Molecular Genetic Pathology CC Exam Module Study Guide

Registrants for the MGP only or combined Primary/MGP CC exam must take the mandatory general MGP module plus any elective HEM modules selected during registration.

CC MGP – General Molecular Genetics I (Mandatory 75-Question Module)	
• assay validation; sensitivity;	• next generation sequencing
• CAP lab accreditation; phase II deficiency	• nucleic acid hybridization reactions
• CFTR phenotype	• oligodendroglioma, FISH
• CLIA proficiency testing; alternatives	• oligodendroglioma; chemotherapy response
• CML; BCR/ABL1	• oncogenes; KRAS activation
• coding sequences; percent of genome	• paternity testing
• CPT coding; mutation panel	• PCR assay performance and validation
• DNA melting temperature	• PCR; annealing temperature
• electropherograms showing mutations	• PCR; exonic vs intronic primers
• extraction controls	• PCR; preanalytical errors
• FISH interpretation	• PCR; viral CNS infections
• FISH validation	• pedigree; Huntington disease
• fragile X	• pharmacogenetics; warfarin response
• frameshift variant	• positive predictive value
• Friedreich ataxia; risk	• Prader-Willi
• gene nomenclature	• primer preparation; calculations
• genetic imprinting	• procedure manuals standards
• gestational trophoblastic diseases; ploidy	• QA; calibration curve; degrading primer
• Human Genome Organization nomenclature	• QC; quantitative RT-PCR
• ICD coding	• quality control; causes of false negative results
• introns; dinucleotides	• quality control; restriction enzyme digestion
• loss of heterozygosity	• quantitative PCR and RT-PCR
• mantle cell lymphoma	• ROC curves
• maternal cell contamination studies	• RT-PCR for t(11;22)
• meiosis; nondisjunction	• RT-PCR; dynamic range
• methylation; epigenetics	• single nucleotide polymorphisms
• microRNA	• single stranded nucleic acids composition
• mitochondrial disorders; neuromuscular	• standard nomenclature, cDNA
• mRNA sequence change; mutation effect	• stop codons
• multiplex ligation dependent probe amplification	• TERT gene
• multiplex PCR vs Southern blot	• tissue identity testing
• mutation effect; amino acid sequence	• Tm; double stranded DNA
• myotonic dystrophy	• viral load precision
• neuroblastoma; microarray	• whole genome sequencing; inherited variants

CC MGP – General Molecular Genetics II (Elective 75-Question Module)	
• APL; retinoic acid resistance	• HPV types; head and neck cancers
• assay validation; precision	• HPV; cervical cancer risk
• BCR/ABL	• HSV PCR on CSF; indications
• bone marrow engraftment analysis	• ion channel defects
• comparative genomic hybridization interpretation	• lab safety; liquid nitrogen
• control of gene expression; alternative splicing	• laboratory specimens for research; IRB
• CSF PCR assays for viral infections	• LOH; false positive results
• DHCR7 gene analysis	• method comparisons; precision
• disease prevalence calculations	• method validation
• Duchenne muscular dystrophy	• mismatch repair enzymes
• effect of gene point mutations	• mitochondrial DNA polymorphism analysis; indications
• EGFR mutations in lung cancer	• MLH1, HNPCC
• engraftment testing; samples	• mRNA sequence change effect
• epigenetic silencing	• nonsense mediated decay
• ethics of notifying estranged family member of new dx	• nucleotide vs nucleoside
• etiologic diagnosis of intellectual disability; array CGH	• odds ratio calculations
• familial hypertrophic cardiomyopathies	• parentage index calculations
• fragile X; inheritance	• paternity testing; interpretation of results
• fragile X; PCR interpretation	• PCR amplification interference; specimen collection
• gene mutation hot spots	• PCR; exonic vs. intronic primers
• gene sequence, human telomere	• PCR; missed mutations
• Genetic Information Nondiscrimination Act of 2008	• pharmacogenetics; codeine metabolism
• gliomas; resistance to tyrosine kinase inhibitors	• progressive multifocal leukoencephalopathy
• Hardy Weinberg	• protective mutations
• HCV types; response to therapy	• quality; preanalytical errors
• heavy chain gene rearrangements; clonality	• risk calculations
• hemochromatosis; genetic testing of children	• STR profiles; forensics
• HER2 - cell membrane tyrosine kinase	• STR; molar pregnancies
• hereditary thrombophilia testing	• succinate dehydrogenase mutations in tumors
• histone acetylation	• TaqMan
• HIV assays; interpretation	• Tm; double stranded DNA
• HIV viral load	• tyrosine kinase inhibitors; GIST
• HLA alleles	• validation; RT-PCR
• HLA matching for transplantation	• Wilson Disease; laboratory findings
• HPV molecular testing; HSIL	• X inactivation

