CCMG Genetic and Genomic Diagnostics

Second Specialty Training Guidelines and Requirements

1. The second specialty training programs in either Clinical Cytogenetics or Molecular Genetics as described below is open to CCMG fellows certified in Clinical Cytogenetics, Molecular Genetics, Biochemical Laboratory Genetics, Biochemical Clinical Genetics or RCPSC Medical Genetics and Genomics/ CCMG Clinical Genetics or equivalent (please refer to the GGD Training Guidelines for provisions regarding examinations and training completion dates).
2. The minimum training duration is one-year full-time equivalent (52 weeks).
3. The second specialty training in Clinical Cytogenetics or Molecular Genetics is organized in a series of units covering related topics in genomic diagnostic. Please refer to the GGD Training Guidelines for detailed information on the content, assessment and learning objectives of the required training units.
4. Individuals training in Clinical Cytogenetics are required to complete the following units (with suggested duration) and the accompanying assessment plan as outlined in Table 1:

    - General Genetics Concepts – Part 1 (12 weeks)
    - Prenatal Genetics (4 weeks)
    - Copy Number Variation (4 weeks)
    - Non-Mendelian Genetics (2 weeks)
    - Structural Variation and Advanced Chromosome Analysis (10 weeks)
    - Cancer Genetics – Part 1 (10 weeks)
    - Transition to Practice (4 weeks)
      TOTAL: 46 weeks

5. Individuals training in Molecular Genetics are required to complete the following units (with suggested duration) and the accompanying assessment plan as outlined in Table 2:

    - General Genetics Concepts – Part 2 (4 weeks)
    - Prenatal Genetics (4 weeks)
    - Copy Number Variation (4 weeks)
    - Single Nucleotide Variation and Residual Risk (8 weeks)
    - Next Generation Sequencing (10 weeks)
    - Non-Mendelian Genetics (4 weeks)
    - Cancer Genetics – Part 2 (8 weeks)
    - Transition to Practice (4 weeks)
      TOTAL: 46 weeks

6. The remaining 6 weeks of training are to be used for remedial training as needed, electives, special projects (e.g. test validation), conference attendance, etc.

7. Second-specialty trainees must demonstrate hands-on/technical experience in 100 cases and interpretative/consultative experience in 200 cases; both recorded in the appropriate section of the CCMG GGD program logbook.
8. The specific requirements outlined in Section II.1.a, b and c of the GGD Training Program Guidelines and Requirements must be met.
9. The margin between molecular and cytogenetic testing has become blurry. It is recognized that some of the requirements for one specialty involve methodologies from the second specialty and that trainees may already be familiar with their technical or interpretative aspects. Nonetheless, exemption to any part of the second specialty training program will not be granted.
10. The second specialty training program must take place in a CCMG-accredited centre. A trainee is allowed 25% of core training in a non CCMG-accredited centre for a total duration of up to 10 weeks.
11. A minimum of one ITER (specific to the unit) must be completed per training unit. The GGD training program ITERs must be used. For training units that overlap between both Clinical Cytogenetics and Molecular Genetics (Prenatal Genetics, Copy Number Variation, Non-Mendelian Genetics), the trainee will be assessed only on the applicable learning objectives.
12. The GGD training program Final In-Training Evaluation Report (FITER) must be completed at the end of the training. Please note that the FITER is additional to the ITER covering the last training unit(s).

Table 1 Clinical Cytogenetics Second Specialty Training – Summary of technical and consultative assessment <u>minimum</u> case requirements for required training unit.

