



Logbook Guidelines for Diplomates in the LGG Alternative Certification Pathway – Adding Molecular 2021 Laboratory Genetics and Genomics Examination

Purpose:

The purpose of the logbook is to document that the applicant has had direct and meaningful involvement in the processing, analysis, interpretation, and reporting of molecular genetics and genomics laboratory tests and has received ongoing and appropriate laboratory supervision. The logbook cases must provide evidence of the required clinical laboratory bench experience and evidence of well-rounded experience with a wide variety of molecular genetic techniques, involving all molecular genetic laboratory testing categories. Cases included in the logbook should demonstrate a broad spectrum of molecular genetic diagnoses including, but not limited to, postnatal and prenatal diagnostic single gene or panel testing, presymptomatic testing, carrier screening, and hematologic malignancies and solid tumors.

Requirements:

Logbooks must be completed in accordance with the instructions provided in this document with cases compiled using the ABMGG Logbook Excel Spreadsheet Tool. Completed logbooks must be submitted to the ABMGG for review and approval. While the ABMGG anticipates ongoing review of cases between the trainee and laboratory director supervising training, the applicant should assure that all requirements have been fulfilled before submitting the final logbook for review.

Case Selection:

1. All specimens must have been processed in a laboratory that is part of an ABMGG or CCMG-accredited training program in clinical molecular genetics and genomics OR in a laboratory that is supervised by a current ABMGG-certified clinical molecular geneticist.
2. None of the 150 cases may be collected until a diplomate's application for the alternative certification pathway has been approved by the ABMGG.
3. At least 75 cases must be obtained through on-site training and no more than 25 cases may be obtained in a single week.
4. Each logbook entry must document the applicant's role(s) in the testing and reporting process, including sample processing, analysis, results interpretation, and/or communication of the test results.
5. Only cases for clinical diagnosis or confirmatory analysis may be included in the logbook. Experimental or control cases, historical material, proficiency testing, or cases that are part of laboratory quality assurance activities will not be accepted. In laboratories where state regulations do not permit unlicensed individuals to generate a clinical laboratory result,

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parallel testing of clinical samples between a licensed technologist and trainee may serve to fulfill this requirement.

6. A given patient or family may appear only once in an individual's logbook, regardless of the number of specimens processed on the patient or family.

Description of Logbook Headings/Columns:

- **Entry Number**: The logbook spreadsheet allows a trainee to enter an unlimited number of cases while in training. For the final logbook, you must select 150 cases to submit that fulfill all of the defined requirements. The applicant must be able to identify each case by its entry number if questions arise about a logbook entry. Patient names and bona fide hospital or clinic numbers may not be included anywhere in the logbook that may be submitted to the ABMGG. Logbooks containing specific information regarding the identity of any patient will not be reviewed.
- **Date**: The date in month/day/year [MM/DD/YYYY] format identifies when the sample was received in the laboratory or, if relevant, the date the patient was evaluated clinically.
- **Primary Laboratory Testing Category**: For each case, use the numbers 1 through 6 as outlined below to identify the single category that best describes the indication for the clinical molecular genetic test. Observe category limits as specified below.

Category 1 **Diagnostic evaluation, postnatal**: testing performed postnatally to confirm or exclude a suspected clinical diagnosis (e.g., fragile X testing on a male with developmental delay; hereditary hemochromatosis testing on an individual with elevated iron levels).

Category 2 **Carrier testing**: No more than 35 cases may be obtained in this category. Testing is performed to identify asymptomatic male or female carriers of autosomal recessive disorders (e.g., cystic fibrosis, Tay-Sachs disease) or female carriers of X-linked disorders (e.g., hemophilia).

Category 3 **Prenatal diagnosis**: A minimum of 5 cases must be obtained in this category. Testing is performed on fetal specimens (e.g., amniocytes, chorionic villus biopsy, percutaneous umbilical cord blood [PUBS]). Communication of results obtained by an outside laboratory may be included in this category

Category 4 **Presymptomatic testing**: A minimum of 10 cases must be obtained in this category. Testing is performed on asymptomatic individuals for the purpose of identifying patients at risk for developing later-onset hereditary conditions. This type of testing is usually performed on individuals who have a family member with a genetic disease but who have no features of the condition at the time of testing (e.g., Huntington disease, autosomal dominant polycystic kidney disease, factor V (Leiden), hereditary hemochromatosis, BRCA1/2).

Category 5 **Identity testing**: No more than 35 cases may be obtained in this category and no more than 5 cases may involve paternity testing. It is *recommended that* a minimum of 5 cases be obtained in this category. Identity testing involves the analysis of polymorphic genetic markers but is not used to detect gene mutations associated

with disease (e.g., paternity testing, forensics, zygosity, transplantation, maternal cell contamination).

Category 6 **Pharmacogenetic testing:** It is *recommended* that no more than 20 cases be obtained in this category. Pharmacogenetic testing involves the analysis of gene variants that give rise to variable drug metabolism or response.