CC MGP – Genetics I (Elective 25-Question Module)	
• achondroplasia	• locus heterogeneity
• allele frequency calculation	• maternal contamination of villous sample
• Angelman syndrome genes	• parentage index
• Belmont report; 3 ethical principles	• pedigree; autosomal dominant inheritance
• CHARGE syndrome	• pedigree; autosomal recessive inheritance;
• cleft palate; mode of inheritance	• pedigree; dominant inheritance
• congenital long QT syndrome mutations	• pedigree; incomplete penetrance
• cystic fibrosis reflex testing; 8 poly-T polymorphism	• pedigree; modifier genes
• cystic fibrosis; parent unexpected test results	• pedigree; patterns of inheritance
• deletion analysis, MLPA	• pedigree; risk of disease
• hemophilia A	• Tay-Sachs incidence
• Huntington disease	• transplant donor identification; HLA testing
• identity testing; twins	• Trisomy 18 by chromosomal microarray

CC MGP – Genetics II (Elective 25-Question Module)	
• apolipoprotein E genotypes and genotyping	• loss of heterozygosity; principles
• CFTR mutations and CF treatment	• mitochondrial disorder testing; specimen types
• CLIA; performance characteristic of a new genetic test	• NGS; results interpretation
• Gaucher disease	• oculopharyngeal muscular distrophy
• genetic exceptionalism	• paternity index calculations
• HFE gene; hemochromatosis	• pedigrees; patterns of inheritance
• HLA nomenclature	• pre-symptomatic genetic testing of minors; ethical considerations
• HLA; allele coding sequence	• pseudogenes and mutant alleles
• HLA; parental and sibling haplotypes	• sibling HLA match; transplantation
• Huntington disease	• trinucleotide repeat disorders
• identity testing	• variant assessment; Integrative Genomis Viewer
• linkage analysis	• zygosity

CC MGP – Infectious Diseases I (Elective 25-Question Module)	
• antibiotic resistance genes	• pertussis; diagnostic molecular tests
• cat scratch disease	• positive predictive value calculations
• CSF viral testing	• post transplant CMV PCR
• CSF; multiplex test for viruses	• prevention of nosocomial MRSA
• enterovirus meningitis	• QC; sample contamination
• hepatitis C	• renal allograft recipients; viral testing
• HIV viral load informing patient management	• respiratory syncytial virus; diagnosis and molecular detection
• HIV; acute phase viral load	• respiratory viruses; amplicons; mortality risk
• HIV; RT-PCR	• RT PCR, norovirus
• HIV-1 resistance testing	• RT-PCR, validation study
• mycobacteria; isoniazid resistance; gene mutations	• single stranded RNA viruses
• mycobacteria; NAAT interpretation; CDC guidelines	• WHO international unit for HCV viral load reporting
• penicillin resistant Strep pneumo	

