3 will be evidenced by a portfolio of work including case reports, answering viva voce and written examination questions interpreting the output of data to assist in the diagnosis and management of conditions</p>

Content	Outcomes	Indicator
	<ul style="list-style-type: none"> • Lipoproteins and lipids • Gamma glutamyl cycle • Membrane transport • Defects of connective tissue • Endocrine disorders <p>BG3.3 – IEM treatment. Explain the treatment of metabolic disorders including, but not limited to:</p> <ul style="list-style-type: none"> • Dietary management • Metabolic inhibitors • Metabolic activators • Scavengers e.g. benzoate, glycine • Cofactors • Plasma exchange/plasmapheresis • Chelation • Organ transplantation • Enzyme replacement • Chaperone therapy • Gene therapy • Stop-codon read-through • How biochemical genetic testing can be used to monitor response to treatment <p>BG3.4 – Developing and reporting a professional opinion</p> <ul style="list-style-type: none"> • Interpret and synthesise laboratory data, aided by validated bioinformatic and database resources and peer reviewed published data where appropriate • Recognise non-genetic causes that may mimic an inborn error of metabolism • Recognise non-specific findings that may require reflex testing on the same sample • Recognise diagnostic patterns of metabolites and use of metabolite ratios, patterns in carriers vs affected • Recognise limitations of testing e.g. normal levels of metabolites in treated patients or non-acute samples • Identify artefacts due to diet, medications, sample degradation, etc • Assess the significance of biochemical genetic results in conjunction with other results: e.g. routine biochemistry, haematology, imaging and other genetic testing (CNVs, long contiguous stretches of homozygosity, assessing pathogenicity of variants). • Add concise, meaningful comments to reports where appropriate • Provide recommendations for additional investigations required to confirm or exclude a diagnosis 	

Content	Outcomes	Indicator
	<ul style="list-style-type: none"> • Use the laboratory information system to design algorithms for investigating and reporting different clinical scenarios • Use algorithms and other appropriate tools to highlight results requiring additional attention and develop protocols for reflex testing • Report in accordance with appropriate legislation and frameworks 	
BG4 – Clinical investigations	<p>BG4.1 – Describe and apply guidelines for laboratory investigations to determine the metabolic causes of clinical presentations, including but not limited to:</p> <ul style="list-style-type: none"> • Acute encephalopathy • Acute life-threatening event • Acute psychosis • Brain imaging abnormalities • Cardiomyopathy • Congenital lactic acidosis • Developmental regression • Dysmorphic features • Eye disease • Exercise intolerance • Hepatosplenomegaly • Hyperammonaemia • Hypoglycaemia • Intellectual disability • Liver disease including acute liver failure • Metabolic acidosis • Movement disorders • Myopathy • Nutritional status and growth failure • Peripheral neuropathy • Rhabdomyolysis • Renal disorders including stone formers, the Fanconi syndrome and tubular acidosis • Seizures • Skeletal disorders • Skin disorders • Unexplained infant death • Unusual urine and body odour 	BG4 will be evidenced by answering viva voce and written examination questions

Clinical Laboratory Standards – Part II – Biochemical Genetics

Standard
<p>Fellows of the Faculty of Science will:</p> <p>Apply the theoretical and technical expertise in laboratory techniques required to lead the activities of a Biochemical Genetics laboratory, including one specialised area of Biochemical Genetics</p>

Content	Outcomes	Indicator
<p>BG5 – Advanced laboratory techniques in Metabolite Analysis</p>	<p>BG5.1 – Detail your experience and contribution with an advanced laboratory technique used in Metabolite Analysis relevant to Biochemical Genetics. Examples may include targeted and non-targeted analyses</p> <p>BG5.2 – Describe the development of an advanced technique used in your field of expertise and its application to the analysis of a pathological disorder. Evaluate the science &/or technology underpinning the technique and describe the contributions of some key authors involved in its development</p>	<p>All Part II outcomes in BG5 to BG8 (Advanced laboratory techniques) will be evidenced by answering viva voce examination questions & submitting Faculty of Science Reports to the satisfaction of the Principal Examiner.</p>
<p>OR</p> <p>BG6 – Advanced laboratory techniques in Newborn Screening</p>	<p>BG6.1 – Detail your experience and contribution with an advanced laboratory technique used in Newborn Screening. Examples may include, but are not limited to:</p> <ul style="list-style-type: none"> • Significant improvements to existing screening methods • Pilot studies for new tests and disorders • Introduction of second-tier testing • Use of advanced bioinformatic procedures to improve the newborn screening process <p>BG6.2 – Describe the development of an advanced technique used in your field of expertise and its application to the analysis of a pathological disorder. Evaluate the science &/or technology underpinning the technique and describe the contributions of some key authors involved in its development</p>	
<p>OR</p> <p>BG7 – Advanced laboratory techniques in Functional Biochemical Genetics Analyses</p>	<p>BG7.1 – Detail your experience and contribution with an advanced laboratory technique used in Functional Biochemical Genetics analyses. Examples may include, but are not limited to:</p> <ul style="list-style-type: none"> • Measurement of enzyme activity or other protein functions • Protein expression and structural studies • Organelle and whole cell metabolism studies • Organelle and whole cell structural studies • Loading and tracer studies in patients 	

Content	Outcomes	Indicator
	<p>BG7.2 – Describe the development of an advanced technique used in your field of expertise and its application to the analysis of a pathological disorder. Evaluate the science &/or technology underpinning the technique and describe the contributions of some key authors involved in its development</p>	
<p>OR</p> <p>BG8 – Advanced laboratory techniques in Bioinformatics</p>	<p>BG8.1 – Detail your experience and contribution with an advanced laboratory technique used in Bioinformatics relevant to Biochemical Genetics. Examples may include, but are not limited to:</p> <ul style="list-style-type: none"> • Procedures to analyse and summarise complex instrumental data • Procedures to identify disease patterns in complex data sets • Development of databases and procedures to improve the storage and interpretation of Biochemical Genetics data <p>BG8.2 – Describe the development of an advanced technique used in your field of expertise and its application to the analysis or prevention of a pathological disorder. Evaluate the science &/or technology underpinning the technique and describe the contributions of some key authors involved in its development</p>	
<p>PLUS</p> <p>BG9 – Instrumentation</p>	<p>BG9.1 – Describe the principles of operation of an advanced system or apparatus in your field of expertise</p> <p>BG9.2 – Explain the significance of this instrument to a specialised area of Biochemical Genetics</p>	<p>Answer viva voce examination questions to the satisfaction of the Principal Examiner, describing scientific principles supported by appropriate formulae and statistics, limitations, error detection and troubleshooting, along with how the apparatus or system has advanced Biochemical Genetics</p>
<p>AND</p> <p>BG10 – Advanced pathology science</p>	<p>BG10.1 – Describe the genetic basis of a group of disorders caused by mutations affecting a common pathway, organelle or a common pathogenic mechanism</p> <p>BG10.2 – Describe the pathogenic mechanisms linking the mutations to dysfunction at the cellular and clinical levels and the implications for the investigation and diagnosis of this group of disorders</p>	<p>Answer viva voce examination questions and submit Faculty of Science Reports to the satisfaction of the Principal Examiner</p>

