



## **Certification in Laboratory Genetics and Genomics**

### **Logbook Requirements for**

### ***2027 Examination***

#### **Purpose:**

The purpose of the logbook is to document that the applicant has had direct and meaningful involvement in the processing of cytogenetic and molecular specimens, analysis of data, interpretation of test results, communication of test results, and has received ongoing and appropriate laboratory supervision. The logbook cases submitted must provide evidence of clinical laboratory bench experience and evidence of well-rounded experience with a wide variety of techniques drawn from all testing categories. Logbook entries should include the broad spectrum of genetic diagnoses.

#### **Requirements:**

A 200 case logbook must be completed in accordance with the instructions provided in this document with cases compiled using the ABMGG Logbook Excel Spreadsheet Tool. While the ABMGG expects ongoing review of cases by the program director (PD), the applicant should ensure that all requirements have been fulfilled before submitting the final logbook to their PD for review. The PD must attest to the ABMGG that all logbook requirements for this specialty have been fulfilled and are clearly reflected in the logbook. The ABMGG reserves the right to audit a logbook to confirm that all requirements have been fulfilled. In this event, an applicant will be notified that they have been selected for audit and must submit the logbook as completed up to the date of the audit, using the ABMGG Logbook Excel Spreadsheet.

#### **Case Selection:**

1. All specimens must have been processed in an ACGME-accredited training program in the specialty of laboratory genetics and genomics (LGG).
2. Supervision for cases must be provided by faculty who are certified by the ABMGG, ABGC, CCMG, or ABP-MGP. For cases obtained during outside laboratory rotations, e.g., state newborn screening laboratories, it is *recommended* that supervisors be certified by their appropriate certifying board(s).
3. **All 200 cases** must be obtained during the inclusive dates of the applicant's LGG training.
4. Logbook entries must reflect at least two (2) years of clinical laboratory experience. No more than 30 cases may be obtained during any 30-day period.

5. Each logbook entry must document the applicant's role(s) in the sample processing, data analysis, results interpretation, and/or communication of results.
6. Only cases evaluated for clinical diagnoses or confirmatory analyses may be included in the logbook. Experimental or control cases, historical or archival material, proficiency testing, or cases that are part of laboratory quality assurance activities are unacceptable as logbook cases. In laboratories for which regulations do not permit unlicensed individuals to generate a clinical laboratory result, parallel testing of clinical samples between a licensed technologist and trainee may serve to fulfill this requirement.
7. A given patient or family may appear only once in an applicant's logbook, regardless of the number of specimens processed from the patient or family.

### **Description of Logbook Headings/Columns:**

- **Entry Number:** The logbook spreadsheet allows a trainee to enter an unlimited number of cases. For the final logbook, 200 cases should be selected that fulfill all 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, laboratory, or clinic numbers may not be included. Final logbook submission should be without PI.
- **Date:** The date month/day/year [MM/DD/YYYY] format identifies the date of receipt 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 7 outlined below to identify the category that best describes the indication for the clinical test. Observe category limits, as specified below.

Category 1 **Prenatal studies:** (e.g., amniotic fluid, chorionic villi, percutaneous umbilical blood or product of conception). A minimum of 20 cases must be obtained in this category. It is *recommended* that results from at least three, but no more than 10, prenatal cases are diagnostic follow-up testing due to abnormal cell-free DNA (cfDNA) screening. These reviews should include at least one positive case confirmed by karyotype, FISH, or chromosomal microarray and one case for which the trainee participates in counseling or observes the communication of results to the patient. cfDNA results should be recorded in the results field but specific nomenclature is not required.

Category 2 **Diagnostic testing:** (postnatal, non-oncology). At least 40 cases must be obtained in this category. Testing should be performed to confirm or exclude a suspected clinical diagnosis. Cases can be obtained using either cytogenetic or molecular methodologies.

Category 3 **Carrier testing:** At least 10 cases must be obtained in this category. Testing should be performed to identify asymptomatic carriers of autosomal recessive disorders (e.g., cystic fibrosis, Tay-Sachs disease), female carriers of X-linked disorders (e.g., hemophilia), or carriers of cytogenetic abnormalities.

Category 4 **Presymptomatic testing:** At least 10 cases must be obtained in this category. Testing should be 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, or BRCA1/2).

Category 5 **Pharmacogenetic testing:** No cases are required. It is recommended that 5 cases be obtained, but no more than 20 cases may be obtained in this category. Pharmacogenetic testing involves the analysis of gene variants that influence drug metabolism or response.

Category 6 **Identity testing:** No cases are required. It is recommended that 5 cases be obtained, but no more than 10 cases may be obtained in this category. Identity testing involves the analysis of polymorphic genetic markers but is not used to detect gene pathogenic variants associated with disease (e.g., paternity testing, forensics, zygosity, transplantation, or maternal cell contamination studies).

Category 7 **Oncology testing:** (e.g., bone marrow, leukemic blood, lymph node, or solid tumor). At least 60 cases must be obtained in this category. At least 30 of these cases must demonstrate use of cytogenetic methodologies (karyotype, FISH or chromosomal microarray) and 30 cases must demonstrate use of molecular methodologies (e.g., Sanger sequencing, Next Generation Sequencing [NGS]).

▪ **Laboratory Testing Methodology:** Specify the laboratory test/methodology performed for each case by entering the Methodology number and associated letter (if any) outlined below. Trainees must participate in a broad range of laboratory testing methodologies. Observe case limits as specified.

1. **G-banding:** At least 50 cases must include G-banding but no more than 20 cases may involve G-banding alone. - The remaining cases should document a complementary method.