CC MGP – Infectious Diseases II (Elective 25-Question Module)	
• assay measurement range determination	• malaria; molecular identification
• BK virus testing	• molecular respiratory virus testing panels
• CNS infections; sensitivity of molecular tests	• molecular testing interpretation; latent infections
• cryptococcus	• multiplex endpoint PCR interpretation
• false positive HIV tests	• positive predictive value
• Hepatitis B virus mutations	• pre-analytical specimen issues
• HIV-1 mutations	• qualitative real-time PCR testing; interpretation
• HPV testing	• quality control charts; interpretation
• HPV types; cervical cancer	• real-time PCR interpretation
• human herpesvirus 6	• stool specimens for molecular testing
• laboratory acquired infections	

CC MGP – Oncology/Hematology I (Elective 25-Question Module)	
• ankyrin mutations	• hairy cell leukemia
• APL	• Imatinib resistance
• B-lymphocyte development	• immunoglobulin gene rearrangement sequence
• capillary electrophoresis; mantle cell lymphoma	• leukemia, KIT
• cDNA sequence; ABL kinase; CML	• major molecular response; CML; qRT-PCR
• clonal T-cell gene rearrangements; PCR; capillary gel electrophoresis	• MALT lymphomas; translocation
• CML; cytogenetic evolution	• mantle cell lymphoma; FISH; immunophenotype
• FISH interpretation	• myelodysplastic syndromes; CGH array interpretation
• FISH, TEL/AML1	• PCR; BM post-transplant; engraftment
• FLT3 fragment analysis; interpretation	• RT- PCR; minimal residual disease; CML
• FR1/FR3 IGH variable region genes	• translocations, lymphoid neoplasms
• genetic abnormalities in myeloproliferative disorders	

CC MGP – Oncology/Hematology II (Elective 25-Question Module)	
• acute lymphoblastic leukemia	• follicular lymphomas
• acute promyelocyte leukemia	• germline organization of heavy chain locus
• ALK	• hemochromatosis; genotypes
• AML with abnormal marrow eosinophils	• hyperplasia vs lymphoma
• AML; SNP array interpretation	• IGH rearrangements
• anaplastic large cell lymphoma	• PCR electropherogram interpretation
• B lymphoblastic leukemis/lymphoma	• PCR; mutations in acute myeloid leukemia
• chronic lymphocytic leukemia	• pre-analytical errors
• chronic myelogenous leukemia	• T-cell large granular lymphocytic leukemia
• embryonal tumor; FISH; prognosis	• TCR gamma; subtypes
• ETV6/RUNX1	• types of mutations
• FISH interpretation	

CC MGP – Oncology/Solid Tumors I (Elective 25-Question Module)	
• BRCA 1 and 2; breast cancer risk	• inherited vs. somatic mutation
• BRCA2 familial cancer syndrome; pedigree	• lung cancer; EGFR mutations; response to therapy
• CNS tumors; prognostic genetic alterations	• malignant rhabdoid tumor
• colon neoplasia; sequence of genetic lesions	• microsatellite instability
• colorectal cancer; MLH-1 IHC	• microsatellite instability
• engraftment analysis; PCR	• neuroblastoma; microarray analysis; FISH
• familial polyposis; genetics	• oligodendroglioma; chemotherapy response
• genes frequently mutated in colon cancer	• retinoblastoma
• gene-syndrome associations	• STR genotype, colon cancer
• germ cell tumors; iso12p	• synovial sarcoma; translocation
• GIST; mutations	• T-cell clonality; FFPE tissue
• glioma; FISH; loss of heterozygosity	• thyroid cancer; molecular testing
• gliomas; IDH1 mutations	

CC MGP – Oncology/Solid Tumors II (Elective 25-Question Module)	
• adenomatous polyposis coli	• lung cancer; molecular testing; mutations
• Birt-Hogg-Dube syndrome	• melt curve analysis; interpretation
• BRAF mutations	• most common somatic mutations
• ClinVar	• non-cutaneous melanomas
• CNS tumors	• oligodendroglioma
• EGFR mutation testing; indications	• reverse transcription PCR; interpretation
• electropherograms; interpretation	• rhabdoid tumor
• EWS-TW1	• ROC curves
• glioblastoma	• TERT
• inherited oncogenes	• Wilms tumor