ABMGG
American Board of Medical Genetics and Genomics

Cytogenetics and Genomics Logbook Guidelines for Diplomates in the Laboratory Genetics and Genomics Alternative Certification Pathway

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 cytogenetics 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 cytogenetic and genomic techniques involving all cytogenetics laboratory testing categories. Cases included in the logbook should demonstrate a broad spectrum of cytogenetic diagnoses. Abnormal karyotypes should reflect a range of clinical indications including, but not limited to, prenatal diagnosis, congenital malformations, developmental or cognitive delay, infertility, pregnancy loss, 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-accredited training program in clinical cytogenetics and genomics OR in a laboratory that is supervised by an ABMGG diplomate certified in clinical cytogenetics and genomics.

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, 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 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 that may be requested for audit, 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 is 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 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 4 as outlined below to identify the single category that best describes the indication for the clinical cytogenetics test. No more than 50 cases may be obtained in any primary laboratory testing category. Observe category limits as specified below.

  Category 1 **Prenatal studies,** e.g., amniotic fluid, chorionic villi, percutaneous umbilical blood; a minimum of 5 cases must be obtained in this category.

  Category 2 **Pediatric and other postnatal studies,** e.g., peripheral or cord blood; a minimum of 25 cases must be obtained in this category.

  Category 3 **Hematology and oncology,** e.g., bone marrow, leukemic blood, tumor tissue; a minimum of 25 cases must be obtained in this category.

  Category 4 **Miscarriage and other monolayer-cultured studies,** e.g., tissue; a minimum of 10 cases must be obtained in this category.

- **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. With the exception of G-banding, no single laboratory testing methodology should be over-represented (i.e. 75 cases of microarray will not be acceptable). Observe limits per category as specified.

  1. **G-banding:**
     a. **G-banding alone**
     b. **G-banding and FISH:** At least 25 cases must be obtained in which FISH is used in conjunction with G-banding
     c. **G-banding and microarray**

  2. **FISH**: No more than 10 cases may be obtained for which only FISH is performed. At least 25 cases must be obtained for which FISH is performed in conjunction with G-banding.

3. **Microarray:** No more than 30 cases, with or without FISH, may be obtained using this technology.

4. **Other:**
   a. **Chromosome breakage** (e.g. Fanconi anemia; ataxia telangiectasia)
   b. **Other staining/banding** (e.g. C-banding, NOR-staining, R-banding, Q-banding). It should be noted that this does **not** include cases for which microarray and FISH are performed.
   c. **Noninvasive Prenatal Screen (NIPS)**: It is *recommended* that results from at least three, but no more than 10, NIPS cases be reviewed with a clinical provider (cases may include those for which testing is ordered by local clinicians or genetic counselors). These case reviews should include at least one positive case confirmed by karyotype, FISH, or chromosomal microarray analysis, and one case in which the diplomate participates or observes the communication of results to, and counseling of, a patient. These cases should be included in Primary Laboratory Testing Category 1.

- **Results:**

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 100 cases may have a "normal" karyotype. The 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 to use current ISCN karyotype designations.**

- **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 100 cases must involve Roles 1, 2, 3, or 4, as defined below. For roles 1 through 6, it is *recommended* that the applicant include at least 25 cases per role. A minimum of three roles must be specified for at least 140 cases. Observe specific limits per role when specified.

1. Cell culture:
   a. Amniotic fluid, chorionic villi
   b. Blood
   c. Bone marrow/unstimulated peripheral blood/solid tumor
   d. Skin, products of conception, other tissue

2. Culture harvest with slide preparation or processing of specimens for microarray study

3. Karyotype analysis at the microscope or analysis of microarray results using appropriate software

4. Karyotype preparation, including digital image capture

5. Interpretation of findings

6. Written report

7. 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 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 each case.