2. **FISH:** At least 30 cases must include FISH, but no more than 10 cases may involve FISH alone. The remaining should document a complementary method. It is recommended that at least 10 cases are panels with multiple probes and at least 10

cases involve metaphase FISH.

3. **Chromosomal Microarray:** At least 40 cases must be obtained using this technology. It is recommended that at least 20 chromosomal microarray cases have another technology. Note, targeted microarray testing falls under section 4h.
4. **Variant analysis:** At least five cases must be obtained for each of at least four of the different analysis methods (a-k) listed below:
  - a. PCR fragment size analysis
  - b. Restriction fragment length analysis
  - c. Quantitative PCR
  - d. ddPCR
  - e. Methylation testing (non-sequencing)
  - f. Triplet repeat-primed PCR
  - g. RNA analysis
  - h. Targeted microarray analysis for exon level deletion/duplication
  - i. MLPA
  - j. High resolution melt analysis
  - k. Other (must specify)
5. **Sequence Analysis:** At least 40 cases must be obtained in this laboratory method, with a minimum of 10 cases using Sanger sequencing and a minimum of 20 cases required using NGS. Of the NGS cases, it is *recommended* that at least 5 are genome sequencing analysis and 5 are exome sequencing analyses.
  - a. Sanger sequencing (single gene or panel)
  - b. Pyrosequencing
  - c. Methylation sequencing
  - d. Next Generation sequencing
    - i. Panel (PCR, capture based, or hybrid)
    - ii. Exome sequencing
    - iii. Genome sequencing

**Results:** A maximum of 100 cases may have normal laboratory findings; the results of identity testing cases must be counted as normal. Sequence and copy number changes interpreted as variants of uncertain significance should be counted as abnormal.

**Nomenclature:** For cytogenetic results, record the nomenclature for each case using the ISCN that was current at the time of analysis and filling in as much of the ISCN as space allows. If you require additional space, provide the full ISCN along with the associated logbook entry number on a separate sheet of paper (if audited, this will need to be submitted with your logbook). No more than 30 cases may have a normal karyotype. For this purpose, well-accepted structural polymorphisms are considered normal. Logbook cases should demonstrate experience with a variety of cytogenetic abnormalities, e.g.: aneuploidy; mosaicism; balanced, unbalanced, *de novo*, or inherited rearrangements. The

check box should only be marked if the result is abnormal.

Examples: chromosomes and array: 47,XY,+21.arr(21)x3

chromosomes and RNA fusion panel:  
46,XX,t(1;22)(p13;q13)[9]/46,XX[11], RBM15::MKL1 fusion  
detected

chromosomes and FISH: 46,XX,inv(16)(p13.1q22)[17]/46,XX[3].nuc  
ish(CBFBx2)(5'CBFB sep 3'CBFBx1) [96/200]

FISH panel:

AML FISH panel, 7 probe sets, negative  
MDS FISH panel, 3 probe sets, nuc  
ish(D7Z1x2,D7S486x1)[20/200]

For molecular results, 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. 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.

ISCN nomenclature should be used for microarray-detected CNVs that extend beyond a single gene. Intragenic deletions and duplications detected by targeted microarray, MLPA, or other methods should be reported using HGVS nomenclature. See examples below.

Examples: HTT, Huntington disease: Allele 1: 40 CAG repeats,  
Allele 2: 20 CAG repeats

BCR/ABL1 qPCR, p210 positive: BCR::ABL1 (p210)  
fusion transcripts were detected by RT-qPCR

F5, hereditary thrombophilia: c.1601G>A, p.Arg534Gln,  
heterozygous

DMD, Duchenne muscular dystrophy, hemizygous deletion of exons  
45-50: NM\_004006.3:c.6439-6614\_7201-7309del, Hemizygous

Array analysis, intellectual disability, arr[GRCh37]  
6q22q24(113900000\_149100000)x1

NGS panel, hearing loss, 70 genes, negative

NGS panel, developmental delay, 60 genes, UBE3A heterozygous  
NM\_13083.5:c.2475\_2478del, p.Lys826Metfs\*34, Heterozygous

For tests that include a panel of genes or probes, the test name and result can be listed in the logbook. All pathogenic sequence variants or abnormal findings detected must be listed in the logbook. If using more than one testing methodology, include nomenclature for both tests.

**Trainee's Role(s):** Check all of the boxes that indicate role(s) in testing, interpreting, and reporting. A breadth of experience must be reflected. A minimum of 100 cases must involve at least one of Roles 1-7, as defined below. A minimum of three roles must be specified for at least 180 cases. Observe specific limits per role when specified.

1. Cell culture
2. Culture harvest or slide preparation
3. Karyotype assembly, including digital image capture
4. Nucleic acid extraction and/or preparation for analysis
5. Wet lab procedures for variant detection assays (see section 4 and 5 of Laboratory Testing Methodology)
6. Result analysis (e.g., analysis of metaphase chromosomes by brightfield microscopy; analysis of FISH by fluorescence microscopy; analysis of microarray or sequencing data using appropriate software)
7. Interpretation of laboratory results, including description of findings using appropriate nomenclature (e.g., variant classification)
8. Written report
  - a. It is *required* that at least 100 cases involve this role.
9. Oral communication of results to health care providers:
  - a. At least 20 cases are required; at least half of these cases must involve abnormal results.
  - b. It is recommended that at least 30 cases involve this role.
10. Oral communication of results to patients:
  - a. At least 10 cases are *required*; at least half of these cases must involve abnormal results.
  - b. If institutional liability considerations prohibit trainee's communication with patients, then the trainee's presence during such communication will satisfy the

requirement.

**Supervisor:** Include the full name, degree(s), and type of certification of the supervisor responsible for activities involving each case.