## 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: **Single Nucleotide Variation and Residual Risk Calculation**

Unit Start Date: _____ Unit End Date: _____

Training Site: _____ Supervisor: _____

| **Learning objectives associated with this unit:** | Below expectations | Meets expectations | Exceeds expectations |
|---|---|---|---|
| **ME 1.3** Apply knowledge of the pathophysiology of inherited genetic disorders and the concept of human variation in the context of genetic diseases with recurrent variants | | | |
| **ME 1.3** Apply knowledge of the concept of founder variants in the context of variant interpretation | | | |
| **ME 1.3** Apply knowledge of residual risk to germline or constitutional diagnosis, and of recurrence risk | | | |
| **ME 1.3** Understand the statistical concepts of positive and negative predictive values, detection rate, specificity, sensitivity in the context of newborn and population-based screening and differentiate between DNA-based screening and diagnostic assays | | | |
| **ME 1.4** Apply knowledge of various methods for the detection of sequence variation (including qPCR, allele-specific PCR, mass array, bead array, Sanger Sequencing, restriction enzyme digestion) and assess their benefits-limitations/ advantages-disadvantages | | | |
| **ME 3.2** Understand the methodological basis of former single nucleotide variant scanning techniques (e.g. high resolution melting, pyrosequencing) and describe their advantages and limitations | | | |
| **ME 3.2** Perform Bayesian analysis and residual risk calculation | | | |
| **ME 3.4** Analyse Sanger sequencing traces to identify variants, recognize sequencing artifacts, flag unusual results and interpret complex sequences | | | |
| **ME 3.4** Select appropriate reference sequences for sequence analysis | | | |
| **ME 5.1** Link specific, inherited sequence variation with a patient's response to a pharmacological agent | | | |
| **COM 2.3** Assign the correct HGVS nomenclature | | | |
| **COM 2.3** Interpret and classify variants according to current guidelines in use [e.g. CCMG guidelines, ACMGG guidelines, disease-specific guidelines (ClinGen, etc.)] | | | |

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|---|---|---|---|
| **S 3.2** Utilize disease-specific databases, general population datasets and in silico algorithms to support variant interpretation | | | |

| **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 _____