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 laboratory tests and has received ongoing and appropriate laboratory supervision. The logbook cases must provide evidence of at least two years of well-rounded clinical laboratory experience with a wide variety of techniques drawn from all laboratory testing categories. Logbook entries should include the broad spectrum of genetic diagnoses.

Requirements:
Logbooks 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 anticipates ongoing review of cases between the trainee and their program director, the applicant should assure that all requirements have been fulfilled before submitting the final logbook for review. The program director will be required to attest to the ABMGG that all specific logbook requirements for this specialty have been fulfilled and are clearly reflected in the logbook. When applying for credentialing to sit for the certification examination in this specialty, the ABMGG reserves the right to request a copy of the logbook for audit and confirm that all requirements have been fulfilled. In this case, an applicant will be notified that they have been selected for audit and will be required to submit the 200 case logbook using the ABMGG Logbook Excel Spreadsheet to the ABMGG within five business days for detailed 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 laboratory genetics and genomics.

2. Supervision for case encounters must be provided by faculty who are ABMGG-certified, ABGC-certified, or CCMG-certified in the appropriate specialty. For cases obtained during rotations in approved outside laboratories, e.g., state newborn screening laboratories, it is recommended that laboratory supervisors be certified by their appropriate certifying board(s). All supervisors must be identified in the training program’s accreditation documents as members of the training faculty.

3. All 200 cases must be obtained during the inclusive dates of the applicant’s medical genetics training, as indicated in the ABMGG Trainee Information Form.

4. Logbook entries must reflect at least two years of clinical laboratory experience. No more than 30 cases may be obtained over any 30-day period.

5. Each logbook entry must document the applicant’s role(s) in the testing and reporting process.
process, including sample processing, analysis, result interpretation, and/or communication of test results.

6. Only cases evaluated for clinical diagnoses or confirmatory analyses may be included in the logbook. Use of experimental or control cases, historical cases, cases related to proficiency testing, or cases that are part of laboratory quality assurance activities will not be accepted. In laboratories for which state 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. Results for 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.

8. For applicants also seeking certification in Clinical Genetics and Genomics and/or Clinical Biochemical Genetics, a given patient may only appear in a single logbook, regardless of the number of specimens processed or methodology used.

**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 that may be requested for audit, you must select 200 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 is submitted to the ABMGG for audit. Logbooks containing specific information regarding the identity of any patient will not be reviewed.

- **Date:** Using [MM/DD/YYYY] format, indicate the date of sample receipt in the laboratory or, if more appropriate, the date the patient was evaluated in clinic.

- **Primary Laboratory Testing Category:** For each case, use the numbers 1 through 7 as outlined below to identify the single 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 products of conception). A minimum of 30 cases must be obtained in this category. It is *recommended* that results from at least three, but no more than 10 *Noninvasive Prenatal Screening* (NIPS) cases be reviewed (cases may be obtained from local ordering clinicians or genetic counselors). These reviews should include at least one positive case confirmed by karyotype, FISH, or chromosomal microarray and one case for which the fellow participates in counseling or observes the communication of results to the patient. NIPS results should be recorded in the results field but the use of specific nomenclature is not required.

Example: 47,XY,+21 NIPS positive for +21

**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 known 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 more than 20 cases may be obtained in this category. Pharmacogenetic testing involves the analysis of gene variants that give rise to variable drug metabolism or response.

Category 6  **Identity testing:** 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 mutations associated with disease (e.g., paternity testing, forensics, zygosity, transplantation or maternal cell contamination studies).

Category 7  **Hematology and oncology:** (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 microarray) and 30 cases must demonstrate use of molecular methodologies (Sanger sequencing, Next Generation Sequencing [NGS], etc).

- **Laboratory Testing Methodology:** Specify the laboratory testing methodology performed for each case by entering the Methodology number and associated letter (if any) outlined below. It is expected that trainees participate in a broad range of laboratory testing methodologies. Observe limits per category as specified.

1. **G-banding:** At least 50 cases must include G-banding but no more than 20 cases may involve G-banding alone. Any additional cases should involve a concomitant cytogenetic or molecular method.

2. **FISH:** At least 30 cases must include FISH, but no more than 10 cases may involve FISH alone. Any additional cases should involve a concomitant cytogenetic or molecular method.

3. **Whole Genome Chromosomal Microarray:** At least 40 cases must be obtained using this technology, but no more than 10 cases may involve microarray alone.

4. **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. Quantitative PCR
d. Methylation PCR
e. Triplet repeat primed PCR
f. Southern blot
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 NGS and 10 cases using Sanger sequencing.
   a. Sanger sequencing
   b. Pyrosequencing
c. Methylation sequencing
d. Next Generation sequencing
   i. Next Generation sequencing panel (PCR or capture based)
   ii. Next Generation whole exome sequencing
   iii. Next Generation whole 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 clinical significance should be counted as abnormal.

**Nomenclature:** Record the karyotype for each case, using the ISCN that was current at the time of analysis, 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 (this will need to be submitted with your logbook if audited). No more than 30 cases may have a "normal" karyotype. Logbook cases should demonstrate experience with a variety of cytogenetic abnormalities, e.g.: aneuploidy; mosaicism; balanced, unbalanced, *de novo*, and inherited rearrangements. The check box should only be marked if the result is abnormal. **NOTE:** Be sure use appropriate ISCN karyotype designations.

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.

Be sure to use the latest ISCN designations when describing microarray results. For CNVs that extend beyond a single gene (detected by microarray), ISCN nomenclature should be used. Intragenic deletions and duplications (detected by targeted microarray, MLPA, or other methods) should be reported using HGVS nomenclature. Please see examples below. Failure to follow appropriate nomenclature guidelines may require resubmission of the logbook.

**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[GRCh37(or38)] (1-22,X)x2 for normal females
   arr[GRCh37(or38)] (1-22)x2,(XY)x1 for normal males
   arr 6q22q24(113,900,000-149,100,000)x1 for a loss at 6q22 to 6q24
47,XX,+21, NIPS positive for +21
NGS panel, hearing loss, 70 genes, negative
NGS panel, developmental delay 60 genes, UBE3A heterozygous
c.2475_2478delACTT

For tests that include a panel of genes (more than five), the test name and result can be listed in the logbook. All deleterious mutations detected must be listed in the logbook.

**Trainee’s Role(s):** 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 100 cases must involve 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 and slide preparation or microarray
3. Karyotype preparation, including digital image capture
4. Nucleic acid extraction and/or preparation for analysis
5. Mutation analysis (see section 4 and 5 of Laboratory Testing Methodology)
6. Result analysis (karyotype or FISH analysis using a microscope, microarray or sequencing analysis using the appropriate software)
7. Interpretation of laboratory results
8. Written report; it is **required** that at least 100 cases involve this role.
9. Oral communication of results to health care providers who requested the testing or their designated contact: at least 10 cases are **required**; at least half of these cases must involve abnormal results. It is **recommended** that at least 30 cases involve this role.
10. Oral communication of results to patients: at least 20 cases are **required**; at least half of these cases must involve abnormal results. 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 who was directly responsible for your activities for each case. Remember that all supervisors must be identified in the training program’s accreditation documents as members of the training faculty.