Innovation, Development and Leadership Standards

Standard
<p>Fellows of the Faculty of Science will:</p> <ul style="list-style-type: none"> • Apply, implement and evaluate strategies that guarantee quality assurance, compliance, safety and efficient use of resources fundamental to the operation of a Genetic Pathology laboratory • Demonstrate a commitment to the continual improvement and advancement of Genetic Pathology • Apply the principles of Evidence Based Laboratory Practice (EBLP) to inform health care decisions

Content	Outcomes	Indicator
<p>I 1 – Evaluate laboratory policies and practices to meet quality management, compliance and safety standards</p>	<p>1.1 Maintain and evaluate a quality assurance system under ISO 15189</p> <p>1.2 Evaluate current practices to ensure compliance with NPAAC standards as appropriate or international equivalent</p> <p>1.3 Synthesise quality assurance, quality control and safety, and Total Quality Management policies to meet NATA accreditation or international equivalent</p> <p>1.4 Act with accountability to facilitate workflow, teams, decision making, and communication in management and planning of services and/or departments</p> <p>1.5 Evaluate and improve workplace safety through proactive management practices, employing laboratory information systems and reporting mechanisms where appropriate</p> <p>1.6 Develop or review the processes of validation and verification of methodology used in the laboratory</p>	<p>Answer written examination and viva voce questions that demonstrate competence in these aspects of management required to lead a laboratory</p> <p>PLUS</p> <p>Satisfactory completion of the RCPA Laboratory Management modules (online)</p>

Content	Outcomes	Indicator
<p>I 2 – Demonstrate leadership and innovation in developing the practice of Genetic Pathology</p>	<p>2.1 Maintain an evidence base to support advice provided to clinicians</p> <p>2.2 Design, adapt and implement analytically valid and traceable routine tests, underpinned by reference materials and documented methods</p> <p>2.3 Evaluate new methods as fit for use</p> <p>2.4 Assess business opportunities for validity where appropriate</p> <p>2.5 Provide strategic direction for laboratory including management of change</p> <p>2.6 Support and promote the education of colleagues, co-workers, students, and the public through a variety of strategies including formal/ informal teaching, educational material development, and mentoring</p> <p>2.7 Reflect on your engagement in Continuing Professional Development (CPD), and personal benefits</p> <p>2.8 Define and model ethical practices in handling/ reporting patient information, interacting with others and seeking opinion, conflict of interest, financial probity, and managing errors</p> <p>2.9 Identify your role in professional societies/ colleges and contribute to its activities</p>	<p>Answer viva voce questions and document activities in the portfolio that demonstrate leadership and innovation in these aspects of laboratory practice, supported by specific personal contributions.</p> <p>review or develop educational materials for non-scientists e.g. Lab Tests Online Australasia</p> <p>Satisfactory completion of the RCPA Ethics and Confidentiality modules (online)</p>
<p>I 3 – Demonstrate the ability to make informed decisions by accessing and integrating the most current, relevant, valid and reliable evidence available</p>	<p>3.1 Identify knowledge gaps during practice and construct focussed, answerable questions to address these gaps</p> <p>3.2 Use an appropriate search strategy to answer identified questions through existing evidence</p> <p>3.3 Critically evaluate the relevance, currency, authority and validity of all retrieved evidence including scientific information and innovations</p> <p>3.4 Apply the appraised evidence appropriately to practice by informing decisions in the given context</p> <p>3.5 Use reflective and consultative strategies to evaluate the EBLP process</p>	<p>Faculty of Science Reports submitted by the candidate should demonstrate principles of EBLP AND</p> <p>Answer written examination and viva voce questions</p>

SECTION 3 – ASSESSMENT POLICY

This section explains the specific requirements and assessment policy for the Faculty of Science Genetic Pathology program. It should be read in conjunction with the **RCPA Trainee Handbook Administrative requirements**, found on the College website. (<http://www.rcpa.edu.au/Trainees/Curriculum>)

Part I – Requirements

Assessment in **Part I** is by:

1. Formal examinations
2. A portfolio of evidence indicating completion of a sufficient number and type of workplace-based activities and assessment
3. Satisfactory progress (Supervisor Reports)

See Assessment Matrix in **Appendix 4 & 5**.

The aim of the **Part I** assessments is to ensure that Trainees have spent time in the laboratory, acquired requisite knowledge and skills and participated in a community of practice, such that they can appropriately mix the laboratory/scientific and clinical elements of Genetic Pathology.

1. Formal examinations

There will be a written examination and an oral examination, held in designated examination centres on dates specified by the College.

The written examination will require short answer and/or extended responses to questions from the Clinical Laboratory (Part I) and Innovation, Development and Leadership components of the curriculum. The research component is assessed separately at Part II level.

The practically oriented structured oral examination will consist of multiple stations of 10-15 minutes duration. The focus of the oral examination will be on demonstrating practical aspects of Laboratory Standards (Part I) and Laboratory Innovation, Development and Leadership Standards such as the interpretation of test results, measurements and calculations, problem solving and reporting, quality control and laboratory management, although the discussion will often be much broader. Where relevant all candidates will be given reading material to evaluate before entering the exam stations.

2. Portfolio requirements

In addition to various formal examinations, assessments carried out in the workplace (i.e. Directly Observed Practical Skills, short case reports, Case-based Discussions) and evidence of other learning activities should be recorded in a Logbook and portfolio. Together, these 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. It is the Trainee's responsibility to keep the logbook up to date and meet the additional portfolio requirements.

Appendix 1 details the Genetic Pathology Portfolio Requirements for both Part I and Part II.

