



## 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: **Structural Variation and Advanced Chromosome Analysis**

Unit Start Date: \_\_\_\_\_ Unit End Date: \_\_\_\_\_

Training Site: \_\_\_\_\_ Supervisor: \_\_\_\_\_

Learning objectives associated with this unit:	Below expectations	Meets expectations	Exceeds expectations
<b>ME 1.3</b> Apply knowledge of the clinical implications and reproductive and recurrence risks for a carrier of a structural chromosome anomaly			
<b>ME 1.3</b> Apply knowledge of the implications of chromosome mosaicism in performing a thorough investigation and reporting appropriately			
<b>ME 1.3</b> Apply knowledge of recurrent microdeletion and microduplication syndromes, their underlying genomic architecture and the mechanism contributing to their recurrence in the context of report writing and recommendation for parental studies			
<b>ME 1.3</b> Apply knowledge of the origins and clinical effects of X and Y chromosome aneuploidy and structural anomalies, including the effect of X-inactivation			
<b>ME 1.3</b> Apply knowledge of the genetic mechanisms of DNA repair to the disorders resulting from their defects as well as the principles of chromothripsis and chromoanasythesis in the context of clinical cytogenetics			
<b>ME 1.4</b> Recognize structurally abnormal chromosomes in metaphases, karyotypes and inferences from chromosomal microarray results with appropriate follow-up test			
<b>ME 1.4</b> Recognize well characterized cytogenetically visible unbalanced structural anomalies and describe their associated clinical features			
<b>ME 2.2</b> Demonstrate the ability to distinguish between recombinant and derivative chromosomes and understand their different clinical significance			
<b>ME 2.2</b> Demonstrate the ability to use chromosomal microarray SNP genotype (allele difference or B allele frequencies) to identify regions of homozygosity and describe their clinical significance (e.g. UPD, parental relationship), and to interpret complex rearrangements, mosaicism, chimerism, sample contamination, etc.			
<b>ME 3.1</b> Describe the diagnostic testing methods for chromosome breakage syndromes, the associated recurrent chromosome findings, and their major clinical features			
<b>ME 4.1</b> Coordinate the use of multiple diagnostic investigations to define a chromosome abnormality or heteromorphism, including appropriate use of			

different staining or molecular cytogenetic methods so as to ensure complementarity and efficiency			
<b>COM 2.3</b> Apply proper use of the most recent ISCN to describe a chromosome anomaly			

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