In-Training Evaluation Report – Genetic and Genomic Diagnostic Specialty

NAME: Last Name _____ First Name _____

Date Training Started: _____ Full Time ☐ Part time ☐

Training Stage: **Core** Unit: **Copy Number Variation Analysis**

Unit Start Date: _____ Unit End Date: _____

Training Site: _____ Supervisor: _____

Learning objectives associated with this unit:	Below expectations	Meets expectations	Exceeds expectations
ME 1.3 Understand the mechanisms whereby recurrent structural abnormalities arise (e.g. aberrant pairing, recombination at repetitive elements, etc.)			
ME 2.2 Understand and evaluate common referral reasons for CMA including common genetic deletion and duplication syndromes, referrals in the postnatal versus prenatal settings, etc.			
ME 2.2 Demonstrate the ability to interpret CMA data consistent with aneuploidy and common genetic deletion and duplication syndromes, and recognize and interpret material of unknown origin identified by chromosome analysis			
ME 2.2 Demonstrate the ability to design appropriate primers for qPCR and interpret data consistent with deletions and duplications			
ME 2.2 Demonstrate the ability to interpret MLPA data, including requests for appropriate confirmatory testing, and describe its use for confirmation of CNVs detected by Next-Generation Sequencing			
ME 2.2 Demonstrate knowledge and use of literature and guidelines relevant to CMA, MLPA and qPCR			
ME 3.1 Apply knowledge of the different CMA platforms used in clinical laboratories and describe their limitations and benefits in the context of clinical testing			
ME 3.2 Describe the best approach for the follow-up of CMA findings in relation to the clinical scenario			
ME 3.2 Describe the benefits and limitations of karyotype and CMA in both the pre- and post-natal contexts			
ME 3.2 Describe different types of qPCR and MLPA, their advantages, limitations and clinical utility			
ME 3.4 Apply knowledge of the steps involved and methodological basis of sample preparation, hybridization, washes and data analysis for different types of CMA used for clinical testing			
ME 3.4 Describe the methodological basis of qPCR including the use of reference genes and control samples in the context of clinical testing			

ME 3.4 Describe the methodological basis of MLPA and methylation-sensitive MLPA in the context of clinical testing			
COM 2.3 Use correct ISCN and Human Genome Variation Society (HGVS) nomenclature to describe CNVs			
L 1.1 Apply knowledge of validation/verification approaches for the establishment of CMA, qPCR and MLPA assays and their respective analysis software if applicable (e.g. original installation versus versioning upgrade)			
S 3.3 Apply knowledge and understand the inherent biases of the bioinformatics tools for the interpretation of CNVs, including control population and patient databases (e.g. DGV, DECIPHER, ClinVar/ClinGen, OMIM, Genome Browsers)			

Longitudinal Competencies:	Never	Rarely	Sometimes	Usually	Always
ME 1.3 Apply knowledge of the main clinical features of genetic disorders in the context of choice of testing procedure, result interpretation and report writing					
ME 1.6 Demonstrate insight into limits of expertise and seek consultation as necessary					
ME 2.1 Prioritize specimens and testing based on clinical indication and impact on medical management					
ME 2.2 Select ancillary tests in a resource-effective and ethical manner that balances costs with potential utility of results					
COM 4.1 Prepare clear, concise, comprehensive, and timely written reports for genetic tests that incorporate personal and family history and results from other relevant testing in answering the clinical question					
COL 1.2 Discuss trouble-shooting issues with colleagues in the genetic laboratory including laboratory members					
COL 1.2 Work effectively with laboratory technologists and laboratory assistants, directing their assistance as appropriate					
COL 2.1 Respond to requests and feedback in a respectful and timely manner					
L 1.1 Actively participates in quality control, quality assurance, and quality improvement initiatives					
L 3.1 Review quality control data, and take appropriate action for deficiency follow-up, including possible sample mix-up					
HA 1.3 Understand the clinical implications of incidental findings, approaches to minimize the chance of finding them, and policies for reporting					
S 1.2 Identify opportunities for learning and improvement by regularly reflecting on and assessing personal performance					
S 2.4 Participate in available learning activities					
P 1.2 Demonstrate a commitment to excellence in all aspects of laboratory practice					
P 3.1 Adhere to the relevant codes, policies, standards, and laws governing laboratory practice including accreditation, standard operating procedures, training and competency, safety, and privacy					

Technical and Interpretative requirements have been completed for this unit Yes ☐ No ☐
If no, justify in the section below.

Summarize the trainee's performance for this unit and formulate recommendations for future improvement

Name/Signature of evaluator(s) _____

Date _____ _____

Name/Signature of Program Director _____

This is to attest that I have read this document Signature of Trainee _____