Logbook

A sample page of what will become a logbook for recording workplace-based activities can be found in **Appendix 2**. **Every formal learning** activity should be recorded here. Only those outlined below should be documented in more detail. Opportunities in the development and assessment of communication skills should also be recorded.

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

Short case reports

Trainees must complete a total of three or more short case reports (~1000 words).

The trainee should discuss with their supervisor before selecting a topic/case for the report. The focus of the report could be on a specific technical aspect covering any of the content areas specified in the Part I Laboratory Standards. The discussion should include a focussed review of the relevant literature.

The Trainee should select a suitable assessor, who should be an RCPA Fellow but does not need to be the listed supervisor. The assessor could note this as a quality activity in their annual Continuing Professional Development Program (CPDP) submission. Short case reports will be evidenced by the assessor completing the assessment form, included as **Appendix 3**. Please include the completed assessment form and the report in the portfolio. Trainees are encouraged to present their case reports at scientific forums as oral presentations or posters.

Case-based discussions (CbD)

Trainees must complete a total of five or more Case-based discussions (CbD). CbDs will be evidenced by the supervisor completing the relevant CbD form.

Doing CbD assessments is excellent preparation for the **oral examinations** for trainees. CbD assessments provide feedback about the trainee's ability to interpret and relate laboratory results to opinions and conclusions, including about case circumstances; to plan appropriate investigations, and to provide advice on decisions related to investigations, including decisions with ethical and legal dimensions. The purpose of the CbD assessment is also to provide feedback to Trainees about their progress by highlighting strengths and areas for improvement, thereby encouraging their professional development.

The Trainee should initiate each CbD assessment. At the time of the assessment, the Trainee should select two (2) recent cases with 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 assessor could note this as a quality activity in their annual Continuing Professional Development Program (CPDP) submission. The Trainee should request a mutually convenient time to meet 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 included in **Appendix 2**. 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.

Directly Observed Practical Skills (DOPS)

Trainees will be required to demonstrate competence in their day-to-day work by performing Directly Observed Practical Skills. In Genetic Pathology, Trainees are required to satisfactorily complete DOPS from the 12 content areas listed below for either the Medical Genomics or Biochemical Genetics strands. Content from the Common Core Material (G1-G3) will be assessed in both strands. Once proficiency is achieved in each DOPS area (to be assessed by at least one instance of observing the trainee in each DOPS and giving feedback) the Supervisor should complete a DOPS assessment form included in **Appendix 2**. **DOPS from the first nine (9) content areas must be completed before a candidate is eligible to sit the Part I examination.** DOPS for the final three (3) content areas must be completed before sitting the Part II examination. The Supervisor should be guided by the outcomes in the Clinical Laboratory Standards section of the handbook for the scope and level of proficiency required.

The 12 DOPS assessment categories for both the Medical Genomics and Biochemical Genetics strands are below:

Medical Genomics

1. Sample reception/handling, data entry, test assignment, storing, checking, sample retrieval and referral procedures
2. Cell separation/purification, culture set up, maintenance and harvesting, nucleic acid isolation by manual and automated approaches, quantification/quality assessment, storage/archiving
3. DNA labelling for FISH, microarray and other hybridisation-based procedures
4. Microscopy (bright-field and fluorescence), banding and karyotype analyses, FISH analysis
5. PCR-based analysis (end point, quantitative, real-time and methylation specific PCR)
6. Other Genotyping methods (e.g. single nucleotide extension, microsatellite analysis)
7. Gel-based hybridisation analysis including the use of methylation sensitive restriction enzymes
8. Electrophoretic fragment separation, sizing and analysis
9. DNA sequencing analysis electrophoretic for somatic and constitutional genomic variants including bisulphite sequencing
10. Array technologies and analysis for detection of CNVs and LESH/LOH
11. DNA sequencing (massively parallel) and analysis for somatic and constitutional genomic variants
12. RNA analysis for splicing assessment and gene expression assessment

Biochemical Genetics

1. Sample reception/handling, data entry, test assignment, storing, checking, sample retrieval and referral procedures
2. Handling and processing of biological fluid samples and simple preliminary investigations e.g. spot/diptests, note haemolysis.
3. Handling and processing of cells and tissues (cell separation/purification, culture set up, maintenance and harvesting; tissue homogenisation and subcellular fractionation)
4. Routine automated/stat/urgent biochemistry
5. Amino acid analysis
6. Organic acid analysis
7. Acylcarnitine profile analysis
8. Enzyme analysis
9. Newborn screening analysis
10. Specialised mass spectrometric methods and analysis
11. Specialised bioinformatics analysis
12. Another analytical method and analysis not addressed in assessed categories (e.g., electrophoretic, immunochemical, radiometric, fluorimetric, luminometric, proteomic, metabolomic, genomic, etc)

Once proficiency is achieved (to be assessed by at least one instance of observing the trainee in each content area and giving feedback) the supervisor should complete the relevant DOPS competence form. The supervisor should be guided by the outcomes in the relevant laboratory standards sections for the scope and level of competence required. Trainees should only use the DOPS form of their own discipline.

Other Evidence

Trainees should ensure that they are engaged in a variety of learning activities related to teaching, scholarship and leadership throughout training. These may include presentations (oral and posters), writing abstracts, staff presentations, conferences, teaching, and developing educational material. A suggestion for educational material development is the Lab Tests Online Australasia editing process, please email your details and discipline to ltoau@aacb.asn.au to participate.

These activities develop written and oral communication skills. Whilst each activity should be recorded in the logbook, documented evidence of a minimum of 5 from a variety of activity types per year should be made available upon request over the training period.

3. Supervisor Reports

The supervisor must review and sign off the *completed portfolio forms* and the *logbook* on the **Supervisor reports**. The supervisor must also rate the trainee according to their professional judgement in a range of competencies including in laboratory skills, research, innovation and leadership, and professional attitudes and behaviours. The behaviours to be rated and the rating scale with anchors are provided in the supervisor report.

Trainees must submit a Supervisor Report for each year of training (and period of rotation if applicable) to the RCPA Registrar. Trainees who are sitting the **Part I** oral examination must submit an additional pre-examination Supervisor Report. A cumulatively updated **Portfolio Summary Sheet**, documenting the portfolio of workplace based activities and assessment, must be appended to the pre-examination Supervisor Report and sent to the RCPA Registrar prior to the **Part I** oral examinations at a time determined by the RCPA. 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. The Supervisor Report form can be found at: <http://www.rcpa.edu.au/Trainees/Training-with-the-RCPA/Supervisor-Reports>

The portfolio summary sheet will be reviewed by the Registrar, Board of Education and Assessment or delegate and the Principal Examiner. The signatories and Trainee may be contacted to confirm evidence of satisfactory completion.

