

Certification in Clinical Biochemical Genetics Logbook Requirements for 2025 Examination

Purpose:

The purpose of the logbook is to document that the applicant has had direct and meaningful involvement in the processing of biochemical 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 biochemical genetic techniques and interpretation of results involving all major biochemical genetic laboratory testing categories. Conditions included in the logbook should represent a broad spectrum of biochemical 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 ABMGG expects ongoing review of cases by the program director (PD), the applicant should be sure 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. When reviewing applications to sit for the certification examination in this specialty, 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 150-case logbook using the ABMGG Logbook Excel Spreadsheet to the ABMGG within five business days.

Case Selection:

- 1. All specimens must have been processed in an ACGME-accredited training program in the laboratory specialty of clinical biochemical genetics.
- Supervision for cases must be provided by faculty certified by the ABMGG, ABGC or CCMG. 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). <u>All</u> supervisors must be identified in the training program's accreditation documents as members of the training faculty.
- 3. All 150 cases must be obtained during the <u>inclusive</u> dates of the applicant's clinical biochemical genetics training.

- 4. Logbook entries must reflect at least one (1) year of clinical <u>laboratory</u> bench experience. <u>No</u> <u>more than half</u> of the cases may be obtained in any four-month period and no more than 25 cases may be obtained in any 30-day period.
- 5. Each logbook entry must document the applicant's role(s) in the testing and reporting process, including <u>sample processing</u>, data analysis, result interpretation, <u>and/or communication of results</u>.
- 6. Only cases for <u>clinical analysis</u> 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 where 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 <u>once</u> 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. For the final logbook, 150 cases should be selected that fulfill all the 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 <u>not</u> be included. Logbooks containing specific information regarding the identity of any patient will <u>not</u> be reviewed.
- <u>Date</u>: The date in 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.
- <u>Primary Laboratory Testing Category</u>: For each case, use the numbers 1 through 5 as outlined below to identify the category that best describes the indication for the clinical biochemical genetic test. Observe category limits as specified below.
 - Category 1 **Diagnostic testing (postnatal):** Initiated because of clinical symptoms and/or family history. It is *recommended* that this includes at least 5 cases of integrated molecular and biochemical testing for diagnosis or carrier status detection. Up to five cases may represent the interpretation and/or communication of results from molecular-based (DNA) carrier testing of a metabolic disorder.
 - Category 2 **Prenatal diagnosis:** Primary testing of prenatal samples, interpretation, and/or communication of prenatal diagnosis results generated by a referral laboratory,. It is *recommended* that at least 5 cases be obtained in this category. Up to five cases may represent the interpretation and/or communication of results from samples referred to another laboratory for biochemical and/or molecular-based (DNA) testing of a metabolic disorder.

- Category 3 **Newborn screening:** Primary testing in a newborn screening laboratory, or interpretation and communication of newborn screening results from a state laboratory to health care providers and families. This category also includes follow-up testing of abnormal newborn screen. No more than 35 cases may be obtained from a state screening laboratory. It is *recommended* that at least 10 cases be obtained in this category.
- Category 4 Management/continuing care: For follow-up testing of previously diagnosed individuals with a biochemical condition.
- Laboratory Test/Methodology: Specify the laboratory test/methodology performed for each case by entering the Methodology number outlined below. Observe limits per method where specified.
 - 1. Amino acid analysis, quantitative: e.g., amino acid analyzer, liquid chromatographytandem mass spectrometry, or related method.
 - 2. Organic acid analysis: e.g., gas chromatography/mass spectrometry.
 - 3. Acylcarnitines/Acylglycines: e.g., tandem mass spectrometry or related method.
 - 4. Screening tests (not including newborn screening): e.g., urine screening for mucopolysaccharides or oligosaccharides; metabolomics, and other testing. It is recommended at least 5 cases be obtained in this category.
 - 5. Enzyme assays: it is *recommended* that a minimum of 10 cases be obtained in this category.
 - 6. **Other analyte analysis**: e.g., galactose-l-phosphate, carnitine, succinylacetone, very long-chain fatty acids.
- **Diagnosis:** For each case, enter the diagnosis or condition being evaluated, using the guidelines below. Logbook cases must demonstrate experience with a variety of biochemical methodologies and conditions. It is *recommended* that no more than 20 cases be for any one diagnosis and strictly *required* that <u>no more than</u> 35 cases be any one condition, such as phenylketonuria or galactosemia. A maximum of 100 cases may have normal laboratory findings. Note: "normal" implies that no major, clinically significant abnormalities were found. Cases being tested for management of a known diagnosis that have normal results due to treatment or clinical status (e.g., a known VLCAD deficiency patient when asymptomatic, a patient with intermittent MSUD) may be counted as "abnormal."

For primary metabolic conditions: Enter the diagnosis using the OMIM name or an OMIM alternative title. All cases representing the <u>same</u> condition should be entered using the <u>same</u> diagnosis name. For example: Enter all PKU cases as "PKU," not "PKU" for some and "phenylketonuria" for others).

For results that are abnormal but not diagnostic for a specific metabolic disorder. Enter the most likely condition underlying the abnormal result, using consistent terminology from case to case. Indicate that the results were abnormal.

Examples: Dietary artifact

Liver immaturity Physiologic-Ketosis Peroxisomal disorder, unspecified Mitochondrial myopathy Methylmalonic acidemia/homocystinemia SSADH deficiency vs. drug artifact Hyperglyceroluria (primary vs. secondary)

For results that are normal following testing for a specific metabolic disorder, initiated because of clinical symptoms, positive family history or abnormal screening result: Enter the disorder being evaluated using the OMIM name or an OMIM alternative title. For newborn screening cases with more than one disorder possible, enter the newborn screening result using standard terminology.

Examples: Elevated C3 Elevated C5 Elevated C5-OH

For results that are normal following metabolic testing for an unspecified condition: Enter "No abnormality detected" in the diagnosis field.

- Trainee's Roles: Check all boxes indicating your role(s) in the testing. interpretation, and reporting process. A breadth of experience must be reflected in the logbook. It is *recommended* that a <u>minimum of three roles</u> be specified for at least 140 cases. Observe specific limits per role where specified.
 - 1. **Sample preparation:** Preparation of the sample for analysis, including dilution, analyte extraction, and other preparative steps. At least 75 cases must involve this role.
 - 2. **Sample analysis:** Programming and running of instruments, chromatographic baseline review, peak identification, performance of manual assays. At least 75 cases must involve this role.
 - 3. **Interpretation of results:** Evaluation of the clinical significance of findings including, where appropriate, gathering additional clinical and/or laboratory data, generating a <u>differential diagnosis</u>, recommending further testing, and generating a draft or final version of the written report. <u>At least 100 cases must involve this role.</u>
 - 4. **Oral communication of results to health care providers:** at least 20 cases are *required;* at least <u>half</u> of these must involve abnormal results. It is *recommended* that at least 30 cases involve this role.
 - 5. **Oral communication of results to patients:** at least 10 cases are *required;* at least <u>half</u> 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 <u>full name</u>, degree(s), and type of certification of the supervisor responsible for activities involving each case.