- **Laboratory Testing Methodology:** Specify the laboratory testing methodology performed for each case by entering the Methodology number and associated letter (if any) outlined below. Observe limits per method where specified. Note that multiple testing methods can be specified for each case.

1. **Mutation analysis;** at least five cases must be obtained for each of at least four of the different mutation analysis methods (1a-1k) listed below:

- a. PCR fragment size analysis
- b. Restriction fragment length analysis
- c. Forward and reverse ASO
- d. Quantitative PCR
- e. Methylation PCR or methylation-specific MLPA
- f. Triplet repeat analysis
- g. Southern blot
- h. RNA analysis
- i. Genotyping microarray analysis
- j. Exon-focused array CGH
- k. Other (you must specify)

2. **Gene specific mutation scanning:** It is *recommended* that a minimum of 10 cases be obtained in this laboratory method.

- a. High resolution melt analysis
- b. Other (you must specify)

3. **Sequence Analysis** At least 40 cases must be obtained in this laboratory method, with a minimum of 10 cases using NGS and 10 cases using Sanger sequencing.

- a. Sanger sequencing
- b. Pyrosequencing
- c. Methylation sequencing
- d. Next Generation sequencing (NGS)
 - i. Next Generation sequencing panel (PCR or capture based)
 - ii. Next Generation whole exome sequencing
 - iii. Next Generation whole genome sequencing

- **Test Performed and Results:** The logbook should demonstrate experience with a variety of molecular genetic abnormalities (e.g., classical single gene disorders, mitochondrial disorders, imprinting disorders, common risk genes, malignancy, identity testing). Infectious disease testing may not be included. FISH may not be included. No more than 20 cases with the same test, gene, and/or diagnosis may be included. A maximum of 100 cases may have normal

laboratory findings; the results of identity testing cases must be counted as “normal.” Sequence changes interpreted as variants of uncertain clinical significance should be counted as abnormal.

The gene symbol (HUGO gene nomenclature) must be listed first, followed by the name of the genetic condition or test, and then the result as shown in the examples below. Abbreviations for the name of the disorder are not acceptable. Current HGVS nomenclature should be used for describing variants. If needed for clarification, the common name can also be listed in parenthesis. The check box should only be marked if the result is abnormal.

Examples: CFTR, cystic fibrosis, negative for mutations analyzed
CFTR, cystic fibrosis, p.Phe508del (delta F508) heterozygote
HTT, Huntington disease, 0 and 46 CAG repeats
BCR/ABL1, chronic myelogenous leukemia, positive
F5, hereditary thrombophilia, c.1601G>A (p.Arg534Gln) homozygote
DMD, Duchenne muscular dystrophy, deletion of exons 45-50
Array analysis, intellectual disability, negative.
arr(1-22,X)x2 for normal females
arr(1-22)x2,(XY)x1 for normal males
arr 6q22q24(113,900,000-149,100,000)x1 for a loss at 6q22 to 6q24
NGS panel, hearing loss, 70 genes, negative
NGS panel, developmental delay 60 genes, UBE3A heterozygous
c.2475_2478delACTT

For tests which include a large panel of genes, the test name and result can be listed in the logbook. All deleterious mutations and variants of uncertain clinical significance detected must be listed in the logbook. Sequence changes interpreted as benign variants do not need to be listed in the logbook.

- **Trainee’s Roles:** Check all of the boxes that indicate your role(s) in the testing, interpretation and reporting process. A breadth of experience must be reflected in the logbook. A minimum of three roles must be specified for at least 140 cases. No more than 10 cases can be tested in an outside laboratory. At least 100 cases must involve roles 1 or 2. Observe specific limits per role where specified.
 1. **Sample processing;** it is *required* that at least 50 cases involve this role, which is comprised of the following:
 - a. DNA extraction (performing manual extraction or observing automated extraction)
 - b. RNA extraction; it is *recommended* that at least 5 cases involve this sub-role.
 2. **Sample analysis;** it is *required* that at least 50 cases involve this role. The trainee must have directly performed the laboratory testing methodology.
 3. **Interpretation of laboratory results;** it is *required* that at least 25 cases involve this role. The trainee must have reviewed laboratory data and determined whether the results were normal, abnormal, etc.
 4. **Written report of results;** it is *required* that at least 50 cases involve this role, which is comprised of the following:

- a. The trainee must have generated a laboratory report using standard interpretive paragraphs; it is *required* that at least 10 cases involve this sub-role.
 - b. The trainee must have generated a novel report due to unusual laboratory results or circumstances; it is *required* that at least 10 cases involve this sub-role.
 - c. The trainee must have reviewed a report generated by another laboratory staff member.
5. **Oral communication of results to health care providers who requested the testing or their designated contact:** at least 10 cases are *recommended* and at least half of these cases must involve abnormal results.
- **Supervisor:** Include the full name, degree(s), and type of certification of the supervisor who was present and was directly responsible for your activities regarding that case.