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

Summary of assessment requirements for Part I

Item	Completion	Assessed by	Comments
Written examination: short answer and/or more extended responses	At the end of three years of training	Marked by two (2) examiners with appropriate experience	Questions set by a panel of examiners
Oral examination: Multi-stationed set of structured interviews, with practically-oriented questions	After submission of pre-exam supervisor report and portfolio summary sheet	Two (2) examiners with appropriate experience per station	Questions set by a panel of examiners
Portfolio items (see Appendix I) to be signed off by supervisor or delegate e.g. DOPS, CbDs, Short Case Reports	To be completed before Part I oral examination	Portfolio summary spreadsheet is checked for completeness by RCPA. If incomplete, the candidate may be required to undertake further activities.	Portfolio items are to be reviewed by the supervisor when preparing the supervisor report. (The portfolio should not be sent to the College unless requested for audit)
Supervisors' Reports with portfolio summary spreadsheet.	Annual (end of rotation if applicable) and Part I pre-exam reports	Reviewed by College registrar or delegate	Referral to Principal Examiner if necessary.

Part II – Requirements

Assessment in **Part II** is by:

1. Formal examinations
2. Faculty of Science Reports on Clinical Laboratory Practice
3. Portfolio of evidence indicating completion of a sufficient number and type of workplace-based activities and assessments
4. Research work and reports
5. Satisfactory progress (Supervisor Reports)

See Assessment Matrix in **Appendix 4 & 5**.

The aim of the **Part II** assessments is to ensure that Trainees have spent time in the clinical laboratory, acquired requisite knowledge and skills and participated in a community of practice, such that they can appropriately lead the activities of a genetic pathology laboratory in their area of expertise.

1. Formal examinations

There will be a structured 'oral' examination, consisting of approximately 3 stations of 20-30 minutes duration. The oral examination will normally pose similar questions for all Faculty of Science candidates (other than in the Laboratory Standards). There will be two examiners per station and responses will be marked against pre-determined criteria. The focus of this examination will be evaluation of specific aspects of Genetics Laboratory Standards (Part II), Research Standards, and Laboratory Innovation, Development and Leadership.

2. Faculty of Science reports on Clinical Laboratory Practice

The **Part II** assessment requires four (4) Reports of 3000-5000 words. These should be of a standard publishable in a journal such as *Pathology*.

The Advanced Laboratory Techniques area selected during Part II should be addressed by at least two (2) reports and the Advanced Pathology Science section of Part II should be addressed by at least one (1) report. Medical Genomics candidates should ensure that massively parallel sequencing analyses are a focus of at least one (1) report. Instrumentation (MG/BG 9) by itself is not considered as a specialised area, but the Reports should demonstrate candidate's competence in Instrumentation where relevant.

The focus of the Report could range from a single patient case or case series to a large population depending on the discipline involved and the complexity of the situation under investigation. The Reports should demonstrate the candidate's approach to analysing the clinical/ pathological problem or issue in the case(s) or the population (including a relevant review of the literature) and follow up action/discussion based on principles of Evidence-based clinical Laboratory Practice.

It is also expected that some Reports will demonstrate the candidate's ability to be innovative, assure quality and consider management issues such as staff, instrument and reagent costs. Where applicable a Report should comment on issues such as, but not limited to, method selection, method validation, method development and trouble-shooting.

Based on the above approach, following are some suggestions appropriate as Report aims:

- The introduction or development of a new test or procedure and comparisons with current best practice
- Transference of an existing test or procedure to a new context, sample type or processing protocol and comparing it to current practice
- A study that examines the sensitivity and specificity of a test or procedure, including positive and negative predictive values in a particular population
- A detailed analysis of cumulative laboratory data (including case series)
- A study comparing specialised populations

Please note that the above list is not exhaustive. Trainees may discuss with their supervisor and determine any other aim, and inform the College administration well before planning the work involved. The Principal Examiner will confirm the appropriateness of the aim.

The Reports will be independently marked by two examiners in the relevant discipline and candidates will be provided with feedback. Candidates are encouraged to submit their Reports early in Part II, and at least 2 Reports should be submitted by the end of the fourth year of training.

Any publications arising from the Reports may be used to meet the requirements of the Research Standards component of the curriculum. Candidates are encouraged to publish their Reports subsequent to examination.

While these reports are considered to be Part II assessments, trainees should commence working on them as soon as possible. It is recommended that all Clinical Laboratory Practice Reports be completed and submitted by the month following the Part II Oral Examination.

Please refer to **Appendix 3** – Guidelines for Faculty of Science Reports (Part II)

3. Portfolio requirements

Directly Observed Practical Skills (DOPS)

DOPS for the final three (3) content areas must be completed before sitting the Part II examination. See pages 45 and 47 for details of the Directly Observed Practical Skills requirements. Trainees should only use the DOPS form of their own discipline.

Other Evidence

Trainees should ensure that they are engaged in a variety of learning activities related to teaching, scholarship and leadership throughout training as described earlier. Whilst each instance of these activities should be recorded in the logbook, documented evidence of a minimum of 5 from a variety of activity types per year should be made available upon request over the training period.

4. Research work and reports

At least 2 first author publications, published in the last ten years together with a written discussion that explains the background, interrelatedness and significance of the research, are required. Candidates must provide details of their own contribution to the research. When addressing this requirement, cross reference should be made to all components of item R1 of the Research Standards section of the curriculum (see p. 8), demonstrating how these standards have been met. Those individuals with a PhD, or a Masters (by Research) related to the area of expertise in Pathology conferred by a university recognised by the College, may be exempted from this requirement at the discretion of the Principal Examiner.

Research management would be assessed through a report to be submitted in the portfolio, which would detail the candidate's ability in managing a research project. The report should contain evidence and discussion (~1000 words) addressing the R2 and relevant R1 outcomes. Suggestions for evidence include research proposals and ethics submissions, grant applications made and/or periodic progress/ evaluation reports of successful grants, and end-of-year reports.