Unit	Technical Assessment	No.	Interpretative/Consultative Assessment	No.
General Genetics Concepts – Part 1	Constitutional chromosomes wet lab – Blood set up, harvest, slide preparation and banding	5 cases	G-banded karyotypes/metaphases review and interpretation	30 cases
	Wet lab – Metaphase FISH set-up and washes	5 cases		
	Constitutional chromosomes dry lab – Complete analysis of cases, including: selecting appropriate metaphases at microscope or from scanning results, image capture, analyzing and karyotyping required number of cells, recording on worksheet, determine analysis result	5 cases		
	Constitutional chromosomes dry lab – Karyotyping or metaphase analysis only	10 cases	Metaphase FISH images review and interpretation	10 cases
	Dry lab – Metaphase FISH slide assessment	5 cases		
Prenatal Genetics	Wet lab – Prenatal specimen (amniocytes, CVS, fibroblasts) set up, maintenance, harvest, slide preparation and banding	3 cases	Prenatal G-banded karyotypes/metaphases review and interpretation	10 cases
	Dry lab – Prenatal specimens karyotyping only	5 cases		
Copy Number Variation	Wet lab - Dosage-based tests: processing of specimens for chromosomal microarray	5 cases	Dosage-based tests: data review and interpretation for chromosomal microarray	15 cases
	Dry lab – Data analysis for chromosomal microarray	10 cases		
Non-Mendelian Genetics	Set-up and analysis of testing for uniparental disomy	3 cases	Data review and interpretation for uniparental disomy	5 cases
Structural Variation and Advanced Chromosome Analysis	Dry lab – Karyotyping only	15 cases	G-banded karyotypes/metaphases review and interpretation	30 cases
Cancer Genetics – Part 1	Wet lab – Oncology specimen set-up, harvest, slide preparation and banding	3 cases	G-banded karyotypes/metaphases review and interpretation	20 cases
	Dry lab – Complete analysis of cases, including: selecting appropriate metaphases at microscope or from scanning results, image capture, analyzing and karyotyping required number of cells, recording on worksheet, determine analysis result	5 cases	Chimerism (post-transplant) testing review and interpretation	3 cases
	Dry lab – Karyotyping only	15 cases	Interphase FISH images review and interpretation	10 cases
	Dry lab – Interphase FISH (including FFPE tissue FISH if possible) slide assessment	5 cases		
Transition to Practice			Management of caseload and supervision of the laboratory	

Table 2 Molecular Genetics Second Specialty Training – Summary of technical and consultative assessment <u>minimum</u> case requirements for required training unit.

Unit	Technical Assessment	No.	Interpretative/Consultative Assessment	No.
General Genetics Concepts – Part 2	Nucleic acids extraction from any of 3 different sources (blood, blood spots, buccal, tissue culture, direct amniotic fluid, CVS, saliva or tissue)	5 cases	Repeat expansion disorders - Result review and interpretation	10 cases
			Identity or relationship testing – Result review and interpretation	2 cases
	Identity or relationship testing	2 cases	Linkage analysis - Review of current, archival or exemplary cases	2 cases
	Repeat-primed PCR for expansion repeat disorders	5 cases		
Prenatal Genetics	Wet lab - Rapid Aneuploidy detection assay [(FISH or QF-PCR) and/or NIPS]	3 cases	Rapid Aneuploidy detection assay review and interpretation	10 cases
	Wet lab – Prenatal specimen (amniocytes, CVS, fibroblasts) set up, maintenance, harvest, nucleic acid extraction	3 cases		
Copy Number Variation	Wet lab - Dosage-based tests: qPCR, MLPA, etc. (must include both methods)	10 cases	Dosage-based tests: data review and interpretation for qPCR and MLPA	20 cases
	Dry lab – Data analysis for dosage-based tests (must include minimum of 10 cases for each of qPCR and MLPA)	20 cases		
Single Nucleotide Variation and Residual Risk	PCR-based assay for SNV detection such as qPCR, allele-specific PCR, mass array, bead array, restriction enzyme digest (must include minimum of 2 different methods)	5 cases	Single Nucleotide variant data review and interpretation	10 cases
			Residual Risk Calculation	5 cases
	Sanger sequencing for SNV detection/confirmation	3 cases		
Next Generation Sequencing	Observation of library preparation and NGS panel set-up	4 runs	NGS data review and variant interpretation	25 cases
	Observation of WES/WGS library preparation and set-up	1 run	WES/WGS data review and variant interpretation	5 cases
Non-Mendelian Genetics	Set-up and analysis of testing for imprinting disorders, uniparental disomy, mitochondrial genome disorders (must include minimum of 2 areas)	10 cases	Data review and interpretation for imprinting disorder, uniparental disomy, mitochondrial genome disorder, follow-up from suspected germline or somatic mosaicism of a Mendelian disorder (minimum 2, maximum 10 cases of each type)	20 cases
Cancer Genetics – Part 2	Nucleic acid extraction from FFPE tissue slide or curls	3 cases	Data review and interpretation of hereditary cancer gene(s) or gene panel – one case reported in the context of microsatellite instability and/or immunohistochemistry	10 cases
	PCR-based methods for cancer prognosis and follow-up (RT-PCR, RQ-PCR, clonality, …)	5 cases		
	Observation of library preparation and NGS panel set-up for somatic variant detection	2 runs	Data review and interpretation of familial or founder variant in a hereditary cancer gene	20 cases

			Data review and interpretation of PCR-based tests for cancer prognosis and follow-up	5 cases
			Data review and interpretation of actionable somatic variant from at least two cancer types	20 cases
Transition to Practice			Management of caseload and supervision of the laboratory	