5. Supervisor Reports

Similar to Part I, Trainees who are sitting the **Part II** examination must submit a pre-examination Supervisor Report with the appended copy of the Portfolio Summary Sheet to the RCPA

Registrar prior to the **Part II** examinations at a time determined by the RCPA. Failure to submit by the due date may jeopardise the accreditation of training time or finalisation of examination results. The Supervisor Report form can be found at:

<http://www.rcpa.edu.au/Trainees/Training-with-the-RCPA/Supervisor-Reports>

Summary of assessment requirements for Part II

<i>Item</i>	<i>Completion</i>	<i>Assessed by</i>	<i>Comments</i>
Oral examination: multi-station set of 20-30 min structured interviews	In the fifth year of training	Two (2) examiners with appropriate experience per station	Questions set by a panel of examiners
Faculty of Science Reports: four (4) of a publishable standard to be certified as candidate's own work and signed by supervisor or delegate	To be completed by the month following the Part II oral examination	Assessed by a panel of examiners	Candidates may be required to revise & resubmit if not satisfactory.
Other portfolio items to be signed off by supervisor or delegate e.g. DOPS	To be completed before Part II oral examination	Portfolio summary spreadsheet is checked for completeness by RCPA. If incomplete, the candidate may be required to undertake further activities.	Portfolio items are to be reviewed by the supervisor when preparing the supervisor report. (The portfolio should not be sent to the College unless requested for audit)
Supervisors' Reports with portfolio summary spreadsheet.	Annual (end of rotation if applicable) and Part II pre-exam	Reviewed by College registrar or delegate	Referral to Principal Examiner if necessary.
Research work and reports	One month before Part II oral examination	Assessed by a panel of examiners	Referral to Principal Examiner if necessary.

APPENDICES

Appendix 1 - Portfolio Requirements for Genetic Pathology

The table below sets out guidelines to assist Faculty of Science 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. In particular, the development and demonstration of effective communication skills

Trainees should start accumulating evidence for the portfolio as early as possible in training.

Appendices contain 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 below.

A soft copy **portfolio summary** (Excel spreadsheet) should also be compiled so that trainees can keep track of what they have completed. 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 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, Board of Education and Assessment or delegate and the Principal 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.

Table: Portfolio Requirements for Genetic Pathology.

	Item	Part I	Part II	Evidence
1	Supervisor report/s with brief reflection (maximum 1 page) on the supervisor's comments for each report.	Annual reports (and end of rotation reports if applicable). An additional pre-exam report is required in the year of the Part I and Part II assessments		See Supervisor Report guidelines and forms Appendix
2	DOPS	DOPS from the first nine (9) areas to be completed satisfactorily before Part I examinations	DOPS from the final (3) areas to be completed satisfactorily before Part II examinations	All forms signed as satisfactory by supervisor or other appropriately qualified person as agreed/delegated by Supervisor.
3	CbDs	Five or more Case-based discussions before the Part I examinations		All forms/ reports signed as satisfactory by supervisor or other appropriately qualified person as agreed/delegated by Supervisor. Short case
4	Short Case Reports of 1000 words	Three or more short case reports,		

	Item	Part I	Part II	Evidence
		before the Part I examinations		reports to be included in portfolio.
5	Clinical meetings (laboratory, multidisciplinary) Plus a list of entities presented at each meeting	A combined total of at least five (5) learning activities with a minimum of one (1) in each type per year		Each meeting logged should be signed by the supervisor or another person as agreed/delegated by the Supervisor to verify the trainee's involvement in the meeting.
6	Teaching sessions Sessions conducted for students, colleagues, medical colleagues or other audiences. Educational material development			
7	Scientific forums Plus the abstracts presented at each meeting			
8	RCPA Laboratory Management modules	To be completed satisfactorily before the Part I examinations		Signed as satisfactorily completed by supervisor
9	RCPA Ethics and Confidentiality modules			
10	Research Management Report of 1000 words		To be completed satisfactorily before Part II examinations	Signed as satisfactorily completed by supervisor, report to be included in portfolio.

Appendix 2 – Logbook and Forms

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 include:

- Logbook page
- Short case report assessment form
- Case-based discussion assessment form – Medical Genomics
- Case-based discussion assessment form – Biochemical Genetics
- Directly observed practical skills assessment form – Medical Genomics
- Directly observed practical skills assessment form – Biochemical Genetic

	<h2>Logbook</h2>		
Trainee name:			
Supervisor's name:			
<p>Record the details of each learning activity in the table below. This will form part of your portfolio. This form should be copied as required throughout training.</p>			
Description of learning activity	Date	Comments	Initial
Supervisor's signature:		Date:	

		<h2 style="text-align: center;">Genetic Pathology Short Case Report Assessment Form</h2>	
Trainee nam		Trainee ID (RCPA)	Stage of training Y1 Y2 Y3 Y4 Y5 if > Y5 please specify
Assessor's name		Assessor's position <input type="checkbox"/> Pathologist <input type="checkbox"/> Scientist <input type="checkbox"/> Other (pls specify)	
Please indicate (✓) if each of the following was deemed Satisfactory (S) or Unsatisfactory (U)			
Aspect of Report		S	U
Clear layout of text with appropriate headings and paragraphs. Figures and tables are well planned and easy to understand			
Correct, concise English without spelling or grammatical errors			
Clear introduction, that covers the background of the topic & introduces the rest of the report			
The main body of the report is well organised, easy to read and answers the question that has been set			
A full range of appropriate sources has been used to research the case/topic, including textbooks, journals, websites, personal communications, surveys and/or experiments			
The conclusion accurately summarises the arguments that have been presented			
References are relevant and are cited accurately in the <i>Pathology</i> journal format			
No large amounts of irrelevant material & text			
Please comment on other relevant aspects, especially on aspects for improvement 			
Please indicate the overall standard of the report: <input type="checkbox"/> SATISFACTORY <input type="checkbox"/> UNSATISFACTORY			
Signature of assessor		Signature of Trainee	
Date completed			

Please comment on whether these aspects of the trainee's performance are as expected for the stage of training		S	U	N/A
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 the laboratory tests results to the pathologist or physician.				
Ability to suggest further relevant or more useful tests towards the management of the patient in relation to diagnosis and monitoring including prognostication.				
Understanding of management and financial aspects of the case				
Please comment on the overall skills in effective communication				
Please comment on other relevant aspects, especially on aspects for improvement				
Final outcome (please tick) <input type="checkbox"/> As expected for the stage of training <input type="checkbox"/> Below expected for the stage of training		Date of CbD	Time taken for CbD	Time taken for feedback
Assessor _____		Signature of Trainee _____		
Name (please print)		Signature		Signature
Laboratory 				
Date completed 				

		<h2 style="text-align: center;">Biochemical Genomics</h2> <h3 style="text-align: center;">Case-based Discussion Assessment Form</h3>	
Trainee name		Trainee ID (RCPA)	Stage of training Y1 Y2 Y3 Y4 Y5 if > Y5 please specify
Assessor name and position:			
Please indicate relevant content area/s			
<input type="checkbox"/> Sample reception/handling, data entry, test assignment, storing, checking, sample retrieval and referral procedures		<input type="checkbox"/> Organic acid analysis	
<input type="checkbox"/> Handling and processing of biological fluid samples and simple preliminary investigations e.g. spot/diptests, note haemolysis		<input type="checkbox"/> Acylcarnitine profile analysis	
<input type="checkbox"/> Handling and processing of cells and tissues (cell separation/purification, culture set up, maintenance and harvesting; tissue homogenisation and subcellular fractionation)		<input type="checkbox"/> Enzyme analysis	
<input type="checkbox"/> Routine automated/stat/urgent biochemistry		<input type="checkbox"/> Newborn screening analysis	
<input type="checkbox"/> Amino acid analysis		<input type="checkbox"/> Specialised mass spectrometric methods and analysis	
		<input type="checkbox"/> Specialised bioinformatics analysis	
		<input type="checkbox"/> Another analytical method and analysis not addressed in assessed categories (e.g. electrophoretic, immunochemical, radiometric, fluorimetric, luminometric, proteomic, metabolomic, genomic, etc)	
Focus of discussion (tick as many as apply)			
<input type="checkbox"/> Principles of pathophysiology		<input type="checkbox"/> Significance to clinical management	
<input type="checkbox"/> Diseases and their diagnostic features		<input type="checkbox"/> Instrumentation	
<input type="checkbox"/> Research relevance		<input type="checkbox"/> Quality control	
<input type="checkbox"/> Application of evidence based practice		<input type="checkbox"/> Advanced laboratory techniques	
Complexity of case: <input type="checkbox"/> Low <input type="checkbox"/> Medium <input type="checkbox"/> High			
Brief description of case presented, discussed and assessed			
Why was this case selected for discussion?			

<p>Does this case broaden the Trainee's experience by being different from previous cases that have been discussed? <input type="checkbox"/> Yes <input type="checkbox"/> No <input type="checkbox"/> N/A</p>					
<p>Please comment on whether these aspects of the trainee's performance are as expected for the stage of training</p>			<p>Yes</p>	<p>No</p>	<p>N/A</p>
<p>Ability to present case clearly and concisely</p>					
<p>Good understanding of clinical issues relating to the case</p>					
<p>Good understanding of laboratory issues relating to the case</p>					
<p>Depth of understanding and awareness of current literature relevant to this case</p>					
<p>Ability to interpret results in a balanced and rational way</p>					
<p>Ability to provide and clearly communicate well reasoned professional advice</p>					
<p>Ability to clinically correlate the laboratory tests results to the pathologist or physician</p>					
<p>Ability to suggest further relevant or more useful tests towards the management of the patient in relation to diagnosis and monitoring including prognostication</p>					
<p>Understanding of management and financial aspects of the case</p>					
<p>Please comment on the overall skills in effective communication</p>					
<p>Please comment on other relevant aspects, especially on aspects for improvement</p>					
<p>Final outcome (please tick)</p> <p><input type="checkbox"/> As expected for the stage of training</p> <p><input type="checkbox"/> Below expected for the stage of training</p>		<p>Date of Cbd</p>	<p>Time taken for Cbd</p>	<p>Time taken for feedback</p>	
<p>Assessor</p> <p>_____</p> <p>Name (please print) Signature</p>			<p>Signature of Trainee</p> <p>_____</p> <p>Signature</p>		
<p>Laboratory</p>					
<p>Date completed</p>					

		<h2 style="text-align: center;">Medical Genomics</h2> <h3 style="text-align: center;">Directly Observed Practical Skills (DOPS) Assessment Form</h3>		
Trainee name		Trainee ID (RCPA)	Stage of training Y1 Y2 Y3 Y4 Y5 if > Y5 please specify	
Assessor name and position:				
Assessor name		Assessor's position <input type="checkbox"/> Pathologist <input type="checkbox"/> Scientist <input type="checkbox"/> Other (pls specify)		
Instrument or technique (tick the box that applies). Activities 1 – 9 must be completed prior to Part I exam. All activities must be completed prior to Part II examination.				
1. <input type="checkbox"/> Sample reception/handling, data entry, test assignment, storing, checking, sample retrieval and referral procedures 2. <input type="checkbox"/> Cell separation/purification, culture set up, maintenance and harvesting, nucleic acid isolation by manual and automated approaches, quantification/quality assessment, storage/archiving 3. <input type="checkbox"/> DNA labelling for FISH, microarray and other hybridisation based procedures 4. <input type="checkbox"/> Microscopy (bright-field and fluorescence), banding and karyotype analyses, FISH analysis 5. <input type="checkbox"/> PCR-based analysis (end point, quantitative, real-time and methylation specific PCR)* 6. <input type="checkbox"/> Other Genotyping methods (e.g., single nucleotide extension, microsatellite analysis) 7. <input type="checkbox"/> Gel-based hybridisation analysis including the use of methylation sensitive restriction enzymes 8. <input type="checkbox"/> Electrophoretic fragment separation, sizing and analysis 9. <input type="checkbox"/> DNA sequencing analysis (electrophoretic) for somatic and constitutional genomic variants including bisulphite sequencing 10. <input type="checkbox"/> Array technologies and analysis for detection of CNVs and LESH/LOH 11. <input type="checkbox"/> DNA sequencing (massively parallel) and analysis for somatic and constitutional genomic variants 12. <input type="checkbox"/> RNA analysis for splicing assessment and gene expression assessment				
Number of hours spent performing the method(s) prior to DOPS assessment			Has the Trainee completed the laboratory's usual training process for these methods? <input type="checkbox"/> Yes <input type="checkbox"/> No	
Please comment on whether these aspects of the trainee's performance are as expected for the stage of training			Yes	No
Understands the principles of the methods				
Understands and complies with the laboratory documentation, package inserts, manuals, etc.				
Has completed the assays successfully and produced valid results that can be reported				
Able to explain the QC procedures for these methods, including internal and external QA				
Able to discuss anomalies and resolve uncertainties for the methods				
Able to explain maintenance and trouble-shooting requirements for the methods				

* All four types of PCR-based analysis should be attempted, 10 samples in total (minimum) with sufficient coverage of all types.

Please comment on other relevant aspects, especially on aspects for improvement

Final outcome (please tick) <input type="checkbox"/> As expected for the stage of training <input type="checkbox"/> Below expected for the stage of training	Time taken for DOPS	Number of samples	Time taken for feedback
Assessor _____ Name (please print) Signature		Signature of Trainee _____ Signature Date	
Laboratory			
Date completed			

		<h2 style="text-align: center;">Biochemical Genetics</h2> <h3 style="text-align: center;">Directly Observed Practical Skills (DOPS) Assessment Form</h3>		
Trainee name		Trainee ID (RCPA)	Stage of training Y1 Y2 Y3 Y4 Y5 if > Y5 please specify	
Assessor name and position:				
Assessor name		Assessor's position <input type="checkbox"/> Pathologist <input type="checkbox"/> Scientist <input type="checkbox"/> Other (pls specify)		
Instrument or technique (tick the box that applies), Activities 1 – 9 be completed prior to Part I exam. All activities must be completed prior to Part II examination.				
1. <input type="checkbox"/> Sample reception/handling, data entry, test assignment, storing, checking, sample retrieval and referral procedures 2. <input type="checkbox"/> Handling and processing of biological fluid samples and simple preliminary investigations e.g. spot/diptests, note haemolysis 3. <input type="checkbox"/> Handling and processing of cells and tissues (cell separation/purification, culture set up, maintenance and harvesting; tissue homogenisation and subcellular fractionation) 4. <input type="checkbox"/> Routine automated/stat/urgent biochemistry 5. <input type="checkbox"/> Amino acid analysis 6. <input type="checkbox"/> Organic acid analysis 7. <input type="checkbox"/> Acylcarnitine profile analysis 8. <input type="checkbox"/> Enzyme analysis 9. <input type="checkbox"/> Newborn screening analysis 10. <input type="checkbox"/> Specialised mass spectrometric methods and analysis 11. <input type="checkbox"/> Specialised bioinformatics analysis 12. <input type="checkbox"/> Another analytical method and analysis not addressed in assessed categories (e.g. electrophoretic, immunochemical, radiometric, fluorimetric, luminometric, proteomic, metabolomic, genomic, etc)				
Number of hours spent performing the method(s) prior to DOPS assessment		Has the Trainee completed the laboratory's usual training process for these methods? <input type="checkbox"/> Yes <input type="checkbox"/> No		
Please comment on whether these aspects of the trainee's performance are as expected for the stage of training		Yes	No	N/A
Understands the principles of the methods				
Understands and complies with the laboratory documentation, package inserts, manuals, etc.				
Has completed the assays successfully and produced valid results that can be reported				
Able to explain the QC procedures for these methods, including internal and external QA				
Able to discuss anomalies and resolve uncertainties for the methods				
Able to explain maintenance and trouble-shooting requirements for the methods				

Please comment on other relevant aspects, especially on aspects for improvement

- Final outcome (please tick)**
- As expected for the stage of training
 - Below expected for the stage of training

Time taken for DOPS

Number of samples

Time taken for feedback

Assessor

Name (please print)

Signature

Signature of Trainee

Signature

Date

Laboratory

Date completed

Appendix 3 – Guidelines for Faculty of Science (Clinical Laboratory Practice) Reports

The Part II assessment requires four (4) Reports of 3000-5000 words. These should be of a standard publishable in a journal such as *Pathology*.

The focus of the Report could range from a single patient case or case series to a large population depending on the discipline involved and the complexity of the situation under investigation. The Reports should demonstrate the candidate's approach to analysing the clinical/ pathological problem or issue in the case(s) or the population (including a relevant review of the literature) and follow up action/discussion based on principles of Evidence-based clinical Laboratory Practice.

It is also expected that some Reports will demonstrate the candidate's ability to be innovative, assure quality and consider management issues such as staff, instrument and reagent costs. Where applicable a Report should comment on issues such as, but not limited to, method selection, method validation, method development and trouble-shooting.

Based on the above approach, following are some suggestions appropriate as Report aims:

- The introduction or development of a new test or procedure and comparisons with current best practice
- Transference of an existing test or procedure to a new context, sample type or processing protocol and comparing it to current practice
- A study that examines the sensitivity and specificity of a test or procedure, including positive and negative predictive values in a particular population
- A detailed analysis of cumulative laboratory data (including case series)
- A study comparing specific populations

Please note that the above list is not exhaustive. Trainees may discuss with their supervisor and determine any other aim, and inform the College administration well before planning the work involved. The Principal Examiner will confirm the appropriateness of the aim.

In Genetic Pathology the Advanced Laboratory Techniques area selected during Part II should be addressed by at least two (2) reports and the Advanced Pathology Science section of Part II should be addressed by at least one (1) report. Medical Genomics candidates should ensure that massively parallel sequencing analyses are a focus of at least 1 report. Instrumentation (MG/BG 9) by itself is not considered as a specialised area, but the Reports should demonstrate candidate's competence in Instrumentation where relevant.

The Reports will be independently marked by two examiners in the relevant discipline and candidates will be provided with feedback. Candidates are encouraged to submit their Reports early in Part II, and at least two Reports should be submitted by the end of the fourth year.

Format

1. An electronic copy in **pdf format** should be submitted.
2. The first page should have the Trainee's RCPA number and the word count (excluding references). For examination and feedback purposes page numbers should be provided for the whole document and line numbers should be provided for all text.
3. The Trainee's name should NOT be displayed anywhere in the document.
4. Any information and contributions provided by others should be clearly identified. Do NOT give personal or institutional details of the individuals concerned. The Report submitted should be primarily the candidate's own work and any attribution of authorship should take place only at the time of possible publication.

5. The manuscript and reference format should comply with the requirements for the journal *Pathology*. <http://edmgr.ovid.com/pat/accounts/ifaauth.htm>

Marking criteria

1. Demonstrates one or more of the Report aims.
2. Demonstrates appropriate principles of Evidence Based Laboratory Practice
3. Introduction discusses the literature and placement of the study in context.
4. Methodology is appropriate. Method described in sufficient detail to allow the study to be replicated; comments on method selection, method validation, method development and trouble-shooting..
5. Analysis: Quantitative or qualitative
6. Results
7. Discussion
 - i. Interpretation of results or critical analysis of literature
 - ii. Placement of results in context of the available literature
 - iii. Limitations of the study
 - iv. Lessons derived are adequately discussed; implications are related to the candidate's own situation and the broader context of the field
8. Format of the paper
 - i. Complies with the requirements for the journal *Pathology* <http://edmgr.ovid.com/pat/accounts/ifaauth.htm>
 - ii. Reference List
 - iii. Writing style syntax, spelling/ typographical errors
 - iv. Graphs and tables.

Reports will be graded as either Satisfactory or Unsatisfactory. Unsatisfactory reports will be returned to the candidate for revision, addressing of feedback, and resubmission to the RCPA for remarking

Any publications arising from the Reports may be used to meet the requirements of the Research Standards component of the curriculum. Candidates are encouraged to publish their Reports subsequent to examination.

Declaration of originality

Each Report must be accompanied by a signed declaration of originality. Please use the form on the next page and do NOT incorporate the form into the Report, to preserve anonymity. The College's policy is that Trainees who submit work that is not their own will fail and the matter will be referred to the Board of Education and Assessment.

Submitting the report and originality declaration

Please *email* the report and the signed declaration of originality to the College at exams@rcpa.edu.au. The declaration and the report will be kept on file at the College. E-copies of the report will be sent to examiners. Please refer to RCPA website for due dates.



Declaration for Faculty of Science Reports

Trainee Declaration:

I certify that this Report, titled:

.....
.....
.....
.....

is my own original work and that the work documented was completed as part of my personal supervised practice during my accredited training. It has not been previously submitted for assessment and has not been used by any other trainee in this laboratory. I have read and understand RCPA Policy 10/2002 - Plagiarism and Cheating in Examinations.

Trainee NameRCPA ID

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

Supervisor declaration:

As the supervisor for, I certify that the work documented was completed personally by him/her during training. The Report is original and has not been used by any other trainee in this laboratory. I have reviewed this item and read the relevant RCPA requirements and believe it is suitable for submission to the RCPA examiners

Supervisor name

Supervisor signature.....date

Appendix 4 – Faculty of Science Medical Genomics Assessment Matrix

	Outcomes to be assessed <i>(From the Faculty of Science curriculum)</i>	Part I		Part II				Portfolio				
		Written exam (SAQ)	Structure d oral exam	Structure d oral exam	Research thesis	Published articles	Faculty of Science Reports	Short case reports	CbDs	DOPS	Other reports	Suggestions for portfolio evidence of activity
Clinical Laboratory – I	G1 Laboratory practice	Y							Y	Y		1, 2
	G2 Foundations of Genetics	Y	Y					Y	Y			1, 2
	G3 Preparation of samples	Y	Y							Y		
	MG1 Laboratory techniques	Y	Y					Y	Y	Y		
	MG2 Somatic Genetics	Y	Y									
	MG3 Investigation of Genomic disorders	Y	Y					Y	Y	Y		
	MG4 Clinical molecular genetics	Y	Y					Y	Y			
Clinical Laboratory – II	MG5 Advanced laboratory techniques in Constitutional Genetic Testing			Y			Y			Y		
	MG6 Advanced laboratory techniques in Cancer Genetics			Y			Y			Y		
	BG7 Advanced laboratory techniques in Reproductive Genetics			Y			Y			Y		
	BG8 Advanced laboratory techniques in Population Genetics			Y			Y			Y		
	BG9 Instrumentation			Y			P					
	BG10 Advanced pathology science			Y			Y					
Innovation & Leadership	I1 Quality and safety of laboratory practices	Y	P	Y			Y		P			4, 5, 6, 7
	I2 Leadership and innovation in developing the discipline	P	P	Y	P	P	Y	P			P	8, 9
	I3 Evidence Based Laboratory Practice in decision making	Y	P	Y			Y	P	Y			1, 3
Research	R1 Conducting Research			Y	Y	Y	P					
	R2 Research Management & administration			Y	P						Y	
	R3 Research Communication			Y	P	Y						1, 2

Y = Yes P = Possibly

* Portfolio categories: 1. Attendance/ presentations at laboratory/ multidisciplinary meetings; 2. Attendance/ presentations at scientific forums e.g. conferences; 3. Teaching sessions; 4. Attendance at management meetings; 5. Quality activities; 6. Incident reports; 7. RCPA Management module; 8. RCPA Ethics module; 9. Educational material development

Appendix 5 - Faculty of Science Biochemical Genetics Assessment Matrix

		Part I		Part II				Portfolio				
		Written exam (SAQ)	Structure d oral exam	Structure d oral exam	Research thesis	Published articles	Faculty of Science Reports	Short case reports	CbDs	DOPS	Other reports	Suggestions for portfolio evidence of activity
Clinical Laboratory – I	G1 Laboratory practice	Y							Y	Y		1, 2
	G2 Foundations of Genetics	Y	Y					Y	Y			1, 2
	G3 Preparation of samples	Y	Y							Y		
	BG1 Metabolism	Y	Y									
	BG2 Laboratory techniques-routine	Y	Y					Y	Y	Y		
	BG3 Investigation of Inborn errors of metabolism	Y	Y					Y	Y	Y		
	BG4 Clinical investigations	Y	Y					Y	Y			
Clinical Laboratory – II	BG5 Advanced laboratory techniques in Metabolite Analysis			Y			Y			Y		
	BG6 Advanced laboratory techniques in Newborn Screening			Y			Y			Y		
	BG7 Advanced laboratory techniques in Functional Biochemical Genetics Analyses			Y			Y			Y		
	BG8 Advanced laboratory techniques in Bioinformatics			Y			Y			Y		
	BG9 Instrumentation			Y			P					
	BG10 Advanced pathology science			Y			Y					
Innovation & Leadership	I1 Quality and safety of laboratory practices	Y	P	Y			Y		P			4, 5, 6, 7
	I2 Leadership and innovation in developing the discipline	P	P	Y	P	P	Y	P			P	8, 9
	I3 Evidence Based Laboratory Practice in decision making	Y	P	Y			Y	P	Y			1, 3
Research	R1 Conducting Research			Y	Y	Y	P					
	R2 Research Management & administration			Y	P						Y	
	R3 Research Communication			Y	P	Y						1, 2

Y = Yes P = Possibly * Portfolio categories: 1. Attendance/ presentations at laboratory/ multidisciplinary meetings; 2. Attendance/ presentations at scientific forums e.g. conferences; 3. Teaching sessions; 4. Attendance at management meetings; 5. Quality activities; 6. Incident reports; 7. RCPA Management module; 8. RCPA Ethics module; 9. Educational material development